THE PERCEIVED INFORMATION NEEDS OF GIRLS WITH TURNER SYNDROME AND THEIR PARENTS

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ABSTRACT

The University of Manchester

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Doctor of Philosophy

The perceived information needs of girls with Turner syndrome and their parents

10th December 2012

The age range at diagnosis, complexity of the condition, and sensitive nature of the issues involved in a diagnosis of Turner syndrome (TS), present specific challenges for health professionals in sharing information. Little is known about the perceived information needs of girls with TS and their parents.

A flexible qualitative design, guided by the principles of symbolic interactionism was employed in this exploratory study. This design enabled meanings girls and their parents attached to TS, how they interpreted, shared and valued information to be uncovered. A purposive sample of 15 families with daughters aged 9 to 16 years were recruited from a tertiary paediatric endocrinology clinic. Girls and parents participated in a total of 27 recorded semi-structured interviews. Data were analysed using the framework approach and the constant comparative method. Analysis revealed how girls and their parents interpreted and used information within the context of their everyday experiences of living with TS.

Three activities were described by families: gathering and receiving, making sense of, and using and sharing information. Throughout these activities, themes of uncertainty, normalising and identity were present. A series of tensions described by the girls and their parents illustrated diverse approaches to the management of information. Meanings assigned to TS by girls and their parents influenced when, what and how information was shared with others.

Despite a wealth of information, the girls and their parents described unfulfilled information needs. The interviews were dominated by discussion of the social implications of the condition and more specifically to social functioning, puberty and infertility. Parents were the primary source of information. These findings provide a basis for developing evidence based approaches to information sharing.
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Dedication

This thesis is dedicated to Simon and Alexander for their encouragement, love and support

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CHAPTER ONE: SETTING THE SCENE

1.0 Introduction

Effective communication and exchange of information between practitioners, children and young people (CYP) and their parents is essential if they are to adapt to the implications of living with a chronic condition and manage their lives as independently as possible (Bradford 1997). This qualitative study is the beginning of an inquiry that aims to understand the experiences of girls with Turner syndrome (TS) and their parents to inform practitioners of their specific information needs.

1.1 Inception of a research project

The story of my thesis began with a discussion of a clinical incident with a group of paediatric endocrinology nurse specialists (PENS). At the heart of this critical reflection was a debate about assessing effectiveness of teaching complex and sensitive information to parents. The debate was fuelled with many anecdotes from practice, including TS. There was a general presumption that all parents would have the motivation, knowledge and skills to pass on such information, educating their children as they progressed through each developmental stage. As the nurse specialists continued to discuss TS, I became aware of the many subtleties and variations in the features and characteristics of this condition. The age range at diagnosis, complexity of the syndrome, potentially sensitive and emotional nature of issues involved appeared to present specific challenges for information sharing to healthcare professionals working with these girls and their families. It was not clear from the discussion how information was shared, although the anecdotal evidence suggested a rather ad hoc and informal process, frequently reliant on the quality of the relationship between the professionals, the girls with TS and their parents.

This became the general research area, specific research questions formulating a clear focus were yet to develop. However, research is often rooted in “something that starts out as poorly understood”, and “given considerable theoretical effort [can] convert it into
something which is clearly defined, logical and rational, and could well be of value” (Robson 2002; p55)

An introduction to TS at this early point of the thesis will assist in orientating the reader to the condition that shaped the research questions and framed this exploratory inquiry.

1.2 History of TS

In 1805 in England, Dr Charles Pears recorded characteristics he observed in a 29 year old woman and suggested that there may be some link between them. He noted that her behaviour was unusual; she was short and had no secondary sexual characteristics (Rovet 2004). In 1930 Otto Ullrich, a paediatrician in Munich, Germany, described cases with similar characteristics. However, the condition is named after Henry Turner, an endocrinologist in the USA who described seven patients between the ages of 15 and 23 years in 1938. These cases had similar features to those described by Ullrich but additional features were also identified. The first report identifying the absence of an X chromosome with the features of TS (45X karyotype) was published by Dr Charles Ford (Rovet 2004). In recent years, genetic research has become more sensitive resulting in greater detail and understanding of the condition to emerge. In 1985 the development of biosynthetic growth hormone (GH) transformed treatment for girls with TS enabling their final height to be at the lower end of the normal range for women.

1.3 An introduction to TS

TS is defined as a relatively common chromosomal disorder affecting 1 in 2000 live female births (Turner Syndrome Support Society (TSSS) 2008) and has cardinal features of short stature and ovarian dysgenesis leading to the absence of pubertal development and probable infertility (Donaldson, Gault, Tan and Dunger 2006, Raine, Donaldson, Gregory and Savage 2006). In addition to these key features there are differing presentations and individual problems which reflect complexity in the genetic origin of TS. The resulting broad range of issues impacts on physical, behavioural and psychosocial well-being.
1.3.1 Genetic aspects of TS

Diagnosis of TS is confirmed by chromosome analysis, which identifies the absence or structural change in some X chromosome material, leading to a wide-range of karyotype (Rovet 2004, Donaldson et al. 2006). The loss of the X chromosome occurs “early in embryonic mitotic division” (Bondy 2005) and in the majority of girls the normal X chromosome is maternal in origin (Donaldson et al. 2006). In approximately 50% of girls a diagnosis of ‘classic TS’ is confirmed by a 45X karyotype, which is the most common form of TS.

The second most predominant karyotype accounting for around 30% of the TS population is 45X/46XX, which is described as ‘mosaic’ (Rovet 2004). This means there is an abnormality of the second X chromosome or absence of some X chromosomes from some cells (Bondy 2005). Within the mosaic form of TS a small number of girls have a complete or partial Y chromosome (45X/46XY). Bondy (2005) cautions that information relating to a Y chromosome in a female child can cause families to question the gender identity of their daughter, potentially causing the family great distress particularly as they are already coming to terms with her probable infertility. These girls also need to undergo further investigation for any Y chromosomal material which would require surgical removal because of the risk of gonadoblastoma (Donaldson et al. 2006). Structural anomalies are commonly mosaic with 45X or 46XX cells (Ranke & Saenger 2001). These may include karyotypes with an isochromosome of X. An isochromosome occurs when the sex chromosome splits horizontally rather than transversely (Rovet 2004). This error results in structural abnormalities producing two short arms (Xp) or two long arms (Xq) on the chromosome. The short arm is associated with many of the phenotypic characteristics. Other structural anomalies of TS can include “ring X, deletions, rearrangements or translocations” (Rovet 2004, Hjerrild, Mortenson & Gravholt 2008). Discoveries in genetic research continue to develop understanding of TS as the functions of individual genes are clarified.

As a consequence of the varying anomalies of the X chromosome, affected girls present with a range of features and characteristics which make up the phenotype. However, the link
between genotype and phenotype is generally poor and cannot predict with accuracy the degree to which individual girls will be affected (Donaldson et al. 2006). Nevertheless, in general it is accepted that girls with 45X/46XX karyotype are more mildly affected than girls with monosomy X (classic TS) where key clinical features may be obvious at birth.

Complexity of the genetic make-up of girls with TS means that referral to a geneticist enables the girls and their parents to understand the implications for treatment, and care management over their life-course. No cause for the missing X chromosome has been identified and no risk factors have been reported.

**Figure 1: Main physical features of girls with TS**
The condition affects the skeletal and lymphatic and reproductive system. Girls are diagnosed at different ages because of the range of features and differences in presentation.

If the diagnosis is made in-utero it is usually incidental, following investigation for unrelated reasons, e.g. amniocentesis because of maternal age (maternal age is not a risk factor for TS) (TSSS 2008). Less distinctive features may lead to diagnosis being delayed until children present with poor growth and short stature in childhood or later when there is failure to progress into puberty. These aspects of the condition are detailed in Chapter Two.

1.3.2 Natural history of TS

Having discussed the cardinal features of TS and identified related problems, it is also necessary to have an understanding of the appearance and progression of the main difficulties girls and their parents face at different life stages (Table 1).
Table 1: The natural history of TS and associated problems

<table>
<thead>
<tr>
<th>Stage</th>
<th>Associated Problems</th>
</tr>
</thead>
<tbody>
<tr>
<td>Infancy</td>
<td>- Lymphoedema&lt;br&gt;- Cardiac abnormalities e.g. coarctation of aorta, ventricular hyperplasia&lt;br&gt;- Short and low birth weight&lt;br&gt;- Feeding difficulties, faltering weight&lt;br&gt;- Poor sleeping pattern</td>
</tr>
<tr>
<td>Pre-school</td>
<td>- Short stature&lt;br&gt;- High activity levels&lt;br&gt;- Behavioural difficulties with exaggerated fearfulness&lt;br&gt;- Recurrent middle ear infection (glue ear), hearing loss.&lt;br&gt;- Sleeping problems</td>
</tr>
<tr>
<td>School age</td>
<td>- Height falling away from 3rd centile&lt;br&gt;- Middle ear disease&lt;br&gt;- Obesity&lt;br&gt;- Specific learning difficulties, e.g. mathematics, visuospatial tasks&lt;br&gt;- Social vulnerability&lt;br&gt;- Foot problems&lt;br&gt;- Renal anomalies, e.g. horse shoe shaped kidney, duplex kidney</td>
</tr>
<tr>
<td>Adolescence</td>
<td>- Impaired pubertal growth spurt even with oestrogen induction&lt;br&gt;- Ovarian failure with absent or incomplete puberty&lt;br&gt;- Obesity&lt;br&gt;- Hypertension&lt;br&gt;- Increased prevalence of immune disorders such as autoimmune thyroiditis, coeliac disease, inflammatory bowel disease&lt;br&gt;- Specific learning difficulties&lt;br&gt;- Social vulnerability&lt;br&gt;- Foot problems/lymphoedema of lower limbs&lt;br&gt;- Tendency to put on weight</td>
</tr>
</tbody>
</table>

(Sources: Donaldson et al. 2006; TSSS 2008)
In summary, TS has many features with lifelong implications. Currently it is not clear how information relating to the condition is shared with the girls or how relevant they perceive it to be to their everyday lives. The broad range of problems and associated difficulties mean that not all girls with TS face the same issues, therefore prioritising information to meet the needs of parents and the unique needs of their daughters in a developmentally appropriate manner is potentially problematic for all involved in the process of sharing information. This research explores the perceived information needs of girls with TS and their parents through their experiences and understanding of living with TS or parenting a daughter with TS.

1.4 Structure of the thesis

This thesis presents the research study in eight chapters. Each chapter has a specific function leading the reader from the inception of the project to the final conclusions and recommendations. The first two chapters aim to introduce the research. Chapter One provides a brief overview of the subject area and acquaints the reader with the context in which the study was conceived. Chapter Two reviews and appraises current literature and places this study within context of previous research and clinical interest. Focusing on information needs of girls with TS and their parents this chapter establishes the significance of this research within the context of existing published work, identifying similarities with other research while establishing its unique contribution to the area of study. Chapter Three addresses the philosophical and ethical basis for the decisions made in designing and conducting the study and also presents the methods and research approaches undertaken including the process of analysis.

Chapters Four to Seven are concerned with interpretation of the research. Findings are presented in three separate chapters that reflect information activities described in these data by the girls and their parents. Chapter Four focuses on data relating to gathering and receiving information; Chapter Five presents data which refer to how girls and their parents make sense of information and finally the findings discussed in Chapter Six illustrate how parents and girls use and share information. Chapter Seven considers strengths and
limitations of this study and then examines key themes that emerged in the analysis. These themes are examined and appraised within the context of published literature and evidence. Finally the thesis concludes by revisiting the major findings, drawing conclusions for clinical practice and offering suggestions for further research. Figure 2 provides an overview of the thesis presentation.

Figure 2: Overview of the structure of the thesis

- Chapter One
  Introduction

- Chapter Two
  Literature Review

- Chapter Three
  Methodology

- Chapter Four
  Gathering and Receiving Information

- Chapter Five
  Making Sense of Information

- Chapter Six
  Using and Sharing Information

- Chapter Seven
  Discussion

- Chapter Eight
  Conclusions and Recommendations
CHAPTER TWO: LITERATURE REVIEW

2.0 Introduction

This chapter presents areas of interest pertinent to this study, providing a comprehensive and coherent rationale for undertaking an exploration of the information needs of girls with TS and their parents.

2.1 Literature search

Reviewing and critically appraising the literature will provide justification and context for the proposed research. Investigation of the scope of existing literature relating to the research topic will enable the purpose of the study to be justified (Carnwell and Daly 2001).

2.1.1 Procedures used for searching and selecting literature

Major themes and keywords were identified from the title, aims and questions within the proposed research. These terms were explored further for alternative keywords and for terms related to each concept using a thesaurus search for each theme across different databases. Keyword searching was also useful to complement and broaden the thesaurus search by introducing alternative words. This approach was helpful because there was minimal literature relating to information needs of girls with TS and their parents from their perspective. An initial keyword search of major health databases for published literature since 2000 led to only two relevant articles being retrieved. The time frame was therefore extended to 1990 resulting in two additional papers. A selected review of the literature using a number of related terms was undertaken to find the literature informing this study.

The inclusion criteria were:

- Time frame 1990-2010
- English language
• Related to focus of the research – children, adolescents, parents, information needs and TS
• Peer reviewed

Additional search strategies were utilised including truncation of words such as parent, child, adolescent; and citation searching, hand-searching and snowballing. These less systematic approaches were helpful in recognising themes relating for example, the use of information in coping and adaptation to chronic illness. Relevant and frequently cited papers in more recent studies that were outside the date band were also included. This approach also enabled papers to be identified where information need was not a primary focus and therefore relevance of a paper was not identifiable through electronic searches. Grey literature included published information available from national support groups provided to parents and girls. Related papers recommended by supervisors were also included. Despite these different search strategies, a limited literature reflecting the specific research questions relating to parent and child perceptions of TS was retrieved.

2.1.2 Search engines

Databases were accessed through EBSCO and ISI Web of Knowledge. The general healthcare databases and subject specialist databases included AMED, CINAHL Plus, MEDLINE, British Nursing Index and PsycINFO. Specialist review sources such as Cochrane Library were also accessed. The primary aim of the search is to learn what is already known about the topic, ensuring the study does not duplicate previous research and instead contributes to a cumulative body of knowledge (Silverman 2005). This literature review positions the study and highlights its original contribution. It also contributed to the design of the research, making it as sensitive as possible to girls with TS and their parents.

2.1.3 Identification of selected themes for the review

From the above strategy two main areas of relevance to this study became evident. Firstly, papers about the information needs of families of girls with TS. The literature in this area is limited, therefore information needs of children with other long-term conditions and their
parents were included. The second area of relevance included broad interrelated themes associated with information relating to the medical management, and psychosocial implications of living with or parenting a daughter with TS. TS is a complicated condition with multi-system implications. An initial search of the literature demonstrated wide-ranging and diverse interests of academics and clinicians within medicine, psychology and social science. Four major areas of interest were identified from the literature: medical research; psychosocial aspects; medical management; educational issues for girls, parents and professionals (Table 2).

Table 2: Initial review: scoping the literature

<table>
<thead>
<tr>
<th>Medical research</th>
<th>Research relating to psychosocial aspects</th>
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<tr>
<td>• Aetiology</td>
<td>• Personality traits</td>
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<td>• Brain morphology</td>
<td>• Social anxiety</td>
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<td>• Genetics, chromosomes, phenotypes</td>
<td>• Needs of adolescents</td>
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<td>• Growth</td>
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<td>• Hormones</td>
<td>• Behavioural issues</td>
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<td>• Cerebral development</td>
<td>• Psychological disorders</td>
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<td>• Motor development</td>
<td>• Self-esteem and body image</td>
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<td>• Neurological development</td>
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<td>• Immune defects</td>
<td>• Social functioning</td>
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<td>• Cancer incidence</td>
<td>• Spatial awareness</td>
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<tr>
<td>• Uterine development</td>
<td>• Language and verbal skills</td>
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<tr>
<td>• Fat mass</td>
<td>• Experience of diagnosis</td>
</tr>
<tr>
<td>• Ocular motor indicators</td>
<td>• Truth telling and disclosure</td>
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</tbody>
</table>
### Medical management
- Cardiac monitoring
- Auditory function
- Skeletal problems: kyphosis, slipped epiphyses
- Motor performance
- Epidemiology
- Precocious puberty
- Puberty
- Infertility
- Referral and diagnosis
- Ascertainment bias
- Health supervision across lifespan
- Health problems in women with TS

### Educational issues for girls, parents and professionals
- Issues with maths
- Behavioural issues
- Oral characteristics
- Parents’ knowledge
- Increasing awareness of health professionals about TS

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<td>Epidemiology</td>
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**2.2 Information needs of girls with TS and their parents**

Four research papers (Starke and Moller 2002; Starke, Albertsson-Wikland and Moller 2002; Williams 1995; Mullins, Lynch, Orten and Youll 1991) directly related to the information needs of parents with daughters diagnosed with TS were retrieved. No studies from the perspectives of girls with TS were found. These studies are now discussed.

**2.2.1 Information received at diagnosis and family communication**

Over 20 years ago, Mullins at al.(1991) identified information provision for families of girls with TS was variable. Some families received verbal and written information as well as counselling and lay support from other families but some received minimal information or support (Mullins et al. 1991). Twenty-one families from a total of 50 were invited to
complete a needs assessment survey using a likert-style questionnaire. The families all had daughters who attended a paediatric endocrinology clinic for TS and were between 1 and 16 years. The mean age at diagnosis was 3 years. The disappointing response rate was thought to reflect insufficient networking by the researchers before the study commenced, and the wide geographical spread of potential participants. The questionnaire was completed by different family members; some girls independently, mothers and fathers together and mothers alone, therefore this relatively small sample size for a survey also had high potential for self-selection.

The findings reported responses to communication of the diagnosis, emotional responses to diagnosis, and family communication about TS. Emotional responses such as fear and shock were reported, but the majority of families described relief. Parents judged the content of information given to their daughters against their age and development, but there were discrepancies between some parents and their daughters about the timing of specific information. Parents thought they should be given information relating to short stature earlier than when the girls wanted to discuss it, and the girls wanted to discuss peer related and sexually related issues earlier than their parents. General agreement across all respondents was that between 10 and 15 years of age all aspects of the condition should have been discussed with the girls. This is a wide age-range developmentally and the study did not indicate what specific content, or when and how information was to be shared.

Mullins et al. (1991) concluded that families of girls with TS want education relating to the condition and its implications, including social support. At the time in the USA a national TS organisation was in its infancy and 18 out of 21 respondents indicated an interest in joining, supporting the finding that there was a need for psychosocial support. Although this study collected data from girls and parents, the results are not clear as to the ownership of responses. The description of the sample is vague and there is no breakdown of the age of the girls. It would have been useful to know the particular type of information in which girls at different ages were interested. Chronological age alone is not an indicator of ability in TS and further details relating to the cognitive and behavioural aspects of the sample would
provide insight into the identified needs of the families. Specific recommendations included a programme of interventions at specific time periods using task objectives related to developmental events, such as school transition or pubertal development. The low response rate may suggest other families with girls with TS have different needs.

2.2.2 Knowledge and experiences of receiving the diagnosis

A national multidisciplinary project in Sweden commenced in 1994 and completed data collection between 1995 and 1997. The overall aim of the project was to improve psychosocial care of girls and women with TS. Researchers had professional backgrounds in social work and psychology who worked within the Paediatric Growth Research Centre, Goteborg University, Sweden. The study group comprised 33 mothers and 11 fathers from different families, whose daughters had a confirmed diagnosis of TS and had commenced GH treatment. From this national project two smaller studies were undertaken by Starke and Moller (2002) and Starke, Albertsson-Wikland and Moller (2002). The first of these two studies explored parents’ need for knowledge concerning the medical condition of their child. It included their motivations and strategies for finding information and its implication for their child (Starke and Moller 2002). The second paper investigated parents’ experiences of receiving a diagnosis of TS (Starke et al. 2002). Both studies were exploratory and retrospective. The daughters in these families had been diagnosed at different ages, ranging from prenatally to 14 years therefore there was “a wide degree of retrospection”.

The first study (Starke and Moller 2002) reports findings from 44 semi-structured interviews, carried out with one parent from each family recruited to the study group described in the previous paragraph. Interviews took place when they attended clinic for an annual examination. The researchers focus was on the motives and strategies of parents in seeking information about their child’s medical condition. The aim of the interview was to explore parental perception of having a daughter with TS focusing on the diagnostic process, parents’ perceptions and experiences, the difficulties and strategies used in managing aspects of their daughter’s development, and interaction with family and professionals in different contexts.
The findings reported parents’ experiences of receiving information during the diagnostic process in addition to information they had sought independently. Twenty-one parents were satisfied with the information and 23 were dissatisfied, emphasising the difficulties for medical staff when providing information to parents who have individual needs. Parents’ motives for seeking information varied depending upon whether they had been satisfied or unsatisfied (n=23) with information provided when the diagnosis was confirmed. Fewer satisfied parents (6 mothers and 4 fathers) sought additional information compared with those who were dissatisfied (15 mothers and 1 father). Dissatisfied mothers used more strategies to seek information than fathers and mothers who were satisfied. The motive of all parents (satisfied and dissatisfied) was to learn more about the condition in order to explain TS to others. A second motive was identified in dissatisfied parents who lacked confidence in the knowledge of the health professionals and therefore wanted some control in what they were told. For example using information they had found to make sure the doctor included all aspects of TS including recent research and recommendations for clinical management.

Differences between mothers and fathers’ information seeking behaviour were identified. Starke and Moller (2002) explain mothers sought information actively and focused on their everyday lives whereas fathers sought information to learn more about the condition in general terms.

The second paper explored parents’ perceptions and experiences of the process leading to the diagnosis of TS (Starke et al. 2002). Semi-structured interviews were undertaken guided by a questionnaire that had been devised from earlier exploratory interviews in a pilot study. The questions were related to the investigation and diagnostic period, asking participants to reflect on information and support received. Different types of questions were used: open-ended, open-ended with answer categories and some requiring yes/no answers. This necessitated different types of analysis for example, open-ended questions were analysed to find patterns and themes whereas statistics were used when comparing answers across
parents, e.g. differences in distribution of phenotypes, age at diagnosis and genotype (Starke et al. 2002).

Comparison was made between parents who had initiated the investigation of their daughter, with parents of girls who had been referred for investigation by health professionals. They report that experience of diagnosis was affected by who initiated the referral. Parents who referred themselves reported feeling more prepared as it confirmed their suspicions that something was wrong, however, families referred by health professionals felt less prepared and expressed fear and sadness. Parents whose daughter was referred by health professionals described information at the point of diagnosis as incorrect or not understood. Starke et al. (2002) suggest this may reflect the level of shock experienced by parents, which would diminish their ability to process and understand information. More than half the group of parents were told their daughter would possibly be infertile and all of these parents identified this as the most difficult piece of information to deal with. They report that mothers and fathers interviewed believed it is harder for the mother than the father to receive this news although no explanation for this assumption is offered.

The amount of information provided about associated physical, cognitive and behavioural problems of TS varied, with some parents stating they were not given any information.

Parental satisfaction, and the term Turner syndrome were also identified themes from the research. The doctor’s knowledge was a key component as was their ability to make the information relevant and understandable. The ability of the doctor to understand parents’ feelings of “grief and worries for the future” was valued. Where these skills were evident, parents had reported the consultations as constructive. However, this had not been the case for approximately half of participants. The diagnostic term TS was viewed by some parents as negative. The concept of “syndrome” was associated with Down syndrome, which they felt would lead to people thinking their daughter was “mentally retarded”.

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Parents made suggestions for healthcare providers about how and what information should be given to parents at the time of diagnosis. Issues such as understanding investigations, timing of information, the positive and negative implications of TS, and access to ongoing support from psychologists and support groups were included.

The use of a qualitative exploratory approach was appropriate for these studies, the purpose of which was to find out more about a subject where there is little known (Starke and Moller 2002). Robson (2002) claims exploratory research is relevant when the aim of the research is to “seek new insights and potentially generate ideas and hypotheses for future work” (p59). Retrospective studies are potentially at risk of memory bias as participants recall past events. This limitation is highlighted by the authors in both studies, who caution that memory bias makes it difficult to determine what is real and what may be “created” memory. However, they add that although this prevents any conclusions being drawn about the circumstances of the past, data are still important because it is the memory and understanding of the past on which parents “handle their present situation including relationships with health professionals” (Starke et al. p353). These papers contribute to an increased understanding of how the delivery, content, presentation and approach to information sharing impacts on the lives of girls with TS and their parents.

2.2.3 Information to help with parenting

A qualitative study carried out in the USA aimed to “identify specific childrearing problems and management behaviours used by mothers of daughters with precocious puberty and TS” (Williams1995, p.110). Six mothers of girls with TS (mean age 10 years) were recruited from the endocrinology clinic and interviewed either at clinic or at home according to their preference. Mothers were selected rather than other family members because of their role as main caregiver in day-to-day aspects of parenting. The stimulus for the study had come from mothers requesting help from nurses in solving parenting problems relating to school, peer and family life. Recorded interviews lasting approximately 45 minutes focused on three general areas of concern: problems with school work, getting along with other children, and behaviour at home. Mothers were also asked to discuss the strategies used in
dealing with these issues. Strategies described were action-orientated for example, going into school to discuss their daughters’ needs with teachers, organising friendship groups through their own friends or accessing recreational groups, in addition to modifying their daughter’s appearance by finding clothes that reflected her age rather than her size. The study concluded that paediatric nurses had a role in supporting mothers to manage information in these areas. Suggestions included, help with information about TS for teachers, accessing resources including printed material, and contacting support groups. It suggested that “anticipatory guidance” from nurses whereby these issues are discussed at clinic helped mothers to plan how to deal with them. Although this paper is dated, the themes still dominate the lay literature for parents and girls with TS, as illustrated in a booklet produced by the UK TSSS (2008) entitled *TS Lifelong guidance and support.* Information included is comprehensive covering medical, cognitive, emotional and social aspects of the condition from infancy to adulthood. This publication includes chapters providing practical information to help parents with parenting their daughter which reflect areas of concern highlighted by Williams (1995).

### 2.3 Information needs of parents

There is limited literature about information needs of parents with girls with TS from their perspective. However, studies of parents of children with long-term conditions other than TS can inform the current study. Taanila et al. (1998) states,

> The effect of the disclosure of a child’s diagnosis on parents varies little regardless of what kind of severe long-term illness or disability is in question (p505)

Research about parental need for information largely reflects its function and purpose, therefore it appears in the literature in a variety of guises, for example coping and adaptation, practical knowledge for effective parenting, decision-making in the treatment and management, knowledge and understanding of the condition to help them explain their child’s needs to others in health, education and social care settings. Parents are also cited as a major source of information for their children. The role as information provider in this
context is central to CYP understanding their condition particularly when they are young. These aspects of using information will now be explored.

2.3.1 Information about coping and adaptation

The literature relating to coping and adaptation strategies of parents of children with health concerns is extensive. The following studies are included in this review because they provide relevant comment on the significance of information for parents in coping and adapting to a diagnosis of a lifelong condition or disability in their child. Taanila et al. (1998) used questionnaires and interviews to compare experiences of disability and disclosure between parents of children aged 7 to 9 years and those of young people aged 12 to 17 years in Finland. Eighty-five families participated, including 84 mothers and 76 fathers. Findings suggested that the initial information provided to parents of physically and/or intellectually disabled children and the way in which it is provided, potentially forms parents’ view of their child and their child’s future. She concludes that this affects parents’ attitude and interactions with their child. As in earlier studies discussed the data were retrospective with some parents recalling events from 17 years previously. Parents of younger children reported being more satisfied with disclosure than parents of the older children, however, it is not clear as to whether this was due to improved diagnosis and medical management of conditions, improved education of health professionals or an improved attitude toward the future for disabled children.

However, timing and judging the amount of diagnostic information to provide to parents is complicated. Hedov, Wikblad and Anneren (2002) in an assessment of clinical goals regarding first information and support provided to parents of newborns with Down syndrome analysed 168 parent experiences. They found that 75% of parents were given the diagnosis within 24 hours of birth and although 50% were satisfied with the timing of the information others reported it as too soon or too late. This emphasises the difficulty for clinicians in meeting individual needs. Parents also reported they received too much negative information, that the knowledge of health professionals varied and communication
skills could be better. Nevertheless, presentation and delivery of initial information can reduce uncertainty and facilitate coping and adaptation as illustrated below.

Information can reduce stress in two ways, by acting as a guide to available resources enabling parents to deal with specific aspects of their child’s condition or as a coping strategy as parents seek out information to help them.

2.3.1.1 Seeking information as a coping strategy

Sallfors and Hallberg (2003) carried out an exploratory grounded theory study to explore parental experiences of living with a child between 7 and 17 years who have juvenile chronic arthritis. Analysis of open-ended interviews using the constant comparative method led to a core category: parental vigilance. Two related categories included emotional challenge and continual adjustment. These categories described how parents managed the demands of their child’s illness, their child’s vulnerability, and the misunderstandings of others toward their child. “Looking for information” was a subcategory of “continual adjustment”. The sample of 22 including 6 fathers, reported a need for information to ensure the best type of care, to find their own solutions to problems, and to increase their knowledge about treatment. Parents also commented that a lack of information led to anxiety and doubt about their child’s treatment, however, they also believed that most of the responsibility for their child’s illness “lies in their [parents] hands”.

2.3.1.2 Quantity and quality of information

The issue of quantity and quality of information is important because inadequate information has been shown to contribute to parental stress, however, too much information is reported as confusing and may lead to stress, anxiety and lack of confidence in parents to manage their child’s long-term condition (Fisher 2001). The concern of over- or under-informing parents recurs in the literature and the difficulty is compounded by some long-term conditions being more complex and difficult to understand than others, hence the variation of information needs of parents and CYP. Eight research studies analysing the needs and experiences of parents with at least one chronically sick child were critically
reviewed by Fisher (2001). The need for information was one of three main themes to emerge from the studies. The other two were the need for normality and certainty and the need for partnership. Synthesis of the studies identified parents managing the care for children and young people with a wide-range of conditions including chronic liver failure; cancer, cystic fibrosis, haemophilia, congenital heart disease, Lowe disease; Duchenne muscular disease, spinal atrophy type II, metachromatic leucodystrophy, Rhett syndrome, cerebral palsy; type 1 diabetes, juvenile arthritis, asthma and children who were ventilator dependent. This indicates the pivotal nature of information to families caring for children across the spectrum of lifelong conditions, and reveals common themes relating to information needs across their experiences. These included:

- difficulty in obtaining information
- being given insufficient information
- receiving information too quickly
- inaccurate information

Dissatisfaction with the information received triggered an information-seeking response from additional sources such as support groups. This response was seen in the parents of girls with TS (Starke and Moller 2002). All the studies reviewed by Fisher (2001) were carried out in the USA and Canada. It appears that parents across these countries and those in the Swedish studies share a common instinct in seeking information as a coping strategy to adapting to their children’s long-term conditions. Fisher (2001) also argues that nurses working within child-health have an important role in being vigilant to the information needs of families of CYP with chronic illness echoing the role of the nurse in providing information to parents of girls with TS identified in Williams (1995).

2.3.1.3 The experience of other families

Some parents believed that contact with other families may help with their specific challenges through sharing experiences (Van den Borne, van Hooren, van Gestel, Rienmeijer, Fryns, Crufs 1999). This is exemplified in a study of parents’ perspectives of
coping with Duchene muscular dystrophy (Webb 2005). Twenty-three semi-structured interviews were undertaken with 7 couples; 6 mothers and 1 father. Parents stated that detailed knowledge about the condition helped them manage their son’s condition but that talking to other parents to gain practical knowledge was invaluable. Webb (2005) states parents concluded “knowledge is power” and valued other parents as a source of expertise.

2.3.1.4 The internet

A growing number of studies indicate that when parents were uncertain and needed information they used the internet to access information independently of health professionals. Roche and Skinner (2009) interviewed 100 ethnically diverse families and described how parents of children referred to genetic services searched the internet for information, and how they interpreted and evaluated that information. Barriers encountered by the families were also reported. The internet as a source of information is increasingly used and these researchers estimated a range of 20–65% e-health usage by American parents. Barriers to effective use include the nature of the diagnosis and the skills of parents in carrying out searches. Those parents who receive a diagnostic label for their child’s condition are able to use this as a search term, however, less clearly defined diagnoses can limit usefulness of internet searching. Tuffrey and Finlay (2002) reported 84% of parents who used the internet before clinic evaluated the information as useful and easily accessible. However, health professionals are reported as having concerns about the quality and accuracy of information accessed this way. Plumridge, Metcalfe and Coad (2007) carried out a survey to determine what advice was available to families of CYP with genetic and long-term conditions. They analysed 12 websites relating to genetic conditions and 30 relating to other long-term conditions, concluding that there was a “mismatch between the information parents need and that provided by the website” (p225). The focus of information provision was on content rather than process. The researchers acknowledge this was a limited review within one search engine and focused on UK webpages.
2.3.2 Sharing information with their children

Sharing information with their daughters about potentially complicated and sensitive information associated with TS poses many parents with a challenge. General advice relating to sharing information with CYP is that it is developmentally appropriate, cognitively and psychosocially (Bibace and Walsh 1980). Nurses were considered well placed to help families to share information with their children in an analysis of how parents share genetic information with their affected and unaffected children (Gallo, Angst, Knafl, Hadley and Smith 2005). Parents described the process as “unfolding” and based their assessment of what to tell and when to tell, on their child’s developmental stage and interest. The children had a range of genetic conditions including phenylketonuria, sickle cell disease, neurofibromatosis, haemophilia, thalassaemia, Marfan syndrome and Von Willebrand disease. Inclusion of this broad range of conditions was to “capture the diversity and range in parents’ information sharing experience”. Parents’ strategies were found to be flexible and responsive to helping their child to adapt to the condition with the aim of normalising their lives. Parents were generally self-taught using information they were familiar with. Gallo et al. (2005) argue that health professionals may not appreciate the sharing of information between parents and their children as an important aspect of their care. However, in minimising omissions, misunderstanding or inaccuracies in information given to children, it is clearly an important area to explore with parents.

Parents are frequently the main source of information, particularly for young children (Beresford 2000), therefore they need a good understanding of their child’s condition, how it is treated, managed and the lifelong implications for health and well-being. A meta-synthesis of 17 research studies concerning communication about inherited genetic conditions between parents and their children, found what and when to provide information was a difficult aspect of information giving for parents (Metcalfe, Coad, Plumridge, Gill and Farndon 2008). Within this study three researchers independently identified key concepts which were used to identify similarities and differences between the studies. The resulting narrative framework identified various areas of concern for parents in sharing information with their children. These included an assessment of appropriate levels of information to
develop children’s understanding and the need for more help from health professionals for parents enabling them to manage their own emotional needs and those of their affected and non-affected children. In some instances parents may withhold information if they perceive it to be in the best interests of their child (Young, Dixon-Woods, Windridge and Heney 2003).

TS does not have a genetic inheritance component to it, however, some aspects of the clinical features including cognitive and social functioning are considered to be particularly sensitive.

### 2.3.3 Differing needs for information

Meeting parental need for information is difficult for health professionals. An exploration of the complexity of parents’ information needs about the treatment of their chronically ill child and the evaluation of information provision was undertaken by Hummelinck and Pollock (2006). This qualitative study involved semi-structured interviews with 20 sets of parents of children with chronic conditions recruited from a children’s unit in a district general hospital. The chronic conditions included asthma, cystic fibrosis, diabetes, epilepsy, leukaemia, other cancers and severe eczema. Their findings included that the need for information varied greatly between individuals and across time especially related to diagnosis, management plans and prognosis. In summary parents wanted to know everything so that they could prepare for the future; parents expressed multiple needs for information including information that would enhance their ability to manage their child’s illness and understand decisions made. Thereby re-establishing some control over their situation enhancing their ability to cope, to be able to fully answer their child’s questions and to come to terms with the diagnosis. However, there were differences across the diagnoses for example, in those families requiring multidisciplinary care and secondary care management, e.g. cystic fibrosis, parents reported “information overload” whereas those mainly managed in primary care, e.g. epilepsy reported insufficient information irrespective of the severity of the child’s illness. They also suggest there is a tension for parents between
wanting to seek out information independently that could either corroborate or undermine professional judgement and trusting and accepting professional expertise.

2.4 Information needs of Children and Young People (CYP)

Literature pertaining to the information needs of CYP can be divided into different areas of interest which largely reflect policy and professional initiatives relating to their participation in all aspects of their healthcare. Information as a fundamental right for children and young people as defined in Article 12 of the United Nations Convention on the Rights of the Child is accepted within this study (UNCRC 1989). Two broad areas of interest to the current study are now examined: knowledge and developing understanding about their chronic condition.

2.4.1 Knowledge about their medical condition

Developmentally, self-care is an essential skill for children that will promote their self-esteem. Mayall and Hood (2001) suggest that children wish to prepare for adulthood and are keen to learn new skills and acquire knowledge about their health and any issues that affect them. Doctors and parents are reported as key sources of information for CYP with different chronic conditions.

2.4.1.1 CYP’s experiences of the medical consultation

One of the main roles of doctors is the provision and exchange of information (Beresford and Sloper 2003). This study focused the experiences of 11-16 year olds communicating with health professionals in hospital to identify factors which hindered or facilitated the use of health professionals as a source of information. The innovative use of role play complemented the semi-structured interviews. Material produced from a “brainstorm” exercise in the preparatory session for the role play was the data used. The 63 adolescents taking part in this study had been diagnosed for at least a year with one of five chronic illnesses including cystic fibrosis, Duchene muscular dystrophy, epilepsy, juvenile chronic arthritis and diabetes. It was thought this sample size and inclusion of different conditions
would provide a sample that would represent most medical and psychosocial issues experienced by adolescents with chronic illness. Individual interviews were carried out prior to group discussion meetings bringing children with similar conditions and of similar ages together. The adolescents were recruited from different geographical areas, which strengthen the findings because of similarities demonstrated in the experiences of participants.

The participants identified areas of importance including wanting choice about who was present in the consultation, e.g. medical students, parents and also choice about the gender of their doctor. The adolescents’ reported that accessible use of language, focusing on them rather than their condition, and engaging them in the dialogue, would help to readdress the power relations in the consultation. Continuity was important in facilitating communication with medical staff and they preferred talking to someone that they knew, adding that the communication skills of staff were also important for themselves and the doctor. Developmental differences in the younger and older adolescents were evident with the older participants raising concern about privacy and confidentiality whereas the younger participants were more concerned with the high status of the doctor. This theme has been supported in other studies with adolescents with and without chronic illness (Klostermann, Slap, Nebrig, Tivorsak and Britto 2005).

The role of parents was ambiguous, being inhibitory in information exchange for some yet facilitative for others. For some CYP they were considered supportive but for others they were perceived as constraining the development of their child’s relationship with the doctor. Parents have been cited as gatekeepers of information guarding the types of information given to their child (Young et al. 2003). Beresford and Sloper (2003) claim that their results support other research into adolescent-doctor relationships which question the effectiveness of communication skills training for health professionals and believe that general cultural issues within UK health services relating to the rights and adequacy of CYP has to be questioned if their health encounters are to be improved.
A key point from their data was that sometimes there were areas relating to the young person’s condition that they did not want discussed. The adolescent was “making a deliberate choice, at the time of the study at least, not to address an information need in order to protect themselves both from distress and the need to assimilate and adjust to new or more detailed information” (p178). This indicates that the agenda for the consultation should not be driven by adult or medical focus if CYP are to be meaningfully engaged in finding out about aspects of their condition they are interested in.

The desire of CYP to be included and informed in their medical consultation was supported in a grounded theory study exploring the experiences of six mothers and four disabled children (Garth and Aroni 2003). Their results also supported the point that children did not always want details relating to their medical management because it may worry them. However, mothers expressed frustration and felt there was often insufficient information regarding options that they needed to make informed decisions about the future care and management of their child. Children in this study also identified parents as the main provider of information about procedures and aspects of examinations. This poses as a dilemma for medical staff and may in part explain why CYP report being marginalised in these encounters with conversations directed at parents, giving priority to parents’ accounts over the child’s (Savage and Callery 2007). These issues are compounded when a child has any impairment in communication because of the external pressure on time within a clinic environment. The issue of information provision by healthcare professionals and more specifically medical staff directly to CYP is complex and influenced by their personal knowledge, attitudes and abilities in communication, the expectations of children and their parents, and external organisational pressures (Coyne 2006; Gabe, Olumide & Bury 2004).

2.5 The multi-layered nature of information about TS

Ten review articles and one seminar paper discussing TS were selected from the literature search. They are the most commonly cited reviews and provide insight into areas of medical management where there is professional agreement as well as those which are debated.
Listed in chronological order in Table 3 are the selected articles which provide an overview of the fundamental aspects of TS with particular reference to key issues relating to diagnosis, management and treatment.

Table 3: Summary of selected papers

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<th>Date</th>
<th>Authors and title of paper</th>
<th>Purpose of paper</th>
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<tr>
<td>2000</td>
<td>Ross J., Zinn A., McCauley E. Neurodevelopmental and Psychosocial aspects of Turner syndrome</td>
<td>To explore the hypotheses that TS associated psychosocial problems are most likely linked to core neurocognitive deficits and do not reflect a separate component of the syndrome.</td>
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<td>2001</td>
<td>Ranke M.B., Saenger P. Turner’s syndrome</td>
<td>Seminar Paper To provide an overview of the condition and supports the need for multidisciplinary care.</td>
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<td>2003</td>
<td>Kelnar J.H. Growth hormone therapy for syndromic disorders</td>
<td>To examine the rationale and evidence supporting decisions to prescribe GH to children with syndromes where short stature is a feature</td>
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<td>2004</td>
<td>Gravholt C.H. Epidemiological, endocrine and metabolic features in Turner syndrome</td>
<td>To discuss epidemiological, endocrine and metabolic aspects of TS</td>
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<td>2004</td>
<td>Rovet J.</td>
<td>To review the evidence on the physical, psychological and neuroanatomic characteristics of individuals with TS.</td>
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<td>Turner syndrome: Genetic and hormonal factors contributing to a specific learning disability profile</td>
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<td>2005</td>
<td>Bondy C.A.</td>
<td>To update views of TS relating to less severe phenotypes, approaches to cardiac evaluation, HRT regimens and offer suggestions for further research into safety of assisted pregnancy.</td>
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<td>New issues in the diagnosis and management of Turner syndrome</td>
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<td>2006</td>
<td>Donaldson M.D.C., Gault E.J., Tan K.W., Dungar D.B.</td>
<td>To systematically discuss the array of problems faced by affected subjects and to outline their optimal management from infancy to 18 years of age.</td>
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<td>Optimising management in Turner syndrome: from infancy to adult transfer</td>
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<td>2008</td>
<td>Christopoulos P., et al</td>
<td>To report on a review of the research literature clinical aspects of the syndrome and the beneficial effects of hormonal therapy in these patients.</td>
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<td>Psychological and behavioural aspects of patients with Turner syndrome from childhood to adulthood: a review of the clinical literature</td>
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<td>2008</td>
<td>Hjerrild B.E., Mortensen K.H., Gravholt C.H.</td>
<td>To review new insights into genetics, epidemiology, cardiology, endocrinology and metabolism from recent studies.</td>
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<td>Turner syndrome and clinical treatment</td>
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These papers highlight the multi-layered nature of information needs for anyone trying to understand the full significance of the condition, i.e. physical, cognitive, behavioural and social. In view of the wide-ranging implications of TS for individual girls and their families a multidisciplinary approach to management of the condition, under the direction of a paediatric endocrinologist is advocated (Saenger et al. 2001; Donaldson et al. 2006).
Although short stature and ovarian failure are the main features shared by the majority of girls with TS there are other significant health issues to be investigated, monitored and managed. These features of the condition are now explored.

2.5.1 Multi-system implications of TS

The multi-system nature of TS is reflected in the need for a multi-professional approach to its management and care. A diagnosis of TS triggers wide-ranging medical screening for all girls regardless of the age at which the diagnosis is made. Following confirmation of the genetic profile and full assessment of the child’s growth, a raft of additional investigations occurs including:

- cardiac assessment because 50% of girls are reported to have congenital defects some of which will require surgery;
- renal assessment to identify malformations such as horseshoe kidney or duplex collecting systems, which are reported to affect up to 30% of the TS population;
- audiology screening to assess potential conductive and sensorineural loss which worsens with age;
- thyroid and liver function are assessed as girls over 4 years are at risk of developing hypothyroidism as well as other autoimmune disorders such as coeliac disease
- high blood pressure is thought to be present in about 20% of girls and its association with aortic dissection, a rare but fatal condition, needing a proactive approach to management
  (Donaldson et al.2006; Bondy 2007; Saenger et al.2001 ).

Potential psychosocial issues relating to the girls’ behaviour is also explored as many girls have specific learning difficulties and behavioural issues that can impact negatively on their cognitive and social functioning. Good communication across professionals engaged in the initial assessment is essential as families develop their understanding of all the issues involved hence the advocated central role of paediatric endocrinologists (TSSS 2008).
The life-long nature of TS requires on-going monitoring of the health status of girls which is an important part of their lives, therefore frequent contact with health professionals becomes inevitable for them and their parents. Four to six-monthly consultations with doctors provide opportunities to exchange information about current and future management of health and associated psychosocial issues.

Additional problems may be identified at different ages and stages of development (Table 1) illustrates associated problems from infancy to adolescence), which have implications for the girls and their parents. TS is associated with increased morbidity and mortality (Hjerrild et al. 2008), therefore an early diagnosis and lifelong management is considered essential to optimise their health and well-being. Bondy (2007) summarises:

This disorder presents the clinician with a challenging array of genetic, developmental, endocrine, cardiovascular, psychosocial and reproductive issues (p10)

This section highlights the diverse and complex nature of TS which presents a challenge for clinicians in delivering complicated information to girls and their parents.

2.5.2 The use of GH to treat Short Stature in girls with TS

GH treatment became readily available following the development and licensing of biosynthetic GH in 1985. This was a significant change to clinical practice because since the 1950s the limited supply obtained from the pituitary glands of cadavers meant that the use of GH treatment had been very selective. This new source made it a potential treatment option to promote linear growth for all girls with TS. Approximately 95–99% of girls with TS experience short stature (Gravholt 2004) with a mean untreated height of 143cm which is approximately 20cm less than that expected for the family (Kelnar 2003). It is considered to be the most common diagnostic feature always present even in the absence of other clinical features (Cave, Bryant and Milne 2003). However, it is worthy of note that in a small minority of girls if their remaining or damaged X chromosome is from a tall parent the girl’s height may fall within normal population parameters.
Some girls with TS have normal GH (GH) levels and therefore it is thought that girls have reduced sensitivity to GH, resulting in an impaired response to it rather than being GH deficient (GHD). Therefore unlike children with GHD who are prescribed physiologically normal levels of GH, girls with TS are prescribed supraphysiological levels. Medical management involves administering this extra GH subcutaneously at night to enhance the growth response. The increase in growth velocity can potentially normalise a girl’s final height to the lower end of the range recognised for the general population. Various factors influence its efficacy such as compliance with the treatment plan, parental and child expectations and variations in individual response (Hjerrild et al. 2008; Donaldson et al. 2006).

There is some debate as to the optimum age for starting GH treatment and therefore practice between consultants and specialist centres may vary. Donaldson et al. (2006) conclude that there is minimal evidence to suggest starting GH treatment at 2–3 years of age will improve final height and advocate starting when the child enters school at 4 or 5 years of age. However, a more recent randomised controlled study reported girls with TS aged 9 months - 4 years, had their growth failure corrected, and 93% of the girls achieved the normal height range at 2 years of age. The researchers conclude that if a child has an early diagnosis between 1–2 years of age then GH treatment should begin early (Davenport et al. 2007).

This is an important aspect of management because it affects the length of time the girls will require injections. GH is now prescribed into adulthood, posing dilemmas around not only when to commence treatment but also when to stop. Long-term use of GH is considered beneficial both physically and psychologically but as yet there is no conclusive research relating to the implications for the long-term use of high levels of GH in girls with TS (Sandberg and Voss 2002).

2.5.3 Benefits of growth hormone treatment for girls with TS

Research in psychology in the past two decades has focused on function and performance improvements for girls with TS related to GH treatment. The psychosocial implications of
short stature are discussed in the next section. The overall aim of GH treatment is to normalise height. Additional suggested benefits are summarised by Hull and Harvey (2003) as including improvement to “body composition, cardiovascular function, and metabolism” (p.327), and Kelner (2003) argues there are improvements to bone mineral density and body composition.

Unlike conditions such as type 1 diabetes mellitus where the consequences of not administering insulin would be life-threatening, girls omitting to take their GH would not see any immediate effect. Equally, they do not notice any immediate benefit. In addition younger girls (2-7 years) developmentally may not perceive themselves to be short and comparison with peers for them is not yet important (Lagrou et al. 1998). This has implications for adherence in younger age groups where children may be unwilling to have the injections and lack understanding of the rationale for treatment. Developmentally as body image and appearance become increasingly important, motivation to adhere to treatment is high as the treatment offers hope to girls to keep up with their peers. However, this can in some cases raise expectations unrealistically, therefore realistic goals need to be reiterated by health professionals involved (TSSS 2008).

Discussion of the potential treatment options relating to the management of short stature and the consequences in the short and long-term with the girls and their parents requires open and sensitive information sharing.

2.5.4 Does being short matter to children?

An important dimension in the clinical decision to commence girls with TS on GH treatment is the potential for increasing their quality of life (QOL). The literature, however, indicates that there are significant difficulties in interpreting data on GH treatment in relation to QOL of life issues. Controversy begins with definitions of QOL and health-related QOL (HRQOL) which Eiser and Morse (2001) claim are difficult concepts to operationalise. This difficulty creates subsequent problems in the way QOL is then measured and interpreted. There is general agreement between researchers that the language
and content of QOL tools should be appropriate to the experience and developmental needs of children and young people if it is to accurately measure their subjective experience (Eiser and Morse 2001).

QOL tools fall into two key categories. The first category measure the well-being of children and young people generally and aims find out about their subjective perception of health and well-being with specific interest in physical, emotional, mental and social life domains. The second group of tools are designed to focus on a specific group of children with the intention of evaluating the impact of their condition or disability on their experiences. No specific QOL tool was found in the literature search for girls with TS. However, girls with TS have participated in studies using measures designed for children with short stature.

A critical evaluation of eight generic and seven condition specific tools available to assess the HRQOL in children and adolescents with GHD or idiopathic short stature was undertaken by Levke Brutt et al. (2009). One of the conclusions from this evaluation was that current measures neglect the patient and family perception of the child’s height versus the child’s actual measured height. They recommend that development of new HRQOL measures should include self-report and parent report versions in any psychosocial assessment. They also suggest that peers could be approached as a source of information reflecting the reported link between short stature and negative peer relationships.

Being tall is perceived as a positive characteristic whereas being short is seen as less desirable. Social stereotypes which are established at an early age relating to height are considered to result in social stigma (Sandberg and Voss 2002). However, not all children and young people with short stature have negative psychosocial experiences. The contradictory results from studies exploring QOL may in part be related to the cause of the short stature because those children with a medical condition or disability will have additional issues to deal with, which may influence social encounters apart from short stature (Erling 2004). Another difficulty is that the concept of “short stature” is relative and in some cases it is a normal variation of general population heights (Bullinger et al. 2009).
Erling (2004) argues that research to test models of risk and resilience are required to determine factors of importance to the children. However, she does describe “juvenilisation” as a risk factor for the emotional and social development of children with short stature. There may be lower expectations of their abilities by adults, they may be subject to bullying or isolation by their peers and have more limited opportunities to develop independent living skills, which in turn erodes their confidence and sense of autonomy (Erling 2004).

Sandberg and Voss (2002) caution that although studies have demonstrated that short stature is associated with negative stereotypes resulting in psychosocial stressors such as teasing, evidence of maladaptive coping has not been demonstrated. They continue that studies investigating the psychosocial impact of short stature are methodologically weak and therefore “as an isolated physical characteristic appears to hold little value as a predictor of an individual’s quality of life” (p459).

2.5.5 Induction of puberty

In adolescence the medical management shifts its emphasis from growth to the induction of puberty as the treatment focus for girls. Height is still an important consideration and pubertal induction needs to be timed to enable maximum growth but an additional treatment aim is to facilitate puberty to occur at the same time as the development of their peers. Boman, Bryman and Moller (2004) concluded in their study that as long as pubertal induction occurred in adolescence its exact timing was not associated with any negative psychological outcome. On occasions when growth is a priority perhaps because of late diagnosis then induction of puberty may be delayed because the induction of puberty with oestrogen limits longitudinal bone growth as a result of fusion of the epiphyses (Saenger et al. 2001). The majority of girls with TS will undergo medically induced puberty. The perception and understanding of the induction of puberty from the girls perspective has not yet been fully articulated in the published literature. Different experiences are reported with some girls feeling they were more prepared for puberty than their peers whereas others report feeling excluded and on the periphery of their peer group (Boman et al. 2004).
minority of girls do have functioning ovaries and are able to sustain and progress through puberty without any assistance.

The induction of puberty also impacts on transitional care with girls usually remaining within paediatric services until completion of puberty. Saenger et al. (2001) recommend that the transitional period should be gradual with involvement of endocrinologists and gynaecologists to advise on fertility issues.

2.6 Social consequences of TS

The previous section identifies a range of physical problems encountered by girls with TS, however, Saenger et al. (2001) caution that there are also “significant psychological risks associated with TS, including social, behavioural and educational components” (p.3064). Several studies suggest that women with TS have an increased frequency of psychosocial problems (Boman, Bryman, Halling and Moller 2001). This section aims to explore some of these risks in order to provide a more comprehensive and holistic understanding of aspects of TS that may impact on information needs.

2.6.1 Neurocognitive and behaviour profile

It is suggested that some of the specific educational and social difficulties associated with this population have a relationship with the karyotype. From a preliminary investigation into cognitive functioning in girls with TS, Skuse et al. (1997) suggested that there is a “genetic locus for social cognition, which is imprinted”. This has implications for social cognition, and language development depending on whether the girl has deletion of maternal or paternal X chromosome. Skuse et al. (1997) believe that further to neuropsychological and molecular investigations, girls with karyotypes with paternal deletions had better verbal and high order skills, and those with maternal deletions were more vulnerable to developmental disorders of language and social cognition such as autism.

Girls with TS are individual and as such have their own unique learning needs. However, there are some aspects of learning and behaviour patterns thought to be specific to these
girls (Rovet 2004). Girls with TS are generally articulate, however, specific learning issues can include difficulty in concentration, problems with short-term memory, poor coordination, and lack of stamina. Abstract concepts and spatial awareness problems may result in a number of difficulties relating to maths, spelling, understanding of time, drawing, copying, ordering, handwriting and changing point of view. Rovet (1993) states that these selective learning problems are a major concern for parents because of the nature of the difficulties which can be misinterpreted as girls not trying or being deliberately disruptive, by ill-informed teaching staff.

Ross et al. (2002) suggest that

visuospatial organisation and visual memory deficits persist into adulthood probably because these deficits are linked to the loss of genetic material on the X chromosome. (p220)

They conclude there may be improvement in abilities such as perceptual judgement through adolescence which implies there may be hormonal influences on cognitive functioning and Saenger et al. (2001) suggest that some of the identified neurocognitive deficits may be improved by oestrogen therapy. Rovet (2004) stated that longitudinal studies looking at such change at different stages of childhood would be useful in further exploring this hypothesis.

Specific behavioural attributes are associated with girls with TS who are described as socially immature compared with their peers and “out of synch” (McCauley et al. 2001). The difficulties with social cognition may lead to bullying and teasing at school and low levels of self-esteem (TSSS 2002). Eiser (1993) introduced the need for social support as a mediator in coping with the effects of chronic illness. The coping literature is complex and general principles seem to be applied very broadly (Schmidt, Petersen and Bullinger 2003). Issues relating to friendship, family relationships, education and trust of health professionals are central themes. Girls with TS are considered to be on the fringes of friendship groups and may become socially isolated as a defence mechanism (Rovet 2004). The degree to which this may affect adaptation to their condition is not clear.
As adults many women with TS appear to be more dependent, living at home with parents and demonstrate less autonomy than non-TS women (Ross et al. 2000). Boman et al. (2001) cite several studies to support this and conclude that many women with TS have fewer friends, limited social support and delayed or infrequent relationships with partners. Their own cross-sectional multidisciplinary study of 63 women with TS investigating psychological well-being, self-rated health and social life, did find that women with TS have more difficulties in social and partner relationships with 71% living alone and 29% cohabiting. However, they also found no evidence of impaired psychological well-being.

Schmidt et al. (2006) state that “shyness and social anxiety” can be identified from studies involving women with TS with the condition’s physical characteristics, deficits in social cognition, ovarian failure and infertility proposed as the main source of this behaviour. They conducted a study to compare psychosocial distress in women with TS, women with premature ovarian failure and healthy women. Increased shyness, social anxiety, depression and decreased self-esteem were identified in the women with TS and women with premature ovarian failure compared with the control group of healthy women. Although there are limitations from the study design such as a self-selecting sample group which may not be representative, the researchers conclude that these findings indicate infertility to be a major factor in developing this behaviour.

Sutton et al. (2005) undertook a qualitative interview study of 97 girls and women with TS and 21 parents to explore concerns experienced by girls and women with TS across the lifespan from 7 to 59 years of age. They found that infertility was a major concern across childhood (7–13 years of age), adolescence (14–19 years), adulthood (20–39 years) and mature adulthood (40–59 years of age). In the childhood age group (n=7) 36% of the parents who were interviewed with the children stated that the inability to have a biological child had distressed the children. In the adolescent group (n=18) there was a mixed response with some respondents considering it an issue for the future and anticipating future upset whereas others thought about it as a current issue. This aspect of the condition is clearly an important and sensitive area for information provision.
2.7 Implications for the current study

Research into the information needs of girls with TS and their parents is scant and no UK studies were located. The US studies were dated, however, themes are still relevant and reflected in current TSSS information for families. The Swedish studies focused on the investigative and diagnostic period and continuing information needs throughout childhood and adolescence were not considered. Suggestions proffered by parents relating to good practice in giving information at diagnosis reflect some of the recent guidance on breaking bad news. The review of the aspects of TS demonstrates the multifarious nature of the condition and its multisystem involvement emphasising complexity and potential difficulty in the information sharing process between girls, parents and health professionals. As with children with other chronic illnesses there is a need to explore the contribution and involvement of girls with TS in managing and understanding their condition. Issues relating to the function and purpose of information across a range of circumstances such as coping and adaptation, controlling health professionals, self-efficacy in managing the condition, access and response to different sources, and their usefulness are of specific interest to professionals working with CYP with chronic illness and their families. These issues have not been fully examined within the context of TS. The international sources of the papers included in the review indicate that issues and responses to information needs appear to be similar for parents and children across different medical conditions, health systems, socioeconomic groups and cultures.

Many of the studies used retrospective data which can only report the way things were perceived and not how they may be now or in the future. However, such data is valuable as a basis to understand an individual’s experience on which they may base current thoughts and actions. Changes in the management of TS since the availability of GH in 1985 included an increased emphasis on supporting the educational and social needs of girls. The introduction and use of GH may suggest that women who did not receive such support or GH when they were growing up may have a different perspective on information needs than girls growing up today. One study identified normal HRQOL in young women with TS
when they had age-appropriate induced puberty and reached normal height range (Bannink, Raat and Mulder 2006). This generational divide indicates that a study into the information needs of girls who are currently being treated and their parents is timely.

Studies report on what should be shared, access to and evaluation of resources, however, less attention is paid to the specific interaction of sharing sensitive information between parents and their children. No studies were found that indicated how children with TS had their knowledge and information needs assessed or developed over the course of their time in paediatric endocrine services. This study will aim to engage girls between 8 and 16 years to gain insight into what their needs and preferences are for information from their experience of living with TS. In addition, the information needs of their parents will be explored from the perspective of experience of parenting a daughter with TS. Sources of information for girls and their parents and the usefulness of these for managing TS will also be investigated.

Methodological issues raised and their implications for the current study are further explored in Chapter Three.

2.8 Summary and conclusion

This review has aimed to examine issues that are pertinent to the research questions. The value of the perspectives of parents and their daughters is necessary as parents engender attitudes toward the condition and the girls’ response to it. From the literature it is clear the primary source of information for CYP about lifelong conditions is their parents. Nevertheless parents express concern with significant parts of the information process. Common themes run across different studies that are relevant to the design of this research. It is argued that “accessible information” is central to partnerships between parents, children and professionals (Mitchell and Sloper 2002). Girls with TS and their parents are experts in their information needs and therefore a study that explores these needs from their perspective will contribute to understanding how clinical practice can respond effectively to meet them.
CHAPTER THREE: METHODOLOGY

3.0 Introduction

The literature review revealed that although there are publications relating to the medical management and psycho-social implications of TS, there is little direct evidence from girls themselves as to their understanding and knowledge of the condition or evaluation of the usefulness of available sources of information to them. There are also limited studies examining parents’ perspectives of their ongoing information needs. This study is therefore designed to contribute to the development of research that informs practitioners of the information needs of girls with TS and their parents by exploring in depth the understanding, knowledge and experience of living with TS throughout childhood and the experiences of parenting a daughter with this condition. To support the research process through preparation, data collection and analysis an advisory panel was set up. The individuals were invited to offer personal and professional perspectives about TS and included the founder of the TSSS (Arlene Smythe), teenager sub group of the TSSS, paediatric endocrine nurse specialists working in the tertiary centre in which recruitment took place, and a consultant paediatric endocrinologist working externally to the specialist tertiary clinic. The involvement of these individuals is described in the following discussion of the research design at the points where they were engaged in and supported specific aspects of the process.

This chapter will provide justification of particular decisions “from the outset to the conclusion” of this research study (Clough and Nutbrown 2002). Examination of practical decisions taken in its design and construction will enable articulation of the values and assumptions that provide the foundation for this work. The process of data analysis underpinning the research findings which are presented in Chapters Four, Five and Six is also described.
3.1 A qualitative research design

A qualitative approach was taken because it has particular relevance in studies where research questions are aimed at examining the subjective meanings that events and circumstances have for the participants (Flick 2002). This enabled me to explore meanings that girls and their parents attach to TS, and how information was interpreted, shared and valued by them. The literature review identified that the process of information exchange for families of children with lifelong conditions involves many situations and interactions with health professionals and others. Therefore the research needed to take account of the context of any information exchange and its subsequent interpretation within each family in developing their understandings about TS, which could be influenced by their own experiences of TS and its effects of their lives. A strength of qualitative research can be “contextual positioning of data” (Flick 2002).

A qualitative approach is appropriate for exploratory studies of this nature and can enable subtle differences, similarities and complexities of the participants’ individual lives to provide context to their reported experiences and understandings of information. There are various perspectives within qualitative research with different theoretical assumptions to conceptualise the ways in which the participants’ experiences relate to the context in which they are being studied (Flick 2002). A useful strategy of inquiry to explore issues involving social interactions, meanings and interpretations is symbolic interactionism (Charon 2010). This perspective was implicit in the approach of this study; drawn upon to provide a theoretical foundation for the research design and identified as the key organising framework as data were collected and analysed.

3.1.1 Principles of symbolic interactionism applicable to this study

Symbolic interactionism was drawn upon to provide a theoretical foundation for exploration of the perceptions of girls with TS and their parents’ of their information needs. This approach recognises the importance of understanding what individuals know and what they
believe to be important. Symbolic interactionism accepts the participant’s point of view but also draws attention to processes by which views are developed (Benzies and Allen 2001).

Symbolic interactionism was established as a theory by Blumer (1969), who identified three basic assumptions about the way in which human beings can be viewed from this perspective:

- They act toward things on the basis of the meanings that the things have for them
- Meanings of such things is derived from or arises out of the social interaction that one has with others
- Meanings are modified through an interpretive process used by the person in dealing with the things he encounters (Blumer 1969, p2)

These basic assumptions indicate that there is social interaction between individuals, but in making sense of situations there is also internal interaction as individuals define the environment and situation in which they find themselves; therefore individuals are viewed as active and not merely responsive in any given situation.

These assumptions guided the approach of this study in discovering what girls of different ages and their parents experience and understand in living with TS, what information they perceived as necessary and how it was used. Social interactions occur in the process of general information sharing, however, in the exchange of information relating to TS these encounters have a specific context for example, they occur between particular individuals (parents, daughters, doctors, nurses, support groups, teachers); and in specific contexts (hospitals, home, school or support group meetings). In addition the meanings these individuals hold toward social structures including health services, schools, family and childhood will inform their subsequent interpretation and response to information. As such these social interactions and contexts needed to be acknowledged as the context for information needs about TS.
As well as the interactions between individuals, symbolic interactionism is a perspective that encourages the examination of individual meanings (Charon 2010). This approach orientated the study to the exploration of the different ways in which girls aged 8 to 16 years, and their parents invest meaning into their experiences and responses to information that impact on their day-to-day lives and future expectations. Focusing on the detail of individual experiences and meanings enables multiple perspectives relating to information needs to be revealed across the participants.

In line with Blumer’s (1969) assumption that action towards things is related to interpretation of meaning, in this study TS information was perceived as relevant or important only when that was the interpretation assigned to it by the participant. Central to this investigation was exploration of how girls and their parents interpreted information, focusing on what was important to them and how social meanings differed across girls of various ages and in different social situations.

The information published for girls with TS and their parents, is largely focused around the medical management of the condition and there is some information relating to social and educational issues (TSSS 2008). In recognition of girls with TS as active in interpreting situations for themselves and creating meaning from these encounters with information, it was an important aim of this work to ensure that exploration of their information needs was from their perspective. This would enable adult concerns regarding information provision to be redefined and framed within the perspective of the girls. Importance of information to girls and parents may vary and the need for information may fluctuate over time. This is supported by the view within symbolic interactionism that objects are defined and redefined according to the importance of that object at particular times (Charon 2010). As girls grow and their developmental needs change, it is likely that information needs for them will also change. Parents and girls may be able to absorb information more readily in different circumstances and situations, reinterpreting information as they come to experience parenting a girl with TS or recognising themselves as a girl with TS.
Symbolic interactionism provided a focus throughout the research process on the context of information sharing thereby illuminating the experience of receiving and using information from the perspective of the participants. The approach was to interpret information needs from the perspective of individuals recognising that these could differ within families and at different times in their lives.

3.1.2 Qualitative methods utilised within this study

A flexible qualitative approach was utilised in the design of this study to meet the objectives. The methods were selected to promote sensitivity to the lives and circumstances of the girls and their parents. As a novice researcher I needed help and guidance for data management and analysis to illustrate how interpretations are made. Critics of symbolic interactionism suggest a limitation of the approach is that there are no clear processes identified (Charon 2010; Benzies and Allen 2001). However, there are two modes of inquiry articulated by Blumer (1969): exploration and inspection.

Exploration focuses on explaining in detail what is going on in a social situation, identifying all elements and developing a focus of interest. Inspection requires the identified elements to be isolated and subsequently clarified, compared and contrasted in a variety of situations leading to central concepts that can be illustrated in different situations (Stryker 1981). To provide clear process for data management and analysis the design incorporated the Framework approach (Ritchie and Spencer 2002) and principles and procedures of grounded theory methods of analysis (Charmaz 2009). The Framework approach provided a systematic way of organising data by case (girls and their parents) and by theme as the analysis progressed. The principles of grounded theory also provided structure for analysis. As a novice it was helpful to use procedures from these two approaches to guide development of themes, mapping them against different cases and developing my confidence as I progressed with interpretation of the data.
3.1.2.1 Contribution of the Framework Approach to this study

Framework approach was adopted in this study because of the applied nature of this research (Ritchie and Spencer 2002) and to manage complex data from multiple sources (Ritchie and Spencer 1994). It is a flexible approach providing a clear structure through which data can be organised and displayed. Benefits of using the framework approach are that the structure provided by the five interconnecting stages (Figure 3) enables the process of data management and analysis to be clear to those outside the study as well as enabling the researcher to move back and forth across the stages as the analysis develops. The application of this approach to the management of data in this study is described in detail later in this chapter. Each stage facilitates exploration of the context of data developing an “all-inclusive review of data” within and across cases (Ritchie and Spencer 2002). A strength of the Framework approach is its clearly described stages which enabled data to be clearly recorded and managed. This offered me a pragmatic solution to the lack of process that comes with using a symbolic interactionist foundation for this study.

Figure 3: Framework approach
Easy retrieval of data supports the ‘exploration’ mode of inquiry within symbolic interactionism with a primary concern for describing what is happening in a particular setting. Processes of indexing, charting and mapping helped to illustrate interpretations formed during the ‘inspection’ mode of inquiry (Blumer 1969).

However Framework Analysis may appear at odds with symbolic interactionism, and the inductive nature of this study. Framework analysis is initiated from the aims and objectives of a research study, and the use of these specific questions can be considered as deductive, promoting a priori thinking in the researcher. In contrast a symbolic interactionist perspective requires detailed exploration of the social world and rejects the notion of a priori thinking with its defined purpose. To guard against a priori thinking data interpretations which emerged inductively were incorporated into the initial thematic framework rather than using the research questions in isolation.

3.1.2.2 Contribution of grounded theory methods

The second mode of inquiry defined in symbolic interactionism is ‘inspection’. The principles of grounded theory enabled a clear process to guide the development of themes, mapping them against different cases and developing further interpretation. Initial and selective coding, memo writing, and constant comparative method were utilised. Examples of these principles in action are included in the data collection and data analysis sections of this chapter. Moving across datasets for each family comparing similarities and differences was instrumental in refining the topic guide, focusing data collection and ultimately identifying major themes which were then discussed and applied within empirical evidence. These methods reflect the view in symbolic interactionism that within the data elements need to be isolated and subsequently clarified, compared and contrasted in a variety of situations (Charon 2010).

3.1.3 The Semi-structured interview

Interviews are social encounters in which participants reflect and create meaning from their experiences, and is a method consistent with the principles of symbolic interactionism.
There were alternative methods that could have been used. Focus groups are considered a useful method in exploratory studies and where opinions of participants about a particular topic are sought, in this case information needs. However, the focus of this study was individual experience rather than collective experience. It was anticipated there may be similarities across those experiences, however, by using individual interviews the participants may be more open, particularly with regard to sensitive topic areas. Within TS the issues of pubertal induction and probable infertility were considered sensitive topics for parents and their daughters, therefore privacy within the interview situation was perceived as beneficial.

On-going assessment of the interview event is important in an ethical research process for example, being sensitive to participants’ verbal and non-verbal cues may suggest that they do not wish to progress with the topic area under discussion. Differences between interviewing adults and children have been reported particularly when addressing sensitive issues. A study interviewing CYP, aged 9–19 years whose parents had HIV, identified different responses of interviewers (Cree, Kay and Tidsall 2002). The researcher was observed to “back[ed] off” when they sensed the child did not wish to continue talking about sensitive topics, however, during adult interviews the same situation resulted in the researcher seeing an opportunity “to dig deeper”.

Separate interviews for parents and daughters also offered an opportunity for both parties to speak openly without sensing a need to protect one another. Questionnaires would in part promote privacy and have been a useful method in terms of identifying content of information sharing. However, participant interpretations and hidden processes of information exchange would less likely be exposed with the more rigidly controlled questions. Semi-structured interviews are more flexible offering a degree of openness where participants can talk about issues important to them and at the same time the interviewer clarifies meaning or probes further to enhance understanding (Robson 2002, Flick 2002, Kortesluoma, Hentinen, Nikkonen 2003). The level of literacy is not an issue, which was a consideration when collecting data from children with different developmental and
cognitive ability. The reported risks of using semi-structured interviews are in the researcher being judgemental or directive (Flick 2002). Therefore the focus was maintained in the interviews on specific areas of interest, exploring what these mean to the participant. Planning the structure and format of the interview enabled strategies to be developed and utilised to minimise these risks and encourage active listening skills.

This study involved both child and adult participants therefore an interview style that offered structure and flexibility was optimal. Semi-structured interviews are widely used in qualitative research and when used with children are considered to be a valuable tool to elicit information regarding their thoughts and views (Ireland and Holloway 1996).

3.1.3.1 Field Notes

Data collected were influenced by interactions between participants and the researcher. Relationships that developed within the interview situation and the context of interactions between them created meanings. Therefore, in addition to recording the interview, data such as the nature of the encounter and how relationships may have affected data generation were recorded as field notes. Deciding what is important in the situation is difficult. The researcher is advised to note down anything that creates an impression “to react rather than to sift out” (Huberman and Miles (2002, p15). However, to apply some structure I made notes including what the participants were doing, how they were doing it, how they talked about information needs, assumptions made about TS, my observations from the setting. During data analysis I considered what was added by the notes taken (Silverman 2005). This information was valuable later in the research process when transcribing interviews and during data analysis. Recordings and text in the transcripts could be considered and explained within their context.

3.1.3.2 Location of ‘self’

Definition of self and the perception of self through and by others is a central idea within symbolic interactionism. Meanings created through social interactions shape ‘self’ and behaviour (Charon 2010). The interaction between myself and the participants was mediated
through socially constructed symbols that represented shared meanings for example, they knew me as a children’s nurse and we assumed a shared knowledge of the health service processes engaged with and described by them; the term TS was used with shared understanding of its broad parameters. In addition to specific professional role identity, parents and older girls also sought personal information, asking questions such as whether I had children and if I knew the health professionals involved in their daughters care. Interaction is thought to begin with “one’s self to develop a perspective that guides the individual in certain situations” (Burbank and Martins 2009). Part of my interactions within the interview situation was guided by experience, age, ethnicity, gender and roles for example, as a woman, wife, mother, daughter, nurse and teacher. The interview situation is a specific encounter in which I was actively involved. My actions were determined by interpretation and meaning I created within the situation from a variety of sources such as participants’ words and behaviours, the physical environment of the interview, proximity of others, and perceived sensitivity of topic areas.

In summary a qualitative approach was selected for this study to facilitate the collection of data focusing on the interactions that participants engaged in with others, which in turn developed their individual perspective and meaning of TS. The consequences of those interactions and resulting strategies to meet their information needs were made explicit. The interview situation was a social encounter derived through communication and interaction of the participants. My participation and approach in the interview affected the encounter with them because my ‘self’ determines what I see and how I see it. The perceived consequences of this were recorded and discussed in the analysis.

3.1.3.3 Power and Status

Power differential within the research interview situation is generally recognised by researchers. However, symbolic interactionism does not consider issues of power relating to addressing an imbalance of power, but it is interested in how power is enacted through social processes (Dennis and Martin 2005). The power differential between adults and children experienced through social interaction is understood in terms of expected roles
engaged in by one another. Mayall (2000) argues that this power differential is clear to children who believe adults have power over their lives on a day-to-day basis. She concludes that rather than ignore this fact, it is more important that researchers invite children to help them to develop adult understanding of their experiences. Christensen (2004) believes power to be more complex than a mere matter of social position and concludes from her ethnographic studies with children that they will test the reliability and genuine engagement of the adult researcher within the research process. She explains that researchers should not attempt to be child-like but instead present themselves as an unusual type of adult who is seriously interested in the child’s perspective. This will prevent any ambiguity as to the purpose of the researcher being there. The written information explained my professional background and my position as a part-time student engaged in this study. Verbally I expanded on this to provide background about how I had become interested in this topic area.

Being clear about the aim and purpose of the study and my connection to professionals who were involved in their care was an important aspect in developing open dialogue with all participants. I told the girls I was interested to hear whatever they could tell me about TS and which aspects of having TS were important to them thereby avoiding the concept of right and wrong answers. It is an important part of the interview process because children are very adept at recognising tokenism which could subsequently undermine their trust in the situation and potentially affect their participation.

Practical considerations in the interview procedure can give children some power in the situation. Familiarity with recording equipment, for example, practising turning it on and off can help children maintain some control. Time was allocated prior to commencement of the interview to familiarise children with the equipment. Although the girls had agreed to be interviewed I explained they could refuse to answer specific questions or withdraw from the interview at any time. To facilitate this, for younger children a traffic light system was introduced. Three coloured cards: red, orange and green were made available to the girls prior to the interview. If they did not want to answer a question or wanted to stop the
interview they could hold up the red card. If they were unsure about answering and wanted to stop the interview for a short while they would hold up an orange card. Only when they subsequently held up a green card would I continue the interview. These approaches were an attempt to enable the interview to be interactive and facilitated the girls to exercise some control in the interview process. It was not my intention to control the agenda (Cavet and Sloper 2004) hence the need to make the interviews child-centred, informal and reciprocal. Christensen (2004) explains reciprocity enables children to demonstrate engagement with the process. Following an ethnographic study on child-health and self-care she observed that researchers should “facilitate a continuing dialogue” enabling children independence in their conversations. Through sharing the communication experience she concludes there was a sense of “equality and commonality in (their) interactions”.

In conclusion semi-structured interviews were selected as an appropriate method within this research design and were developed to ensure they were used as a specific method of data collection and not a generic information gathering tool (Flick 2002). Sub section 3.5.1 examines the format, structure, content and procedure of interviews utilised in this study.

3.2 Ethical approval

Approval for the study was obtained from a Local Research Ethics Committee (LREC) in September 2006.

3.2.1 Ethical considerations

Traditionally ethical concerns to be addressed when undertaking qualitative interviews are informed consent, the right to privacy and protection from harm (Fontana and Frey 1994). Issues specifically related to these and appropriate for this study are now discussed.

3.2.1.1 Informed consent

Consent is pivotal in ensuring legitimate research (Hagger and Woods 2005). Informed consent is supported by the provision of accurate information about the study provided by the researcher. In gaining ethical approval it was necessary to provide well written
information sheets about the research to all participants (Appendices 1,2,3). The leaflets included information about the purpose of the study, why potential participants had been selected, what their participation would involve, how any information would be protected and used within the study. The information also stated categorically that parents and/or their daughters may withdraw from the study at any time without giving any reason and with no implications for their future healthcare services or treatment. Competence to decide whether to take part in research is based on principles of understanding all aspects of the research study and participating voluntarily. To facilitate understanding of this study information was developed in accordance with expected levels of cognitive development. It was anticipated that girls between 8 and 11 years of age would be thinking in terms of concrete realities and therefore use their everyday experiences to make sense of the study information. Wording was important and therefore information both written and verbal was framed within usual experiences and language of children of this age. Information leaflets were therefore prepared for girls from 8 to 11 years and for girls aged 12 to 16 years to meet their differing developmental needs.

The study design acknowledged parental responsibility (DH 2004) and that parental consent was essential to enable their daughter to participate. However, in line with published research with CYP, the girls were involved in the consent process through the concept of assent, demonstrating respect for their developing autonomy.

3.2.1.2 Assent

Parental consent in this study enabled subsequent assent (a child’s informed agreement to participate) of the participating girls to be secured. Assent emphasised the role of parents acting in the best interests of their child whilst acknowledging the girls as active participants in the process (Lind, Andersen and Oberle 2003). Once written consent was secured from parents (Appendix 4) I explained to the girls all aspects of the research and how they could participate. I had an assent form (Appendix 5) which they signed if they wished to continue.
Consent and assent were not considered to be a one-off process and the concepts were kept as a “live issue” (Cree et al. 2002; Lindeke, Hauck, and Tanner 2000). Viewing consent as a process rather than an event minimises any coercion to continue (Birbeck and Drummond 2007) with the research. Parents and the girls were reminded that they could withdraw from the interview at any point without anyone being cross or disappointed and that a reason for withdrawal was not required. Sensitivity to body language, particularly when interviewing younger girls was also necessary because CYP may not state verbally that they are uncomfortable with questions or feel reluctant to answer (Cree et al. 2002). The traffic light system described earlier was used to assist children in this situation.

3.2.1.3 Anonymity and confidentiality

As far as possible all identifying elements were removed from these data. In this study any reference to names of people (family and professionals), locations, e.g. towns, cities, schools or hospitals were removed in the transcripts. Anonymity was achieved by assigning a code to the interview transcripts and pseudonyms were used for family members and participants. The small number of endocrinologists, particularly female endocrinologists made them more identifiable than other groups therefore it was necessary to ensure references to them were gender free. [Dr] replaced any reference to him or her, she or he in the transcripts.

The girls in this study were interviewed in their own homes. The girls and their parents were consulted about where the interviews would take place and how confidentiality and privacy during the interviews was to be assured. I detected that privacy of the girls was compromised on two occasions, one where the mother requested the door was left open (Case H) and a second where parents joined their daughter and me before she had completed her individual interview (Case I). This is discussed in Findings chapters.

Responsibility to protect the girls’ confidentiality was balanced with child protection policies. How this issue would be dealt with had to be clearly stated in the participant information and in the documentation completed for ethical approval.
3.2.1.4 Protection from harm

Support following interview was arranged with the two PENS with whom the families were familiar and who were aware of the research aims. Families were aware that the PENS knew they had been approached to participate in the study. They were able to act as a resource for the families. Information relating to accessing support groups for girls with TS and their families was also provided offering alternative support from outside health services. The TSSS were also aware of the project’s aims.

3.3 Sampling method

Securing an adequate and appropriate sample is fundamental to any research design. The sample in this study was targeted to explore broad questions of interest with a small number of participants. The age range for this study was selected for the following reasons:

- Their experiences span aspects of development where key treatment decisions are made, e.g. administration of GH and induction of puberty
- Old enough to participate in an interview situation
- To provide insight into the issues that girls consider to be important to them growing up with TS

Purposive sampling was utilised because it is a method of sampling participants who have specific characteristics to fulfil the aims of the research (Robson 2002) and is reportedly favoured within interpretive research methods (May 2002). The exploratory nature of the research and the need to access participants with key experiences and knowledge of information-related issues makes this sampling strategy appropriate. With an incidence of 1 in 2000 live female births (TSSS 2008) TS is not considered rare, although it is relatively uncommon, therefore, there is a need for girls to attend a tertiary paediatric endocrinology clinic throughout childhood and adolescence for specialist care. This clinical setting provided a large population from which to select a sample.
Some criteria for selection were identified. The principal inclusion criteria were English speaking girls who had a diagnosis of TS and were aged between 8 and 16 years. This was a pragmatic decision reflecting the time frame, costs, and the limitations of using translators. Girls with TS have a broad range of features and in some cases may have moderate to severe learning difficulties. The principal exclusion criteria were girls with moderate to severe learning difficulties because I did not have the necessary communication skills to enable their full participation. However, in reality no girls were excluded on these grounds because they did not present to clinic within the timeframe in which I was recruiting.

Bias is reported as the main threat to this type of sampling and the tendency toward convenience sampling and self-selection needed to be avoided. A convenience sample could have been selected for this study by recruiting participants from the membership of the TSSS. This organisation was supportive of the study’s aims and easy access to this group made it a tempting option as it provided a large population from which to recruit. However, it was thought that the population would be largely made up of those families who had identified with the diagnosis and had selected to be part of a group representing the interests of girls with TS and their families. Sampling from this group would have potentially excluded individuals with different attitudes to the diagnosis which could have limited the range of experiences reported by participants. A purposive sample from a tertiary clinic where all girls attend out of necessity of medical management of the condition enabled access to a broader range of participants to be accessed.

3.3.1 Identifying potential participants

The process of recruitment began by identifying girls with TS between the ages of 8 and 16 years from an existing database of all children in this growth clinic who were receiving GH treatment. Girls with TS are treated with GH from 4–5 years of age therefore using this database meant girls within the 8–16 year age band would be on it. Patients seeing all the paediatric endocrinologists working in the tertiary clinic were on this list. The intention was to recruit five girls between ages 8 and 9 years; 10 and 11 years; 12 and 13 years and 14 and 16 years, however it proved difficult to recruit from the youngest age band because of the
few girls available in this group, therefore an even spread across pre-pubertal (8–11 years) and post-pubertal (12–16 years) age groups was aimed for. This division of age had already been introduced in the study through preparation of information leaflets designed for those under and over 12 years. The initial interest in subdividing the sample into four age groups was to enable identification of how girls understand their condition across the ages.

3.3.2 Recruitment procedure

Recruitment took place in a growth clinic in a tertiary paediatric endocrinology clinic using the prepared list of potential recruits referred to in the previous paragraph. An overview of the recruitment procedure is illustrated in Figure 4 over the page. There were three stages to recruitment. Stage One involved providing information about the study to families in clinic and recording a contact number so that I could contact them 7 days later. This gave the families time away from clinic when they could consider the implications of the study and decide whether or not they wished to be involved. In stage two I telephoned families to confirm whether or not they wished to be interviewed. Where agreement to interview was given a convenient date, time and venue was arranged. The third stage of the recruitment procedure was immediately prior to the interview when parents consented to the interview and for me to access their daughter’s medical notes, and written assent to be interviewed was obtained from the girls.

Within qualitative research there is no definitive method for determining sample size. However, ethics committees require an indication of the number of participants to be involved. It was estimated that a sample of approximately 20 girls and their parents would be sufficient to account for variation in the analysis and category saturation. Interviewing and recruitment were concurrent. It is suggested that once recurring issues are emerging in the interview data then further recruitment is not necessary hence 15 families were finally recruited to this study.
Figure 4: The recruitment process

Parents and Girls given information leaflets in growth clinic
n=22

Declined to participate
n=5

Agreed to further contact
n=17

Contacted by researcher via telephone 7 days later
n=17

Agreed to participate
n=15

Interview date, time and venue arranged

Declined to participate
n=2
3.3.2.1 Stage One: providing information about the study

I attended clinic to provide written information to girls and their parents and also to be available to answer any questions about the study. On two occasions I was unable to attend and the PENS agreed to provide a family with an information pack on my behalf. This pack contained a letter of invitation, age-appropriate participant information and study information for parents and contact details. Two other families were given information by their consultant endocrinologist, one family was subsequently recruited and one family declined to participate (Table 4).

Table 4: Summary of recruitment

<table>
<thead>
<tr>
<th>Stage One: providing information</th>
</tr>
</thead>
<tbody>
<tr>
<td>Information given to 22 families in total</td>
</tr>
<tr>
<td>17 families agreed to be contacted 7 days after receipt of information</td>
</tr>
<tr>
<td>5 families declined to be involved</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Stage Two: agreement for participation</th>
</tr>
</thead>
<tbody>
<tr>
<td>17 agreed to be interviewed: dates and venue negotiated</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Stage Three: consent and interview</th>
</tr>
</thead>
<tbody>
<tr>
<td>2 families withdrew prior to interview</td>
</tr>
<tr>
<td>15 families consented to be interviewed</td>
</tr>
<tr>
<td>15 girls gave assent to be interviewed</td>
</tr>
</tbody>
</table>
In clinic girls had their height and weight recorded and following this they were shown to the waiting area where they stayed until their appointment with the consultant endocrinologist. It was during this time that I provided a brief verbal introduction to the study supported by written information. In providing initial verbal information I did not refer to TS to ensure that their diagnosis was not identified in this public arena. I had access to a private room where I was able to talk to any families who had specific questions about the study. This maintained their privacy and assured confidentiality for any medical or personal information they disclosed. Usually parents read the information whilst waiting for their appointment and then sought me out after their medical consultation. Some stated this was preferable because they did not want to miss their appointment slot. Five families reported they had asked their consultant about the study.

3.3.2.2 Stage Two: agreement to participate

Parents and their daughters were generally enthusiastic. Five out of 22 who were given information declined to participate. Two who agreed to be contacted and arranged their interview subsequently cancelled on the day of interview. Following the cancellation I repeatedly contacted the families at their request; however, on each occasion they were unable to identify a convenient time in the future to be interviewed due to school and family commitments. They stated that they were still willing to participate, however, I felt repeated follow up telephone calls could be considered intrusive by the families and therefore after two follow up calls I suggested that they contact me within 2 months if they were able to be interviewed. No further contact was made by them. Although providing a reason for non-participation is not required and families were assured of this verbally and in the written information, five families who declined to participate freely offered explanations which are recorded in Table 5.
### Table 5: Reasons for non-participation

<table>
<thead>
<tr>
<th>Age of daughter</th>
<th>Reason given for non-participation</th>
</tr>
</thead>
<tbody>
<tr>
<td>8 years</td>
<td>Too young to answer questions, <em>she’s shy</em></td>
</tr>
<tr>
<td>10 years</td>
<td>Had participated in another study and did not want to get involved in another one at the moment.</td>
</tr>
</tbody>
</table>
| 11 years        | Mother said her daughter did not like to talk about TS, it was her way of coping with it.  
                  | The daughter said she liked the leaflet |
| 12 years        | Would have liked to participate but very busy time with impending family wedding |
| 12 years        | Girl was very keen to participate. She was accompanied by grandmother who gave me mother’s contact number. Mother declined because she said they had “too much on” |

Motivations of those who agreed to participate are reported as part of the findings in Chapter Four.

#### 3.3.2.3 Stage three: consent and assent to interview

Prior to the interviews with parents and the girls I reviewed the purpose of the study with them and formally took written consent or assent from them. This concluded the recruitment procedure. Details of the participants now follow.
3.4 Participants

There are fifteen cases in this study.

3.4.1 Girls with TS

The initial grouping of pre and post-pubertal girls was further refined in recognition of the need for girls with TS to have medical induction of puberty. This significant event for girls may influence their information needs which should take account of their overall development. Table 6 identifies participants in the pre-oestrogen group (n=9).

Table 6: Summary of cases: pre-oestrogen group

<table>
<thead>
<tr>
<th>Family ID</th>
<th>Interview participants</th>
<th>Daughter’s age (yrs) at interview</th>
<th>Age at diagnosis</th>
<th>Age GH commenced</th>
</tr>
</thead>
<tbody>
<tr>
<td>C</td>
<td>Parent (a) Daughter (pp)</td>
<td>10</td>
<td>8 mths</td>
<td>4 yrs</td>
</tr>
<tr>
<td>G</td>
<td>Parent (a) Daughter (pp)</td>
<td>10</td>
<td>Birth</td>
<td>5 yrs 6 mths</td>
</tr>
<tr>
<td>H</td>
<td>Parent (a) Daughter (a)</td>
<td>11</td>
<td>8 yrs</td>
<td>8 yrs</td>
</tr>
<tr>
<td>J</td>
<td>Parents (dp) Daughter (pp)</td>
<td>10</td>
<td>1 week</td>
<td>4 yrs</td>
</tr>
<tr>
<td>K</td>
<td>Parents (a) Daughter (a)</td>
<td>9</td>
<td>3 days</td>
<td>5 yrs</td>
</tr>
<tr>
<td>L</td>
<td>Parent (a) Daughter (a)</td>
<td>12</td>
<td>18 mths</td>
<td>4 yrs</td>
</tr>
<tr>
<td>M</td>
<td>Parent (a) Daughter (pp)</td>
<td>12</td>
<td>6 yrs</td>
<td>6 yrs</td>
</tr>
<tr>
<td>N</td>
<td>Parents (a) Daughter (pp)</td>
<td>10</td>
<td>In-utero</td>
<td>4 yrs</td>
</tr>
<tr>
<td>P</td>
<td>Parent (dp) Daughter (pp)</td>
<td>10</td>
<td>2 weeks</td>
<td>4 yrs</td>
</tr>
</tbody>
</table>

(a): interviewed alone; (pp): parent present during daughter’s interview; (dp): daughter present during parents’ interview
The age range for the pre-oestrogen group was 9 to 12 years. With the exception of two girls, the diagnosis had been made early. One girl was diagnosed prenatally, four within the first two weeks of life; one at eight months old; and one at eighteen months old. Two later diagnoses took place at ages 6 and 8 years. This contrasts with the post-oestrogen group (Table 7), where all girls were older when the diagnosis was confirmed, between 6 and 13 years and 8 months.

Table 7: Summary of cases: post-oestrogen group

<table>
<thead>
<tr>
<th>Cases</th>
<th>Interview participants</th>
<th>Daughter’s age at interview</th>
<th>Age at diagnosis</th>
<th>Age GH commenced</th>
<th>Age oestrogen commenced</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>Parents (a) Daughter (a)</td>
<td>14 yrs</td>
<td>7 yrs</td>
<td>7 yrs</td>
<td>13 yrs 4 mths</td>
</tr>
<tr>
<td>B</td>
<td>Parents (a) Daughter (a)</td>
<td>12 yrs</td>
<td>6 yrs</td>
<td>6 yrs</td>
<td>12 yrs 4 mths</td>
</tr>
<tr>
<td>D</td>
<td>Parents (a) Daughter (a)</td>
<td>16 yrs</td>
<td>9 yrs</td>
<td>9 yrs</td>
<td>13 years</td>
</tr>
<tr>
<td>E</td>
<td>Mother (a) Daughter (a)</td>
<td>15 yrs</td>
<td>13 yrs 8 mths</td>
<td>13 yrs 8 mths</td>
<td>14 yrs 8 mths</td>
</tr>
<tr>
<td>F</td>
<td>Mother (a) Daughter (pp)</td>
<td>14 yrs</td>
<td>6 yrs</td>
<td>7 years</td>
<td>13 yrs</td>
</tr>
<tr>
<td>I</td>
<td>Parents (dp) Daughter (20mins then parents present)</td>
<td>14 yrs</td>
<td>8 yrs</td>
<td>8 yrs 6 mths</td>
<td>13 yrs</td>
</tr>
</tbody>
</table>

(a): alone; (pp) parent present; (dp) daughter present

Five of the six girls in the post-oestrogen group commenced oestrogen therapy at the ages of 12 or 13 mirroring their peers’ commencement of puberty. However, one girl with a late diagnosis at age 13 years and 8 months had induction of puberty delayed to optimise her growth potential. The two 12-year-old girls in the pre-oestrogen group had discussed at clinic the possibility of starting oestrogen after their next consultation (i.e. in their twelfth year).
3.4.2 Parents

Fifteen mothers and nine fathers participated in the interviews.

Table 8: Parent participants

<table>
<thead>
<tr>
<th>Case</th>
<th>Mother</th>
<th>Father</th>
<th>Reason offered for non-participation</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>✓</td>
<td>✓</td>
<td></td>
</tr>
<tr>
<td>B</td>
<td>✓</td>
<td>✓</td>
<td></td>
</tr>
<tr>
<td>C</td>
<td>✓</td>
<td>✓</td>
<td></td>
</tr>
<tr>
<td>D</td>
<td>✓</td>
<td>✓</td>
<td></td>
</tr>
<tr>
<td>E</td>
<td>✓</td>
<td></td>
<td></td>
</tr>
<tr>
<td>F</td>
<td>✓</td>
<td></td>
<td>Difficult to find time outside work</td>
</tr>
<tr>
<td>G</td>
<td>✓</td>
<td></td>
<td></td>
</tr>
<tr>
<td>H</td>
<td>✓</td>
<td></td>
<td>“not interested in getting involved in this type of thing”</td>
</tr>
<tr>
<td>I</td>
<td>✓</td>
<td>✓</td>
<td></td>
</tr>
<tr>
<td>J</td>
<td>✓</td>
<td>✓</td>
<td></td>
</tr>
<tr>
<td>K</td>
<td>✓</td>
<td>✓</td>
<td></td>
</tr>
<tr>
<td>L</td>
<td>✓</td>
<td></td>
<td>Caring for siblings</td>
</tr>
<tr>
<td>M</td>
<td>✓</td>
<td></td>
<td>Difficult to find time outside work</td>
</tr>
<tr>
<td>N</td>
<td>✓</td>
<td>✓</td>
<td></td>
</tr>
<tr>
<td>P</td>
<td>✓</td>
<td>✓</td>
<td></td>
</tr>
</tbody>
</table>

Where a couple had agreed to participate they were interviewed together. Six mothers were interviewed alone. In Cases F, H, M, and L mothers spontaneously offered an explanation of
why the father had not participated (Table 8). Two mothers made no comment (Cases E, G). With the exception of one family all parents were sharing the family home. The parents who lived separately arranged to be interviewed together and only after the interview did they explain they lived apart. I aimed to interview the girls alone but responded to the preferences of the participants.

3.5 Data Collection

This section describes in detail the planning and execution of the data collection process beginning with considerations for the interview procedure.

3.5.1 Structure and format of interviews with girls and their parents

There are benefits and limitations to using group or individual interviews with children. Coyne (1998) emphasised the alien environment of the interview situation to children and suggested they may feel more secure interviewed with children from their own age group. Peer support may help to minimise the power differential between the adult/researcher and the child. Skills required by the interviewer vary with differing demands of different types of interview for example, managing groups rather than one to one. Hill, Laybourne and Borland (1996) compared their experiences of group and individual interviews. They found that children’s responses in groups were more spontaneous whereas individual interviews yielded more thoughtful responses. Privacy, they state was assured leading to expression of deeper feelings and quieter children who were reticent in the group situation were more able to respond. Some children were more reserved in the one to one situation. Girls with TS are reported to be less comfortable in social situations and also there was potential for sensitive topics to be raised therefore it was decided to undertake individual interviews.

Parents were generally interviewed first which provided insight into their daughter’s condition and enabled exploration of social processes relating to family communication and information sharing for example, in one case the mother specified discussion about infertility with her daughter was not permitted. It was hoped interaction with parents would
foster their confidence in me and the interview process thereby increasing the possibility of allowing their daughter to be interviewed separately.

Interviews with the girls were structured around their everyday experiences to make the situation more relaxed for them and to encourage participation (Backett and Alexander 1991). Where possible the interview was led by the girls. This was because the purpose of the interview was to elicit their story highlighting areas of interest to them which may not be the same as that of adults or even be a focus on their lives at all. The intention was to construct and format interviews that created an environment where there was no pressure for answers and where answers were not considered right or wrong. Planning the predictable and ‘known’ elements of the interview situation is essential, however, there also needs to be a flexible approach to the families at the time, responding to the unpredictable environment and unexpected requests of the participants. The estimated timing for the interviews with the girls and parents was one hour; however, I was sensitive to the individual situation and responded by shortening or lengthening the time.

3.5.1.1 Preparation of supporting materials

Subtle social and learning skill deficits are part of the TS profile (TSSS 2008) and needed to be accounted for in order to facilitate participants’ participation in the interview process. Preliminary decisions were made relating to the predictable aspects of the interview process (Table 9).
Table 9: Preparation for interviews

<table>
<thead>
<tr>
<th>Known requirements for interviewing parents and girls with TS</th>
<th>Resources used/ prepared for interview</th>
<th>Source of guidance in development and preparation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Record interview</td>
<td>Digital recorder</td>
<td>Supervisor, experienced colleagues</td>
</tr>
<tr>
<td></td>
<td>Tape recorder with sensitive microphone for group interview</td>
<td></td>
</tr>
<tr>
<td>Develop topic guide</td>
<td>Topic guide for parents</td>
<td>Published literature including TSSS information leaflets</td>
</tr>
<tr>
<td></td>
<td>Topic guide for Children and young people</td>
<td>Pharmaceutical leaflets</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Supervisor, research colleague</td>
</tr>
<tr>
<td></td>
<td></td>
<td>*TSSS Mother and daughters group; TSSS Teenagers group</td>
</tr>
<tr>
<td></td>
<td></td>
<td>*PENS</td>
</tr>
<tr>
<td></td>
<td></td>
<td>*Paediatric endocrinologist.</td>
</tr>
<tr>
<td>Develop/ identify supporting material</td>
<td>Paper, pens, crayons, pencils</td>
<td>Published literature</td>
</tr>
<tr>
<td></td>
<td>“About Me” booklet</td>
<td>Experienced research colleague</td>
</tr>
<tr>
<td></td>
<td>Strategies for pictorial representation of information needs for use with younger children</td>
<td>Previous learning from Researching with Children course at the Social Policy Research Unit, York University</td>
</tr>
<tr>
<td></td>
<td>Coloured cards for traffic light system to use during interview with young children</td>
<td></td>
</tr>
<tr>
<td>Identify style of questions and prompts</td>
<td>List for reference</td>
<td>Published literature</td>
</tr>
</tbody>
</table>

*Denotes individuals from the advisory panel
A booklet “About Me” was developed to support the interview. TSSS (Arlene Smythe) suggested this type of activity may help girls to engage with the interview process as it would act as a mediator of information. Although the documented information was about them we would discuss what was written as a starting point for discussion thereby avoiding direct questioning which some girls with TS find uncomfortable. The booklet contained sections that mirrored the topic guide for example, asking about friends and family, school activities and achievement, hospital experiences relating to management of TS and things they already knew or would like to know about TS. It is beneficial to structure the questions around everyday experiences to provide familiarity (Backett and Alexander 1991).

3.5.1.2 Topic Guide and questioning technique

Using open-ended questions such as ‘how’ rather than ‘what’ or ‘why’ encourages the participant to talk around subject areas avoiding the risk of one word answers. However, a series of direct questions when interviewing young children can be useful in providing opportunities for the child to speak and reveal information which the researcher can subsequently explore (Irwin and Johnson 2005). To augment the interview process with children it is suggested that visual methods are employed as ice breakers and promote dialogue between the researcher and child. Such approaches it is argued help to facilitate discussion and enable children to be active participants (Pridmore and Bendelow 1995). Crayons, pens and paper were used during the interviews with younger girls and pictures when discussing what they knew and what they might want to know more about. This helped to reduce a sense of being ‘tested’.

The topic guide was grouped into familiar contexts; experiences at home, experiences at school, experience of hospital. Added to these were issues such as understanding TS, and issues related to growing up. Issues about growing up such as pubertal development were discussed in an age-appropriate way using Personal, Social and Health Education (PSHE) syllabus, which indicates specific content relating to relationships and puberty for each school year and forms part of the national curriculum. This was useful in orientating me to
the expected level of knowledge of girls relating to these topic areas and offered reassurance to parents.

The topic guide on the next page, (Table 10) incorporated themes found in the content of the published literature. In recognition of the potentially sensitive nature of these topics I attended a TSSS mothers and daughters weekend to develop insight and seek further guidance. The aims of the study were presented to the main group of attendees. Following this, two groups volunteered to scrutinise the topic guide and information leaflets. The teenager group examined information for girls aged 12–16 years and a group of four mothers and their daughters under the age of 11 years considered the information for 8–11 year olds. Comments were noted and information amended accordingly. They stated that they thought the study was worthwhile and relevant to them. The topic guide (see over page Table 10) identifies key areas to explore across the two age groups but related verbal questioning was adapted to the age, developmental level and response of each child.
Table 10: Initial topic guide for girls

<table>
<thead>
<tr>
<th>Experience at school</th>
<th>Growing up</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Typical school day</td>
<td>• Current interests</td>
</tr>
<tr>
<td>• Likes</td>
<td>• Puberty</td>
</tr>
<tr>
<td>• Dislikes</td>
<td>• Body image</td>
</tr>
<tr>
<td>• Social activities/ Clubs/ friendships</td>
<td>• Height and weight</td>
</tr>
<tr>
<td>• Exercise</td>
<td>• Sharing information</td>
</tr>
<tr>
<td>• Support</td>
<td>• Choice/ decision-making</td>
</tr>
<tr>
<td></td>
<td>• Independence</td>
</tr>
<tr>
<td></td>
<td>• Friends and peer relationships</td>
</tr>
<tr>
<td><strong>Life with family and friends</strong></td>
<td><strong>Understanding TS</strong></td>
</tr>
<tr>
<td>• Activities outside the home</td>
<td>• What do you understand by TS</td>
</tr>
<tr>
<td>• Typical non-school day</td>
<td>• When did you first find out about it</td>
</tr>
<tr>
<td>• Domestic activities engaged in</td>
<td>• Growth hormone</td>
</tr>
<tr>
<td>• Relationship with parents/ siblings/ others</td>
<td>• Nutrition</td>
</tr>
<tr>
<td></td>
<td>• Sources of support</td>
</tr>
<tr>
<td></td>
<td>• Information sources and available resources</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Experience of hospital</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>• Relationship with staff</td>
<td></td>
</tr>
<tr>
<td>• Environment</td>
<td></td>
</tr>
<tr>
<td>• Examinations and questions</td>
<td></td>
</tr>
<tr>
<td>• Involvement in decision-making</td>
<td></td>
</tr>
<tr>
<td>• Treatment regimen</td>
<td></td>
</tr>
</tbody>
</table>

As data collection and analysis progressed the topic guide was altered to add issues that arose, for example, gender of medical staff.
3.5.1.3 Location

Common places cited for interviews are in the child’s home, school and in hospital. Positive and negative issues of these locations were identified. Girls with TS are experts in their lives and insight into their experience of living with TS will be valuable to adults supporting them. Selecting the most appropriate environment for them to participate will optimise their contribution to the study. Girls with TS prefer familiarity therefore their own home was selected as a suitable location. Hill et al. (1996) reflect on interviews within children’s own homes and at school. They conclude that individual interviews undertaken at school were the ones in which the children appeared the most relaxed and open, surmising that children were less anxious because they knew that parent(s)’ could not overhear their responses. However, privacy is not always desired by children and therefore researchers need to be more responsive to the individual situation.

There were different scenarios for the presence of parents during their daughter’s interview and for daughter’s presence during their parent’s interview. In the pre-oestrogen group, six were interviewed with parents present and three (Case H, L, K) were interviewed alone. Three girls requested that their parents remain in the room with them (Case C, N, P); one of the girls joined her parents and the interview continued as a family with all parties contributing (Case M); one set of parents stayed because their daughter had learning difficulties (Case J); in Case G the mother requested to stay because of her daughter’s young age (10 years). Parents offered helpful prompts and were supportive without being directive. In the post-oestrogen group, with the exception of one girl who requested that her mother should stay, all girls were interviewed alone. In Case I parents were preparing a meal. The interview started with their daughter and they came to join us after 20 minutes.
3.5.1.4 Creating datasets

Data collected was put into datasets developed for each family and referred to as *cases*. These cases included:

- Verbatim interview transcripts
- Supporting materials from the interview such as ‘About Me’ Booklets or pictures drawn by children
- Field notes
- Supporting demographic and medical information
- Synopsis of the interview written by me to order information chronologically

3.6 Data analysis

Symbolic interactionist modes of inquiry, exploration and inspection, are integral to this data analysis process (Blumer 1969). To enable this to occur, framework analysis and grounded theory methods were employed.

Framework approach stages were utilised as a structure for managing data (Ritchie and Spencer 2002). The constant comparative method occurs throughout the analytical process. It began as each transcript was coded. Each idea and incident that was assigned a code was compared with similar examples, and new categories were applied to previously coded data. It is a complex activity that involves developing and integrating categories at the same time. Coyne (2008) describes it as “trying to put the pieces of a puzzle together without having the picture available”. It was a process that was integral to the whole analysis.

An overview of the process, procedures and researcher’s activities throughout the analysis are illustrated in Figure 5. This figure depicts the processes of coding and the constant comparative method which requires constant movement back and forth between each stage of the analysis. At times during the analysis these processes were occurring simultaneously. Presentation of this abstract process in an apparent linear style is difficult, however, the aim is to capture some of this movement for the reader through examples from the analysis.
Figure 5: Overview of processes and procedures

<table>
<thead>
<tr>
<th>Process</th>
<th>Procedure</th>
<th>Researcher Activities</th>
</tr>
</thead>
<tbody>
<tr>
<td>Assemble primary data</td>
<td>Data Collection</td>
<td>Transcribe Interview data</td>
</tr>
<tr>
<td>Sift and sort data</td>
<td>Data Management</td>
<td>Collate Field notes; demographic data &amp; medical information</td>
</tr>
<tr>
<td>Identify initial ideas</td>
<td></td>
<td>Literature for girls and parents produced by TSSS &amp; pharmaceutical companies</td>
</tr>
<tr>
<td>Detect patterns</td>
<td>Descriptive Analysis</td>
<td>Framework Approach</td>
</tr>
<tr>
<td>Seeking explanations</td>
<td></td>
<td>Familiarisation: listening, transcribing, reading</td>
</tr>
<tr>
<td>Wider application</td>
<td>Explanatory Analysis</td>
<td>Conceptual Framework: using topic guide and initial recurrent ideas</td>
</tr>
<tr>
<td>Explicate meanings</td>
<td></td>
<td>Indexing: early common ideas</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Initial Coding: line by line; in vivo</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Selective Coding: focused, mapping themes across cases, developing charts</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Memo writing</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Clarifying context of themes across cases: general and specific</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Illustrating similarities and differences across cases/ girls / mothers/ fathers</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Theoretical integration with examples outside TS/ theory and published research</td>
</tr>
</tbody>
</table>

Adapted from Charmaz (2009); Ritchie & Spencer (2002)
3.6.1 Data Management

The framework was utilised in this study to manage and organise data. It has five interconnecting, non-linear stages (Figure 3). These five stages allowed data to be explored broadly and kept the initial framework flexible, enabling new ideas emerging from the data to be easily added. It also promoted an iterative process from the outset, which is essential in qualitative analysis as a range of broad views expressed by the participants were built up. In summary this approach incorporated structure and flexibility which was useful when working with datasets from parents and girls with TS.

3.6.1.1 Familiarisation

This initial stage involves the researcher immersing themselves in data with the aim of reviewing the range and diversity of information collected. I was able to develop an overview and appreciation of the scope of data. Familiarisation with data meant a systematic process of sifting and sorting material developed preventing a sense of being inundated by the amount and diversity of data generated. Initially data in this study was looked at within each family, firstly the parents’ transcripts and then their daughter’s. As interviewing and transcribing progressed I was able to also recognise similarities and some differences across families, similar ideas amongst parents and similar ideas among the girls. This approach enabled data to be considered within cases and as a whole. Added to this was data from field notes. The process of familiarisation involved initially listening to the recordings and later reading repeatedly the transcripts and field note summaries. At this stage anything that appeared interesting was highlighted. In the first transcript 205 different pieces of text were highlighted. For illustration the table below illustrates some of these in no particular order of importance.
<table>
<thead>
<tr>
<th>A lot of individual things</th>
<th>It just described her</th>
<th>Like a weight falling off your shoulders</th>
</tr>
</thead>
<tbody>
<tr>
<td>She likes familiarity</td>
<td>It’s difficult to know if she’s really understood what you’ve said</td>
<td>I like (Dr). comes out at your level</td>
</tr>
<tr>
<td>Everyone cleared their own little bit</td>
<td>Totally out of me depth</td>
<td>(Dr) directs the answer to Anna</td>
</tr>
<tr>
<td>She’s lovely, dead lovely</td>
<td>Kids will always find it difficult to talk to parents about relationships</td>
<td>We’ve been seeing a lot of different doctors...it’s difficult for her cos she’s got to then start up another relationship hasn’t she</td>
</tr>
<tr>
<td>Not good with people she doesn’t know</td>
<td>Me Dad’s got Alzheimer’s it’s the same sort of thing</td>
<td>She can’t articulate what’s going on in her head</td>
</tr>
<tr>
<td>Genetics are complicated</td>
<td>Misfit</td>
<td>That worries us the most how she’d cope with demanding social situations</td>
</tr>
<tr>
<td>Anna’s mind, well you can’t get in there its shut and even when she’s desperately unhappy we can’t find out</td>
<td>We were really wary of her going and getting lost in this big school</td>
<td>As long as she’s happy knows where she wants to be .we’ll give her all the support she needs</td>
</tr>
<tr>
<td>Real problems at school, didn’t want to listen</td>
<td>Dr (consultant Endo) did explain things to her</td>
<td>Just forgets things</td>
</tr>
<tr>
<td>(School work) It was a battle, it was unpleasant</td>
<td>She’s less receptive to some of the doctors</td>
<td>Telling siblings</td>
</tr>
<tr>
<td>We didn’t understand</td>
<td>Shock and sadness</td>
<td>Few disasters with cartridges and she (nurse)was saying how much they cost you think oh my god</td>
</tr>
<tr>
<td>Immature compared with other girls</td>
<td>She’s not one for mixing</td>
<td>Patient literature we first got was a godsend</td>
</tr>
<tr>
<td>Social skills worry us the most</td>
<td>mind blowing</td>
<td>Stubborn, single minded</td>
</tr>
</tbody>
</table>
3.6.1.2 Developing a thematic framework

The next step using Framework analysis involved developing a thematic framework. The framework was developed from common ideas identified in the familiarisation process and the research aims which had been introduced to the participants through the interview topic guide (Ritchie and Spencer 2002). Broad areas of interest were assigned a numerical number as demonstrated in the early index/thematic framework.

Table 12: Early index

<table>
<thead>
<tr>
<th>Number</th>
<th>Category</th>
</tr>
</thead>
<tbody>
<tr>
<td>1.0</td>
<td>Sources of information</td>
</tr>
<tr>
<td>2.0</td>
<td>Sharing information</td>
</tr>
<tr>
<td>3.0</td>
<td>Knowledge and understanding of TS</td>
</tr>
<tr>
<td>4.0</td>
<td>Identity</td>
</tr>
<tr>
<td>5.0</td>
<td>Growing up</td>
</tr>
<tr>
<td>6.0</td>
<td>Social relationships</td>
</tr>
</tbody>
</table>

The index at this stage enabled the large mass of data to be divided into “manageable ‘bites’ for subsequent retrieval and exploration” (Ritchie and Spencer 2002). For this reason Ritchie and Spencer (2002) caution about being, “over elaborate in detail because the aim is to develop an overview of these data and not be interpretive at this early stage. As the index was applied to further interview transcripts it became clear that refinements needed to be made which moved the process into a descriptive stage of analysis.

3.6.2 Descriptive analysis

3.6.2.1 Initial coding

Familiarisation with the transcripts had involved reading, re-reading, highlighting or writing in the margin any word or phrase that appeared interesting or relevant to the parameters of the study. Continued application to subsequent transcripts further developed and extended the index as new ideas emerged, for example transcript H had a strong issue about visibility
to others and concealing information so these were added. Transcripts previously coded were also revisited in light of any new emergent ideas reflecting the back and forth nature of the qualitative research process.

The transcripts were now coded line by line and assigned a numerical code that corresponded to the index. The purpose of this was to break down the data in order to begin categorisation. This line by line approach focuses the researcher on the raw data and therefore the temptation to introduce preconceived ideas is avoided (McCann and Clarke 2003). However, this was not a straightforward exercise because the early index was ‘subject’ orientated and the ideas presented by parents and girls were often about how they felt at a particular moment within information sharing or they described actions taken in light of information received. Therefore subcategories were developed within the index to accommodate these data and opening up the broad early categories. Strauss and Corbin (1998) describe this technique as axial coding where data are assembled by categorising and articulating how the subcategories link to the categories. They were tentative categories but they enabled data to be organised more purposefully. It was important not to force any ideas into the index but to keep the index fluid and responsive to what I was hearing and reading within the interview data. The subcategories demonstrated the wide-ranging nature of each category. Table 13 illustrates an early category of Sources of information and the subcategories that developed within it,
Table 13: Early Category: sources of information

<table>
<thead>
<tr>
<th>1.0 Sources of Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>1.1 Timing- when information was received or gathered</td>
</tr>
<tr>
<td>1.2 Information received</td>
</tr>
<tr>
<td>1.3 Information found independently</td>
</tr>
<tr>
<td>1.4 Ease of access</td>
</tr>
<tr>
<td>1.5 Perceived value of information</td>
</tr>
<tr>
<td>1.6 Misinformation/ misunderstanding</td>
</tr>
<tr>
<td>1.7 Lack of knowledge/ information</td>
</tr>
<tr>
<td>1.8 Suggestions for improving information sharing</td>
</tr>
<tr>
<td>1.9 Information transfer between professionals</td>
</tr>
</tbody>
</table>

**3.6.2.2 Constant Comparative Method**

This grounded theory method occurs throughout the analytical process. It began as each transcript was coded. Each idea and incident that was assigned a code was compared with similar examples and new categories were applied to previously coded data. It is a complex activity that involves a process of developing and integrating categories at the same time. Coyne (2008) describes it as “trying to put the pieces of a puzzle together without having the picture available”. Although introduced here it was a process that was integral to the whole analysis.

Once all the transcripts had been coded, the aim was to develop a picture of the data as a whole by considering the range of attitudes and experiences in each of these early categories. I began to develop tables for each category and subcategory. As I moved text into this tabular format it became clear that there were overlapping categories and integrated ideas. This made it difficult to decide where they should be included. In some situations it was possible to separate the issues and assign them to a suitable category, however, this was
done with caution to guard against losing the meaning and/or context of what had been said. It was important not to code on the basis of content, e.g. GH or short stature but to ensure the context and meanings of what was said remained at the core of coding decisions to ensure they were faithfully represented. At this point in the analysis it became clear that there was an increasing amount of interlinking and overlapping of ideas. An example of this was ‘misunderstanding of information’ and ‘misinformation’ which was described similarly by some parents and differently by others. The supervisory team prompted a further examination of data in its original context to determine how the terms had been used and described by the parents. Both of these ideas were being associated with complexity. Ideas that linked subcategories and/or categories together were briefly recorded often just as bullet points and kept on file for future reference as the analysis progressed.

**Table 14: Linking subcategories and categories**

<table>
<thead>
<tr>
<th>Complexity</th>
</tr>
</thead>
<tbody>
<tr>
<td>• <em>Around diagnosis there is an emphasis on understanding what TS is and the</em></td>
</tr>
<tr>
<td><em>implications of the diagnosis</em></td>
</tr>
<tr>
<td>• <em>There is then complexity for parents in trying to understand what TS</em></td>
</tr>
<tr>
<td><em>means for their daughter specifically relating to health, education and</em></td>
</tr>
<tr>
<td><em>social functioning. A shift from the general to the specific individual</em></td>
</tr>
<tr>
<td><em>information</em></td>
</tr>
<tr>
<td>• <em>Later in the sequence i.e. once the parents have learned what TS</em></td>
</tr>
<tr>
<td><em>means for their daughter some parents seem less concerned about the</em></td>
</tr>
<tr>
<td><em>complexity of many characteristics, e.g. chromosome analysis, and focus</em></td>
</tr>
<tr>
<td><em>on what they do know and what they can influence, e.g. monitoring</em></td>
</tr>
<tr>
<td><em>aspects of management, e.g. growth</em></td>
</tr>
</tbody>
</table>

This was a useful practical strategy to map the development of thinking about specific ideas and later inform memo writing.
3.6.2.3 Selective Coding

Coding now moved to a new phase, away from ‘sifting and sorting’, open and axial coding to selective coding. This change in the analysis developed sensitivity to patterns and ideas that appeared connected and was the beginnings of explanations for these. One method of facilitating this activity was the writing of detailed memos. Having described the data, the purpose of memo writing was to now start to perceive the data theoretically.

3.6.2.4 Memo writing

To move the analysis to a higher level of description questions were asked of the data. Memos were useful in exploring the identified categories by developing ideas about them. Their purpose is to prompt the researcher to analyse codes early and then through successive memos to increase the level of abstraction (Charmaz 2009). The supervisory team suggested that initial explanations for the categories should be written. Some early memos were still descriptive in part whereas others were more detailed aiming to define the nature of the categories and the relationships between them. Table 15 provides an excerpt of an early memo relating to sources of information.

Table 15: Excerpt from an early memo

<table>
<thead>
<tr>
<th>Memo: Initial category ‘Sources of Information’ (Parent interviews)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1.0 Sources of Information</td>
</tr>
<tr>
<td>1.1 Timing</td>
</tr>
<tr>
<td>1.2 Information received</td>
</tr>
<tr>
<td>1.3 Information found independently</td>
</tr>
<tr>
<td>1.4 Ease of access</td>
</tr>
<tr>
<td>1.5 Perceived value of information</td>
</tr>
<tr>
<td>1.6 Misinformation/ misunderstanding</td>
</tr>
</tbody>
</table>
1.7 Lack of knowledge/information

1.8 Suggestions for improving information sharing

1.9 Information transfer between professionals

These categories relate to the content element of the respondents’ narrative. They do not explicitly illustrate the process of information acquisition including the involvement of others, e.g. professionals, support group.

Coded data prompts questions e.g. at what point in time do parents seek information; why do they seek information at different times; who is involved in finding sources of information; what makes some parents actively seek information and others to be more passive; do mothers and fathers respond to seeking and receiving information differently; where do they get information from; how accessible is it both physically and intellectually; why are some sources more valued by parents. Different dimensions relating to the acquisition of information are explored broadening the initial category and incorporating not only where information comes from but also the processes involved.

**Timing**

- Some parents sought information prior to diagnosis trying to make sense of aspects of their daughters’ behaviour or health in determining that there was something wrong.

- Some parents sought information prior to seeing the consultant endocrinologist

- Some parents continue to seek information responding to daughter’s developmental stages; some are vigilant and want to be better informed; lack of trust in the health system

- Some parents did not seek information but waited to be informed of what they needed to know by paediatric endocrinologist

- Mothers and fathers may need information at different times and delivered in different ways. Parents should not be perceived as “a job lot”, they may have different needs and ways of accessing information.

- Timing is also an important component of giving information to others. These ‘others’ may be family members, friends or the girls themselves. “When” to tell, and also what aspects of the diagnosis are determined as suitable to share. This may be socially or developmentally influenced.

So there appears to be an active–passive continuum, with some parents actively seeking information from a variety of sources and others waiting to receive relevant (as perceived by health professional) and filtered information from official sources.

Writing these explanations emphasised relationships between the initial categories and also the consequences they had for one another and independently. Memos are useful in
examining processes (Charmaz 1995) which were essential in exploring perceived information needs, for example, explanation relating to the early categories led to consideration of the process of information giving and receiving, the conditions in which this occurred, how parents and girls engaged with the information process and subsequent consequences to them of that process.

Some of the connecting ideas between the categories documented earlier in the analysis process also became the subject of memos. Selection and importance of categories was strengthened through the development of charts that provided a clear visual reference of data across cases, and individuals within cases. Some categories were strengthened by the amalgamation of subcategories which refined and redefined their initial parameters. These categories were now identified as initial concepts.

3.6.2.5 Charting

Another method employed in developing the descriptive analysis was the use of charts. As stated above as data were compiled into tabular format they prompted further memo writing. Charting was utilised in different ways. Initially within each case to identify what had been said in relation to the category and then later to develop a picture of the data as a whole by considering the range of responses across cases. Transcribed text from the interview transcript was moved, the line number was identified so that the text could be read within the context and structure of the interview to ensure it remained grounded in the participants’ meaning. Table 16 summarises parental responses to one topic area. It illustrated in an excerpt from Case B for the theme coded as 1.5 which represented *Perceived value of information.*
Table 16: Index Code: perceived value of information

<table>
<thead>
<tr>
<th>ID</th>
<th>Index code</th>
<th>Line</th>
<th>Text</th>
</tr>
</thead>
<tbody>
<tr>
<td>B</td>
<td>1.5</td>
<td>29</td>
<td><em>R1:</em> (Dr) sat down with us and drew all the diagrams about the X chromosome and how it’s affected and what happens to the X chromosome and how it affects growth bits that affect it and [Dr] said that if I wanted to see chromosome experts at (hospital) [Dr]’d arrange for that for us and anything we wanted to do</td>
</tr>
<tr>
<td>B</td>
<td>1.5</td>
<td>34</td>
<td><em>R1:</em> We did but there seemed to be so much because it seems like it affects growth it could affect this it could affect that it all depends on the individuals</td>
</tr>
<tr>
<td></td>
<td></td>
<td>36</td>
<td><em>R2:</em> Mind blowin’ wasn’t it?</td>
</tr>
<tr>
<td>B</td>
<td>1.5</td>
<td>272</td>
<td><em>R2:</em> I remember finding it well it was quite technical to me and I don’t like anything technical, I found that daunting you looked at probably a lot more didn’t you?</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td><em>R1:</em> Well it was very hit and miss I feel because it depends on various degrees</td>
</tr>
<tr>
<td>B</td>
<td>1.5</td>
<td>279</td>
<td><em>R2:</em> All that flawed me at the beginning</td>
</tr>
<tr>
<td>B</td>
<td>1.5</td>
<td>299</td>
<td><em>R2:</em> When as the years have gone on we’ve had a book from Dr (endocrinologist) didn’t we from the early days with the little pen that lights up, its too complicated, [Dr] gave me that from when she was about seven. And I’ve just opened it probably again last year maybe and its still too complicated for her.</td>
</tr>
</tbody>
</table>

Verbatim text was inserted where the participants’ meaning was clearly representative of the category, particularly insightful or was considered unique. For example in the above text I did not want to lose the power of phrases such as “mind blowing”, “hit and miss” or “all that flawed me at the beginning”.

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Explanations as to why data had been assigned to each category enabled subtleties of similarity, difference and connectedness to emerge into what appeared to be cross-cutting themes. These themes were:

- Individualisation
- Complexity
- Gender
- Uncertainty
- Normalisation
- Limited recognition of long-term health risk

These emergent themes prompted further charting. Theme charts were useful in making explicit the characteristics of these early themes and also enabled the views of fathers, mothers, and girls to be separated. Girls’ views could be looked at in terms of age and/or stage of development, e.g. pre-pubertal and post-pubertal. Thereby, the use of tables and charts illustrated a range of responses from different participants. The data was beginning to take shape and would ultimately form a response to the research questions.

Supervision meetings at this time contributed to the development of rationales and relationships between emergent themes. The relationship between the initial concepts and emergent cross-cutting themes was complex.

For example the category ‘Sources of information’ had collated data associated with receiving and seeking information; all issues relating to the acquisition of information. This involved the process of parents getting information; from seeking out information themselves and receiving information from others. The categories sharing information and understanding TS involved parents assimilating information, making sense of what TS meant for their daughter and deciding what to tell others, who to tell and how to share the information. Identity, growing up and social relationships had coded data illustrating how parents used information. This included, for example how they shared it with others to benefit their daughters, e.g. information for teachers; whether parents decided to reveal or
conceal information; how they communicated information with their daughters and taught them about different aspects of the condition; meeting key developmental tasks; how they valued the on-going support of medical staff in helping them make decisions. The cross-cutting themes had some relevance for each of these categories but to a greater or lesser degree. They were not of equal importance to each category. Therefore it seemed more useful to consider them in terms of how they influenced parental responses to the domains of the information process. That is, at this stage, it was more useful to view them as modifiers.

Consequently three activities relating to information exchange became apparent:

- Getting and Giving
- Processing and Managing
- Using and Sharing

The next stage was to clarify the context of the themes within each of these domains, explain their characteristics and consequences.

### 3.6.3 Explanatory analysis

#### 3.6.3.1 Clarifying context of themes across cases

Connections between these cross-cutting themes were defined and discussed at length with the supervisory team. The cross-cutting themes had some relevance to a greater or lesser degree for each category but they were not of equal importance in each category. The properties of each theme were examined further and characteristics defined and refined, developing explanations that described the relationships between them. This resulted in a change and amalgamation of some themes. For example, gender had been raised in three different contexts, firstly with reference to the gender of medical staff and the impact of this on the consultation and examination experience of girls. Secondly, gender differences between some mothers and fathers with regard to their need for information and thirdly, there appeared to be a gender difference in the emphasis parents placed on fertility issues
for their daughter. Further comparison of each of these dimensions of gender within each case and across the themes revealed overlap of these issues. These different dimensions were subsumed into individualising, normalising and uncertainty. Limited recognition of long term health risk as a theme did not stand alone either. Returning to the transcripts for context of this theme illustrated it was a more complex issue than might appear at a surface level. The meanings across the participants differed and were better illustrated in themes of normalising and uncertainty.

Patterns of activity were also recognised in these data. The category sources of information had collated data associated with all issues relating to the acquisition of information. This involved the process of parents getting information; from seeking out information themselves and/or receiving information from others. The second activity was evident in categories sharing information and knowledge and understanding TS, involving parents assimilating information, making sense of what TS meant for their daughter and deciding what to tell others, who to tell and how to share the information. The third activity found within the categories of identity, growing up and social relationships, had coded data illustrating how parents used information, for example, providing information for teachers; revealing or concealing information; how they communicated information with their daughters and taught them about different aspects of the condition; how they valued the ongoing support of medical staff in helping them make decisions. In summary, three broad activities in relation to information exchange were identified: getting and giving information, processing and managing information and using and sharing information. Further refinement came within the supervisory team. Lengthy discussion seeking supportive evidence by mapping examples across the cases, clarified the activities which were concluded to be:

- Gathering and receiving information
- Making sense of information
- Using and sharing information
3.6.4.2 Answering the research questions

The second mode of inquiry within symbolic interactionism is inspection, a procedure described as “flexible, imaginative, creative and un routinised” (Stryker 1981, p10). This stage of the analysis determines how the data had addressed the initial research questions. The methods drawn upon in this research design provided a framework in which this creativity was structured and systemised, guiding the cyclical process of data examination, reduction and explanation. Ritchie & Spencer (2002) claim this is the most difficult part of data analysis to explain because it appears mechanical but in reality making “conceptualisations and connections” depends upon “leaps of intuition and imagination” (p321). Constant comparison facilitated on-going interpretation of the emerging themes facilitating the development of meaning out of the experiences of the participants. Corbin & Strauss (1990) believe creativity of the researcher depends on their analytical ability and sensitivity to the actions and interactions described in data (p19).

Supervision meetings at this time contributed to the development of rationales and relationships between emergent themes. Verification of the themes was supported through the process of explaining to others. It was necessary to test conclusions drawn about the patterns identified in these data and test the explanations developed. The supervisory team prompted the looking for negative evidence and sought rival explanations. This iterative process continued and re-structuring through further abstraction by examination of the relationships of the early themes led to the emergence of three major themes:

- Uncertainty
- Normalising
- Identity
3.7 Summary and conclusion

The design for this study has been described to demonstrate the decision-making involved and systematic approach adopted in carrying out this research. Symbolic interactionism maintained my focus throughout the research process on the context of information sharing, which guided an approach that focused on ‘exploration’ and ‘inspection’ of all elements of information need from the perspective of the participants. In-depth semi-structured interviews were appropriate to the philosophical framework and encouraged the participants to share their perspective and experience of TS. The next three chapters illustrate in detail the findings from these data. How major themes were identified is demonstrated through the deconstruction of these data. The findings are presented using the three information activities which were identified by the participants:

- Gathering and receiving information
- Making sense of information
- Using and sharing information

Chapter Seven provides detailed discussion of the major themes by synthesising the findings in Chapters Four to Six.
CHAPTER FOUR: FINDINGS: GATHERING AND RECEIVING INFORMATION

4.0 Organisation of Findings Chapters

The research aims were:

- To identify what girls aged 8–16 years old know and understand about TS
- To explore their experiences of living with TS
- To discover what parents and girls with TS perceive their information needs to be
- To investigate sources of information and support, and their usefulness

There are three Findings Chapters which present data to elucidate the perceived information needs described in this study by girls with TS and their parents. Chapter Four presents data relevant to sources from which information is gathered and received; Chapter Five addresses how families describe processing and making sense of information, and finally Chapter Six reports data about how information is used and shared, between parents and girls with TS, with family and friends and across health, education and social settings.

4.1 Introduction

Data illustrating how girls with TS and their parents received and gathered information that contributed to their knowledge and understanding of TS are presented. The interviews explored from where information was gathered and how useful and relevant it was perceived to be. This chapter initially focuses on sources of information identified by parents and girls with TS. The subsequent section presents findings related to the usefulness and relevance of information sources to the participants at different points in time, i.e. from the initial diagnostic period to confirmation of diagnosis, referral into specialist endocrinology services and on-going care management. The overall aims of this chapter are:
• to identify all sources of information that are considered useful to parents and their daughters in developing an understanding of TS;
• to present the sources of information specifically accessed during the initial diagnostic period including the initial consultation with an endocrinologist;
• to examine the different types of information that meets their individual needs.

4.2 Medical and nursing sources of information

Medical and nursing sources of information cited by the girls are presented below. The vertical axis is the number of girls identifying the source.

**Figure 6: Sources of information identified by girls: medical and nursing**

The two most commonly cited sources of medical information across all girls and their parents were paediatric endocrinologists and PENS. All the girls made some reference to the doctors at clinic and were familiar with the consultant endocrinologist identified by gender, by name or referred to as “my doctor”. Parents stated they also received information from other doctors as listed in Table 17. With the exception of the specialist registrar (SpR) in endocrinology who was seen in the absence of the consultant
endocrinologist, these other doctors had been involved in the process of getting to a diagnosis of TS. The ‘types’ of medical staff giving information about TS to parents in this period varied across the pre and post-oestrogen groups (Table 17).

Table 17: Medical staff involved in the diagnostic period

<table>
<thead>
<tr>
<th>Medical Sources</th>
<th>Pre-oestrogen group</th>
<th>Post-oestrogen group</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>n=9</td>
<td>n=6</td>
</tr>
<tr>
<td>Obstetrician</td>
<td>1</td>
<td>0</td>
</tr>
<tr>
<td>Paediatric Registrar (NNU)</td>
<td>1</td>
<td>0</td>
</tr>
<tr>
<td>Paediatric Registrar (Local Hospital)</td>
<td>1</td>
<td>0</td>
</tr>
<tr>
<td>Community paediatrician</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>General Practitioner (GP)</td>
<td>0</td>
<td>1</td>
</tr>
<tr>
<td>Consultant paediatric physician</td>
<td>5</td>
<td>4</td>
</tr>
<tr>
<td>Consultant paediatric endocrinologist</td>
<td>9</td>
<td>6</td>
</tr>
<tr>
<td>Specialist Registrar (Endocrinology)</td>
<td>2</td>
<td>1</td>
</tr>
<tr>
<td>Geneticist</td>
<td>9</td>
<td>6</td>
</tr>
</tbody>
</table>

There are some differences between the pre and post-oestrogen groups. The age at diagnosis determined which professionals were involved in the initial investigation and diagnosis, e.g. within the pre-oestrogen group parents in Case N were told by an obstetrician their unborn baby had TS; for two families who alerted health professionals that something was wrong soon after the birth of their daughter paediatric registrars provided initial information. Within the post-oestrogen group two girls had heart defects detected at birth had been followed up within local paediatric services. Their general
growth and development was being monitored in relation to this diagnosis and only subsequently were they referred to specialist services. In other cases, doctors working within primary care provided initial information, for example, a general practitioner (GP) in Case I and in Case M the community paediatrician investigated the girl’s short stature following referral by a school nurse. All girls were referred to a paediatric endocrinologist for confirmation of diagnosis, treatment and management.

All families were referred by the endocrinologist to a geneticist for further explanation of their daughter’s specific karyotype. Therefore all parents cited this professional as a source of information whereas only one girl in the post-oestrogen group referred to the geneticist. This girl had been diagnosed 13 years 8 months and had seen a geneticist with her parents at this time.

Not all parents saw the geneticist around the time of diagnosis and offered various reasons for this. In Case C the girl had been diagnosed at 2 years of age and parents stated “we turned it down because I thought I didn’t feel I needed it at that time when the doctor said it was Turner’s, but it [the karyotype] was quite unusual”. As their daughter grew up they became increasingly confused receiving inconsistent information from clinicians outside endocrinology services who were involved in their daughter’s care “they’re all saying slightly different things” about her unusual karyotype. At 7 years the parents attended an appointment with a geneticist for clarification. Other parents saw the geneticist without their daughter who they considered too young at the time but reported they had been told their daughter could have a consultation when she was older. The perceived usefulness of the information provided by the geneticist is explored later in this chapter.

All girls interviewed in this study were being treated with GH. PENS taught all aspects of the administration of GH and all families had contact with them for this purpose. Personal qualities and approachability were identified as an important part of this role by parents and some girls. The senior nurse in the tertiary growth clinic was also identified by one mother
as providing practical information about how the family could store the GH during a family holiday.

4.3 Non-medical Sources of Information

The tables below illustrate non-medical sources of information identified by girls (Figure 7) and parents (Figure 8). Each of these sources is now examined.

Figure 7: Sources of information cited by girls: non-medical

Figure 8: Sources of information cited by parents: non-medical sources
4.3.1 Parents: a source of information

In fourteen families the parents were married; in one family the parents did not live together but actively shared care of their children and were interviewed together. Fourteen girls identified their mothers as providing them with most information about TS. Fathers were always linked to mothers with reference to information giving therefore all girls who included their father as a source of information stated “Mum and Dad”. In the pre-oestrogen group seven children included both parents as main providers of information, and two specifically identified their mother only (Case G,H). In the post-oestrogen group four girls identified both parents as providers of information, and two cited mothers.

Two explanations were offered where fathers were not a cited source of information. The first explanation was that the father had suffered severe health problems of his own therefore the mother was the most accessible person for information (Case E). The second related to specific information about puberty. In Case F the mother reported the father was uncomfortable in clarifying information following his daughter’s sex education sessions at school or after discussions about pubertal development in clinic (Case F). One father (Case B) commented that now his daughter had reached puberty his wife was naturally the one with whom his daughter would discuss these issues. Although a small sample it appears that there may be a developmental shift that influences the type of information that is shared between parents and their daughters.

4.3.2 Turner Syndrome Support Society (TSSS)

The TSSS was accessed by twelve families, eight from the pre-oestrogen group and four from the post-oestrogen group. TSSS is a national charity caring for the needs of those with TS. It provides a range of resources for parents, girls, teachers and health professionals, they organise an annual conference involving professionals and families and a residential weekend for mothers and daughters. Personal support via telephone helpline is provided by its founder member (Arlene Smythe), the mother of a girl with TS now in her early twenties. Five mothers and three couples referred to her directly as a personal point of
contact and support. Three girls in the pre-oestrogen group also referred to her by name and stated they could ask her for information. These girls had met Arlene because their parents were active members of TSSS. Two of the families who had attended a conference had also attended the mothers and daughters weekends. TSSS resources accessed by girls and their parents are identified below.

**Figure 9: Utilisation of TSSS resources by girls and their parents**

Information from this source was described by parents as having several functions (Table 18). The three girls who had attended the conference and/or mother and daughter weekends stated they had enjoyed being with other girls with TS. Specific examples of this are provided in Chapter Five and Six.
Table 18: Functions of information gathered from TSSS

<table>
<thead>
<tr>
<th>Function</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Provision of factual information about TS</td>
<td></td>
</tr>
<tr>
<td>Emotional support through talking to others about their daughter’s experience of TS</td>
<td></td>
</tr>
<tr>
<td>Sharing information to help them make sense of TS</td>
<td></td>
</tr>
<tr>
<td>Social opportunities for their daughters to mix with other girls with TS</td>
<td></td>
</tr>
<tr>
<td>Practical help for parenting a daughter with TS</td>
<td></td>
</tr>
</tbody>
</table>

Written material produced by TSSS most frequently used by parents included:

- an information leaflet providing guidance for teachers
- a newsletter ‘ASPECT’ which is published quarterly provided day-to-day advice about living with TS as well as medical, research and education issues
- a comprehensive book entitled ‘TS Lifelong Guidance and Support’ which claims to provide “everything you need to know about TS”.

Six of the fifteen girls mentioned the book and two of these had also read the newsletter. In twelve cases parents had accessed the TSSS website for contact details and information about TS however none of the girls cited the website as a source of information. Three fathers and one mother stated that the TSSS was not as established ten years ago when they were trying to access information as it is today emphasising the retrospective nature of some of these data with parents reflecting on their experiences relating to initial diagnosis and management of their daughter’s condition. Three families (Case E, G, I) had not accessed TSSS although they were aware of it. In one case (E) the mother stated her intention to contact them. All families who had used TSSS were white caucasian; the two asian families interviewed had not accessed it.
4.3.3 Lay sources of information

There was some reference to lay information from others with experience of the condition and also of parents offering their experience to others. Two families had contacted local support groups (Case B and N). Case B had met other parents and arranged to meet with one family outside the group meeting. Cases N and P had also offered themselves through the TSSS to be a resource for other families with a girl with TS. Another family (Case D) had also been told of a girl in their daughters’ school who also had TS and they had met up. Another example of sharing lay information came from an incidental meeting at ‘Sure Start’. The mother (Case L) met a lady who had recently adopted two children. The lady explained that she was unable to have her own children because she had TS. They had discussed this and arranged that the daughter could contact her in the future.

The outcome of the encounters for Case B, N and P were not reported as positive. Case B had not found the meeting constructive and ceased to attend the group meeting and did not contact the other family again. Case N had invited a mother and her daughter to their home to share their experiences however the visiting family were negative toward the diagnosis which was counter to the attitude of the host family. Case P also experienced negativity toward the diagnosis. The girl in Case D comments that she had nothing in common with the other girl with TS and that she was not like her

She doesn’t wear make-up, she’s quiet whereas I’m the opposite, I’m confident (DG: L382)

The individual nature, interpretation and perception of TS described in these data are examined further in Chapters Five and Six.

4.3.4 The Internet as a source of information

The internet was reported as providing both medical and non-medical information. The girl diagnosed at age 13 years 8 months was the only one to state she had used the internet to find out about TS and this was with her mother prior to seeing the paediatric
endocrinologist. Parents in five of the six families in the post-oestrogen group had used the internet before they saw the endocrinologist. Once the term TS had been mentioned they searched the term. At this stage parents in the post-oestrogen group used general search engines such as Google, reporting an immense amount of information being found. This contrasted with the parents in the pre-oestrogen group, where two mothers and one father stated they had used the internet prior to confirmation of the diagnosis by the endocrinologist. However, five used it subsequently. Following an appointment with the paediatric endocrinologist four families (three pre and one post-oestrogen group) reported being given recommended sites to look at by their consultant.

The lack of use of the internet by the girls to seek out information was an unexpected finding because all the girls stated they enjoyed spending time on their computers. They mentioned regularly searching for information to support school work, accessing social networking sites such as MSN or to play internet games.

4.4 Information Trajectory

Identifying sources of information within the cases also provided data relating to an information trajectory that parents and their daughters followed. Each family had their individual experience of acquiring information although they also shared similar processes of finding out information, the beginnings of which involved getting to a diagnosis. As already identified there are differences between parent experiences in the two groups (pre and post-oestrogen) in getting to a diagnosis. The main aspects that differed included the range of professionals involved in diagnosis and the triggers for referral to specialist services.

4.4.1 Pre-oestrogen group: getting to a diagnosis

This section focuses on information involved in getting to a diagnosis. Appendices 6&7 present a summary of the concerns that triggered referral and the route to accessing specialist paediatric endocrinology services. Comments made by parents relating to
interactions with health professionals leading to confirmation of diagnosis are also included. Similarities and differences across these cases are subsequently presented.

4.4.2 Recognising early signs of TS

A key clinical feature of TS is lymphoedema of hands and feet in the newborn. In four cases in the pre-oestrogen groups parents had raised this as a concern and in one case it was noticed by a paediatric registrar. In Case J there had been no indication of any problem during the pregnancy apart from passing comments made by a GP who the father recalls was an “ex NHS consultant, obstetrician whatever and she said ‘baby feels a bit small, the baby feels a bit small’ but the scans were fine” (JP/R1:L21–22). This infant was transferred to special care baby unit within 24 hours of delivery where a paediatric registrar noticed her swollen hands and feet, and that she was short. Diagnosis was made within a week. In four of the cases where the infant presented with lymphoedema the diagnosis was made within the first days and weeks of life therefore information about TS was received by parents very early in their daughter’s lives.

The significance of lymphoedema was not always recognised. Midwives in the hospital did not recognise this characteristic as important and neither did one of the paediatricians. Within primary care a GP also did not consider this clinical feature important. The impact of missing this clinical sign and delaying the diagnosis was significant for these parents (Case C), and in particular for the mother, who reflects that she continued to raise concerns about her daughter’s swollen feet with her GP for six months,

… [G] said to me I was a paranoid mother, I had a fat child and had a chubby daughter ‘cos it was her legs that were swollen and like my son, he was three then and he’d been quite chubby at a few months old but not the way her feet looked, swollen I knew it wasn’t and because [Dr] said I was paranoid I think I just wanted to prove [Dr] wrong. I don’t think [Dr] should have said I’m paranoid (CP/R1: L175–179)

However, a community midwife who had visited in the first ten days following discharge home acknowledged that it was unusual and also commented upon the infant’s small toe
nails. Although not understanding the significance of these features she acknowledged it was unusual and encouraged the mother to seek a second opinion if the swelling persisted.

4.4.3 Specialised circumstances

Two families received the diagnosis before or soon after birth in specialised circumstances. In Case G the girl was born prematurely at 26 weeks gestation with ambiguous genitalia, and was confirmed as a girl with TS. Case N was diagnosed in-utero at 24 weeks gestation by an obstetrician following amniocentesis for anomalies identified on the routine 19 week scan,

[Dr] said well either your baby’s got Down’s syndrome or Turner syndrome but that’s unlikely or your baby has just got an extra growth of skin around its neck and it could just grow into that as its growing (NP/R2:L19–21)

The parent’s response was to seek out information and prepare for a diagnosis of Down’s syndrome by contacting support groups. They had decided that whatever the diagnosis they would continue with the pregnancy. The mother described their sense of losing control of decision making when in receipt of such serious information

I was so shocked and you were so shocked that, end up getting carried along in this stream of, people make decisions for you and you agree with it. If that happened now I wouldn’t agree to that [amniocentesis] (NP/R2:L23–25)

When receiving the results they again experienced an inability to control information they were receiving. The first piece of information shared with the parents was “it’s not fatal”. Receipt of this information clouded decision making and consensus about the next piece of information being revealed especially for the mother

I couldn’t speak because at no point had anybody said it could be, it hadn’t entered our heads and then from that statement [Dr] went on to say would you like to know the sex of your baby. Well I couldn’t get over the first statement, I couldn’t speak and I remember you saying [father] yeah OK and I’m thinking in my head I don’t know if I do, I don’t know if I do but I couldn’t speak and then [Dr] said ‘well you’re having a little girl and she has Turner syndrome, and we were just made up (NP/R2:L34–38)
This notion of fatality linked to TS was reported by other parents where diagnosis was made close to birth. When the diagnosis was made in older girls there was some anxiety associated with cardiac assessment but the girls were perceived as generally healthy with a height problem. In Case G the concerns about fatality were related to prematurity and not TS.

Following the initial diagnosis by the obstetrician the parents were referred to a paediatrician who expanded on what TS was, however the parents reported much of this information was incomplete or inaccurate for example,

we don’t know what she’ll look like but she may have a twisted foot, all twisted round we don’t know but I’ve (paediatrician) seen pictures of that in an encyclopaedia, this is what [Dr] said and she may not have a womb erm we just don’t know (NP/R2:L47–49)

These were the only parents interviewed who had an in-utero diagnosis, they comment positively that having all the information before she was born enabled them to be thoroughly prepared. They contacted TSSS and attended TSSS annual conference, “we wanted to see a variety of girls and see what they looked like”. Early information they explained meant that every observation or decision they made from then on was framed within TS. TS as part of the girls’ identity is further examined in Chapter Five.

4.4.4. Recognising later signs of TS

Three cases in the pre-oestrogen group were diagnosed later because of short stature, a cardinal clinical feature of TS. Two diagnoses were direct responses to parental concern (Case L and H) about their daughter’s growth, one girl a toddler and the second girl at aged 8 years. In the third case (Case M) the School nurse referred the child to the community paediatrician because of concern about her height however this mother had raised concerns several times prior to this official confirmation reinforcing her own concerns.
In the four cases referred from primary care settings to paediatric services in secondary care the parents report associated clinical features that had not been identified for example, poor feeding, recurrent ear infections, poor growth. This emphasises the subtle nature of the clinical features which individually may be explained as common childhood illness and only when put together lead to diagnosis of TS.

These parents also report having other children enabled them to see that there was something wrong with their daughter at birth, or in her subsequent growth and development that may have otherwise gone unnoticed. In the post-oestrogen group comparisons with siblings had reassured parents there was nothing wrong.

### 4.4.5 Parental response to diagnosis

Emotional response to the diagnosis varied across families reflecting their personal circumstances. Their response was also influenced positively or negatively by the approach taken by the health professional providing them with information. Where families had raised concerns and provided information about their daughter to health professionals that had not been promptly responded to there was a certain sense of relief. They were right and there was something to be concerned about (Case C,H,M), “I wasn’t imagining things”. This feeling however was also mixed with sadness about the diagnosis (Case H,L); “devastation” (Case M), “they said yes it is and I started crying” (Case P); and shock “we were pretty shell-shocked” (Case K,L,J). Case G was complex and at the point of diagnosis of TS their focus was on their daughter’s survival from complications of prematurity.

### 4.4.6 Post-oestrogen group: getting to a diagnosis

The age range at which diagnosis was made in this group is 6 years to 13 years and 8 months. Within this group the referral to specialist services had been triggered in all cases by professional concern related to height. In only one case was the professional a non-health professional. In this case it was a games teacher at school who was concerned about the girl’s short stature and poor coordination in comparison to her peers.
4.4.7 Response to diagnosis

In the post-oestrogen group diagnosis was unexpected for four of the six families, who describe feeling shocked because they had not thought anything was particularly wrong with their daughter. For two cases (E,I) there had been some parental anxiety about the daughter’s growth. Mother in Case E had long standing concerns about her daughter’s height but had been reassured by the GP on several occasions that there was no problem and she was just small. In Case I the parents were anxious about their daughter’s height but had been reassured by family and friends that she was just petite however they made an appointment to see the GP who had referred them to the paediatrician.

In four of the cases (A,B,D,F) parents had noticed their daughters were small but had justified it in different ways because other aspects of development had not caused any concern. Rationale for short stature included being a small family, something to do with her diet, or anticipation of a late growth spurt. In this group comparison with siblings had offered some explanation for example, being a twin, or expectation of being petite in comparison with an older brother.

The main emotional response reported by parents to the diagnosis was shock. Although this was also an emotion identified in parents in the pre-oestrogen group there was the added dimension for these parents and girls of knowing their daughter and themselves as a ‘non-TS’ girl for several years. Once the diagnosis was made parents became familiar with information about features, characteristics and behaviour associated with the syndrome. How these individual families made sense of the diagnosis incorporating this new aspect of their daughter’s identity is examined in the next chapter.

Frustration was another emotional response expressed in Case A and mother in Case E. Some characteristics of the condition that were referred to at diagnosis had been questioned earlier in their daughter’s lives by the mothers themselves, however, at that time they were
not considered as significant by doctors they had consulted. The next section of the chapter examines data related to the perceived usefulness of information sources.

4.5 Initial diagnostic phase: perceived usefulness of information

This section focuses on the period of time from initial diagnosis to consultation with the paediatric endocrinologist. To present the similarities and differences in the perceived usefulness of each identified source of information across each case, this section follows the sequence in which parents and their daughters’ obtained information, how they utilised different sources and how they gathered further information to enhance their understanding.

Table 19 illustrates the information trajectory described by parents when gathering and receiving information, the sources of information, the responses of parents relating to gathering information. Data supporting this came from cases identified.
Table 19: Information trajectory

<table>
<thead>
<tr>
<th>Sources</th>
<th>Pre-diagnosis</th>
<th>Initial diagnosis</th>
<th>Confirming diagnosis</th>
<th>Clarifying individual profile</th>
<th>On-going management</th>
</tr>
</thead>
<tbody>
<tr>
<td>GP</td>
<td>Parents</td>
<td>Paediatrician</td>
<td>Paediatric Endocrinologist</td>
<td>Paediatric Endocrinologist</td>
<td>Paediatric Endocrinologist</td>
</tr>
<tr>
<td>Parents</td>
<td>School nurse</td>
<td>Community</td>
<td></td>
<td>Geneticist</td>
<td>Pediatrict Endocrine Nurse Specialist</td>
</tr>
<tr>
<td>Teacher</td>
<td>Midwife</td>
<td>paediatrician</td>
<td></td>
<td>TSSS</td>
<td>Specialist Registrar in Endocrinology</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Obstetrician</td>
<td></td>
<td></td>
<td>Parents</td>
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<td>TSSS</td>
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<td>TSSS</td>
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<table>
<thead>
<tr>
<th>Response</th>
<th>Unsystematic gathering of information</th>
<th>Systematic gathering of information</th>
<th>Focused information gathering</th>
</tr>
</thead>
</table>


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4.5.1 Information provided by paediatricians to parents

In the pre-oestrogen group eight families were given verbal information by paediatricians in a secondary care setting and one family within a primary care setting by a community paediatrician. Paediatricians gave various explanations of TS. The reported detail and accuracy of information from these professionals varied and this influenced the actual and perceived relevance of the information to parents. The personal qualities of the professional were commented upon when information imparted was limited or uncertain. In Case C parents felt the paediatrician had not handled things very well, they were advised not to tell any family or friends which contrasted with how the parents were feeling, “I didn’t feel it was something to be ashamed of”. In Case N the parents concurred “a lovely [Dr] and very caring but really didn’t have the information” (NP/R2:L40).

All families in the post-oestrogen group were seen by a consultant paediatrician in their local hospital and at this point TS was mentioned for the first time. The paediatrician was identified as the first specialist source of information for parents. The key information recalled was the abnormality of an X chromosome; their daughter would be short and she would not be able to have children. How this information was received varied and some parents reported misunderstandings and omissions in the information. Three families thought shortness meant dwarfism and this perception only changed at their first consultation with the paediatric endocrinologist. Written information was valued by parents, as was time was spent by endocrinologists in explaining what TS meant. This father reflected:

[Dr]sat down with us and drew all the diagrams about the X chromosome and how it’s affected and what happens to the X chromosome and how it affects growth bits that affect it (BP/R2:L29–33)

Not all parents received this considered approach. One mother states that at the end of their initial consultation with a paediatrician TS was mentioned to the medical student who was present and then as she and her daughter were leaving the doctor commented
“I don’t think Ellie has got it but we’ll test her anyway” (EP: L32)

All families were subsequently referred to a consultant paediatric endocrinologist at a tertiary paediatric endocrine centre.

4.5.1.1 Unsystematic gathering of information

Once the term TS had been mentioned the majority of the parents reported a period of unsystematic information seeking. In the case referred to above, having been given the term TS the mother and her daughter (Case E) accessed the internet for information on their return home. However, more generally the initial experience of hearing their daughter had TS did not appear to influence whether or not the internet was used to find out more information. All families had a computer at home and access to the internet. Mothers in Cases H and L commented that the internet provided easy access to information and afforded them privacy in accessing information.

Parents in both groups who did use the internet reported using general search engines such as Google; no one was aware of recommended sites in this initial phase of seeking information. Within the pre-oestrogen group a father comments

[You] get an explanation from a doctor who perhaps didn’t fully understand what it could be and then you sort of go and look on the internet (KP/R2:L64)

The desire to seek out information as soon as the term TS was used was also experienced by parents in Case P however their source was an old 1950’s medical text book. The mother explained

there was this list, absolutely worse case scenario, mental retardation, stops growth, web neck, fingers swelling, aorta wonky (PP/R1:L23–24)

In the pre-oestrogen group fewer families had sought information independently at this stage than the post-oestrogen group. Four of the six sets of parents (Cases A, D, E, and F) in the post-oestrogen group used the internet to research TS prior to their appointment with the consultant paediatric endocrinologist at the tertiary centre. However, one family did not
seek any further information before their consultation with the endocrinologist. There were similar responses across the two groups to the information accessed. They found this source of information as “mind blowing” (BP/R2:L37), “horrifying really cause you get a picture of potentially sort of terrible problems and deformities” (Case F).

The unsystematic gathering of information for these families was instinctive and only partly useful depending upon what they found. Several had experienced a degree of distress. At the first consultation with the endocrinologist several families learned that the complexity of TS meant that much of what they had found on the internet or in the dated literature did not apply to their daughter and was therefore of limited value.

For some parents the internet was used immediately after their first appointment (Case F). All these families shared the same experience of finding a vast amount of complex information available on the internet.

One mother commented,

    got onto the internet and it just mind blows you really, (FP:L34)

She reiterates,

    It was just mind boggling (FP: L39)

A father explained,

    [We] started looking round at the websites the technical stuff was totally out of me depth as we read all these studies (AP/R1:L24).

Another mother reflected,

    I remember finding it well it was quite technical to me and I don’t like anything technical, I found that daunting (DP/R1: L389)
And her husband concluded,

Well it was very hit and miss I feel because it depends on various degrees of, and I don’t think the research is there yet to say exactly what is, whether this is going to affect your, your growth or affect some other part of you (DP/R2: L390–92)

With complexity came questions of whether information was relevant to their daughter and a note of caution,

But the thing is, what is on the computer is quite over the top as well for us by looking at that it could be quite worrying for a few parents (DP/R1:397)

For these families information was independently and unsystematically sought once they had heard the term TS. However, one family did not seek information at this point. They had the initial diagnosis from a paediatrician in their local hospital, and did not access further information prior to the consultation at the specialist centre. They waited for their appointment with the paediatric endocrinologist, reporting that they preferred to have the information provided by the specialist. The father stated at the first meeting with the endocrinologist “the doctor printed out some pamphlets on TS” (IP/R1:L78), he continued “we were really happy when we, we had it to read and that alone” (IP/R1:L84). This was the only family in the post-oestrogen group where English was not their first language. They stated that having pamphlets in English was not a problem for them but may be for others,

Information available in their language whether it’s Gujarati, whether it’s Hindi, whatever it is I think it would be a good thing but for us it wasn’t serious at all” (IP/R1:L606–607)

The father also made an important point that within the family there are potentially different language abilities, so that although the younger generations may understand English other family members do not. This potentially leaves parents to translate complex and sensitive information.
4.5.2 Sifting information: relevance by karyotype

Variation in karyotype of the girls meant terms such as mosaic or classic were reported as being used by paediatricians when explaining the diagnosis to families. However, this information was prior to the parents’ consultation with the geneticist and therefore information and/or understanding was limited. The usefulness of the information was assessed against ‘types’ of TS and the ‘type’ of TS that parents perceived their daughter to have. Parental perception of the type of TS their daughter had was shaped by their interpretation of this information. Data relating to this are presented in Chapter Five. Mother in Case E stated that information sourced on the internet provided details about girls with “full TS” and because her daughter has a mosaic form many of the aspects of the condition described on the websites did not apply to her. Two other parents also referred to the degree to which their daughters’ were affected by TS for example, this father explains

   er what do they call it I’m trying to think of the er…the term where it’s not completely missing mosaic, there’s different percentages though isn’t there?

He continues,

   [TS] could affect this or that it depends on the individual (BP/R2:L35-37),

A mother adds,

   there are so many different types of Turners (FP:L37).

4.5.3 Summary of information gathered and received in the initial diagnostic period

The initial diagnostic phase is dominated by parents’ experience of information receiving and gathering because the girls in this study were not included in the initial information gathering that took place around the diagnostic period. This was partly due to their young age at diagnosis or the unexpected nature of their diagnosis. There was one exception, the girl diagnosed at 13 years 8 months who became aware of the suspected diagnosis at the first consultation with a paediatrician. She explored TS with her mother on the internet. She described the internet as “sometimes helpful” but was unable to identify any websites that
she had found particularly useful and was unsure about reliability of sites she and her mother had visited. They had not looked at the TSSS website although her mother had the address and stated her intention to do so. Table 20 summarises the sources of information and their usefulness in this initial diagnostic period.

Parents realised that information they had gathered was general and encompassed all aspects of the condition. Information they had received from paediatricians varied with some parents receiving specific information relating to their daughter and others receiving more general information which created the potential for misunderstanding.
<table>
<thead>
<tr>
<th>Source of information</th>
<th>What was useful</th>
<th>What was not useful</th>
</tr>
</thead>
</table>
| **Paediatrician**     | - Unhurried, detailed explanation supported with diagrams  
                         - Warm approach “he was lovely, really didn’t have the information”  
                         - Recognition of clinical features of TS | - Unsubstantiated comments  
                         - Limited knowledge  
                         - Not listening to parents concerns |
| **Leaflet**           | - Provided an explanation of what TS is | - Brief “skims over it all” (FP:L506) |
| **Book**             | - A starting point for information | - Out dated material  
                         - Includes everything and not all of information is relevant |
| **Internet**         | - General overview of the condition  
                         - Can access from home, private | - General search - too much information  
                         - Complexity  
                         - Unsure of relevance  
                         - Unsure of reliability |
| **TSSS**             | - Emotional Support from Arlene  
                         - Pictures on the website so parents could see what girls looked like | - |
4.6 Confirming the diagnosis: perceived usefulness of specialist information

The next stage of the trajectory for these families was their first appointment in specialist endocrinology services. The primary source of information for parents was identified in all cases as the paediatric endocrinologist. All parents knew their daughter had TS at this first meeting. The girl with a late diagnosis also knew (Case E). The initial meeting with the paediatric endocrinologist provided information about the different features and characteristics involved in TS and the lifelong nature of the condition.

4.6.1. Quantity and complexity of information

Parents expressed some difficulty in receiving a wealth of detailed, complex information in the diagnostic period. There is a tension here for the clinician between ensuring families are fully informed yet not overwhelming them with information. Parents in Case P comment

[Drs] live in that world don’t they…it’s a bit embarrassing to keep saying I don’t know what you’re saying to me (PP/R2:74–75)

As they became more experienced as parents of a girl with TS, they also became more selective and systematic in gathering information which they then needed to manage and apply to their individual circumstances. With all long-term conditions there is an element of monitoring and surveillance alongside management and treatment decisions. For these families the process of gathering and receiving information was an ongoing process that involved varying degrees of activity by parents and their daughters. A developing source of information for parents was the girl with TS herself. Parents gathered information through observation of their daughters’ development, behaviour and response to treatment. Parents also received information directly from conversations with their daughters which indicated to them the girl’s level of understanding about the condition, its management and treatment.

All the girls in this study were treated with GH. The main source of information relating to the administration of GH for all families was the PENS. They described the nurses’ visit positively. They were all taught at home and felt able to carry out the procedure before the
nurse left. Data exploring the administration of GH and specific teaching of this skill is explored in Chapter Five as parents and their daughters start to make sense of GH as part of the treatment of their condition.

4.6.2 Information received from the endocrinologist

Paediatric endocrinologists were cited by all parents as the main source of useful information as they describe their information needs moving from developing a general understanding of TS to developing a specific understanding of what TS meant to their daughter.

4.6.2.1 Parents’ perspective

Data already presented indicates the management of TS required a multidisciplinary approach and consultant endocrinologists were identified by parents for clinical expertise, knowledge of the condition, orchestration and interpretation of the outcomes of multiple investigations. They were perceived by parents and medical colleagues as the expert in the clinical management of TS. One mother commented that the paediatrician at her local hospital had said,

it was above his field and he would hold us back, so he got us some information and then he referred us to [specialist centre] (FP:L22)

They had expertise for determining individual problems one father states,

TS, fine that is one thing, now the second complication was Coeliac…. We are very fortunate that the doctors, they did a wonderful job, all credit to them (IP/R1:L55)

In Case P the paediatrician told them the endocrinologist was the one who could tell them why it happened, and what may or may not be wrong.

Finding the parameters of their individual TS profile involved undergoing a significant number of investigations, including physical examination to assess visible stigmata; assessment of cardiovascular system; renal system; growth; hearing; ovaries and uterus.
Donaldson et al. (2006) states 50% of girls with TS have cardiac abnormalities. Two of the girls in this sample had known heart defects which were diagnosed at birth however for the children with no known abnormality these tests were particularly anxiety provoking. This was similar for all parents irrespective of girl’s age at diagnosis,

once we’d got all the tests back about her heart and we knew it wasn’t a life threatening syndrome we just kind of got it in perspective (FP:L93–94)

[Dr] was brilliant but it was just all the tests which we had to do, X Rays, and blood tests and everything like this and different things,[Dr] gave us a lot of information (BP/R2:L40)

each one (tests) was like a mini Everest, you go and have the kidneys done and there was the heart wasn’t there and then something else and each one you think we’ve passed that one and having got a relatively clean bill of health from that you sort of, it’s a massive wow it’s not so bad we can cope with this and you know it’ll be alright (KP/R2:L75–80)

like getting a trophy(PP/R2:L98)

Investigations to determine the parameters of the condition for each girl were organised and undertaken. The results of these were the start of individualising the condition.

Gathering information from the results of these investigations enabled paediatric endocrinologists to determine the features that were part of the individual TS profile of each girl. Information provided to parents was now of direct relevance to their daughter and was the beginning of understanding what TS meant for her. As illustrated in the above quotation there was relief, “a massive wow” when the results were satisfactory.

4.6.2.2 Girls’ perspective

The girls attended the specialist clinic every four to six months. All the girls in this sample knew they had TS and cited parents as their main source of information. Paediatric endocrinologists and other doctors in the specialist clinic were also cited as providing information. Parents shared with their daughters’ written information they had received from the doctors. Its usefulness to the girls varied.
The initial meeting with the paediatric endocrinologist was remembered by two of the girls interviewed. One who was diagnosed at 9 years old recalled that the doctors provided her information but not in detail. She stated

[Dr] explained about chromosomes and how I’d got one missing erm I don’t think they went into that much detail about it ‘cos they didn’t want to frighten me at the time (DG: L90)

When recalling her response to the information she received at this first meeting with the endocrinologist she stated

I can always remember going home and Mum and Dad said we could have a DVD and they were like a bit shocked and I was like it’s alright, Mum, Mum stop being so - its fine but it wasn’t until I was older I realised maybe it was a bit more serious a bit more well not serious but I dunno (DG:L90–92)

This highlights the developmental nature of providing information to children with ongoing conditions.

The second girl had a late diagnosis aged 13 years 8 months. She appeared reluctant to share her experience of this first meeting apart from confirming she knew what was going on and there was a decision made to commence GH which she wanted to have.

4.6.2.3 Recognising TS is an uncommon condition

TS affects 1:2000 live female births (TSSS 2008) and as such is a relatively uncommon condition. However, there was some dissatisfaction with the level of knowledge of doctors expressed by some parents as they developed their own knowledge and understanding of TS. Information gathered, received and interpreted by endocrinologists enabled them to put together all the features and behaviours associated with TS in the context of their daughter’s presenting features. For example, within the post-oestrogen group as parents developed their understanding there was a sense of dissatisfaction that doctors had not put the information together to make an earlier diagnosis,
She had hearing problems, she had low hair line, she had a high roof palate, she was sick all the time, she wouldn’t sleep, it was like her toe nails were like classic Turners, her finger nails were really narrow, every single, it’s just like everything now we know was connected to TS but nobody would listen (FP:L73–75).

R1: we didn’t realise at the time but when she was born she was very poorly and she was all bloated and reading back on the symptoms now, puffy eyes she was a real she wasn’t when she first was born, was she, but then she sort of exploded like a big balloon. So they were testing for kidneys whatever they didn’t twig at that stage though but looking back now when we received the information that was one of the signs that can indicate that there’s a problem. (AP/R2:L37)

… doctor was absolutely appalled she’d got to 13 without being diagnosed because she was only small birth weight baby, full term, normal size mother, she should’ve been tested at birth (EP:L37)

This dissatisfaction is aimed at services outside specialist endocrine services emphasising the expertise of the paediatric endocrinologist as illustrated here

We feel that [Local Hospital], is not being adequate, its second rate to [specialist centre] they just don’t know enough about it they are not specialised enough (DG:L60)

Only two mothers in the pre-oestrogen group shared this dissatisfaction because generally the diagnosis had been made early in this group of girls and where parents had raised concerns there had been a timely response by the health professionals involved. In Case H the diagnosis was not made until their daughter was eight years however the parents in Case H felt that as soon as they verbalised concern about their daughter’s height action was taken whereas in Case C and M the mothers report not being listened to. Through the mother’s persistence in raising concern about her daughter’s swollen feet in Case C and in response to a school nurse (in Case M) who alerted medical staff to the girl’s short stature was action taken. The mothers’ confidence and competence in assessing their child had been undermined by medical staff. It was also stated that some health visitors had little or no knowledge of what TS was and parents had passed information they had gathered to them.
Parents also viewed endocrinologists as an excellent source of information not only about the condition but also in providing guidance on accessing additional sources such as websites as discussed earlier or support groups such as TSSS. One family reported that their consultant endocrinologist had given them information and also guided them to additional material for further reading

[Dr] emailed me websites to go to and look at so we had that to hand, ‘go and have a look at this’, ‘read up a bit more on this information’ (BP/R2:L44-5)

Booklets and leaflets received from the paediatric endocrinologist were perceived as generally useful. The following quotation suggest for these parents it was essential to understanding the condition and how they could use that information to help their daughter,

the patient literature which we first got was a Godsend now we’ve taken that on-board and used it to understand how we need to approach Amy and help her really (AP/R1:L476)

However, one set of parents were given the TSSS book entitled ‘TS Lifelong Guidance and Support’ and the mother commented “I think you still need something in layman’s terms” (FP:L35). This book is aimed at parents and is produced by the support group however her comments may reflect the comprehensive nature of the information in this book, which requires parents to filter out the relevant aspects to their daughter. This issue of relevance was identified by the eldest girl interviewed who did not find the more general information booklets and leaflets helpful indeed the information had alarmed her. She explained,

I didn’t find them useful at all ‘cos they’re talking about really, really serious stuff and well about things that don’t concern me really like about heart problems and kidney problems where I’m not like that so it kind of scared me a bit (DG:L338–40)

There were conflicting views from two families about a book published by a pharmaceutical company that was given to them by the endocrinologist. It was an interactive text that checked understanding. One family reported that the book was “too complicated, he gave us that she was only seven…. It’s still too complicated for her” (BP/R1:L299–300). Their daughter was now 12 years old. However, another family had
found it useful, their daughter was 10 years old. The ten year old enjoyed the activity of using the pen which would light either red or green in response to a wrong or correct answer however it was not clear what understanding had taken place. This suggests it would be useful to assess the appropriateness of resources given to girls taking into account their age, abilities, readiness and preferences for learning.

4.6.3 Information received from the geneticist

One of the first professionals to whom parents were referred by endocrinologists was the geneticist. The general pattern was that parents had initially received some information about chromosome analysis from paediatricians, then in more detail from paediatric endocrinologists and finally specific focused information from the geneticist. These data illustrated that many parents were unsure about their understanding of this topic area. For example, “Said that er the mosaic and the X and Y yeah they said that right at the beginning”. One mother stated she could not remember but had a record of it in case it was needed (Case H). Some parents were clearer about the chromosome analysis “she’s XO, not mosaic, she’s completely without an X, she hasn’t got a broken X” (Case P) however the implications of the analysis were less understood as seen in these two quotations

mosaic seems to have the worse problems … so we were happy she didn’t have worse case scenario (PP/R2:L94)

and yet other parents believed

she fits into more of a mosaic … maybe a misunderstanding from ours but give us a bit of false hope that maybe she would be able to have children (DP/R:L42–3)

The complexity of the genetic make-up of girls with TS leads to an array of differing presentations. This information provided detailed karyotype for the individual girl and explained some of the documented TS traits. However, faced with multiple investigations the usefulness of the information from the geneticist was considered limited by some families. One mother (Case P) stated “chromosomes took a back seat ‘cos that’s not going to change but what can they do about the rest”. However, once all the investigations had
been completed she concluded that knowing about the chromosomes still had limited value because TS means so many different things,

    even now if people ask what’s wrong we go through symptoms that describe her not ‘TS’ chromosomes they don’t mean anything (PP/R1:L140–41)

This is supported by other parents, who concur,

    we know she’s missing whole or part of an X and that’s as far as it goes…it isn’t going to change anything (KP/R2:L711)

Usefulness for these parents seems to be linked to whether it is something that they can influence or change. As there is no action for them to take that can influence this aspect of the condition then the information was perceived not to be as important as information that could be acted upon particularly in the early investigative stages. A different view came from the family with the prenatal diagnosis. This couple commented that because they had spent time researching TS and discussing the condition at length with Arlene (founder of TSSS) much of what they were told by the geneticist they already knew

    We’d spent a lot of time with Arlene on the phone and at various meetings erm the level of support he gave was what we’d already had (NP/R2:L998–990).

However, there had also been personal benefit for each parent in meeting with the geneticist. The mother had blamed herself for the condition and the geneticist reassured her that she was not to blame. The father explained “he said it was my fault [laughing] he did say that, I was like OK”. The father expressed the information about the “whole gene thing” meant that he could understand the cognitive and behavioural aspects of TS

    [Dr] came and did the whole gene thing explained it really well and that made a difference to me to really understand it and made a real impression I could see why certain things were difficult [for daughter] (NP/R1:L1000–1002)

Two girls had received information and were aware of their chromosome analysis at this stage.
4.6.4 Information from the TSSS

In addition to gathering and receiving information from medical staff parents also sought information to help them with parenting a daughter with TS. The most commonly cited source for this purpose was the TSSS. For twelve families TSSS was a valuable source of practical information about living with TS such as how to deal with feeding problems, behavioural issues, difficulties at school.

For some families this sense of belonging to a community of girls with TS was highly valued and families involved developed strong supportive networks. Further data relating to understanding TS and using information from TSSS to benefit their daughter is presented in Chapters Five and Six. However, some families did not want their daughter to be defined by her diagnosis of TS and therefore expressed that joining TSSS was not something they had thought would be beneficial. One family felt it was not culturally appropriate for them to join. This family (Case I) described themselves as from the ‘Asian community’ and were keen for information about other children with TS with their shared cultural background:

Sort of er Turner’s Syndrome Society or yes my son is a Turners Syndrome your daughter is a Turners Syndrome fantastic the two of them could be friends y’know, in our society it doesn’t happen like but I hope it could I would like to be amongst those parents who would probably arrange to have a meeting with other Turner syndrome kids....With the same cultural background that’s what I’m trying to say (IP/R1:L482–87).

TSSS provides information about day-to-day living with TS. However, they confirm that the majority of the membership is white British families. This potentially limits its usefulness in the provision of psychosocial support to families who do not share the same background or day-to-day experiences. Their daughter concurred with her parents, that she would like to meet other girls with TS from the same background as herself. The family had decided to approach the PENS, “we hope she will know of other families of our background” (IP/R1:L489). The two Asian families interviewed in this study were not members of TSSS. There is reference in the above quotation to “my son is a Turners syndrome” which suggests misunderstanding of the genetic basis of the condition.
One girl provided insight into her experience of postal delivery of information from TSSS stating that this had caused her particular upset. She felt her diagnosis had been revealed outside the family. Her mother had requested leaflets from the TSSS and the postmark had TS on it, she stated,

I don’t think they should do that in a way, well I mean it kind of gives it away, I mean the postman’ll obviously see it” (DG:L333)

Parents had commented that the use of the internet and written material provided some privacy because they could access that information in their home however for this girl there is a sense that her right to privacy is threatened by the needs her parents have for finding out about the condition.

4.6.5 Summary of information sources following referral to specialist services

Table 21 summarises the sources of information described in the diagnostic period following referral into specialist services, what was and was not useful.

**Table 21: Usefulness of sources identified by parents following referral to specialist services**

<table>
<thead>
<tr>
<th>Source</th>
<th>What was useful</th>
<th>What was not useful</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Paediatric endocrinologist</strong></td>
<td>Expert knowledge and clinical expertise</td>
<td>Provided reading material inappropriate to age</td>
</tr>
<tr>
<td></td>
<td>Explanations of all the investigations and findings</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Able to explain relevance of information to individual girl</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Able to recommend different sources of information: TSSS, leaflets, websites</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Positive personal qualities</td>
<td></td>
</tr>
<tr>
<td>Source</td>
<td>What was useful</td>
<td>What was not useful</td>
</tr>
<tr>
<td>------------------------------</td>
<td>--------------------------------------------------------------------------------</td>
<td>------------------------------------------</td>
</tr>
<tr>
<td><strong>Geneticist</strong></td>
<td>Some understanding of the chromosome anomaly</td>
<td>Too complex</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Timing</td>
</tr>
<tr>
<td><strong>PENS</strong></td>
<td>Personal qualities: friendly, patient</td>
<td><em>Nothing stated by participants</em></td>
</tr>
<tr>
<td></td>
<td>Able to explain all aspects of storage and administration of GH</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Took time to teach girls and parents – did not feel rushed</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Can call them if you have a problem with GH</td>
<td></td>
</tr>
<tr>
<td><strong>Written Information</strong></td>
<td>Able to read it in private</td>
<td>Not age-appropriate</td>
</tr>
<tr>
<td>(received in clinic)</td>
<td>Can revisit information</td>
<td>Too complex</td>
</tr>
<tr>
<td><strong>Internet</strong></td>
<td>Recommended websites</td>
<td>Some studies were too complicated</td>
</tr>
<tr>
<td></td>
<td>TSSSS</td>
<td></td>
</tr>
<tr>
<td><strong>TSSS</strong></td>
<td>Someone to talk to who has had similar experiences</td>
<td>Culturally specific</td>
</tr>
<tr>
<td>(personal contact)</td>
<td>Knowledgeable</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Friendly</td>
<td></td>
</tr>
<tr>
<td></td>
<td>“describes” daughter</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Can always ‘phone back and as often as needed</td>
<td></td>
</tr>
</tbody>
</table>
4.7 On-going management: useful sources of information

Data suggested the most prolonged period requiring information for parents and their daughters is the on-going management and treatment of this lifelong condition. During this time parents develop their expertise and become more knowledgeable at dealing with the condition.

Continuity with the endocrinologist was considered key to some families,

> I mean continuity of care it’s very important to parents very, very important (JP/R1:L431)

On-going management was the period where parents made sense of the individual nature of the condition and data supporting this is presented in Chapter Five. Sources already identified as useful continued to be so however there were changes in their involvement.

Table 22: Summary of main sources accessed to assist on-going management and parenting of girls

<table>
<thead>
<tr>
<th>Source</th>
<th>What was useful</th>
<th>What was not useful</th>
</tr>
</thead>
<tbody>
<tr>
<td>Internet</td>
<td>Recommended sites</td>
<td>Being exposed to negative stories (Mother Case F)</td>
</tr>
<tr>
<td>Written material</td>
<td>Reading about the experiences of other girls</td>
<td>Reading general information that did not apply to individual girl’s condition/ features</td>
</tr>
<tr>
<td>(leaflets, DVD or book from hospital or TSSS)</td>
<td>Good information about GH</td>
<td>Not age-appropriate</td>
</tr>
<tr>
<td>Consultant Endocrinologist</td>
<td>Making complex material accessible and understandable</td>
<td>Not seeing the same doctor at clinic</td>
</tr>
<tr>
<td>Source</td>
<td>What was useful</td>
<td>What was not useful</td>
</tr>
<tr>
<td>----------------</td>
<td>--------------------------------------------------------------------------------</td>
<td>-----------------------------------------------------------------------------------</td>
</tr>
<tr>
<td>Parents</td>
<td>Can ask parents anything</td>
<td>Potential for embarrassment for fathers and daughters re puberty discussion and/or examination (Cases B, F, I)</td>
</tr>
<tr>
<td></td>
<td>Can explain so their daughter understands</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Available to repeat information</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Acquire and provide written material that parents consider age and developmentally appropriate</td>
<td></td>
</tr>
<tr>
<td>PENS</td>
<td>Friendly and approachable</td>
<td>Nothing stated by participants</td>
</tr>
<tr>
<td></td>
<td>Supportive when problem arises with GH administration</td>
<td></td>
</tr>
<tr>
<td>Nurse in clinic</td>
<td>General information relating to storage of GH</td>
<td>Nothing stated by participants</td>
</tr>
<tr>
<td></td>
<td>Can telephone and they are able to pass you on to doctor</td>
<td></td>
</tr>
<tr>
<td>TSSS</td>
<td>Practical advice: information to help with issues in each stage of growing up, e.g. feeding, school, puberty, infertility</td>
<td>Comprehensive nature of the information</td>
</tr>
<tr>
<td></td>
<td>Comprehensive booklet, “TS lifelong guidance and support”</td>
<td></td>
</tr>
</tbody>
</table>
4.8 Summary and Conclusion

This chapter has focused on sources from which parents and girls receive and gather information about TS. Three broad categories of information needs for parents have been identified from these data,

- **Physical**: what the condition is; defining physical parameters for individual girls; management and treatment of physical features; co morbidity and predicted outcomes for future health

- **Psychosocial**: coping and adapting to diagnosis; strategies to manage behavioural aspects; cognitive abilities and school achievement; social relationships; personality and TS traits; implications of infertility for parents and daughters; hopes for the future.

- **Practical**: living with a diagnosis of TS; solutions to common difficulties, e.g. advice on feeding; buying shoes and clothes that take account of lymphoedema and small stature; advice for school teachers; egg donation.

These findings have identified sources of information to meet the cognitive, emotional and social needs of parents across an information trajectory from getting to a diagnosis to ongoing management. The initial diagnostic period and exposure to information varied depending upon the presentation of the girls. The earlier the diagnosis and quicker the family accessed specialist services the less time available for parents to be given or seek out misleading and/or inaccurate information. For some parents seeking out information from readily available sources such as the internet or books was something they could actively do however because of the complexity of the condition much of the information gathered proved to be anxiety provoking.

Parents valued a warm and honest approach from paediatricians prior to referral into specialist services although on occasions these professionals did not have detailed
information that parents required. The individual nature and complexity of the condition means the most valued source of information for all parents particularly in the diagnostic period was reported as the paediatric endocrinologist. It is through this source they acquired the factual information they needed to understand individual parameters and implications of their daughter’s TS. However, there was a tension for the endocrinologist between potentially overwhelming parents with information and ensuring they were fully informed. Parents report TSSS publications and personal contact with Arlene (founder of TSSS) through the helpline was valuable and a reliable source of information. TSSS had a role in clarifying biomedical information in addition to providing practical guidance on parenting issues and insight into potential social aspects of TS.

When girls are diagnosed in infancy or early childhood the gathering and receiving of information for parents begins earlier than for their daughters. For parents the starting point of the information trajectory begins with the first time they become aware of TS as a possible diagnosis. For their daughters the starting point varied, largely dependent upon when and what information was shared by the adults involved in their treatment and management and most importantly by their parents. The girl who was diagnosed as an adolescent gathered and received information at a similar time to her mother.

Gathering and receiving information is not a linear activity but an on-going process. These data identify that triggers for further seeking of information for these families included change connected to physiological and developmental need, and social and educational issues.

The next chapter presents findings associated with how parents and their daughters make sense of this wealth of information.
CHAPTER FIVE: MAKING SENSE OF INFORMATION

5.0 Introduction

This chapter focuses on how information, gathered and received, by girls and their parents was assimilated and processed by them. The individual nature of a diagnosis of TS means that information has to be interpreted and understood by girls and their parents in order to identify its relevance to their particular circumstance. Data presentation in this chapter shares the experiences of girls and their parents in making sense of complex and diverse information, examining the similarities and differences across cases illustrates how evolving knowledge and understanding enabled them to manage ‘their’ TS.

The chapter begins by considering factors identified by the girls and their parents that facilitated understanding of TS. This is then followed by data relating to how they made sense of TS on a day-to-day basis. Findings about making sense of information in relation to the girl’s identity as a girl with TS are then presented. The chapter concludes with data providing insight into how parents and their daughters have made sense of TS in thinking about the future.

5.1 Making sense of TS: factors identified in developing understanding

Not all girls with TS face the same issues as there is a broad range of existing and potential health problems including associated psychosocial difficulties. Prioritising information to meet the needs of parents and the unique needs of individual girls in a timely and age-appropriate manner requires expertise from a range of health professionals. The literature review (Subsection 2.5.1) identified a multidisciplinary approach to the management of TS under the direction of a paediatric endocrinologist is advocated by health professionals and support groups (Saenger et al. 2001; Donaldson et al. 2006). Long-term monitoring of the health status of girls with TS is an integral part of their lives requiring regular contact with specialist endocrine services (TSSS 2008). The most valued professional relationship identified by parents and girls in this study, was that with doctors working in the specialist
The paediatric endocrinologist was referred to most frequently. Expert knowledge and positive personal characteristics were identified as important aspects of this potentially (depending on age at diagnosis) long-term relationship.

5.1.1 Paediatric endocrinologist: the clinical expert

Parents reported initially being reliant upon doctors in the specialist clinic to mediate information they gathered and/or received from various sources such as the internet or TSSS. The paediatric endocrinologist was cited by all parents as mediating complexity to a greater or lesser degree by explaining and clarifying aspects on an individual basis thereby helping to make sense of the features and characteristics in the context of each girl’s specific circumstances.

Common characteristics for all girls with TS are short stature and infertility however these also have to be interpreted and managed on an individual basis. The individual response to GH treatment cannot be predetermined and treatment options for infertility will vary. Expert knowledge was cited by parents as essential in ensuring correct application of complex information to the specific and individual on-going management of their daughter’s condition. Eleven families out of fifteen of the cases expressed a preference for receiving information directly from the endocrinologist. For some this appeared to be linked to a lack of knowledge demonstrated by other doctors who had provided limited information or misinformation. In the following quotation parents express how the endocrinologist relieved their anxiety following confirmation of the diagnosis. They express their anxiety was not only about what may be wrong but also of the process of acquiring appropriate expertise and accurate information. Their anxiety is heightened because the condition is uncommon and other doctors are reported as not knowing what it was and were unable to tell parents anything about it,

The whole thing was a dreadful thing really because we just weren’t told what was going on you were just left to your own devices to sort it out, and because nobody at the hospital, we didn’t see an endocrinologist at the time, nobody knew what it was or could tell us anything (KP/R2:L57–59)
Ten days after the initial diagnosis from the paediatrician these parents met the paediatric endocrinologist

R1: But [Dr] was fantastic really.

R2: [Dr] was the one who said it’s not that bad er she didn’t have er…

R1: There wasn’t any physical deformities.

R2: No, but they can have the whole range of physical things, the wide neck, the low set ears, y’know and she didn’t have a lot of the things she could have initially had and she’s obviously not going to develop those and [Dr] said we’ll get the kidneys scanned, we’ll get her heart scanned, and it was only through seeing [Dr] when she was ten days old we felt that somebody who actually knew something. (KP:L63–71)

The endocrinologist explained the physical and visible features of TS to them and identified which their daughter had and which ones she did not have. General information about the condition was personalised. Having determined the visible features which were ‘fixed’ and would not change over time, the endocrinologist started the process of screening for unseen problems potentially involved as part of this multisystem condition. These parents were reassured that they were now accessing “somebody who actually knew something”.

However, even when accessing the endocrinologist and explanations had been given one family remained unclear about the implications for their daughter,

Well no she hasn’t that’s what I don’t understand, they tell you webbed neck, puffy eyes, finger nails, and horse shoe kidneys and that, she’s got nothing like that and so I don’t understand that. (LP:L367–68)

This mother expressed uncertainty in making sense of the diagnosis considering the lack of visible and physical TS features expressed in her daughter.

The perception that only paediatric endocrinologists knew enough about the condition through expert knowledge and clinical experience was evident in data from the eleven families referred to earlier and is illustrated in the following quotations
..it’s about having someone that’s got knowledge that’s built up over a considerable period of time, got the T shirt etc (KP/R1:L662)

you feel sometimes you feel when, when you see the registrars, you have to understand it’s a teaching hospital and everything, but sometimes you think well we’re going a long way and we’re getting the B team and certainly when it gets involved either in stopping medication or increasing medication you want to see the top man…(JP/R1:L455-458)

When I see the registrar I always feel they’re trying to justify what they are telling me (HP:L432)

The second quotation above, refers to the long-term nature of the condition and the desire to develop a relationship with the consultant, “we’re going a long way”.

In four cases (F, G, M and P) preference for the consultant endocrinologist was not expressed. They were satisfied with information provided by other doctors in clinic. Case P reports they had seen the endocrinologist in the diagnostic period but had since had consultations with other doctors, they stated,

we saw [Dr] when she was a baby but we’ve been filtered out to [Dr]’s staff (PP/R2:L654).

In Case M the family had consistently seen the same endocrinologist in the early years however following their retirement there had been less continuity. Nevertheless this family stated they valued consultations with all medical staff over the years. However, the doctor who the mother had recently seen was specifically praised for their ability in information giving

[Dr] was actually brilliant, [Dr] was a lot more informative with regard to the oestrogen it was a great consultation, I felt a lot happier when I came out not that I wasn’t happy with the others, I want to make that clear, they’ve all been brilliant but I felt more settled, understanding (MP:L348)

There are educational, behavioural and social dimensions to TS. Some endocrinologists provided information relating to these aspects
[endocrinologist] said to me that Turners’ children were not clued up they struggle more if say a boy was making a pass at her then she wouldn’t understand that a pass was being made at her (HP:L386)

However, this mother did not believe this was relevant to her daughter who she described as “very clued in there”, highlighting the individual nature of the condition and how parents make sense of what is relevant to their daughter and what is not as she grows and develops.

Practical support for education and behavioural issues were also dealt with by some endocrinologists by writing letters to support assessment by educational psychologists or to secure additional time for examinations at school. Case N valued an approach that recognised and helped them to manage psychological and emotional needs of their daughter as well as her physical needs,

[Dr]’s brilliant [Dr] always asks, really supportive of all aspects of Sarah’s care, mental health and physical health which for us is what we need.

Endocrinologists have a pivotal role accessing other professionals

[Dr]’s referred us to see a clinical psychologist who will hopefully give Sarah some tools to help her (NP/R2: L116–118)

Parents’ expectations of the medical consultation varied across the pre and post-oestrogen groups from those preferring to see their consultant to those who did not express a preference of personnel but did express a preference for the way in which information was delivered to them.

5.1.2 Personal qualities and attitudes of doctors

The above quotation identified another component of the relationship between families and endocrinologists, which related to personal qualities of this senior clinician. Across 12 of the cases reference to liking the endocrinologist appeared to be important in this relationship for example,
When I first met [Dr] I remember I came out and I said to (husband) I’m so glad [Dr]’s so nice. That was very important you know we’re going to be spending a long long time and if you haven’t got that um feeling, feeling of trust I suppose then it would be very difficult, if you didn’t like that person or you didn’t have confidence in them. (H:L449)

Other parents commented “[Dr] was very very nice” (EP:L80); “we saw [Dr] was very nice, very kind” (I: L508). Nurturing and reassuring qualities were also demonstrated for example, “put her under [Dr] little wing” (D: L39).

Parents in Case P compared how different attitudes of doctors affected the experience of the family during their clinic appointment including the perceived quality of information given to them and their daughter,

…well I felt we were kind of inflicting ourselves on [Dr]’s time, not that [Dr] ever, but it was like this is how much she’s grown, how much weight she’s put on, are you alright, any problems and then you were out (PP/R1: L644–647)

The focus for the consultant appears to be on monitoring the condition, lacking personal interaction with parents and their daughter. However, they had a different experience when they saw another doctor who took a different approach and personalised the consultation

R2: I like [Dr] an all gives us a bit more information doesn’t [Dr], explains things very well… this [Dr]’s attitude’s a lot better …. bedside manner I s’pose

R1: It’s ‘cos [Dr] talks to Phoebe isn’t it… more like Phoebe how are you…. takes the emphasis off us and onto Phoebe (PP/R2:L670)

Parents stated that consultant endocrinologists were key individuals in helping families assimilate complex information. This father however adds that information needs to be at the appropriate level for individuals

I’m not very good at remembering what doctors tell you cause you come out there and you think what was that about? But [Dr]’s very good I like [Dr].comes to your level and so there are things that, you might be able to remember it. (AP/R2:L375)

In summary for the majority of families a consultation with the consultant was seen as desirable because their status represented their knowledge and clinical expertise. However
for other families the consultation was about the accessibility of information which was more dependent upon attitudes and teaching skills of medical staff. For these families all doctors working within the specialist clinic were perceived as having expertise.

5.1.3 Trust and familiarity

Trust and familiarity was promoted by continuity, i.e. regularly seeing the same consultant in clinic. The following quotation illustrated that continuity was increasingly important over time for these parents

Int. Is it confidence do you think?
R1: It’s trust, and that’s been built up over well nearly eleven years ten and a half eleven years now
R2: And I think experience as well cause [Dr]
R1: [Dr] is good, Jodie trusts [Dr] and likes him, same with [names another endocrinologist] (JP:L458-62)

This quotation emphasised the long-term nature of the condition and long-term relationship with the endocrinologist.

Some families travelled a significant distance to the tertiary clinic, and the four to six month interval between consultations heightened their expectation of seeing ‘their’ consultant,

..of course the best situation would be to have the same doctor because then he knows exactly or she knows exactly what’s happening (IP:L 509)
R2: To me not being under one person to be sort of, I mean we’re under one person now because we said look were not very happy about this we want to be under one person…
R1: Especially when we are coming such a long way it means a day off work for [husband], a day off work for me (DP:L36–38)
we said that we wanted to see [Dr] so we went in and had an appointment with [Dr] and saw [Dr] and said we don’t want to be fobbed off with a different registrar every time not that we’re saying in in isolation but there’s no consistency ..(KP/R2:L537)

The frustration of not having an appointment with ‘their’ endocrinologist appeared to be related to parents’ confidence in the knowledge and expertise of doctors they saw in clinic.

every time we used to go down we would see someone different and they didn’t, actually we were getting mixed reports(DP/R1:L39)

A preference for familiarity is identified as a characteristic within TS population (TSSS 2008). This was commented upon in relation to seeing the same doctor by families with girls across the age range. The first quotation is the experience of parents with a ten year old,

Int: Do you get to see the same Doctor each time
R1: Yes usually
Int: And is that helpful
R2: Good for Sarah ‘cos she stresses out over any change
R1: Over anything
R2: Yeah, anything she hasn’t experienced before (NP:L110–115)

And for this fourteen year old her parents believed inconsistency compounded their daughter’s difficulty in developing new relationships and therefore limited her contribution to the consultation

R1: I do get the impression that Anna likes [endocrinologist] trusts [Dr] er she’s less receptive to one or two of the others.

R2: Well she does know, she knows we’ve been seeing a lot of different ones every time we’ve gone this month different you see and I know it’s difficult for her I think cause she’s, she’s got to then start up another relationship hasn’t she? (AP:L384)

And in addition to their clinical knowledge this mother trusted the endocrinologist to know what was appropriate to discuss,
I certainly would not like to be told or have Hannah [daughter] told anything other than by Dr [endocrinologist], because you know, well I presume [Dr] will see us at all the crucial stages (HP/L437)

‘Crucial stages’ for this mother referred to key decision making points involving new information being given to her daughter particularly relating to infertility. This trust between the mother and the endocrinologist enabled her to have a degree of confidence and control over information discussed during the consultation.

The following quotation illustrates this girl shared her parents’ frustration at not being assured of seeing the consultant

we were jumping from one doctor to the next. Every time we were going down we were having a different doctor and that’s why in the end Mum and Dad arranged for us to see Dr [names consultant] all the time because we were sick of seeing different doctors (DG:L191)

And she stated the importance to her of continuity with one doctor,

[Dr] understands me more and ‘cos you get a bond with them as well,

And on a pragmatic level she adds “they know what’s been going on” (DG:L197).

When explaining from whom she had received information another girl described a warm and reciprocal relationship with the endocrinologist she usually saw. She commented that this is not always the case when she is seen by other medical staff.

R1: Dr [endocrinologist]’s really good ‘cos when [Dr]’s talking to you [Dr]’s talking to me, looking at me, explaining to me. Sometimes other doctors explain to my parents but [Dr] doesn’t [Dr] sits there and explains to me about everything so it’s good really. [Dr]’s like one on one and [Dr] talking to me

Int: So do you feel important in the consultation

R1: Yes (FG/R1:L299)

A father acknowledged that inconsistency was often a result of difficulties in the organisation,
it’s not really a criticism of them as individuals I think it’s just the system. There isn’t enough resource to say right OK here is someone who knows everything about Turners children and they will just see Turners children at every clinic (KP/R2:L611–12)

To circumvent this difficulty one mother tried to arrange private consultations however the consultant she liked preferred to see them within the NHS

if we could see her each time then I would. When we first did all this investigation it was done privately and erm we never saw (endocrinologist) privately. [Dr] said there was no point because everything could be done in the Health Service (HP:L424–425)

Parents and girls described trust and familiarity as valued components in developing relationships with doctors. These aspects of the relationship appear to contribute to parents’ confidence in the medical management of their daughter’s condition.

5.1.4 Aspects of the clinical consultation that assist in making sense of TS

All girls had a clinic appointment every four to six months. Clinic visits provided an opportunity for some parents to observe other girls and make comparisons with their own daughter

I say to Hannah well you just have to count your blessings and we’re not so bad, there are lots of people worse off than we are. You know I suppose in that way going to the hospital is quite grounding (HP:L429)

However, this was a matter of conjecture. Confidentiality prevented discussion of other families with clinic staff. Families usually attended a specific clinic for girls with TS, however, sometimes they would attend non-TS growth clinic and occasionally a girl without TS attended a TS clinic leading to potential misinterpretation relating to diagnosis and reason for clinic attendance. For one of the younger girls in the pre-oestrogen group this had created some confusion because she had seen a child in a wheelchair at a growth clinic and assumed that all girls with TS would have to have a wheelchair at some point. No parents reported speaking to other parents in the waiting room even when they were confident in their assessment that the girl had visible characteristics of TS.
All girls across the age range demonstrated awareness of the process of the clinic visit clearly as summarised in Figure 10.

Mixed responses were obtained from parents and girls as they described their experiences of the consultation. The ability of the doctor to make the level of verbal information accessible to all within the consultation appeared to be an important aspect of making sense of TS for these girls and their parents. Data relating to the quantity of information received during the medical consultation is now presented.
Figure 10: Process of a clinic visit

1. Arrive at clinic and book in
2. Wait
3. Record weight
4. Measure height
5. Wait
6. Consultation
7. Blood tests
8. Make appointment
9. Home or school
5.1.4.1 Quantity of information

As demonstrated in Chapter Four the period around diagnosis involved gathering and receiving a vast amount of information. One father used an analogy that provided insight into the energy expended and initial sense of urgency parents felt in assimilating all the information they were receiving.

Instead of us like running a marathon with information, it was like sprinting all the time and then we got all these answers and we thought right we'll live normally go to clinic every six months, they can monitor it (PP/R2:L709-11)

The issues detailing how to use the information to live normally are revisited in Chapter Six where ‘Sharing and Using’ information is explored.

Consultations provided an opportunity to obtain results of the investigations that started to refine understanding of what TS meant for their daughter. They also provided a chance to seek clarification about information parents had found independently, e.g. from the internet, as these quotations elucidate,

First port of call (internet) because I knew I would get information from it but then it is nice to have the personal touch with someone to clarify everything for you as well. (HP:L472)

or to ask for specific information relating to clinical management

…. we talked about taking oestrogen, or the administration of oestrogen and when it was first mentioned I was totally against it it wasn’t explained that she would still stay on the growth hormone, I thought they would stop the growth hormone and go onto the oestrogen, the last consultation I had was a lot more productive, in respect that it was made clear that she would still stay on the growth hormone and would start to introduce the oestrogen, because I thought she would get so far up and stop it, therefore she wouldn’t get any more growth and as far as I was concerned and her Dad for Maisie we’d get the height for her and then think about the puberty but it’s going to kick in next time (MP:L217)

For some parents the amount of detailed information was difficult to remember. Therefore to manage it they limited what they would focus on. Parents learned to pace themselves
over time as they started to make sense of the implications of living with a daughter with TS

    I feel at the moment what I know now is OK and as time goes by I’ll find out as I need it (GP:L82)

Or limit what information they sought or tried to understand

    Really the medical side of it how much do you need to know (BP/R2:L520)

This echoes the sentiment in the first quotation that the doctors have responsibility for their daughters condition which ends with “they can monitor it”.

The consultation could also be a missed opportunity for information if the approach taken by the medical staff was not interactive or parents felt rushed or distracted. Here a mother expressed feeling responsible for not making the most of the appointment

    It is but like I mean they just, when we go they just talk to us and see Louise and just examine her and then that’s it and by the time we get out we’ve forgot to ask, d’you know what I mean and you can’t really go back, I s’pose it’s our fault (LP:L158)

5.1.4.2 Managing different needs for information during the consultation

Inclusion of all members of the family within the consultation proved to be a challenge for doctors

    R1: [endocrinologist] says ‘have you got any questions to ask me Anna?’ but conversations taken place almost as if she wasn’t there between me particularly and [endocrinologist] so she’s picking up on stuff.

    R2: Well she does, that’s what I like about [endocrinologist] cause if I ask [Dr] something [Dr] will include Anna ‘cos that’s what I tried to do with all the doctors and not all of them do it, if Anna’s not going to answer they look to me and then Anna’s left out you see. But I’m trying to get her to answer for herself ‘cos I mean I don’t know what’s in there not really.
R1: [endocrinologist’s] good cause we ask the question we’d like Anna to ask and Dr [endocrinologist] answers them and [Dr] directs the answer at Anna (AP: L365)

The comment “I don’t know what’s in there” suggested that dialogue between the consultant and their daughter enabled parents to indirectly check out their daughter’s understanding of TS.

Some parents favoured seeing the endocrinologist without their daughter

… sometimes from my point of view I would have appreciated to have that time with the specialist or with my husband on our own rather than with Debbie there because I sometimes think I haven’t really listened and taken it in because I’m too concerned about what her reaction is and maybe I still have that motherly instinct to protect her so I won’t ask the questions that I want to ask because I don’t want to say it in front of her……at the same time I understand the reasoning why they do it (DG/R1: L219)

However, the father preferred the inclusive approach,

I’m quite happy for them to be open in front of her myself, and tell us in front of her, I mean you’re slightly different to me at least it gets it out at the end of the day she knows from that day on rather than two years down the line I’m quite happy for them to sort of mention everything in front of Debbie. (DP/R2:L219)

Their daughter’s response was ambiguous.

Int: You wouldn’t want to see Dr [endocrinologist] by yourself

I would rather be in a room by myself, like I have been more open with this than if my Mum and Dad were here but when we go to clinic I’d rather my Mum and Dad be there ‘cos they need to take in the information as well and if they went separately I’d think they were hiding something from me…..I’d rather know everything I don’t want anyone to hide anything from me. (DG:L428–433)

She was comfortable with her parent’s presence to ensure all information was available to her, not be left with a sense of something being kept from her, however, she also stated that she could be more open if she was on her own. This draws attention to the practical difficulty facing clinicians in meeting the varying information needs of all members of the family at all times.
This is further exemplified in the following quotation which identified differences between parents with regard to wanting different levels and types of information. The first quotation is from a mother explaining her husband’s approach:

Yeah, if there’s something to deal with he’ll deal with it but he doesn’t need to find out all the information out beforehand. I don’t think he, he kind of puts it to the back of his mind ‘cause as long as it’s not not happening but then it happens and then he waits for me to find everything out (FP:L350)

This same approach was described by another father who articulated the difference between himself and his wife:

That’s where people are different because you like to have that pathway whereas I’m more when I need to deal with it I’ll deal with it (BP/R2:L538–39)

The mothers appear to want as much information as possible whereas the fathers in these examples are focused on the most immediate information required for the present.

5.1.4.3 Gender of the doctor

The issue of gender was raised in the first interview undertaken in this study. When discussing her observations of the process of physical examination to monitor pubertal development in her daughter, a mother commented that her daughter was “a bit shy” about this now. When asked if having a female consultant made it easier for her daughter she commented “well she probably does prefer a woman but it is more to do with familiarity”. Data relating to gender of medical staff is limited as I did not explore the issue fully with each family or probe sufficiently, particularly with the younger girls in the pre-oestrogen group. However, the girls who did comment stated preference for a female doctor or a male doctor that they knew.

It was generally mothers who commented on this aspect of care with two fathers referring to the issue of puberty to be something the girls’ mother would ‘naturally’ deal with. During the interviews questions relating to puberty and physical examination by the doctor were met by age-appropriate shyness by girls in the pre-oestrogen group (n=9). They
giggled or were reticent in responding indicating the need for sensitive handling of this information by adults. In this group they were generally accepting of examination by doctors. One commented at preferring a “lady” (Case M: age 12 years); others said they had “got used to it” (Case C: age 10 years) although it was “a bit embarrassing” (Case H: 11 years).

Girls in the post-oestrogen group (n=6) were all currently having their progress through puberty monitored. Comments illustrating the differences and similarities between girls and their mothers in the post-oestrogen group are presented in Table 23. Comments reflect a number of issues that influenced information giving in this situation for these girls. These included concern of mothers about the sensitive nature of the physical examination, the preference for familiarity for girls with TS and the developmental needs of adolescent girls.
### Table 23: Physical examination for pubertal development

<table>
<thead>
<tr>
<th>Case</th>
<th>Parents’ perspective</th>
<th>Girls’ perspective</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>she probably does prefer a woman but I think it’s just that she’s familiar and she’s not very good with people she doesn’t know</td>
<td>No comment made</td>
</tr>
<tr>
<td>B</td>
<td>She used to get very embarrassed doesn’t she ‘cause they still do physical examination that’s one of the other things over the years they’ll have loads of students in, which they will won’t they, but you feel a little bit like a Guinea pig, as she’s got older it’s got more embarrassing as they check for body hair and whatever, growth, and its getting quite embarrassing.(L457) she’s seriously embarrassed over it now, do they really need to check can’t they just ask me? I mean I know, there’s no sign yet, I don’t know I just wonder if they think about the child and the way they feel as much about the medical condition and what I need to know now ‘cause she’s just a funny age this age, twelve.(L465)</td>
<td>In commenting on the physical examination during the consultation she stated “it’s fine” and “you get used to it” and on whether medical students should be present she stated “I don’t care”</td>
</tr>
<tr>
<td>D</td>
<td>she’s not bothered about going to see a male doctor which is good (L:262)</td>
<td>I s’pose I’m more comfortable with a lady but <em>(short pause)</em> I have seen another lady but I’m not too fussy there’s a lot more men. I do prefer a woman but just because of the checks really. Int: You mean the physical examinations when you have to get undressed R: Yeh but so I prefer a woman but if it’s a man well it’s a bit awkward but I wouldn’t say no or anything like that I’d just it’d be alright</td>
</tr>
</tbody>
</table>
|   | Not commented upon and no direct question from interviewer | Int: when you’re with the consultant and doctors, do you have to be physically examined  
R: Sometimes  
Int: How do you feel about that  
R: Not bothered about it really  
Int: No. Is it usually one doctor  
R: Yes just one  
Int: Are there medical students  
R: Occasionally but *(shrugs)*  
Int: You don’t mind?  
R: No |
|---|---|---|
| F | Not commented upon and no direct question from interviewer | Int: Do you like having a lady doctor  
R1: Yeh  
Int: Does it make things easier  
R1: Yeh I’m more comfortable ‘cos I’m a girl myself its so *(doesn’t complete sentence)*  
Int: Does she examine you, do you have a physical examination  
R: Yeh  
Int: How do you feel about that  
R1: OK really ’cos it’s a woman. If it was a man I’d be a bit more thingy but *(doesn’t finish sentence)*  
R2: Sometimes it’s a man though isn’t it  
R1: Yeh, I have to, I haven’t said anything yet but I do feel a bit awkward  
Int: Have you ever said you’d rather have a lady  
R1: Well I think it but I’ve never said anything, I’ve just got on with it |
| I |   | Int: do you prefer seeing a man or a lady  
R: Lady, it’s more comfortable  
Int: Is that when you have to be examined  
R: Yes |
5.1.5 TSSS and others: the lay experts

Thirteen families had contact with TSSS.

In the early years we’d go on the internet or ‘phone Arlene (TSSS) because we didn’t feel we could ask the doctors because they are very clinical and it wasn’t just clinical stuff (PP/R:L282)

This quotation recognises the fundamental components of a diagnosis of TS. The physical aspects can be measured and identified by doctors, however, the psychosocial elements evolve as girls mature and are largely interpreted by parents. Sharing information and experiences with other families who have a girl with TS helped some parents to do this. Some parents refer to Arlene who founded the TSSS as another expert on whom they can rely for emotional support and information that is of practical use in parenting girls with TS at different ages and stages of development and treatment. Figure 12 illustrates the interrelationship between the two ‘experts’ cited by parents and their daughters as the main sources of information. These were the main mediators of complex information that enabled parents and their daughters to make sense of what TS meant to them. Education needs at school are an issue that illustrates the two complementing each other. TSSS guidance for schools was utilised and individual endocrinologists used medical evidence to support the girls’ needs for specific educational support.
5.2 Making sense of TS: Perception of normal

In ten cases, girls and parents articulated they were hesitant in stating that they/their daughter had TS without a qualifying statement as to what it meant in their particular circumstances.

5.2.1 Perspectives of parents and girls: TS vs normal

Parents in Case D whose daughter was diagnosed aged 9 years and at the time of the interview was 16 years old stated that they had always said,

as far as we are concerned Debbie you’re perfectly normal child just like anybody else all your friends and everything. Alright you’re gonna be a bit shorter than them
you’ll have to watch your weight, you may be able to have children you may not, but also out of ten of your friends so many of them won’t be able to have children (DP/R1:L202–204)

There is a comparative element with ‘normal’ girls in this quotation that served to minimise their daughter’s TS features. However, some parents expressed difficulty in determining a TS behavioural characteristic from what is considered normal as illustrated here by one mother

you don’t know what a Turner’s girl is and what a normal girl is because that just could be her (KP/R2:L170)

The father in this case continued

[She’s] a little girl, a normal little girl, she’s intelligent, she’s articulate, she’s good company, she’s got a lovely nature about her so I think sometimes you sort of see ghosts where there aren’t ghosts. (KP/R1:L232)

This is powerful use of language that conveys the dilemma for parents as they seek to separate their daughter’s behaviours and attributes from the features and characteristics of the condition yet the condition is part of her genetic identity.

This ambiguity about ‘being normal’ yet having a diagnosed medical condition that affects many parts of the body commenced at diagnosis for Case P when the paediatrician was reported as saying

does she look any different to a normal baby and we said well no, so, right then stop getting upset, she’s a beautiful healthy little girl really apart from she’s got TS (PP/R1:L34)

you’ve got a healthy baby with a slight problem so what, it’s not the end of the world (PP:L45).

The parents stated they were reassured by this approach. However, by equating physical appearance of the girl with being ‘normal’ or abnormal, healthy or unhealthy the doctor may have risked minimising the complexity and wide-ranging nature of this condition.
5.2.2 Understanding individual parameters of TS

Data provided insight into how parents and their daughters reconciled all aspects of TS. They made sense of the emerging medical profile and notable TS characteristics that together formed the parameters of ‘their’ TS, i.e. what TS meant in their particular circumstances.

5.2.2.1 Perspectives on the term ‘TS’

The term TS was perceived as unhelpful by some as stated by this mother:

I don’t like it, to me she’s so normal, she’s not everything it says and it’s just her height and not being able to have babies really (GP:L321)

As a term to help explain to others it was seen as too vague:

People’ll say what’s wrong with her and we go through symptoms not TS chromosomes, ‘cos I just think that doesn’t mean anything (PP/:L329)

The term TS appeared to have negative connotations for some parents and one girl. The diagnostic label ‘TS’ does not make it clear what is wrong without further clarification because of the wide variation in presentation. This was verbalised in the following statement by one of the girls in the post-oestrogen group:

I think I’d rather just know I’ve got a growth problem and can’t go into puberty myself but TS has got so many different factors and it’s like ticking and crossing the one you’ve got.........So really when you say you’ve got TS you’re saying you’ve got all of them to me (DG:L375)

It appeared TS did not represent who she perceived herself to be.

Parents of the girl with a prenatal diagnosis of classic TS stated that having TS was nothing to be negative about “there’s nothing not to be positive about”. They stated that they wanted their daughter to be,

proud of who she is…. just because she’s been born with a condition doesn’t mean, well she should be proud and think she can talk about it (NP/R2:L5)
Data concerned with motivations and content about what information to reveal or conceal is presented in Chapter Six as parents and girls determine how to share and use information.

Across the cases there were similarities and differences in the interpretation of what a diagnosis of TS meant. There were many comparisons made by girls and their parents between the perception of a normal girl and a girl with TS.

5.2.2.2 Making sense of chromosomes

It would appear that of all aspects of TS, the chromosome analysis was one piece of information viewed as unchangeable. However, although the analysis is clear the information it produced was open to interpretation by parents and had the potential for misunderstanding and misapplication. Understanding was generally scant across all the girls. Seven did not know about them or had never heard of chromosomes. For example, “I’ve heard about it (chromosomes) but I’m not sure” (IG:L185). The girl with the most understanding was 15 years old and stated “like X chromosome’s not divided or something” (EG:L196) clarifying (chromosomes) “some of them were normal and like some weren’t” (EG:L198). Limited understanding included statement such as “there’s a chromosome missing” (DG:L172), “something in your genes is missing” (AG:L95). The girl in Case B was unsure if it was the X or Y chromosome, she states “I think its the Y”.

However, parents in particular used knowledge of chromosomes to try to make sense of the characteristics that their daughters displayed. Parents also used the information to minimise the implications of the diagnosis. The fewer ‘classic’ features identified the ‘less’ Turners she was considered to be. For some, the mosaic diagnosis meant that the girl was not ‘fully Turners’,

We explained she does look slightly different, I suppose but she’s not full blown Turners and she understood that (MP:L180-81)
Parents sometimes had chosen to disengage with the information about chromosomes because they could not change it. They made a judgement about what they should focus their energies on trying to understand as illustrated by this couple,

R2: we know she’s missing whole or part of an X and that’s about as far as it goes really isn’t it?

R1: Yes I suppose yes

R2: We haven’t pursued it any further because it isn’t going to change anything but it it might have given us more understanding and less fear

R1: I think again it might be a bit burying your head in the sand but the more layers you peel off the onion you just there’s just more and more things that you well, we’ve got to a level now where you can manage it, it works there aren’t any big issues

R2: So let’s not create any

R1: Yeah let’s not take the lid off the box sort of thing, leave it where it is erm you know had the symptoms been far worse then we might have been tempted to find out more because we might have needed to do more to deal with them but as we haven’t I think we’ve sort of got to just about the right level of knowledge (KP:L417–26)

Two metaphors highlighted in the text above are about hiding something through covering and one is about gradually revealing. They highlight a tension in taking the decision to limit information between potentially missing something that could have helped them understand TS more and the risk of divulging issues they would rather not know about. Parents in Case P acknowledge this tendency to avoid information but claim that it is important to become knowledgeable about TS “even though it does sometimes come at a cost it’s best to know” (PP/R2:L974).

Two families who had pursued further understanding of the karyotype found the complexity and lack of understanding of a rare chromosome analysis was reflected by some doctors,
Like the surgeon… he wasn’t aware of the diagnosis of Turners so I would say something about Turners and he’d say oh she hasn’t got Turners she’s got this and then maybe I’d see the GP and she’d write another name for it in my red book (CP/R1:L98-100)

This couple also attended a TSSS conference to seek clarity

and we said our baby girls 46XY … she was taken aback, said [they] was a professor and she like couldn’t get her head around that (CP/R2:L102)

The literature review presented papers that suggested some of the specific educational and social difficulties associated with the Turner population have a relationship to their specific karyotype yet parents did not discuss this in any of the interviews. In contrast there was general acceptance of the cognitive and behavioural profile reported in the literature and anxiety around whether their daughter displayed characteristics that may cause social difficulties. Examples of how these were expressed by their daughters were offered,

It’s not just learning its all the other associated factors, how she might struggle with her peer group, if she’s falling behind on her development (BP/R1:L201-2)

Parents and one girl interviewed in this study perceived chromosome analysis as having limited value in explaining what TS meant to them. All these families had the same diagnosis but the girls’ profiles were all very different.

As referred to earlier the complexity of information and the amount of information could be cognitively and emotionally challenging for parents. Parents confirmed they received information around the diagnostic phase some of which was not discussed again or it was forgotten,

R2: There’s other things I s’pose that you’ve got, still got questions she’s got horse shoe kidneys hasn’t she? Does that affect you later in life or so if it doesn’t affect you forget all these there’s so much isn’t there you forget it all.

Int: Its almost like the information’s not really revisited you get a lot at the beginning.

R1: Well that’s right.

R2: Then you forget. And what does that mean again? (BP:L523-28)
Features and characteristics of TS that appeared to be the focus for parents and health professionals were the aspects of TS that need to be considered from a problem solving perspective, i.e. because these needed management and treatment. The next section identifies how girls perceive themselves as girls with TS.

5.2.3 Being a girl with TS

5.2.3.1 Understanding of TS at different ages

When explaining what TS meant to them all girls included problems with growth and the older girls included puberty, some stated this simply, “it affects my growth and puberty and that’s it” (Case B), “I know it makes me small and I haven’t started my periods yet” (Case E); some offered a little more detail “ Well its like er, a gland’s not working for me, I’m still growing but its not providing enough growth hormone for me to grow as much as I should do” (Case I), and one girl referred to hearing as she experienced recurrent ear infections, “it’s when you struggle with your hearing and your growing” (Case N)

Others provided a definition acknowledging its individual nature, key physical aspects but also stressing that it need not restrict future hopes and aspiration,

Int: What does it mean to you? If you had to explain to somebody what would you tell them?

R1: Erm you’re smaller that other people and you don’t develop as well as other girls do and there are things that can help you and eventually you will get to where you want to be and at the end of the day everybody’s different anyway (FG:L244–46)

The commonly reported focus on growth and puberty in these quotations may reflect the central position of these in the management and treatment of girls with TS in childhood and adolescence.

The post-oestrogen group included girls aged 12 to 16 years of age. There are many developmental changes between the ages of twelve to sixteen. It is a period described as
young adolescence bringing with it experiences of physical and cognitive changes, development in social cognition and self-concept as well as alterations within social relationships. All these aspects of development are illustrated by the girls interviewed with added insight into how TS impacts on each of these aspects of growing up.

5.2.3.2 Perception of self

Being small was an accepted part of who they were and was not cited as a particular problem by many of the girls because it was the way they had always been, “I’m used to it because I’ve always been small” (Case C), she continues “I was aware I was the smallest but wasn’t too bothered about it”. In the pre-oestrogen group three girls stated they were not the smallest in their class (Case C, H and P). The majority of girls identified 5 feet or their ‘mother’s height’ as the target for their adult height. This did not mean they would not like to have been taller but generally they were not concerned with their height and although they gave examples of when it was difficult on occasions, they did not perceive it as a negative attribute. For example, one girl explained it was the practicalities of being small that were the issue rather than just being short, “I’d like to be a bit taller but mostly so I can get trousers to fit” (Case D). Another girl described having to get a chair to find chocolate that her mum had put out of reach in a high kitchen cupboard.

Body proportion was a concern to some girls rather than height per se,

you see normal short people and they’re fine because they’re still skinny and I’m short and I try my hardest, I don’t eat junk food or anything like that and I still put on weight and that’s really hard ‘cos if you’re short you can get petite clothes but if you’re short and still quite big that makes it harder (DG:L474-6)

Many of the girls describe themselves as not being typical of girls with TS. They refer to being normal and doing normal things like other girls their age without TS, “I’m just normal, just ordinary”(DG:L398). One girl explains until the diagnosis was made she “didn’t think there was anything wrong with me apart from I was small”(EG:L292).
5.3 Making sense of TS: learning about growth hormone treatment

The administration of GH was taught to all families by PENS.

5.3.1 The role of the paediatric endocrine nurse specialist (PENS)

Parents generally identified the nurse by name and expressed satisfaction with the teaching and support they had received. The PENS was reported by all the girls as visiting their families at home to teach them “all about it”. All girls were prescribed GH. Parents and girls identified learning this skill as a significant event.

Continuing information and support provided by the PENS was based around two main aspects. Firstly to manage problems with the administration of GH, e.g. the procedure, equipment or advice about initial side effects such as headaches, and secondly to provide practical assistance for example, if girls were going on a school trip or holiday. Many of the girls commented on noticing the PENS in clinic but commented they “did not need her anymore” or that “they did not see her”. One girl commented that the nurse offered support to her mother but she did not really know her. In the mother’s interview the PENS was only mentioned in terms of advice and teaching relating to GH administration, no additional support or relationship was commented upon.

5.3.2 Administration of GH

Four out of the six administered GH themselves but two of the girls (aged 12 years and 13 years) wanted their parents to continue to give it to them. They stated they did not want to do it themselves and the 12 year old also added she did not feel confident enough but her Dad had shown her how to do it and encouraged her to do it. This girl also reported her legs were like pin cushions. The six girls interviewed all expressed acceptance that they needed to have GH and one girl (Case E) specifically stated that if given the choice she would have chosen to have it. There was a unanimous belief that GH had been necessary in helping them to grow therefore it was considered a positive aspect of treatment. Nevertheless, this was tempered by reflections on some of the physical (“pin cushions”) or social difficulties
they had or continued to experience. The following extended quotation from the eldest participant interviewed provides insight into how her attitude toward the administration of GH changed over time. She commenced GH when she was nine years old. Initially she was terrified of the injections but she got used to them

R: When I started I was absolutely terrified. I remember having it for the first time and it really hurting and I remember saying it’s only a prick and it’s not ‘til the final years that you realise it is only a prick ‘cos you get used to it

As she started to socialise they became an inconvenience,

obviously I had to think about when I was going to see my mates. I didn’t really mind it at first but as you get older you start going to sleepovers and you’ve got to think about it ‘cos it has to be out for half an hour that’s the pain you can’t just take it out and have it and then it’s over. You have to wait half an hour for it to cool and you start to get annoyed with it

And as she reached puberty her motivation to have it waned as she believed the chance of further growth was minimal

I was really good for past say from nine to fourteen and from fourteen to about fifteen I was, I got sick of it and I started saying do I have to have it anymore ‘cos I don’t see the point, I was, my Mum was saying, told me from the GP to try and make me feel better that he said that I’ve been through so much perhaps they should let me erm let me be lean with it and erm I just said I’m not going to take it anymore ‘cos I wasn’t growing so I got fed up (DG:L115)

In this reflection the response of the adults to her rationale and reluctance to continue suggests she felt a degree of support from them in omitting doses, “by being lean with it”. When she was younger she reports that her father was keen that she should have all her GH

he was more fussed about getting my jab out than I was really, he’d ask when we’d go to clinic, oh how much has she grown and I’d be there and not really mind, if I’d grown I’d grown and if I hadn’t, I hadn’t (laughs). It was good if I had grown ‘cos Dad was happy but really I wasn’t that fussed (DG:L146)
GH injections are a daily invasive procedure and all the girls made some reference to the need to “get on with it” or that they had “got used to it” demonstrating a certain stoicism in some, and resilience in others for example,

Well the only thing is if well you have to have a needle every night but you have to get on with it think about more important things like family and friends and health (FG:L309)

In terms of the process of administration the girls offered some information about the storage “in the fridge”; time of administration “at night”; and preparation of her injection was clearly described by one girl

R: It’s a pen and a cartridge, you put the cartridge in the pen and turn it to zero, press and as you do you like put the needle in it and then there’s a red button at the bottom, you have to click it and then turn it to one point. I’m 2.4 so I do 2.4 (IG:L140–42).

Pain was not a major feature of having GH and only one girl cited the injection as very occasionally hurting. Other descriptions of what it felt like to have the injection include “a tickle” (I:L147) and “not really feel like anything” (BG:L187).

5.4 Making sense of TS: growing up

5.4.1 Friendships and social functioning

All girls stated they had friends. They referred to their friends by name and some girls across both groups stated how many friends they had. Girls identified a best friend or that they were part of a group. Age related activities were reported as being engaged in. The pre-oestrogen group commented on more organised activities involving clubs and groups such as Brownies and Girl Guides. Post-oestrogen group demonstrated independent activities such as going to the cinema with friends and shopping.

Parents commented that having good friends was important for their daughter’s self-esteem,
She’s got some good friends now, they make her feel good about herself (NP/R2:L106)

In Case N the father commented that it was not always the case,

She was desperate to be friends to be accepted, it was horrible (NP/R1:L64–65)

Two sets of parents describe their daughter’s friendship groups in this less positive light

Two groups of girls basically at school, cool and non-cool, she belongs to the non-cool section (EP:L231–232)

you could tell that as a threesome they won’t be the mainstream in the class in terms of friends they looked like three misfits together to be, to be brutal that’s how it seemed to me, each for a different reason but they seemed to be together ‘cause they didn’t fit anywhere else.(AP/R1:L130–33)

The non-affected girls in these friendship groups were in the same position and were not girls with TS. Difficulty in forming friendships cited in TS literature (TSSS 2008) is a characteristic that many of the parents interviewed were anxious about and positive action was taken to promote social interaction with others through a variety of organised activities, this is demonstrated further in Chapter Six.

5.4.2 Induction of puberty

Puberty was mentioned by thirteen out of the fifteen girls. The two girls who did not mention it were two of the youngest girls interviewed, aged ten years. One of the girls had a moderate learning difficulty and developmentally was not considered to be at a stage for parents to discuss puberty with her. In the second case the mother stated she would talk to her soon because she knew it would be discussed at school in Year 6. In the thirteen others one girl aged nine years had found out at clinic and stated she thought the doctor should not have mentioned it because she was too young. Details relating to puberty were limited across all ages. There was reference to changes in girls’ bodies such as development of secondary characteristics and menstruation, and the need for medical help to achieve pubertal development.
Table 24: Girls' statement relating to puberty

<table>
<thead>
<tr>
<th>Case</th>
<th>Age at interview</th>
<th>Girls’ statements</th>
</tr>
</thead>
<tbody>
<tr>
<td>C</td>
<td>10 years</td>
<td>“TS stops me growing; won’t bleed like normal people unless I have a tablet then hopefully I’ll bleed”</td>
</tr>
<tr>
<td>F</td>
<td>14 years</td>
<td>“I have tablets to help me develop and like, like a woman should really “(L217)</td>
</tr>
<tr>
<td>H</td>
<td>11 years</td>
<td>“Have a tablet to help me grow ‘boobies’”</td>
</tr>
<tr>
<td>L</td>
<td>12 years</td>
<td>“I think it’s gonna affect my periods so yeah that’s what [Dr] said (L235)</td>
</tr>
<tr>
<td>M</td>
<td>12 years</td>
<td>“If you has TS from when you were a little baby you would be helped in puberty if you wanted by the doctor” (L187–8)</td>
</tr>
</tbody>
</table>

5.4.3 Making sense of probable infertility

There appeared to be some gender difference in the approach and emphasis parents placed on the issue of infertility (Appendix 10). The daughter of one family had learning difficulties and was unlikely to develop intimate relationships or live independently in the future and fertility was not raised as an issue for these parents. Of the remaining fourteen, ten of the mothers commented on infertility as the most difficult part of the condition to deal with. They expressed in personal terms about how unhappy they would have been if they had not had children,

cos I know that if I couldn’t have children it would be quite a big, even though it’s not the end of the world, it can be the end of the world to somebody can’t it (CP/R1:L)

...if I’d not been able to have children I’d have been suicidal, that was the main thing for me (EP:L70)
...knowing how I wanted them so much and IVF wasn’t straightforward you know. To know that she’s probably got to go through all that. You know the success rates and all that but then it’s another ten years or so and things improve so…

and she concluded,

my initial feeling of complete sorrow for her really (HP:L245)

In the following quotation misunderstanding offers some hope to the mother who also expressed that she is reluctant to accept it

So her womb’s normal but her ovaries aren’t functioning that’s what he said isn’t it? So does that stay like that forever? (BP/R1:L540)

Two mothers related infertility directly to womanhood and express hope for future medical advancement will help in the future,

My fear was she wasn’t going to be a proper girl because she couldn’t have children she’s physically not a proper girl and then not a proper woman (KP/R1: L694)

Maybe in ten years they will have come up with new technologies to help her become a mother one day because I think as a woman it is very important erm even if she was married and very happy the one thing you would want is a child so I do know, I just hope there will be some way (GP:L213-15)

The couple whose daughter was diagnosed prenatally appeared more accepting that infertility was part her condition and any consequences of this would be dealt with when they arose. The father identified puberty and infertility as feminine issues that he found “awkward”, he explained

I get more involved in the psychological side of things rather than the more feminine issues, you [looking at wife] deal with those because I would feel awkward about that

Int: So issues around puberty, infertility

R1: Yeah, [Wife’s name] talks more about that because she understands more than I would about how sensitive that all is (NP/R1:L906–09)
For some of the mothers there was a need to take some action such as egg donation. All the mothers expressed hope that something might be possible in the future and advances in technology would enable their daughters to have children. One father also specifically referred to hope for the future as his daughter was to have a scan of her ovaries at the next clinic appointment. They were the only couple to refer to their faith as support for whatever the outcome might be.

Seven of the nine fathers interviewed commented on infertility. One father had to leave the interview to take his sons to a local football event prior to the discussion about infertility. As already indicated one child was unlikely to become independent and infertility was not raised as an issue by these parents. Six fathers talked of infertility as an issue that should be dealt with in the future (Cases A,B,I,K,M,P). One father stated that he and his wife were ‘distressed’ for their daughter, however, fathers generally discussed it in less emotional terms than the mothers, more of a problem with possible interventions such as egg donation or IVF and various outcomes depending upon their daughter’s preferences, e.g. to pursue a career, or to decide she did not want children.

A common thread across mothers and fathers is the concept of choice for their daughters. This is illustrated in two distinct ways. Firstly taking action to facilitate choice for their daughters in the future, e.g. egg donation and secondly that decision to have children belongs to their daughter, it is her choice, as illustrated below by a father whose wife had donated her eggs

sort of taking more options so she had choice further down the line, but still she may not want to have children you never know but it’s up to her (KP/R2:L444)

One girl stated it was “the biggest blow” of TS.

No parents mentioned aortic dissection a rare but potentially life threatening risk of pregnancy in TS women.
Some parents rationalised their daughter’s probable infertility with suggestions that not all women have children and their daughter may choose this path and another comment suggested that no-one really knows if they are fertile irrespective whether or not they have TS.

5.4.4 Implications of TS for long-term health

Medical management revolves around minimising the effects of the condition on the girls’ lives and promoting their long-term health and well-being, however, it was not clear how issues for the girls’ long-term health and well-being were discussed between doctors and parents and the girls. There was minimal reference by parents to specific long-term health issues.

Four-monthly appointments in clinic attended by all the girls have a role in monitoring all aspects of their health and well-being that may be affected by TS. This also includes planning future management. All girls had undergone a variety of investigations for assessing their potential health needs, yet only one girl appeared to make direct connections or refer to potential health complications that may affect her or that she had been screened for. Complexity of the condition is also identified in the following comment by an eleven year old unaware of the connection between thyroxin and growth,

Have a thyroid tablet but that’s nothing to do with growing (HG:L96)

The majority of parents and girls across the age range were not aware of transitional care services and they were vague as to why their daughters need further follow up.

It’s crept up so quickly and now it’s like, oh I don’t know what will happen when she turns sixteen. (FP:L369)
5.5 Priorities for information

The emphasis of parents’ information needs altered in line with their daughter’s stage of development. For example, when parenting an infant with TS parents’ information needs appeared to focus on physical aspects of the condition. Initially on her physical safety, e.g. confirmation the condition is not life threatening and information about the biomedical consequences of the diagnosis. The second area for information was that which helped with practical difficulties of parenting an infant with TS, e.g. coping with feelings, managing feeding difficulties. In infancy the social consequences related to sharing information with others. The changing emphasis of information needs across stages of development is depicted in Figure 12. The degree of emphasis is represented by the size of the circle and key issues are identified for each aspect.
Figure 12: Changing priorities for information

Infancy

Physical
- Making sense of multiple investigations
- Identifying medical problems

Practical
- Feeding
- Behavioural issues
- Managing feelings

Social
- Sharing information

Pre-School

Physical
- Identifying characteristics and associated problems.
- Monitoring and growth

Practical
- Managing behaviour
- Tips for managing ear infections
- Dealing with sleep problems

Social
- Interacting with other children, at home and with family friends

In the preschool years the physical, social and practical issues were similarly prioritised.
When girls are school age the priorities for physical and social aspects of the condition start to come together, and where they overlap is where the need for practical support occurs. In adolescence the social and physical aspects increasingly overlap reflecting their developmental needs.
5.6 Summary and conclusion

Making sense of information about TS for each family was an individual experience, however, there were similarities in the issues they face, the processes they experience and their need to understand information they receive. There are aspects of the condition that can be identified and quantified through examination and investigations which were managed and explained by endocrinologists, the clinical expert. For many parents TSSS is the lay expert offering emotional and practical expertise for parents in making sense of the less controllable social and behavioural aspects of TS, which emerge over time as their daughter grows up.

Once the parents have learned what TS means for their daughter some parents seem less concerned about the complexity of many characteristics, e.g. chromosome analysis, and focus on what they consider they need to know and what they perceive they can influence, e.g. growth, social functioning.

The girls cited parents as their main source of information supplemented by direct information from the endocrinologist. Girls did not always have the same view of their condition or share their parents’ concerns about the implications for their lives in the future. For example, issues around short stature were a concern for many parents yet for younger girls who have not yet developed their conceptual understanding of height it does not appear to be important, and in the older girls interviewed concern with practical issues relating to height and her body proportion were more important than overall height. Knowledge of specific details of TS appears limited across all girls.
CHAPTER SIX: SHARING AND USING INFORMATION

6.0 Introduction

Data presented in Chapter four illustrated that content of information gathered and received falls into three areas, physical, psychosocial and practical. Chapter Five examined factors involved in the synthesis of complex information from each of these areas and how parents and girls came to make sense of their individual TS profile. Information varied in type and complexity; in immediate relevance and in long-term impact. The purpose of Chapter Six is to examine data referring to girls’ and their parents’ experiences of sharing and using information about TS. Data provides insight into intrinsic and extrinsic motivations and triggers for sharing and using information.

6.1 Managing information: public and private

This section considers data relating to how parents managed the quantity, complexity and sensitive nature of information.

Information relating to short stature, GH and specific learning difficulties were shared both within and outside the family. Half the number of families who discussed behavioural issues within the family had discussed them with others outside the family. Over two thirds of the families shared information about chromosomes and infertility with their families. However, no one reported sharing information outside the family about chromosomes and only one family shared information about infertility. Case K openly discussed infertility outside the family. This mother was politically active in working for a change in the law so that eggs could be stored for longer than 10 years for young girls known to be infertile. Therefore to progress this campaign she had seen a need to be open about her daughter’s infertility. Figure 13 illustrates key topic areas and recipients of information.
The difference in the type of information released within and outside families suggests a public-private divide with information perceived to be more sensitive shared only with the family and close friends.

6.2 Managing the release of information

All parents described controlling information-sharing from the initial diagnostic period continuing throughout childhood and into adolescence. The following sub sections explore what prompted this control of information and how parents went about it.

6.2.1 Parents’ experience of sharing information with family and close friends

The first time parents reported sharing information with others specifically about TS occurred soon after the diagnosis was confirmed.
The main rationale for sharing information across all cases was stated as the need for support, or being “open as a family”. However, deciding what information to disclose varied across cases,

Int: Have you told family and friends about Turner syndrome?

R1: Very few, I don’t think it’s fair to Maisie, because it is her, it’s about her and we wouldn’t want her to be seen as different, or treated differently (MP:L500)

The desire for their daughter not to be seen as different to other girls is a theme across these data from parents and girls. This mother’s hesitancy to share information because of it being unfair to her daughter raises a question about ownership of information implicit in her statement about information being about her daughter “it is her, it’s about her”. For this mother a diagnosis of TS partly defined her daughter’s identity. However, as her daughter grew up, the mother became more confident about the implications of the condition for her daughter. She appeared to become more relaxed and open with the information

at first it was very private and confidential, now it’s just, well, she’s got Turners, so, what, it’s not a problem, we don’t make it an issue, we haven’t brought Maisie up to make it an issue, (MP:L72)

There was concern expressed by the majority of parents across the cases about what the implications of sharing information might be for their daughters. Some parents believed complete openness was a positive approach whereas others did not want the diagnosis revealed at all. In some cases the perceived negative implications of sharing information with family members led parents to conceal specific aspects of the condition. In the following example the family decided to exclude information about infertility when sharing information with elderly grandparents. Their rationale for this decision was the potential upset it could have caused. They felt that this aspect of the condition would have to be faced many years in the future therefore the grandparents would not have to deal with it so they did not want to worry them,

Int: So how do you decide who needs to know then?
R1: Well we took the conscious decision that our parents didn’t need to know

R1: Well they needed to know certain aspects, her having her injections, she had growth problems we just left it at that, our parents, your mum’s not alive now, my parents are in their late eighties so all the other aspects

R2: We just felt that they didn’t need to have to worry about things that they weren’t going to experience cause they weren’t going to be here to see the problems she’ll have later on so, that was a conscious decision that we took which is a difficult one because knew what Mum was like she used to research stuff didn’t she, luckily she didn’t have access to the internet. (BP:L258–266 )

Parents appear to be aware of the difficulties in controlling information as indicated in the statement “luckily she didn’t have access to the internet”, however, the risks of their well-intentioned approach were explained by the mother in the following quotation,

But I remember one night really clearly ‘cos I don’t watch much telly but [Grandmother] was watching Holby City and sure enough there was a little baby on there with Turner syndrome ……she ‘phoned me up my Mum and she said have you seen this? I said I don’t watch Holby City she said well put it on she said there’s a baby on there with Turners and do you know what they’ve just said? And I said ‘oh Mum it’s such a complicated thing it could happen to some and not others’ I thought well I’m alright here having to justify this. (BP/R1:L358–64)

The above extract highlights that trying to control information is difficult in a multi-media society, however, the complexity and individual nature of TS offers some protection; “could happen to some and not others”. The diverse nature of TS potentially helps parents to be selective in revealing certain features and characteristics of TS. However, for other families information was shared openly “each stage they all everybody came with us sort of thing”, “told them everything” (PP/R1:L253).

Some parents were uninhibited about sharing information with others explaining they did not want to bring their child up with a secret (Case C,M,N,P). However, deciding the content of what to share was challenging,

R2: I kind of felt that it wasn’t difficult to tell people but it was difficult to explain what it was and also
R1: Yeah and when they’d say what does that mean for the future and well we don’t know, and I mean when she was sat in her pram she’s a perfectly normal baby and they’d look and say she’s OK, normal, and you’d think well yes but we don’t know about the future, you know once she starts walking and talking and then going to school, well we’ll take each year as it comes and we’ve always been like that we’re still like it now. (PP:L219–22)

The extract above illustrates when sharing information with others parents had to explain complicated aspects of the condition, what it meant to their daughter and also respond to the reaction of those receiving the information. Parents identify different types of uncertainty in this process. Uncertainty associated with the diagnosis, uncertainty in their understanding of the complexity but also uncertainty in how their daughter will develop and what her specific issues may turn out to be. The final comment “we’ll take each year as it comes and we’ve always been like that we’re still like it now” implies there is an emphasis on the present, the here and now rather than planning for the future. There is also on-going uncertainty particularly faced by parents when their daughter was diagnosed at a young age.

Sharing information with close friends was a source of support referred to by mothers. In one case a close friend was given information about infertility that was not shared with family members. No fathers specifically stated talking to friends for support although two fathers stated they would have liked to have had contact with other fathers of girls with TS in the initial diagnostic period (Case K and N). Limiting information sharing to close family and friends suggested a tension between wanting to share information for the support of others yet wanting to maintain privacy.

Responses from family and friends were reported as positive by most families, however, there were some exceptions for example, in Case A the maternal grandmother not supportive of GH treatment, “just let her be small” and in Case F grandmother not supportive of GH treatment or accepting that the doctors can know that her grand-daughter will be infertile. In case E the paternal grandmother was reported as did not accept anything was wrong because her grand-daughter looked normal, “if she can’t see it it doesn’t exist”.

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Case L reported little support from their extended family where the general response “she’ll manage”.

6.2.2 Explaining TS to siblings

Siblings of girls with TS had been made aware of their sister’s condition. One mother reported a range of feelings expressed by her unaffected daughters and their change in attitude toward the daughter with TS once they had the information about how TS affected their sister,

[Sisters] were obviously very upset and they felt very guilty, as we’d say like go and turn the light switch on and she couldn’t reach the light switches, silly things, trying to put hangers in the wardrobe, so they all felt guilty, in respect they’d sort of criticise her for being lazy, but, it was that she couldn’t do it, became very perceptive and understanding (MP:L206–210)

In families where siblings were older or similar age to the girls with TS (n=11), there does appear to be a gendered difference in the reported responses to information about their sister’s diagnosis. Brothers’ were reported as more detached than sisters, “not really bothered about it” (EG:L398), although in one case where the girl with TS had developmental and physical problems (Case J) brothers were cited as helping with their sisters care. There is also an example of discrepancy in parent’s perspective of their son’s response to his sister compared with their affected daughter’s perception. Parents reported their son as “not really bothered”(Case D) about the diagnosis or its implications whereas his sister explained he did show an interest asking about GH. Another girl explained her older brothers treated her “like I’m a normal little sister” (PG:L229)

Sisters’ responses were related to issues of menstruation, infertility, make up and clothes (Case A,F,H,L,M). These data show sisters offered practical support to meet immediate needs and future needs of the affected girl for example, one sister was reported as administering GH in the absence of her mother (Case H) and two families reported sisters had offered or been invited to donate eggs in the future,
Int: What about her and her sister then, does her sister know that she has Turners does she ask questions about that?

R: She did, the only thing C (sister) says encourages her not to worry about it she’ll give F some eggs, so she just thinks it’s that simple. (FP:L213)

R: …because we (3 daughters) watch lots of medical bits and pieces on telly and like, (siblings) ask can she conceive in other ways? Well, you might want to donate an egg and they would ask how does that work? So, we’ve given them the whole scenario of not having any eggs, to adoption, IVF, donorship (MP:L461)

It is not clear the implications of such an offer were fully understood as the siblings were not interviewed. Egg donation by siblings was mentioned by two mothers (Case D and E) who thought their daughter disadvantaged by not having a sister for this purpose,

Plus she hasn’t got a sister, if she had a sister she could have had her sisters eggs you know what I mean rather than a brother” (DP/R1:L489)

Normal sibling rivalries across both genders were reported.

6.2.3 Girls’ perspective of sharing information with friends

Ten girls had told their friends something about TS. One girl (Case N) said she had not told her friends but her best friend had TS. They had met through TSSS; one girl had not disclosed any information outside the family (Case I); one girl had not reached the developmental level of sharing information with friends (Case J), one girl was reticent in answering (see table below), and one girl made no reference to telling friends.

Examples in the table below illustrate how the younger girls provided simple statements of fact, for example, having injections to help them grow. Friends accepted the information and the girls were not questioned by them. There was some anxiety expressed that children other than close friends might use the information negatively, e.g. name calling. Positive comments from friends were made about being brave for having daily injections.
Table 25: Examples of disclosure between friends (8-11 years)

<table>
<thead>
<tr>
<th>Disclosure between friends in 8-11 year olds</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Int:</strong> How did you what did you tell them about it?</td>
</tr>
<tr>
<td><strong>R:</strong> I didn’t really tell them anything about it. I just told them I had it</td>
</tr>
<tr>
<td><strong>Int:</strong> And did they ask you any questions?</td>
</tr>
<tr>
<td><strong>R:</strong> No (BG:L161)</td>
</tr>
</tbody>
</table>

| **Int:** Have you told any of your friends you’ve got TS |
| **R:** Only one or two [names friends] because I know I can trust them |
| **Int:** Would you want anyone else to know |
| **R:** No because they might call you, I don’t want to let too many people know (KG:L249) |

Older girls demonstrated more caution and were more selective about who they told. They thought through possible responses of their peers to the information. These approaches reflect their developmental age thinking more abstractly, seeking acceptance from peers results in anxiety about peer responses.

Table 26 illustrates examples of disclosure between friends in 12–16 year olds
Table 26: Examples of disclosure (12-16 years)

<table>
<thead>
<tr>
<th>Disclosure between friends in 12–16 year olds</th>
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<tbody>
<tr>
<td>R1: I’ve told my best friend in primary and I’ve told J. I’ve not told S specifically but she does know, so I’ve not told her in detail not as much as J kind of thing.</td>
</tr>
<tr>
<td>Int: So what does J know, what did you tell her</td>
</tr>
<tr>
<td>R: Erm I told her basically everything everytime I’ve come back from the hospital, they’ve said this and they’ve said that. We’re really close, she tells me everything and I tell her, I feel I can talk to her about it</td>
</tr>
<tr>
<td>Int: Does she ask you questions about it</td>
</tr>
<tr>
<td>R1: No not really, she well we get talking about it and like when her period starts she’ll say like oh you’re dead lucky you’ve not got anything yet, you’re really lucky, be happy why you can ‘til anything does happen (FG:L 264)</td>
</tr>
<tr>
<td>R: Erm well I haven’t told them everything. They just know my condition because when I first found out well I was only very young and I was foolish, you just tell, I told my mates at primary school and then it gets out doesn’t it</td>
</tr>
<tr>
<td>Int: Yeah</td>
</tr>
<tr>
<td>R: and then when I went to secondary school I was more careful about who I told but because I’d told all my close friends erm because obviously some were the primary school lot so they already knew so I thought well I should tell them, just my main friends, there’s about 7 or 8 of us well 10 of us altogether and its just them that know (DG:L292–299)</td>
</tr>
</tbody>
</table>

A developmental shift in thinking is illustrated in the second example in Table 26 above. The 16 year old reflects on who, what and when she told friends. She had learned that once information had been disclosed to others there was no way to control it and subsequently when she was older she was selective about what she had shared. The quotation below clarifies what she has shared. General information relating to growth was seen as acceptable, however, she did not disclose information about infertility or the chromosomal
nature of the condition consistent with the distinction between public and private information identified earlier in the chapter,

Int: So what sorts of things have you told them

R: Erm I just say I have, I didn’t really go into specific details, I just said I had a growth problem and erm I don’t produce, oh well once in PSHE if you had a condition and you wanted to you could do a talk about it so I said basically there is a gland in my brain that doesn’t release a chemical that makes me grow so I have to inject it myself so that’s basically all I’ve told them. I haven’t told them about chromosomes or anything like that (DG:L283-86)

Case I is a family where culturally it was not the norm for girls to form close friendships outside the family as explained by the father

[In] our community, kids seldom continue the friendship after school (IP/R1:L197) and his daughter confirmed that she had not shared any information with girls from school

Int: Would you like to talk to your friends
R: No, it’s just a fact so I never talk about it (IG:L191)

6.2.4 Sharing information with teachers and others

Parents shared information with others to support girls either at school or in recreational groups. The main recipient of information outside the family was school. Parents were the main informants to teachers. The primary purpose for sharing information with teachers stated by all parents was to support their daughter to achieve her academic potential particularly with maths and with extra time for examinations. For five families in the pre-oestrogen group additional motivation was to secure pastoral support with specific reference to behavioural characteristics (n= 6), developing and maintaining friendships groups (n=5), and encouraging teachers to be proactive in the prevention of bullying (n=2). In summary, parents revealed certain information to maximise their child’s potential to achieve success at school either academically or socially. Information was also shared in an attempt to protect their daughter from any negative attention from other children.
6.2.4.1 Acting to optimise academic potential

In Case K, parents felt they could have greater influence over their daughter’s school experience if they became directly active in school affairs. The father was a parent governor and the mother an active member of the PTA. The following quotation illustrates how they became aware of poor communication between teachers which they quickly resolved by direct intervention of the mother,

I assumed that it would be passed on from teacher to teacher. And when she went into year three she’d had a couple of weeks and I saw her teacher and said ‘how is she getting on with everything?’ She had no idea ‘what are you taking about she is fine isn’t she?’ So she had no idea that anything was wrong with her, no information had been passed on which annoyed me initially but then I thought if a teacher hadn’t identified any issues it’s not that bad. (KP/R2:L18–204)

The mother expresses mixed feelings, on the one hand annoyance that information was not shared with the teacher and on the other that her daughter must be functioning at an appropriate level because the teacher had not identified any difficulties.

The father in Case I was concerned about the way information about the condition could be used,

[she] has Turner syndrome, so ‘take it easy with her don’t press her, don’t push her’, y’know she won’t be able to cope with mathematics all the things, they give them information and is sometimes very very dangerous when some children do have the potential so they are sort of like discriminating basically you are put into a lower group (IP/R1:L306–309)

He suggests identifying difficulties associated with TS creates a tension between maximising academic potential and the potential for underachievement through lowering teachers’ expectations. Recognised profiles of potential learning difficulties associated with TS particularly in mathematics are documented, however, not all girls share the same profile as illustrated by Case E where the girl achieved grade A in her mathematics mock GCSE examination.
6.2.4.2 Attitudes of teachers

Mixed experiences relating to teachers attitudes to the information provided by parents about TS across primary and secondary schools were reported. Primary schools were generally described as being more receptive to information provided by parents, however, two families did not share this experience,

‘cos they didn’t seem to want to listen, we sent them the literature er and some of the correction activities; one of the teachers seemed to get clued in but the guy who was doing special needs didn’t seemed to want to know (AP/R1:L66-68)

Parents invested time informing schools about the potential learning needs of their daughters and relied on the information being cascaded to others. When this did not happen their daughters could be disadvantaged. Parents expressed frustration and anger as demonstrated in the following quotation

I’m not sure which tests they do when they first start and sure enough she’s marked right down on the non-verbal stuff but when we saw the tutor it didn’t sound like the message had got across to the form tutor so in that case can’t say the message gets across to individual tutors and if it hasn’t I’m going to be really annoyed because I spent a long time going through all this (BP/R2:L183-87).

In Case A parents commented on their frustration and anger at inappropriate responses from teachers to their daughter’s behavioural difficulties even after repeatedly providing the school with information, in this case organisational and sequencing tasks,

she came home one day and said the headmaster’s made me sit at the clock and watch the clock for fifteen minutes and I was going no they’re not doing that and we said that to him didn’t we and he said oh yeah it’s quite possible I did because if people want to waste time by not doing the homework they’re wasting our time so we can waste their time. And that was it wasn’t it I lost it I was just so upset by that comment (AP/R1:L478–489)

Sub section 6.2.4.1 reported communication between teachers and across years could be problematic. Across all cases potential and actual communication problems were described. Some parents proactively decided to regularly remind teachers about their daughters needs,
So we give it to every teacher she’s ever had, I think they’re sick of us now at school (PP/R1:L152–156)

The tension for parents illustrated throughout this section is to use information to help their daughter achieve academically and socially by informing others of her needs or placing her in environments and situations that meet those needs and at the same time not share information that may result in her being treated differently to other children.

6.2.4.3 Sensitive topics

Sex education was considered a sensitive topic by seven families and it was hoped that by providing teachers with information about puberty and TS teachers would be able to deal with the topic sensitively. However, there was some concern expressed as to whether information was passed on to the relevant teacher. One family also identified that sensitivity of the information for their daughter may be difficult for her teacher to manage because of gender

doesn’t help cause she’s got a male tutor but even so he needs to be aware of it just to keep his eyes open to see if she does start to have problems (BP/R1:L200–202)

6.2.4.4 Selecting a secondary school

Four families (Cases H, J, K, N) stated the move from primary school to secondary school was an anxious time. Their knowledge of traits commonly associated with girls with TS, such as lack of self-confidence, problems with short-term memory and concentration, poor organisational and spatial awareness skills and potential to be bullied formed part of their selection criteria for secondary school. For some families a smaller school meant that teachers maintained a pastoral role “a school where they could keep a closer eye on her” (HP:L155).

The size, layout of schools and how these impacted on teachers’ abilities to observe and respond to academic, personal and social needs of girls with TS were important issues in the decision making process for these parents as illustrated here,
.... it’s a fantastic school, resources and as a building it’s phenomenal but it’s got a one way system and if you go past your room you have got to go all the way round again which well y’know with a child with poor spatial awareness, getting lost that will be a nightmare. (NP/R1:L397–400)

Two girls (Case F, M) attended secondary schools that siblings attended therefore offering a familiar setting and familiar teachers. This preference for familiarity is a trait commonly associated with girls with TS and was highlighted in Chapter Five within context of continuity with doctors in clinic. Knowledge of the school in the girl’s transition from primary to secondary education was viewed as helpful by parents as demonstrated here

they go and have like half a day, then they go again and they go again and Maisie’s got two sisters that were at the school as well so

Int: And do you think that helped?

Yes, definitely (MP: L123)

One girl was going to go to a school where her mother worked. She had visited on several occasions,

and she knows most of the teachers ‘cos we do concerts and she’ll be able to take all her music, she’s very musical so she’ll be able to shine. (NP/R2:L118)

The next example highlighted the difficulties for parents whose daughter had more complex needs and although the parents were well informed they were unable to utilise this information effectively which had made them feel unsupported in seeking a place at a suitable school,

R1 The biggest worry we’ve had in recent times is Jodie’s pending move to high school that is just tortuous at the moment cause we don’t have an answer at the moment and we are not getting any help from anywhere in terms of getting something suitable.(JP:L128)

Some parents who had experienced anxiety of the move to secondary school found that they were pleasantly surprised by the reality,
she’s settled down so well and we were quite wary of her going and getting lost in this huge great school …. y’know whether the way they teach is such that it reflects her needs” (AP\R2:L87–90)

The tension illustrated throughout this section is to use information to help their daughter to achieve academically and socially by informing others of her needs and at the same time not share information that may result in her being treated differently from other children.

6.3 Deciding what information to share

This section focuses on the content of the information shared with others.

6.3.1 Influencing experiences at school

Subsection 6.2.4 presented data relating to the need to inform teachers about TS and the attitudes of teachers in response to that information. This section provides data about specific content of information about TS that parents provided to teachers.

Parents across all cases told teachers their daughter had a growth problem. In thirteen cases specific learning difficulties associated with TS that may affect learning such as difficulty with mathematics and spatial awareness were discussed. ‘Tips for teachers’, prepared by the TSSS (Appendix 8) was given to the teachers in thirteen Cases. These tips are one part of a booklet that provides more detailed information about all aspects of the condition entitled ‘How to survive and succeed at school’. Three cases made the full booklet available to school. Of the two who did not use it, one girl had no academic difficulties and was diagnosed late (Case E), and the other had learning difficulties and additional support at school was in place (Case J). In addition to these ‘tips’ some parents shared information about physical limitations, puberty and behavioural characteristics associated with TS.

‘Tips for teachers’ suggests suitable techniques for teachers to employ in the classroom to optimise achievement at school for girls with TS. Where it was thought necessary some parents (n=6) had further assessments to ascertain individual learning needs,
I did have an educational psychologist assessment of her and she just thought Hannah was probably, well may be a bit slow but didn’t really have any specific weaknesses. Erm some weaknesses in reading skills but maths wise well not really. (HP:L83–88)

The above quotation again identifies the individual nature of the problems experienced by girls.

### 6.3.2 Growth hormone treatment

This appeared to be the least contentious piece of information shared by parents with others, and by girls with their friends. In some cases it was the only piece of information shared with others and was often used as the defining feature of TS.

### 6.3.3 Induction of puberty and pubertal development

The following quotations identify several points relating to the process of sharing information about puberty and what the implications of this are for a girl with TS.

[School] did sex education and I thought it was strange ‘cos she said well it doesn’t apply to me ‘cos I’m not going to have periods and that’s what I thought was a bit strange because she still needs to know about everything (FP:L103)

The mother in this case explained normal puberty to her two daughters together and then explained to her daughter with TS the differences for her in experiencing menstruation,

you’ll have like a breakthrough bleed you’ll have a pill so you will have some sort of bleed but none of the pains that everybody gets and her friend said you don’t want to go through that so she was quite chuffed (FP:L110)

The following quotation illustrates a more matter of fact approach in explaining what their daughter needed to know,

when you’re old enough you’ll have injections that’ll be to start your puberty, so she says so I’ll be bleeding like Lauren and I said yeah like every month you’ll have the same as Lauren. Oh right she says so I won’t miss them out so I said no (LP:L368)
There is misinformation in the above statement as no injections are involved.

The following quotation illustrates that for some mothers deciding what and when to tell their daughter was difficult and they preferred to be guided by the endocrinologist
time is going to come pretty soon I would have thought but [endocrinologist] says that [Dr] will tell her when the time is right (HP:L304)

Once oestrogen had commenced, the physical development of the girls needed to be assessed during the clinical consultation. This involved discussion and physical examination of girls’ secondary sexual characteristics which was reported by some as an area for concern

Er, she’s seriously embarrassed over it now, do they really need to check can’t they just ask me ? I mean I know, there’s no sign yet, I don’t know I just wonder if they think about the child and the way they feel as much as about the medical condition (BP/R1:L467-70)

There were some examples of parents being unclear about physiology for example, not all aspects of pubertal development such as body hair are controlled by ovaries,

R2: Some of these things that were happening starts you thinking well she’s started to go through puberty after all but I suppose I could have gone and done more research but [Dr] said ‘no that’s the male hormones that do all that; you asked me that didn’t you ? What does that mean? and I said, told you what [Dr]’d said.

R1: Well, I just wanted those tablets started

R2: Testosterone, so as he put it, he said ‘the male hormones that do the nasty bits.’ It is confusing. (BP:L547)

Parents were cited as the main source of information for girls. The accuracy of the information girls receive largely depends upon parental understanding.

6.3.4 Long-term health

Only the eldest girl (Case D) mentioned TS had issues for her long-term health. No parents mentioned talking to their daughters about the long-term health implications of having TS.
6.4 When information was shared

Content and timing of information sharing between parents and daughters was interlinked. What parents considered appropriate for different ages determined what girls were told. The range of information considered to be suitable differed across families. This mother states,

I’d like her to know everything that there is to know then I don’t think you’ve ever sort of got to get to an age and then tell her something, I’d rather she was brought up with it. (CP/R1:L75-77)

One mother took her lead from her daughter responding to questions as they arose, and making a judgement about the level and pace of information depending on her daughter’s responses,

you grow with them, you give them as much information, as not to lie to them, or be deceitful in any way, or form, or closeted, with the information, you give them enough information, that you think, well, they can digest that at the moment and that would be as much as we’d need to give and she’d be quite happy, because I would only stop answering her questions when she stopped asking and once she was happy, she’d ask another question (MP:L494–499)

Some parents appeared to seek reassurance of the appropriateness of their actions,

I’m not sure whether that’s right or wrong, I don’t know whether that’s good parenting or not, it seems to have worked (MP:L501)

6.4.1 Age-appropriateness

A common theme of wanting to give information at a time when it was considered to be developmentally appropriate is evident. However, determining when that was and what specifically parents should tell their daughter could be problematic. A sense of anxiety in getting such judgements wrong is expressed by this mother,

..if she wants to know things she knows she only has to ask us anyway. But then it’s kind of like do we keep saying things to her and open a can of worms if she’s not ready to know or do we just leave it and we think well she’s going to ask us but then she’s waiting for us to tell her so it’s a fine line really (FP:L398)
Equating the release of information to opening a can of worms is a powerful expression that indicated the mother’s anxiety at inadvertently creating problems and upset for her daughter if all aspects of the condition were revealed to her. Information concerned with puberty and fertility was viewed by the majority of parents as the most sensitive. Some parents felt their role included protecting their daughters from receiving information that was not age-appropriate.

R2: We can protect her at the moment can’t we, we can tell her things and explain it to her

R1: Well as best we can

R2: Yeah, we protect her by not telling her everything at once until she gets to an age where we think that it’s like appropriate for her to know (PP:L530)

One mother stated that there was some difference of opinion as to the timing for disclosure of information relating to puberty between herself and her husband. Judgements about what was considered age-appropriate were personal to each family and between couples with a range of approaches illustrated.

One mother was reassured by the endocrinologist that issues would be addressed at the appropriate time, i.e. when their daughter was developmentally ready to discuss issues such as puberty. In this example the mother wanted the endocrinologist to take the lead because the mother felt she would be unable to cope emotionally when sharing sensitive information. However, there was also a desire to know in advance when this might be so that she had some time to prepare her daughter and herself,

Int: So when you share information with her it will be in that environment with [endocrinologist]

R: That’s my understanding yes

Int: And will that be helpful to you rather than have to do it on your own

R: Erm, well maybe to me, with [Dr] saying that I thought it will be better coming from the doctor rather than me because I might be too emotional to tell her. (HP:L308–12)
This mother expressed anxiety at the information to be shared and at her own predicted response.

There was variation in what parents considered appropriate in terms of content but for some it was the level of the information that needed to be appropriate rather than the topic area as illustrated in the following quotation,

I think there’s a lot she doesn’t understand, it’s quite hard with Chloe because you have to tell her what she’s capable of understanding for her age. We keep having to tell her a bit more as she gets older and older because of the XY chromosomes like, Turners like. But she understands she knows that she had her organs, well they weren’t really organs removed when she was born and she knows that she doesn’t have any eggs, she knows that she can’t have any children and she knows y’know I really talk to her about adoption or donor eggs she knows about that side (CP/R1:L112–17)

Data relating to parents and girls responses in making sense of infertility were presented in Chapter Five. What girls were told as they grew up varied from having no information (Case H) to being informed from an early age (Case C).

6.4.2 Finding opportunities to share information

Once a decision had been made to share information with their daughter, parents determined the timing of release of information. From these data the trigger for telling daughters about puberty came from different sources:

- once parents knew it was to be taught at school
- clinic appointment where the induction of puberty had been discussed
- sibling/peers starting periods

Preparing their daughter for induction of puberty was discussed by several mothers. Some planned the teaching in advance,

… in Year five they’ve started doing a little bit so I did a bit with her and in Year six I did quite a lot more cause they get all the talk then they do it quite early now don’t they on periods and everything, so I was quite sad really cause she’s had that explained to her quite matter of fact later at a very young age and it’s sad because
you’re sort of dealing with it as if it’s like really a medical sort of thing (BP/R1:L315–18)

This quotation also suggested a sense of loss of a normal aspect of development that has become medicalised.

Other parents took opportunities that arose and provided impromptu information giving sessions

Well it were over Lauren [sister] really, she’d just started her periods and there was some blood on her bed and Louise started laughing and saying ha- ha, teasing and saying I won’t have to have anything like that and I said but you will (LP:L182–184)

Several mothers described opportunistic approaches being adopted in relation to sharing information with daughters about infertility. In the first quotation the mother is more direct than the second,

From when she was very young I’ve told her she can’t have children of her own (CP/R1:L120)

she’s also seen lots of people who haven’t had children so you can still have a life, a nice life without children. But I know she’s, she’s said in conversation “when I have children” you know, and a few times I’ve just said well not everyone has them – trying to – just put the seed there (HP:L194)

These mothers appeared to be attempting to develop a frame of reference for the future.

And for older girls an introduction to possible options for the future were introduced with varying responses, for example,

we’ll sit and watch, y’ know er is it Robert Winston who does the fertility things, we’ll talk about that and she’ll sit and she’ll say to you ‘I don’t want donated eggs cause that’s not my baby I want my own.’ So to be twelve and to talk like that it’s quite sad isn’t it? (BP/R1:L321–23)

Unplanned interactions required spontaneous responses from parents which some found difficult to cope with,
She found the bit regarding fertility and she came straight out and asked would I not be allowed to have children? That was a bit of a killer (MP:L190–192)

6.5 How information was shared

One mother used available resources from school to teach her affected and non-affected daughters about puberty. She found it useful to discuss normal development and then the differences for her daughter with TS,

.... so that made it easier because I was telling Frances (sibling) what was going to happen when she starts her periods and then tell Flora its like this for you (FP:L283–86)

She had covered the information with her daughter with TS on a previous occasion. This raises the issue of revisiting topics to reaffirm information and check understanding, and also supports the concept of learning as a social activity.

The following extended quotation eloquently demonstrates how decisions about what to tell, when to tell and how to tell are closely entwined. The mother describes sharing with her daughter sensitive information about puberty and infertility. This has been described throughout these data as the most difficult aspect of TS,

... the chat I had with Sarah about puberty whether she had Turner syndrome or not I would have had I just well its just it’s a much more emotional experience, not just passing on of information ‘OK well you know any time or whatever this is going to happen to you’ because actually its saying something very different as well as saying that, so it is a more emotional experience and I think as a mum you’ve got to be ready for that experience as much as the daughter has to be ready for it. I wasn’t ready to do it erm but I had to do it because school were going to do this subject so I thought right I don’t have a choice now so I’ve got to deal with it whether I want to or not and erm we often have baths together and so I thought that’s the best place, no distractions and erm that’s how I did it and we’ve since talked about adoption and things haven’t we [looks and smiles at daughter], talked a little bit about that, lots of little children who need homes and you don’t always have to have, to grow inside a mummy’s tummy to love a child (NP/R2:L923–932)

This emotive quotation illustrates the close association made between information giving related to puberty and fertility for girls with TS. For this mother revealing issues relating to
puberty meant telling her daughter she would not be able to have babies of her own. In normal girls puberty is a sign that the girl is becoming fertile hence the comment “actually it is saying something very different”. When to tell has been placed within developmental needs but here the mother identifies the importance of being emotionally ready to share information and explains that personal readiness may not coincide with external influences.

**6.6 Developing an individual meaning of TS**

As presented in Chapter Five all parents interviewed had accessed information and developed an understanding of specific physical features and behaviours that are associated with the condition. How the visibility of these features and behaviours influenced the parents’ perceived success or otherwise in normalising their daughter’s condition will now be examined.

**6.6.1 Minimising the diagnosis**

In Chapter Five it was identified that the majority of parents (n=11) expressed some unease about sharing their daughter’s diagnosis with others without adding clarification of how she was affected.

In seven cases (B,D,F,G,K,M,P) parents attempted to minimise behaviours and characteristics associated with TS by making comparisons between their daughter and ‘normal’ girls and their daughter with other girls with TS, as illustrated in this text.

> The thing is at the end of the day although she’s got it apart from the height she’s very normal she’s above average with her school work which most people with Turner’s syndrome probably aren’t (DP/R2:L:326–328)

The above quotation suggests that girls with TS are not normally academically able. Comparison with others enabled the parents to determine how severely affected they perceived their daughter to be. The mother continued.
I mean we’re very lucky because Debbie has it very mildly you know what I mean when I say she’s got it some children have a lot more side effects and a lot more things that cause problems (DP/R1292–294)

Another mother stated

it’s something she’s got, Mummy’s got a bad knee, Maisie’s got Turners (MP:L89)

When clinic was busy and parents perceived their daughter’s TS not to be primarily a health problem it made it less easy to perceive their daughter as a patient and led them to feel they should not be taking up the doctor’s time

R1: Phoebe’s not that bad and y’think there’s nothing wrong with her

R2: Yeah don’t like taking their time up knowing how normal she is really (PP:L442–446)

These quotations equating physical appearance of the girl and her aptitude for school work with being ‘normal’ keeps the complexity of TS hidden.

For some parents minimising the diagnosis was illustrated by descriptions of individual behavioural characteristics of their daughter which distanced her from the commonly described behaviours in the patient literature.

For example difficulty with social skills

Int: So, she mixes well with other girls?

R1: Brilliant, she hasn’t got any of those traits at all, I know some Turner girls can find it quite difficult, with behaviour problems, no, she hasn’t got any of that (MP:L281–287)

Or lack of self-confidence and dependency,

I think it’s sort of, because we’ve never made an issue out of having Turner’s Syndrome ..... she’s had to get up and be, get out she sorts herself out whereas I sort the dog and everything else out. And she catches the train .... that’s given her lots of confidence. (DP/R1:L102–107)
And difficulty in maintaining friendships

She’s got some good friends, she had one drop her for someone else but now she’s got some very good friends” (EP:L133–134)

However, minimising the condition to normalise TS can be difficult in practical terms. This father explained

it’s a fine balance in making sure you’re daughter gets everything medically that she needs and at the same time treating her as a normal girl because that’s what she is and that’s very hard to do (KP/R1:L745)

In this statement the father stresses her normality – “that’s what she is” but admits that it is “very hard to do” implying they are inextricably linked. This suggests a need to separate out the TS identity from their daughter’s ‘real’ identity and is seen in other cases.

6.6.2 Using TS as a lens

This section focuses on how parents used the behavioural information as a frame of reference to explain their observations of their daughter’s behaviour. The majority of parents (n=14) made some reference to finding it difficult to determine what a Turner characteristic is and what were their daughter’s natural abilities and characteristics.

TS was categorised by families not according to the chromosomal analysis but rather by the number of characteristics and features that their daughter presented with,

there are some things on the list of all the things, Yes that’s Hannah, and that’s Hannah definitely, no that’s not, that’s not Hannah (HP:L 410)

The interpretation of behaviour either leads parents to decide they have observed a TS trait or explain the behaviour as part of their daughter’s personality or as an inherited trait. In the following quotation the girl with TS is displaying a characteristic that is different to other family members and the mother commented
it’s just her confidence really she’s not, and whether or not because she’s different to me and her sister I don’t know but it takes her a long time to make friends; when she does make friends then the friends are like for ages

She later added

Yeah, but whether that’s because of Turners or whether that’s just Flora, I don’t know. (FP:L154–160)

If the behaviour was recognisable in another family member it was thought it could be an inherited characteristic but uncertainty is still expressed

PE she’s not fantastic at sport but I’m not and art I’m not good at and she’s not, so what’s an inherited characteristic and what’s Turner’s? (KP/R1:L171)

Where constant comparisons and interpretation occur this father expressed a sense of misgiving

R1: Well we’re continually looking for things and trying to sort of think oh that must be all it is but in reality nobody else would ever guess there was anything wrong ..... I mean, alright her proportions are slightly out of balance but she a little girl (KP/R2:L226–235)

The following quotation highlights similar unease

I think sometimes when we think about these things, about school we think like in the back of your mind you are thinking she’s going to have a problem she might have a problem with this year cause obviously it’s there but then she doesn’t so you think well that’s fine I don’t have to worry about it ‘cause she’s not got a problem (BP/R2:L248-51)

Knowledge of general TS characteristics is used as a frame of reference by this father who reluctantly anticipates issues that may occur. The individual nature of the condition means that there is on-going uncertainty about whether these issues will be observed in their daughter.

Girls with TS may be visible through their physical features or behavioural characteristics or both. As presented in Chapter Five all parents interviewed had accessed information and
developed an understanding of specific physical features and behaviours that are associated with the condition. An acceptance of the diagnosis without individual interpretation meant that there was a possibility of less desirable characteristics being associated with their daughter. In the following quotation a mother explained that during a routine developmental assessment by a Health Visitor her daughter was unable to pick up objects as instructed. The mother tells of her upset as the health visitor assumed TS had affected the intellectual ability of her daughter rather than assessed the physical problem of her puffy fingers,

Int: So you felt her judgement was about her intellect not about her physical capability

R2: Yeah and that upset me er that they were already prejudging her, thinking there was something wrong with her mentally rather than physically and it’s not that she couldn’t do it... she couldn’t clasp her fingers together because of the puffiness (PP:L179–186)

This mother fears assumptions about her daughter having impaired intellectual ability.

6.7 Visibility to others

Further data illustrating how the visibility of TS features and behaviours were described by parents are presented. A range of different parental perspectives were communicated about the physical appearance of girls with TS. Table 27 is a list of possible physical attributes identified within the publication ‘TS Lifelong guidance and support’ (TSSS 2008). Some parents report using this list to identify how many of the features and characteristics their daughter had. They equated the number of features and characteristics with how severely affected they thought her to be. However, there is no reliable correlation between the number of expressed features, the girls karyotype and the potential for co-morbidity. The cases where parents had identified these features in their daughter are presented in the right hand column.
## Table 27: Possible visible physical features

<table>
<thead>
<tr>
<th>Characteristic physical feature</th>
<th>Cases commenting on this feature in their daughter</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lymphoedema</td>
<td>A, B, C, J, K, P</td>
</tr>
<tr>
<td>Webbed neck</td>
<td>N</td>
</tr>
<tr>
<td>Small hyperconvex nails</td>
<td>C, F, H, K</td>
</tr>
<tr>
<td>Low hairline</td>
<td>F, N</td>
</tr>
<tr>
<td>Low set ears</td>
<td>None</td>
</tr>
<tr>
<td>Small jaw</td>
<td>None</td>
</tr>
<tr>
<td>Short stature</td>
<td>All Cases</td>
</tr>
<tr>
<td>Increased carrying angle of elbows</td>
<td>C, E, M, K,</td>
</tr>
<tr>
<td>Pigmented naevi</td>
<td>M, N</td>
</tr>
<tr>
<td>Drooping eye lids</td>
<td>None</td>
</tr>
<tr>
<td>Short fingers</td>
<td>P</td>
</tr>
<tr>
<td>Myopia or other eye problems</td>
<td>A, H, J, K, N</td>
</tr>
<tr>
<td>Breast development fails to occur</td>
<td>E, F</td>
</tr>
</tbody>
</table>

### Descriptions of physical appearance

<table>
<thead>
<tr>
<th>Description</th>
<th>Details</th>
</tr>
</thead>
<tbody>
<tr>
<td>Broad chest, stocky build</td>
<td>K</td>
</tr>
<tr>
<td>Short stature and relatively short legs</td>
<td><em>All parents commented on short stature but for some, body proportion was raised as important</em></td>
</tr>
<tr>
<td>Tend to put on weight</td>
<td>A, D, E, H, I, J</td>
</tr>
</tbody>
</table>
Some parents stated that physical stigmata associated with TS were not recognisable in their daughter

    That’s the thing you can look at Beth and you wouldn’t think she’s different (BP/R2:L521)

However, for others as in Case H, the mother seemed particularly troubled by her daughter’s appearance which she considered to be noticeably different from other children,

    She was in a crowd and I said, oh she looks different than all the other children (HP:L251–2)

Her concern was whether other people noticed her daughter’s difference,

    And she does look different, she stands out – but I don’t know whether that’s because I know or whether other people are thinking she looks different. (HP:L253–255)

It is being different that concerns this mother irrespective of whether she is identified as having TS. This comment also suggests that once a parent has knowledge of the features and characteristics they then see their daughter through this ‘lens’ which is sometimes used to make judgements about other people they see, “there’s a girl at John Lewis and I’m convinced she’s got it” (HP:L516).

However, there is a sense of anxiety expressed as to whether other people will or will not notice their daughter’s features. This father tries to reassure his wife claiming it is unlikely because the condition is not well known amongst the general public,

    put that in perspective there aren’t that many Turner’s girls in the population, it’s very small, we would probably know which is Turners, but the vast majority of people haven’t got a clue (KP/R2:917–920)

In Case H the mother explains her concern about her daughter’s difference by direct comparisons between her daughter with TS, her sister and a friend who had come to play,
Look that’s my 13 year old daughter, nearly 14. She is mature and that’s her friend who is tiny. B, Hannah’s twin is as tall as she is, so you look and think well there we are, there is a 13 year old girl who is small so there are (pause)

Int: There is a range

R: Yes there is a range in normality as well but to me now it is just that Hannah looks different (HP:L323–327)

Clearly the mother feels that her daughter’s height on its own does not identify her as having TS. Other features make her “look different”, she talked of girls with TS having a similar shaped head and asked the interviewer “when you see the children do you see a look of them” (HP:L512). The mother in Case K comments that on the TSSS website,

there’s a picture of all the children at the conference and they’re very very alike but it’s hard to say why. I’ve asked several doctors and yes they could identify Turner child in the room (KP/R1:L907)

This mother echoes the concerns of those expressed in Case H. She worries “that people will look at her [daughter] and think what’s wrong with you”. Being singled out as different was a concern expressed in ten Cases. In some cases the initial anxiety had dissipated following treatment to normalise the girls height,

she was gonna have to go through all that in her life and everybody knowing just by looking at her that there was something wrong (FP:L 51)

The mother later conveys relief at her daughter’s good response to GH which means she is now a similar height to other people,

[She] doesn’t look any different to the majority of people. There’s people taller and people smaller, she won’t be the smallest.

Int: So that was your aim

Yes I just wanted to get her to a height where she could go anywhere and not feel she’s the smallest in the room (FP:L255–60)

Height as a defining feature of TS is not absolute for being seen as having TS. Here the parents’ comment that two other girls at school could be singled out on short stature rather than their daughter
R1: But the bottom line is she’s not out of balance with any of anybody of her age group in terms of being very short or…

R2: If you looked at her class and you had to identify the Turner girl there’s at least two who are a lot smaller than she is (KP:L33–335)

In addition to physical aspects of TS, girls could potentially be identified by their behavioural characteristics. Table 28 summarises observable characteristic behaviour of girls with TS sourced from TSSS literature. Figure 14 illustrates a continuum of parental perception of the visibility of their daughter’s features, from those not appearing to have any physical features of TS to those with characteristic stigmata.

**Table 28: Summary of possible behavioural aspects of TS**

<table>
<thead>
<tr>
<th>Characteristic behavioural features</th>
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</thead>
<tbody>
<tr>
<td>Hyperactivity</td>
</tr>
<tr>
<td>Immature</td>
</tr>
<tr>
<td>Specific Learning Difficulties and problems with spatial awareness</td>
</tr>
<tr>
<td>Sleeping problems</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Descriptions of possible behavioural features</th>
</tr>
</thead>
<tbody>
<tr>
<td>Problems with short-term memory and concentration</td>
</tr>
<tr>
<td>May find sudden changes of routine difficult</td>
</tr>
<tr>
<td>Lack self-confidence and have poor self-image</td>
</tr>
<tr>
<td>Easily hurt or discouraged if anyone is unsympathetic</td>
</tr>
<tr>
<td>Not keen to try new things</td>
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<tr>
<td>---------------------------</td>
</tr>
<tr>
<td>Can be articulate with a natural flair for English and other languages but mathematics in particular may cause problems</td>
</tr>
<tr>
<td>Can have problems with fine motor coordination skills, poor hand eye coordination and with short stature this makes Games lessons a problem</td>
</tr>
<tr>
<td>Can find it hard to follow instructions and perform tasks that require several steps</td>
</tr>
<tr>
<td>Stage where she seems immature and finds it hard to maintain friendships with girls of her own age leading to a sense of isolation and loneliness</td>
</tr>
<tr>
<td>Bullying can be a particular problem due to size and appearance</td>
</tr>
<tr>
<td>Difficulty in interpreting the behaviour of others and often find it hard to appreciate what is happening; can be gullible</td>
</tr>
<tr>
<td>Self-organisation may not come naturally</td>
</tr>
<tr>
<td>Low energy and stamina</td>
</tr>
<tr>
<td>May have a high pain threshold</td>
</tr>
</tbody>
</table>

(Source: TSSS 2008)

Some parents used their knowledge of these features to develop practical strategies to manage them, whereas others used them as a frame of reference with which they compared their daughter’s behaviour.
Figure 14: Parental perception of the visibility of TS features to others

She looked very distinctive you’d know straight away she had many of the features (N)

TSSS, it just described her (lists features) (A)

Just petite (I)

Some of the features; not in proportion (K)

She has it very mildly (D)

No physical sign of anything wrong (E)

By looking at her there’s nothing wrong with her besides her height (G)

Same shaped head same mannerisms she looks different to other children (H)

She does look slightly different but she’s not full blown turners (M)

Some of the features; not in proportion (K)

You can look at her and you wouldn’t think she’s any different (B)

Doesn’t look any different to normal baby (P)

Doesn’t look any different to the majority of people (F)

They tell you webbed neck, puffy eyes, fingernails, horse shoe kidneys and that but she’s nothing like that (L)
6.7.1 Identifying with other girls with TS

For two fathers, joining support groups or facilitating their daughter to meet up with other girls with TS was to be avoided. They did not want who their daughter was defined by TS. TS was something she had, not who she was,

he didn’t want it be Flora with Turners Syndrome and, and that’s the only thing we see really he wanted her to be known as Flora, OK she had got this thing and we’ll deal with it, but he didn’t want her going on outings just because they were children with Turner’s syndrome (FP:L43–46)

Similar view expressed by father in Case B

With Turner’s syndrome you could think ‘my daughter’s got a problem’ and you treat them as if they’ve got a problem, we’ve never taken that view have we, and that’s one of the reasons why we’ve stopped getting involved with this [local support group] society because you got that impression (BP/R2:L237–240)

However, later in the interview the mother expressed that sharing experiences of other girls with TS may be beneficial,

R1…there was some case studies as well a lot of girls writing on the internet which probably ask Beth to look at when she’s older, their experience so they were good too but everything was different because it seems to…
R2: Well I don’t even know about asking her to look at that yet because why spook her at twelve y’know what I mean?
R1:Why spook her?
R2:Well it could be you’ve got girls writing about I can’t do this I can’t have that
R1:Well it’s not because they just write about their experiences (BP: L424-30)

This viewpoint was seen in another case,

I mean I think it’d help like explain things y’know what’s the point asking me y’know she might feel better asking someone else who’s got it (LP:L96)

Another difficulty in relation to the range of abilities, features and characteristics is revealed here. The parents have differing perspectives on the value of contacting other girls. The mother feels her daughter would benefit from sharing experiences with other girls like herself whereas the father is concerned at what she may be exposed to. He
uses powerful language in conveying his anxiety “why spook her”. This discussion between parents also highlights the role of parents as gatekeepers to information.

Finding out about the experiences of other girls was helpful to some parents as illustrated here,

when I first found out about Hannah I spoke to the lady at the Turner syndrome Support Society and I think her daughter had just finished her injections and all her treatment the night before, and she was saying she had gone on to do her A levels. So that initial ‘Oh my God what’s going to happen to her’ and then reading other information made me realise it wasn’t the end of the world. (HP:L324)

6.7.2 Concerns for social functioning in the future

The development of social skills was a major concern for these parents,

I think it’s the social skills that worry us most (AP/R2:L185)

The mother clarified,

how she’d cope with what are very demanding social situations as she grows up beyond school, college, further education if she goes that far, if she wants to go that far or work everything’s geared, the social side, interaction with people, and you worry how she’ll cope with that to be honest (AP/R1:L401)

Boman et al. (2004) argue that if girls with TS are involved in less social interaction with their peers then they may be disadvantaged leading to a tendency to socially isolate themselves. Overprotective parenting may compound this problem, however, parents interviewed in this study were generally proactive in seeking out suitable social activities for their daughters.

6.8 Summary and conclusion

Data presented illustrated management of information by parents and the girls which appeared to determine who they shared information with, what was shared, when and how it was shared. Data demonstrated there was a selective release of information related to the subject area, girls age and developmental level, individual TS profile, i.e.
what was perceived as relevant, attitudes toward information sharing and external influences such as sex education at school.

Two components of using and sharing information are evident in these data, content and process. Firstly parents came to understand the content relating to the physical, psychosocial and practical aspects of having a daughter with TS and secondly they made decisions about the process of sharing information in the best interests of their daughter. These decisions are very personal to each family, reflecting their motivations to share information, and usual ways of communicating as a family and with others.

As parents became more knowledgeable about the condition they engaged in the interpretation of information from their observations of their daughter’s characteristics as she grew and developed; and they optimised social and academic opportunities for their daughters. Managing information brought with it certain tensions.

A flexible approach to managing information helped parents in normalising aspects of the condition thereby enabling them to manage TS in the context of their everyday lives and in thinking about possibilities/hopes for her future.
CHAPTER SEVEN: DISCUSSION

7.0 Introduction

The main discussion is centred around uncertainty, identity and normalising which are the three major themes to emerge from data presented in Chapters Four, Five and Six. The aim of the discussion is to develop understanding of the underlying reasons for the information needs identified by girls and their parents in order to provide a sound basis on which to develop approaches to providing information.

7.1 Theoretical framework

This research set out to explore the perceived information needs of girls with TS and their parents. Social interactions during the activities of gathering and receiving, making sense of, and using and sharing information were described in these data and are important in understanding how information can be best presented and delivered in clinical practice. Symbolic interactionism (Blumer 1969) can provide a useful framework for considering such interactions. Central to Blumer’s view was that an individual’s actions occur within a communicative process. This perspective proposes that the individual develops from the process of social interaction. In other words individuals, rather than being merely reactive to certain stimuli within the environment are instead active in interpreting their world view which subsequently informs action.

Symbolic interactionism considers the individual and the context of their environment as inseparable (Charon 2010) in the development of meaning. Data presented across the findings chapters identified different types of information and illustrated how it was incorporated to a greater or lesser degree by the girls and their parents into their personal definition of TS. Various encounters with others in different settings, e.g. clinic appointments with doctors, meetings with teachers, attendance at TSSS events, discussion with family and friends, contributed to the meaning of TS developed by the girls and their parents. Social encounters where information was exchanged either formally or informally, served to reinforce different facets of the condition for example, a visit to clinic or learning to administer GH would focus a girl and her parents on the
physical nature of the condition which required medical management; meetings at school to discuss their daughter’s educational needs, achievements and behaviour would reinforce the psychosocial elements of the condition; parent and daughter interactions enabled an individual and personal interpretation of TS. The nature of the actions undertaken during these social encounters in these diverse settings supported different aspects of TS as illustrated in Table 29.

The broad variation in phenotype of girls with TS meant that these facets of TS had different weighting for each family. This directly influenced their perception of TS which had implications for their information needs. For example, in Case M the girl had a mild phenotype and future potential physical difficulties that girls with TS can experience were not considered important because she did not look like a girl with TS, however, visiting clinic reinforced that she has a medical condition that requires monitoring and a treatment plan.
Table 29: Actions reinforcing a diagnosis of TS

<table>
<thead>
<tr>
<th>Facets of TS</th>
<th>Actions reinforcing view of TS</th>
</tr>
</thead>
</table>
| Physical /biomedical view of TS       | • Measuring  
|                                       | • Examinations  
|                                       | • Treatment plans  
|                                       | • Investigations  
|                                       | • Taking action for the future, e.g. egg donation  
| Psychosocial view of TS               | • Providing information for teachers  
|                                       | • Enrolling in social activities to build self-confidence  
|                                       | • Participation in TSSS events  
|                                       | • Observing friendship groups  
| Personal/ individual view of TS       | • Emphasis on girl’s strengths  
|                                       | • Distance behaviours from undesirable TS traits  
|                                       | • Recognising features of TS  
|                                       | • Reconcile familial traits with TS traits  
|                                       | • Openness about TS diagnosis  
|                                       | • Concealing information about TS  

Personal contact with TSSS provided parents with insight into a world of living with TS, and considers potential implications for the future through communication with other parents. It provided their daughters with social experiences with other girls with TS. Developing mutual perspectives between “social actors” can enable individuals to
see themselves from the standpoint of one another and appreciate the collective attitudes of others (Turner 2008).

Social encounters such as these provide the potential for girls and parents to see themselves as girls with TS and parents of girls with TS. These identities are created and/or reinforced through the responses to them by others. This may help to explain why some families who were ambivalent toward the diagnosis (in that they felt it misrepresented them) were reluctant to engage socially or identify with a wider population of girls with TS. Turner (2008) explains that the perception of others to an individual’s “public appearance” becomes merged with “the self” influencing how the individual perceives themselves. For some parents and girls identifying with others with TS is at odds with their personal perception of themselves as normal. These social processes contribute to girls’ and parents’ interpretation of the meaning of TS and indirectly contribute to their perception of their information need as they subsequently decide which aspects of the condition are relevant to them.

In summary, meanings of TS came from their knowledge of features and characteristics identified in the published literature which were then refined in light of personal experience of having TS and of parenting a daughter with TS. Confirmation of the diagnosis assigned identities of ‘being a girl with TS’ and ‘being the parent of a girl with TS’. How these new or revised identities are experienced by girls and their parents is informed by an interactionist view of identity, and the ‘self’.

A symbolic interactionist view of identity is “the way an individual defines, locates and differentiates self from others” (Hewitt 1992) and integrates both feelings and thoughts (Burke 1980). Western society places emphasis on youth, attractiveness, active lifestyles and independence, therefore any disruption to these ideals through illness or disability is a significant challenge to the sense of self-identity (Turner 2008). In childhood and adolescence there are also developmental changes to the self, the self therefore can be seen to change with “life-course and disruptions”. TS is such a disruption, therefore viewed in the social world as undesirable because it is not reflective of social norms. Goffman’s (1963 reprinted in 1990) seminal work on stigma
has influenced subsequent theoretical discourse on chronic illness as ‘deviant’ and the stigmatisation of individuals (Charmaz 1991). The hidden and less visible features of TS mean for many there is the option of being selective about information they share with others leaving them either at risk of being socially identified or being defined by their difference. However, Charmaz (1991) argues that when individuals with chronic illness “move beyond loss and transcend negative labels” they define themselves as much more than their bodies and illness. The concept of stigma can be useful in considering the varying perception of girls and parents with regard to their management of different aspects of TS. Closely allied to the concept of stigma is the concept of normalisation. Wolfensberger’s (1972) theory of normalisation will be included in the discussion to explore the concept of normalisation within the context of the experiences of girls with TS and their parents. Within healthcare normalisation has long been associated with explaining how families manage chronic illness, and the defining attributes of the concept were clarified by Knafl and Deatrick (1986). Reference to this work will enable exploration of the similarities and differences for these families compared to those families of children and young people with other long-term conditions.

Uncertainty was a pervasive component within the information activities of gathering and receiving, making sense of and using and sharing information. Information seeking behaviour in parents has been identified as a normal activity and a coping strategy. It facilitates rational problem solving where parents recognise a deficit in their knowledge, which motivates them to search for information (Khoo, Boly, Bald, Jury and Goldman 2008; Plumridge et al. 2007; Hayes 2007). Approaches to seeking information vary among individuals. This partly accounts for the variation across the sources and types of information collated by parents in this study, including individual descriptions of how they managed the quantity and complexity of it.

In summary, the following discussion draws upon the principles of symbolic interactionism recognising that individuals will make sense of their own social world. Consequently priority is given to the perspectives of girls and their parents. Analysis focused on the interpretation of these subjective experiences enabling examination of
how the main themes of uncertainty, normalising and identity apply within the context of TS. The conceptual ideas within the stigma literature based on Goffman’s early work will add to the interpretation and explanation of the major themes of uncertainty, identity and normalising within the context of the perception of information needs of girls with TS and their parents.

7.1.1 Terminology
The table below aims to act as a reference point for the reader, clarifying key terms from the findings chapters which are now used in the following discussion.

Table 30: Key terms

<table>
<thead>
<tr>
<th>Information Trajectory</th>
<th>Pre-diagnostic and initial diagnostic period/“Getting to a diagnosis”</th>
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<tbody>
<tr>
<td></td>
<td>Confirming diagnosis and clarifying profile</td>
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<tr>
<td></td>
<td>Ongoing management</td>
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<tr>
<td>Information Activities</td>
<td>Gathering and receiving information</td>
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<tr>
<td></td>
<td>Making sense of information</td>
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<td></td>
<td>Using and sharing information</td>
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<tr>
<td>Themes</td>
<td>Uncertainty</td>
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<td></td>
<td>Normalising</td>
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<td></td>
<td>Identity</td>
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7.2 Information activities
Three activities undertaken by girls and their parents in responding to their information needs were identified from these data and were utilised in providing an organising framework for the findings chapters. However, these activities were not described by parents as part of a linear process, but rather as activities closely interconnected as illustrated below.
Each activity was not an end point in itself and the relationship between them could create action or no action on the part of girls or their parents. For example, some information was shared although not fully understood or used directly such as information for teachers designed by TSSS; some information gathered was made sense of initially and informed their daughter’s TS profile; partial understanding of information triggered further gathering of information; sharing information with others could assist in making sense of it or prompt the need to gather further information. Parents and girls repeatedly moved between these activities.

7.3 Major themes
These data were dominated by the impact of the diagnosis on issues relating to psychosocial functioning and where possible medical aspects, physical features and cognitive-behavioural characteristics of TS were minimised. This is not to suggest that on-going physical health issues were not perceived as important by parents and girls. Parents commented on difficulties in coping with their daughter’s recurrent ear infections and resulting medical interventions such as surgery. In two families where the girls had co-morbidity which impacted on their day-to-day lives management of their medical needs was a key concern (Case I and J) and one girl of 16 years (Case D) commented about her health and well-being in the future. Nevertheless in thirteen families once the early investigations confirmed there was no life-threatening problems the priority and focus of parenting described in these data shifted to managing the
psychosocial consequences of having TS. Three major themes identified in this study and evidenced across the information activities are:

- Uncertainty
- Normalising
- Identity

These themes appear in published research relating to chronic illness in childhood and adolescence (MacDonald & Gibson 2010; Tluczek, Chevalier-McKechnie and Lynam 2010; Woodman 1999). Consideration of similarities and differences TS has with other conditions will provide context for discussion of these three themes within TS.

### 7.3.1 Relationship between the major themes

The themes of uncertainty, normalising and identity were central to the information needs of girls with TS and their parents. Uncertainty is the most pervasive affecting the information needs relating to identity and normalising. The consequences of the uncommon nature of TS and the complexity and diversity of information led to uncertainty in knowing about TS in general terms and uncertainty in knowing how an individual would be affected. Uncertainty created ambiguity, which enabled a degree of flexibility to negotiate a personal view about what TS meant and how it was interpreted as part of a girl’s identity. Genetic information gave certainty of a diagnosis of TS, however, application of this to individual girls provided uncertainty. This enabled parents and girls to create individual interpretations of their profiles and priorities. Uncertainty offered benefits as well as limitations to parents and girls in normalising features and characteristics of TS enabling them to negotiate their personal identity of being a girl with TS. To elaborate and extend understanding of how these themes connect, detailed discussion of each theme now follows.

### 7.4 Uncertainty

Uncertainty emerges from individual interpretation of a situation as positive or negative (Mishel 1990) and has been linked to the mental health of parents of children with long-term conditions (Holm, Patterson, Reuter and Wamboldt 2008).
For families in this study uncertainty was a constant feature of their daughter’s past, her present and her future. Multiple uncertainties leading up to the diagnosis were expressed. Less uncertainty was expressed by parents about how the condition currently impacted on their day-to-day lives, however, they all expressed uncertainty about their daughters’ future social roles described as anxiety provoking (n=15). In response to questions about parental concern for their daughter’s future there was little comment made relating to long-term physical health issues, with the exception of one family whose daughter had significant co-morbidity. Parents and older girls were unsure about transition from children’s to adult health services, or whether there was a need for lifelong healthcare. Mishel (1990) argues that uncertainty results from ambiguity, complexity, lack of information or unpredictability. Table 31 below provides examples of uncertainty expressed by girls and their parents related to these four dimensions.

**Table 31: Dimensions of uncertainty**

| Ambiguity                                      | Non-inherited nature of this genetic condition |
|                                               | Deciphering natural characteristics from TS characteristic |
|                                               | Perceived discrepancy between phenotype and karyotype |
| Complexity                                    | Inherent complexity of topic areas such as genetics, growth and puberty |
|                                               | Ascertaining what level of detail is needed to understand the full implications of TS for an individual girl |
|                                               | Limited knowledge of health professionals outside specialist tertiary services |
| Lack of information                           | Uncommon condition |
|                                               | Forming and maintaining relationships with knowledgeable health professionals |
|                                               | Life-long need for care |
| Unpredictability                              | Developmental changes as daughter grows up reveals how she is affected by TS |
|                                               | Individual response to GH treatment and final height |
|                                               | Anticipating future social roles |
Five types of uncertainty were evident in these data and two broad types of reaction in responding to it were described. The first was an immediate response aimed to reduce uncertainty by finding out as much as possible about TS. The second involved a less immediate reaction whereby information was managed in a more systematic and selective manner focusing on their individual circumstances and related information need.

Table 32 summarises key findings relating to ‘uncertainty’. In the right hand column of the table the five types of uncertainty described within the findings are identified. These types of uncertainty will be discussed individually. Similarities and differences between these findings and published research will be examined leading to new insights into issues involved in the information needs of girls with TS and their parents.

Information is a major component that influences whether parents and girls with TS perceive something as certain or uncertain. Within this study health professionals encountered by parents and information provided by TSSS were instrumental in influencing parents to frame aspects of TS as either positive or negative. The main source of information identified by girls in this study was their parents. They influenced directly their daughter’s knowledge and appraisal of TS. Delivery of relevant and understandable information helped parents to appraise and reappraise what TS meant to them, which in turn influenced their daughter’s perception of the condition.
Table 32: Summary of key findings: uncertainty

<table>
<thead>
<tr>
<th>Information Activities</th>
<th>Key findings for discussion</th>
<th>Types of uncertainty</th>
</tr>
</thead>
</table>
| Gathering and Receiving (Chapter Four) | Initially overwhelmed by nature of information particularly relating to genetics and infertility  
Information received is complex  
Selective and systematic in gathering information | Uncertainty related to understanding complexity and diversity of information  
Sub section 7.5.1 |
| Making Sense of Information (Chapter Five) | Endocrinologist acts as mediator for complicated information and diversity of issues  
TSSS acts as mediator of information relating to social dimension of TS  
PENS role in teaching the administration of GH to families  
Siblings act a source of reference for family characteristics and developmental norms | Personal uncertainty relating to breadth of knowledge required in understanding the broad nature of TS  
Sub section 7.5.2  
Societal uncertainty relating to knowledge about TS  
Subsection 7.5.3 |
| Using and Sharing (Chapter Six) | Parents are gatekeepers of information for their daughters and for others – medical and non-medical information  
Girls are a source of information and parents are interpreters of daughter’s characteristics  
Uncertainty provides an opportunity to negotiate meaning of TS  
Parents seek reassurance that they are doing the right thing  
Girls are selective about sharing information with friends, uncertainty in peer response | Uncertainty about what information is applicable to an individual girl  
Subsection 7.4.5  
Uncertainty about using and sharing information to benefit girls with TS  
Subsection 7.4.6 |
7.4.1 Uncertainty related to understanding complexity of information

TS is an uncommon condition. Only one mother in these fifteen cases had heard of it prior to the diagnosis and her information was very limited

I said well I’ve heard of it but I don’t know much about it, all I know is that the children died or they were infertile (EP:L7–8)

Initial unsystematic searching for information from the sources identified in Chapter Four led to many families feeling overwhelmed by the nature of the information they discovered. These sources of information contributed to how parents and girls (depending on age at diagnosis) began to perceive aspects of TS. Parents (with exception of Case I) viewed information from published sources or on the internet as reliable and were open to the information these sources provided. Early in the information trajectory parents met health professionals working in primary and secondary care settings such as doctors and midwives who in some cases demonstrated scant knowledge of the condition or its implications. Incomplete, inaccurate information contributed to uncertainty about what TS meant. These sources of information had a positive or negative impact on parents. Parents indicated which source had minimised their uncertainty through information provision and which had contributed to increasing uncertainty. Appraisal of how threatening aspects of TS were to them appeared to be significantly influenced by the positive or negative attitude of those providing information in addition to the complexity of the content of the information. Some sources changed from being a negative influence to a positive one for example, once parents used reputable sites on the internet rather than a general search engine, the internet was appraised more positively.

7.4.1.1 Mediating information: doctors

Information provided some parents with a sense of control but others portrayed a sense of bewilderment about the complexity of the information. Many aspects of the content of information about TS were considered by parents to be complex and with complexity came the potential for uncertainty and misunderstanding.
TS is further complicated because of the wide-range of potential features and characteristics that girls may or may not present with. This also provided opportunities for error in parental interpretation of independently sourced information and its relevance to their daughter’s specific case. The subject areas involved are inherently complex for example, genetics, physiology of growth, and puberty. The consultant paediatric endocrinologist therefore had to address a key tension between providing adequate information at the right level to ensure the family were fully informed without overwhelming them. The consultant paediatric endocrinologist was cited as the main source of clinical expertise by all families. They became an important focus in reducing uncertainty and mediating information for girls and their parents from the point of diagnosis onwards. The majority of parents expressed that they were also the preferred professional to be present when any changes to treatment were to be decided. Some families explained this preference as a result of the specialist nature of the condition and the need for an experienced clinician. Theoretical knowledge was important to these families but experiential knowledge through clinical experience was also highly valued:

not to be critical of the people we see who rotate through but y’know it’s about having someone that’s got the T shirt (KP/R1:L662)

And someone in whom they could trust

Its trust, and that’s been built up over well nearly eleven years ten and a half eleven years now (JP/R1:L459)

However, for some parents, clinic itself was considered to be a centre of expertise and they valued the consultation with all medical staff irrespective of status. They placed importance on good communication skills of the doctor. These parents viewed the clinic visits as a monitoring exercise whereas parent seeking the consultant specifically raised issues such as fearing unplanned disclosure of information about infertility by doctors who did not know them or lack of continuity because other doctors would not know their daughter. Personal qualities of the consultant demonstrating kindness, empathy and personal knowledge of individual girl’s circumstances were reported as important aspects in developing and maintaining this long-term relationship.
Chapter Four illustrated that parental response along the information trajectory shifts from unsystematic gathering of information to more selective and focused gathering as they started to make sense of information and filter out what they perceived to be relevant to them. The more expert the source the more relevant the information appeared to become and the more the positive and reliable information was perceived to be. The two expert sources were identified as the paediatric endocrinologist and the TSSS. These two experts could help parents personalise general information about TS to individual girls. As parents became more confident and knowledgeable they also became more selective about whom they considered to be a reliable and knowledgeable source of information.

7.4.1.2 Learning new skills: the role of the PENS

Educating, teaching and providing information to CYP with chronic illness and their families is a recognised part of the nurse’s role (Coyne 1998; Fisher 2001; Hopia et al. 2005). It is a responsibility that is particularly evident in the role of clinical nurse specialists working with CYP with a range of chronic conditions and their families. Swallow, Clarke, Campbell and Lambert (2009) cite Allen (2004)’s observation that nursing work includes functioning as a healthcare mediator. However, in the care and management of girls with TS they had a very specific and defined role relating to one aspect of the condition, the administration of GH. A longitudinal qualitative study of family learning explored nurses’ involvement in teaching families to share chronic kidney disease management of their children (Swallow et al. 2009). Five families and four clinical nurse specialists involved in their care participated in semi-structured interviews, other data sources included four mother/child diaries and 19 post interview case-notes reviews. Analysis revealed five teaching activities which included assessing learning needs, creating learning opportunities, implementing teaching strategies, acting as interpreters and ambassadors and assessing learning progress. Elements of these teaching activities were described by families in the current study but they referred to a discrete area of learning: all aspects of the administration of GH.

Initial contact with the PENS coincided with commencement of GH. GH is generally started when girls are 4–5 years old. The age at which the girls met the nurse and the
point at which nurses entered the information trajectory depended upon the age of diagnosis and the age at which GH started. As a result where girls were diagnosed in infancy there was no reported input from the nurse until the girl was 4–5 years old, whereas girls diagnosed older than 5 years would meet the PENS close to the point of diagnosis because GH was prescribed immediately. This differs with other chronic illnesses in childhood where the clinical nurse specialist is involved from the point of diagnosis to support, educate and manage care with the families.

The PENS was introduced to the family with the express purpose of teaching the family about all aspects pertaining to the administration of GH. Teaching took place in the family home and once this was achieved, nurses were not perceived as being needed unless there were problems connected with GH. Girls and parents report the PENS as being relaxed, and confident, and able to adapt her approach to teaching in response to their learning needs; they stayed until families felt confident and were assessed as competent and they also contacted them to check they were managing everything satisfactorily during the first few days. The girls who were considered old enough or well-motivated were taught to administer GH alongside their parents. Girls who were too young or did not want to do it were taught by their parents at a later date when it was considered the right time by girls and parents. The PENS did not revisit the family so these girls had no formal teaching or information outside the family. There did not appear to be a desired age at which girls would be expected to administer their own GH. The initial experience of learning how to administer GH was considered stressful for some families and the PENS demonstrated flexibility in teaching style to match the abilities and emotions of girls and parents. However, they did not describe them acting in the roles identified by Swallow et al. (2009) of interpreter between families and medical staff or ambassadors with school staff. The personal qualities of the PENS in the current study were described as “good”; “patient”; “lovely”; “supportive”. Across these data it is clear that parents and girls valued the teaching and support from the PENS, however, her role was specific and transient as illustrated below:

... but we don’t need her now but when we did she was really really good, I mean she was here with us all morning, all one Saturday morning. She went through it and through it and not once did you feel that you were y’know putting
on her or she was bored with saying it y’know. I was hurting my daughter with these injections and I really didn’t want to do it. (PP/R1:L650)

This significant encounter between nurse and parent changed the parent’s identity toward one of a parent with a child who needs medical intervention as part of their life. For the girls learning to administer their GH it was a move toward independence in managing a significant aspect of their condition.

Hodgkinson and Lester (2002) identified that nurses could act as mediators. They examined stressors and coping strategies used by 17 mothers living with a child with cystic fibrosis and identified that nurses were the first professionals these mothers turned to when they were concerned. In addition the mothers saw a critical aspect of the nurse’s role as interpreting information, “the bridge between the medical and non-medical worlds”(p381). These families as with families in Swallow et al. (2009) study have children with complex conditions that require families to engage in daily “intensive management” of care which influences stability of the condition and survival of their children. In the organisation of children’s services it may be that the immediate physical health and medical needs of children dominate the quantity and typology of nursing care and girls with TS do not fit this clinical picture.

Girls with TS rarely have periods of hospitalisation related to their condition or daily threat of physical crises as seen in chronic conditions such as asthma or diabetes. Many of their health needs will not manifest until they are possibly into adulthood. Another point to raise here that will be revisited in depth when considering how girls and their parents normalise TS is that not needing a nurse regularly potentially assists in minimising of the medical aspects of the condition. Nurses are traditionally considered as being needed to care for those perceived as sick.

The variable point at which the nurse becomes involved in the care of these girls undermines their effectiveness as credible sources of information outside administration of GH. PENS are specialist nurses within endocrine services but they are not specialist TS nurses. However, further research to determine how PENS in other specialist centres work with girls with TS would be needed to draw any clear conclusions from this observation. There is some suggestion in these data that communication and parents’
understanding could be enhanced by closer input from the PENS. Parents did refer to the complexity of information and purposeful nature of the medical consultation. There was a short time frame for sharing information with the doctor and some parents stated that it would be useful to have someone with whom they could talk things through outside the medical consultation, therefore the role of the PENS is worthy of further detailed exploration.

7.4.2 Personal uncertainty

Closely allied to complexity is the diversity of TS. Inadequate information can contribute to parental stress (Franck, Cox, Allen and Winter 2004), however, too much information can confuse resulting in stress and anxiety and lack of confidence (Fisher 2001). Parents in the current study referred to difficulty in explaining aspects of the condition to others because of the complexity resulting in uncertainty in their personal understanding of the information received.

To develop understanding of the diverse nature of the condition, the quantity and quality of information needs to be relevant and accurate because both have been shown to directly trigger uncertainty (Mishel 1990). There is a wealth of available information about TS and parents expressed uncertainty in assessing what they need to know, how to sift relevant information and feel confident that they know enough.

7.4.2.1 Managing the breadth of available information

Over time parents became more experienced, learning to use information sources more selectively and systematically. Gathering and receiving information was seen as an ongoing process with varying degrees of activity in response to different stimuli such as developmental change, outcomes of treatment, and increasing parental confidence in managing the condition. As already identified, the two expert sources cited by parents as key to developing their understanding were the consultant paediatric endocrinologist who addressed aspects of the syndrome that were physiological and required medical intervention, and the TSSS, which helped to clarify general medical information in more accessible language and provided valuable practical information for parenting a girl with TS. Some parents cited Arlene, the founder of the society, as providing
emotional support particularly around diagnosis. The TSSS also offered social support to families through organised events.

Hummelinck and Pollock (2006) explored the diverse nature and resulting complexity of parents’ information needs about the treatment of their chronically ill child and how they evaluated information. Parents of children with asthma, cystic fibrosis, diabetes, epilepsy, leukaemia, other cancers and severe eczema reported information needs varied greatly between individuals and across time. Parents wanted to know everything so that they could prepare for the future; parents need for information varied considerably and included the ability to manage their child’s illness and understand decisions made in an attempt to re-establish control. Control enhanced their ability to cope, to be able to fully answer their child’s questions and to come to terms with the diagnosis. Parents reported professional communication and information provision to be inadequate, a recurrent theme in the literature (Beresford 1997; Hedov et al. 2002). Differences were reported across the diagnoses for example, in conditions such as cystic fibrosis, parents reported “information overload” whereas those with children with epilepsy reported insufficient information irrespective of the severity of the child’s illness. It was suggested the difference was related to being managed in primary or secondary care. Seeking out information independently had the potential to cause tension as it could be interpreted as corroborating with or undermining professional judgement. My findings echo some of these results but there are also key differences some of which may reflect that all the parents and girls interviewed were attending a specialist clinic in a tertiary centre. These issues are now examined further.

7.4.2.2 Motivations for developing broad understanding

A similarity with Hummelinck and Pollock’s findings was parents’ desire for information they understood so that they could explain aspects of the condition to their child. This was significant as the girls in the current study identified their parents as their main source of information.

Starke and Moller (2002) investigated the motives and strategies of parents in seeking information about their child’s medical condition and the implications this may have for
their child also concur with this finding. It is useful to compare the outcomes of this study with the current findings because there is little published work relating to information needs of this specific group. Other motives for selecting information included lack of information provided by the doctor, doctors’ lack of knowledge about the syndrome, to learn more about the condition, to be able to explain the condition to others, and to control what the health professionals told them including recent research and recommendations (Starke and Moller 2002). Two mothers in the current study demonstrated a need to use information to maintain some control in decision-making with doctors. The first quotation reflects preparing for clinic appointments by reviewing their child’s weight and height since her last visit and checking what investigations had been done at the previous appointment.

you put yourself in their hands but I can’t imagine doing that completely, well I mean you’re guided by them at the end of the day it’s what they decide but you’re more equipped I think.(KP/R2:L608)

The following quotation is from a mother who also indicated information is shared with the endocrinologist so that they can clarify the information’s relevance to her daughter,

Internet, fantastic tool. If I didn’t have that I’d be reliant on doctors. First port of call because I knew I would get information from it but then it is nice to have the personal touch with someone to clarify everything for you as well.

(HP:L372)

7.4.3 Societal Uncertainty relating to knowledge about TS

There were differences between parents in these data with some wanting to raise the national profile of TS, to promote understanding of the condition more widely in society and others who did not want features and characteristics of the condition widely known, because this could lead to misunderstanding about their daughter’s specific profile.

7.4.3.1 An uncommon condition

Diagnostic certainty is considered to be beneficial to the psychological health and well-being of parents and uncertainty rises when a diagnosis is unclear or non-specific (Mishel 1990). However, in a diagnosis of TS there appears to be some benefits
associated with specific aspects of uncertainty. Uncertainty provides parents with an opportunity to individualise TS incorporating certain features and rejecting others.

There were varying degrees of uncertainty expressed amongst parents about the response of others to their daughters once they knew she had TS. Parents generally anticipated negative responses from other children and were concerned as to whether girls would be treated differently by significant adults’ particularly family members or teachers. One mother explained this had happened in relation to the genetic nature of the condition. She reported that people could not understand that it was a genetic but not a hereditary condition. The rarity of TS also had implications for identity which are discussed below.

**7.4.4 Uncertainty about what information is applicable to an individual girl**

Focused appraisal of information in terms of its relevance to an individual girl was undertaken by parents and professionals once the diagnosis was confirmed. The girl who was diagnosed as a teenager also considered the relevance of certain aspects of TS to her and older girls made comparisons between how they perceived themselves and what the literature said about girls with TS. This is examined further in the Section 7.7 about identity. For some families there was a perceived mismatch between their perception and assessment of their individual experience of TS when compared with descriptions of the features and characteristics of TS. As a result in some families ambivalence toward the formal diagnosis of TS was expressed by girls and their parents.

**7.4.4.1 Source of information: daughters with TS**

Parents’ observations led them to develop subjective opinions about how affected their daughter was by TS. Information was used to individualise the diagnosis and parents reported looking for consistent evidence to support and reinforce the diagnosis of TS. When unsure about their daughter’s progress or behaviour, parents used siblings or their daughter’s friends as a source of reference with which to make comparisons. Strategies were subsequently employed to optimise their daughter’s potential and promote coping of immediate and deferred health and social aspects of the condition. Making sense of
information and their observations was frequently a trigger for seeking out further information.

7.4.4.2 TS as a lens through which to view their daughters

The findings illustrated that parents used the information they gathered about TS to review their daughter’s known features and characteristics. Dominating these data was the social dimension of TS. This aspect created anxiety for parents because of uncertainty about their daughter’s actual or potential social functioning and how this could impact on her both now and in the future. Potential social functioning was a significant source of uncertainty, particularly when the diagnosis was made early in the girl’s life. Two families identified it as one of the worst features of the condition.

The later the diagnosis the lesser was the certainty associated with expected characteristics of TS for example, the girl diagnosed as a teenager was good at maths, achieved the highest grade in mock GSCE and was intending to study it at A level. There had never been any judgement made about her ability related to TS, whereas all the parents of girls diagnosed in early infancy or childhood anticipated maths would be a problem and even when it turned out not to be, they still commented on her ability in relation to the diagnosis pointing out it was unusual for a girl with TS not to have a problem. The mother of the girl diagnosed as a teenager had also not experienced any anxiety around her daughter making friends. She did identify two types of girls at school – the cool ones with boyfriends and the non-cool ones without. Her daughter she said was in the non-cool group with other girls who were not yet interested in boys. However, when talking about her daughter going to university, she also made a comment that was the antithesis to the uncertainty expressed by other parents with regard to socialising and potential for girls with TS to struggle in social situations, she stated

I just hope she doesn’t get in with a fast drinking, fast living crowd and erm I don’t know if she’ll be able to have normal sexual intercourse I don’t know exactly about that (EP:L164–5)
Two families in this study were at the extremes of the diagnostic spectrum, one occurred prenatally and the other in adolescence. There do appear to be some differences in these families with regard to their approach, experiences, need for and use of information. For the child with a prenatal diagnosis TS was an integral part of her identity for all of her life whereas the girl with a diagnosis in adolescence stated “until I found out I didn’t think there was anything wrong with me apart from I was small” (EG/L292).

7.4.5 Uncertainty about using information to benefit individual girls

Uncertainty was evident in the process of using and sharing the information. Some information was easier to share than others.

7.4.5.1 Public and private information

Findings in Chapter Six reported a range of views about sharing information about specific aspects of TS with others. Information perceived to be sensitive such as chromosomes and infertility was treated as private information being shared only with family or close friends. Information about short stature, GH, and learning difficulties was shared with others such as school teachers or adults organising recreational activities, e.g. girl guides, dancing classes. Only one mother stated sharing information outside the family about probable infertility as she pursued policy change on donor egg storage.

Decisions about sharing information with their daughters were influenced in part by the need to control this public–private divide. Once children knew something they may choose to disclose information to others regardless of their parent’s wishes. Uncertainty about the response of others made some parents hesitant to share all information relating to TS with their daughter or close friends and family. Social support is important to families in adapting to an unexpected diagnosis of any sort, however, anxiety about the response of others potentially may reduce the support available to families fearing negative responses. Other families were not concerned about negative responses but genuinely did not share information because they did not see its relevance to their daughter.
7.4.5.2 Managing multiple uncertainties: timing, content, ownership

Parents were the most important source of information about TS for girls participating in this study. The point of disclosure of TS to the girls in this study was rarely at the point of diagnosis. Parents had to determine what information they considered suitable to share with their daughters at particular ages and how much detail to provide. As reported in the findings parents wrestled with decisions about who they should share information with, what information was to be shared, when it was appropriate to tell others, and how they should share information. They described their role in terms of interpreting and mediating information they had gathered or received. They knew their daughter best and wanted to provide information in a way she would understand and at a time that was appropriate for her stage of development and that was accurate for her specific circumstances. As parents developed their expertise and understanding of TS they developed different strategies for sharing information with their daughters and others who in turn may help her, e.g. teachers. This experiential approach led them to seek reassurance that they were doing the right thing.

7.4.6 Summary

Dealing with uncertainty varied across families and between members of the family. Different actions were taken by parents that included negotiating and individualising the condition; taking direct action to reduce uncertainties; selectively disclosing information; minimising engagement with complex, or sensitive information including information that may be relevant in the future but was not currently a concern. Parents also sought reassurance that they were ‘doing the right thing’ with disclosure of information to their daughters.

7.5 Normalising

The findings chapters presented data illustrating degrees of normal described by parents and girls with TS ranging from no visible features or sense of being different to those with considerable co-morbidity, visible physical and behavioural features, and moderate learning disability. This has implications for information needs and priorities for information. Three different purposes are offered here to explain the normalising
approaches adopted by girls and their parents described in these data. These are included in the right hand column in the table below and form the structure for the following discussion.

Table 33: Summary of key issues relating to normalising

<table>
<thead>
<tr>
<th>Information Activities</th>
<th>Key issues from findings</th>
<th>Purpose of Normalising</th>
</tr>
</thead>
</table>
| Gathering and Receiving (Chapter Four) | • Determining visibility of features and characteristics  
• Ambiguity about TS status | • Develop a frame of reference for TS |
| Making Sense of Information (Chapter Five) | • Determining usefulness of the diagnosis  
• Minimising the features and characteristics of TS | • Determine what TS means for social identity |
| Using and Sharing (Chapter Six) | • Reveal or conceal: continuum of information sharing  
• Tension between wanting to be given special treatment and not being treated differently; separating health and social needs associated with TS and being a normal girl | • Reconciling TS as part of girl’s identity |

The concept of normalisation offers a way of explaining how families manage chronic illness as an integral part of their lives in order to maintain normal family life and is frequently reported as a valued aim for families (Knafl and Deatrick 1986; Robinson 1993; MacDonald and Gibson 2010). The defining attributes of the concept include:

- Acknowledging the condition and its potential threat to lifestyle.
- Engaging in parenting behaviours and family routines that are consistent with the normalcy lens.
• Engaging in parenting behaviours and family routines that are consistent with normalisation.
• Developing a treatment regimen that is consistent with normalcy.
• Interacting with others based on a view of the child and the family as normal.

(Deatrick, Knafl, and Murphy – Moore 1999)

Normalisation in this form is reported to provide a useful coping strategy whereby families focus on their similarities rather than their differences to other families.

Normalisation is described as a cognitive process that focuses on families perceiving and defining their lives as normal. However, this study suggests normalisation also incorporated the idea of making the girls look as normal as possible thereby reducing the potential of social difficulties. Medical management aims to normalise the girls’ bodies for example, treatment of short stature with GH which affords an opportunity to achieve heights within normal female parameters, pubertal induction coincides with the physical development of peers. Introduction of potential options to enable girls to have families of their own in the future offers hope for future functioning in anticipated normal female roles. In these data normalising was also closely associated with the girl’s identity. How these ideas relate to theories of normalisation more generally are integrated into the following discussion.

7.5.1 Developing a frame of reference for TS

Summarised in the following quotation is a tension at the centre of the theme of normalising between being a girl with TS and being perceived as a normal girl:

It’s hard to reconcile the normal child and treating her normally without trying so hard to make her normal that you’re actually then not making allowances for the Turner’s (KP/R2:L749)

Earlier discussion explored how uncertainty enabled flexibility for interpretation of TS. These interpretations were influenced by how normal parents perceived their daughter to be. The pervasive nature of uncertainty enabled some families to maintain TS in a state of ambiguity which supported their ambivalence toward the diagnosis. Other
families embraced the diagnosis and utilised all available information to optimise their daughter’s learning and social opportunities. These issues are now examined further.

7.5.1.1 Ambiguity in being normal

Ambiguity about what it means to have TS confronted some families in the first few weeks of their daughter’s lives. One set of parents recall the paediatrician who in an attempt to reassure them stated “she’s a beautiful healthy little girl really apart from she’s got TS” (PP/R2:L34). This statement serves to introduce doubt into the relationship between TS and health/ill health. It appears to minimise the medical implications of the condition and also the statement suggests that the girl can “pass as normal”, linking the concept of normalisation with that of stigma. The concept of normalisation has long been associated with stigma and was examined in a recent study to increase understanding about the impact of stigma and normalisation on parents of children requiring complex care (MacDonald and Gibson 2010). In-depth interviews with 47 participants including parents, grandparents, nurses and social workers revealed that normalisation was used as a strategy to counteract stigma rather than as a coping strategy. Data in this study also evidenced normalisation being used in this way. The following overview of key developments in the theory of normalisation helps explain the relationship between these two concepts.

The theory of normalisation originated in Denmark in the 1950s specifically to challenge the organisation of services for people with learning disabilities. Further development saw the emphasis on securing legal and human rights for individuals with learning disabilities in line with other members of society. It was a humanistic approach that emphasised freedom and self-determination in order to maximise quality of life (Nirje 1972; Emerson 1992). At the same time in North America, the influence of the civil rights movement had led to challenges about the use of prolonged institutional care for people with learning disabilities and mental health problems. A detailed definition of normalisation was developed by Wolfensberger (1972), who based his work on then popular theories of deviance. The basic aim of normalisation he proposed was to change the social status of any devalued group, to see all individuals as having the potential for growth and development. To achieve this changed status the individual was required to
conform to certain cultural norms. Critics of Wolfensberger’s theory argue that difference is denied in favour of conformity. In the 1980s Wolfensberger (1983) renamed his normalisation theory calling it the role valorisation model. This followed considerable criticism centred on the concept of normality nevertheless the aim of the theory remained value-laden and focused on making the individual acceptable to wider society through acquisition of socially valued roles in that society.

The symbolic interactionist perspective considers that an individual develops from the process of social interaction. Within any social interaction decisions about acceptance are uncertain, reliant on others in society and their interpretation of normal. The perception of being normal was important to several families and girls interviewed in the current study and the term was utilised in different ways by the parents and girls. In this study normalisation brings together issues relating to family life, life-style and the girls’ bodies, in terms of their visible appearance and function. Parents, doctors and girls made efforts to conform to society’s expectations of family life, behaviour at school and with peers, and use treatments to normalise aspects of the girls bodies both the unseen and seen.

7.5.1.2 Distancing themselves from recognisable features

One strategy described by families was to identify the differences between the girl with TS and the comprehensive profile of TS presented in the literature. The main source for this was the TSSS book (2008). Information and knowledge about TS was important in the process of distancing girls from the perceived negative aspects of the diagnosis. A good understanding of the condition placed parents in a position to recognise aspects of development as normal or abnormal, however, their subsequent interpretation was often equivocal. Utilising information about TS, girls and parents compared their behaviours and abilities with expected or known characteristics of TS. These comparisons contributed to the girls’ self-identity and the identity assigned by those knowledgeable about TS, such as medical staff and the TSSS. Families interpreted the narrowly defined norms for TS assigned by the experts and placed it within their definition of TS. This personalised definition of TS incorporated their daughter’s experience of living with TS
in addition to personal social and cultural expectations of what ‘being a normal girl’ entailed.

Ambiguity was present in all sources of information gathered and received from the pre-diagnostic period onwards as parents and girls identified whether the broad features and characteristics had individual relevance or not. However, uncertainty lessened as the girls grew up and new characteristics particularly relating to behaviour were no longer emerging. Where the girl’s phenotype was perceived as mild by the parents they commented on the differences between their daughter and the stereotypical features and characteristics of girls with TS. They distanced their daughter from the diagnosis explaining why she was not like girls with “full blown Turner’s”. This tendency was also evident in the girls’ descriptions of what TS meant to them. This decentring of the condition is reported in published studies where families with children with other chronic illness avoid the full implications of the condition, in an attempt to minimise the perceived effect of the condition on the child’s self-identity (Bluebond-Langer 1996).

Minimising the diagnosis and its implications for their daughter is reinforced by a dialogue that involved parents stating their daughter is normal and then providing information about features that inadvertently define her as a girl with TS for example:

“she’s a normal girl apart from she’ll be shorter than her friends and she can’t have children”(DP/R2:L19)

Normalising identity in this way seemed to be more important in families who perceived their daughter as almost normal and not completely TS. Robinson (1993) suggests normalisation in this way can have potential costs for the affected individual and their family as potential problems are not shared with health professionals.

This attitude to the diagnosis was also evident in the girls’ data where they described themselves as normal, same as their friends adding a qualifying statement about height or induction of puberty. The word normal was used to describe themselves and was also used by their parents when describing their daughters. This is a value-laden term not easily defined, however, in this context it appears to mean the girl was comparable with peers physically and socially, and had the potential to adopt the normal social roles for
women in the future. The use of the term normal implies that if they are not normal then they are abnormal. The dictionary definition of normal has a health component, which may be why it is used whereas behaviours, and hopes for the future, are less about normality and abnormality and more to do with being ordinary. In a study of children aged 7–17 years with cystic fibrosis, the researchers identified two conceptualisations of normalities from their data, the first was “self-referential” and the second was “socially derived”. Self-referential was based on an individual concept of self which embodied past and current experiences and future hopes and expectations. The socially driven concept included various views and expectations from others (Williams et al.2009). Williams et al. (2009) suggest this may lead to differing definitions and possible tension associated with social expectations and norms. This has resonance with findings in these data and questions the premise of social expectations and norms that only value those who conform to normal roles. This is a criticism of Wolfensberger’s theory, which is perceived by some as supporting the status quo, as it emphasises the need to be similar rather than different and in doing so stresses that being different is less desirable than being normal. The parents (Case N) who received a prenatal diagnosis appear to reject the notion of normalising through conformity and instead celebrated their daughter’s difference. This will be examined further when considering identity.

7.5.1.3 Visible differences

Some parents were concerned about how visibly different their daughter appeared to be to others. They raised concerns about recognisable stigmata that may make their daughter vulnerable to negative attention or that the cognitive and behavioural profile associated with girls with TS may result in her being noticeable to others. However, this was not consistent with descriptions given by the same parents of their daughters as ‘normal’ or their expressed lack of concerns about her becoming independent and forming relationships in the future. This supports the point earlier that ‘normal’ in TS is a qualified normal relating to function and role, to visible differences and unseen differences. It is these differences that separate girls from being normal and on which families and clinicians focused efforts of normalising. The presumption being that these differences would be perceived as negative and limit the girls’ life chances.
However, a study examining the perceptions of fifty healthy children between ages 8 and 12 years indicated that when they were given medical explanations about peers with visible characteristics of illness they viewed them positively across affective, cognitive and behavioural domains as they did children with no visible sign of their chronic illness (King, MacDonald and Chambers 2010). It is suggested that the provision of information invoked sympathy for the affected child. This idea is mirrored in these data where girls who had told their friends about their GH injections reported friends expressing sympathy or telling them they were brave to have daily injections. Older girls experienced support from friends who reassured them during the induction of puberty.

Not all girls with TS have physical stigmata that would be obvious to those who knew nothing about TS. Some girls’ have specific difficulties in recognising facial expression which can make interactions with peers difficult as they may misinterpret responses and react inappropriately (TSSS 2008). Research investigating friendships for children with chronic illness often focus on physical conditions and as a result identify limited physical ability, prolonged or repeated absence from school due to ill health that interrupts social activities, time consuming or complex treatment as problematic (La Greca, Bearman and Moore 2002). These issues do not generally apply to these girls who did not need to control symptoms on a daily basis or experience recurrent exacerbations of symptoms associated with their condition. Administration of GH was not a particularly time consuming activity and parental attitude to administration was flexible so that their daughter could participate in social opportunities without revealing this difference. For example, the findings identified parents occasionally sanctioned the omission of GH injections when their daughter was on sleepovers. Managing the treatment regime to ensure daughters engage in normal activities is an example of parents’ adaptation to the demands of the condition, enabling them to normalise their life-style and family life.

7.5.2 Determining what TS means for social identity

Parents and girls described themselves as doing and desiring the same things as other girls of similar age. Activities perceived as normal for girls without TS were
encouraged. Parents strived to ensure their daughter was exposed to a variety of social activities focused mainly on building her self-confidence and self-esteem, places where she would have opportunities to develop friendships and improve social skills.

7.5.2.1 Usefulness of the diagnosis

Interpretation of the diagnosis by parents was seen to facilitate as well as hinder social development. Collating information was the starting point for parents in comparing and contrasting the objective published literature with the subjective day-to-day realities of their own lives. This resulted in judgements being formed about the severity of the condition for them as individuals and the degree to which they were prepared to sign up for the diagnosis. Older girls also expressed reservation about using TS as a diagnostic label because they felt it misrepresented them in a negative way.

The broad range of features and characteristics fell into two interrelated spheres of influence on the management of TS, the medical and the psychosocial. These appeared to be treated as separate entities by the parents for the purposes of finding out about the condition and as already illustrated were headed up by different types of expert: the clinical expert and the lay expert. Input from these experts assisted in the development of holistic understanding of the condition. However, when parents or girls add their expert knowledge of themselves or their daughter there was not always a connection or relationship between the TS expert and the families particular experience.

Gunther et al. (2004) describe ascertainment bias in current understanding of TS which may offer some insight into this mismatch for some families. They analysed baseline data which comprised of medical history, presence of phenotypic features, length/height and weight from 88 girls between the ages of 9 months and four years old who were randomised into a multicentre GH intervention trial. Girls were assigned to one of two groups, incidental or traditional. The incidental ascertainment group (n=16) was made up of girls diagnosed by prenatal karyotype analysis undertaken for reasons not connected with a suspicion of TS; and the traditional group (n=72) included girls diagnosed following prenatal analysis due to abnormal ultrasound (n=19) or postnatal karyotype undertaken in response to clinical findings suggestive of a chromosomal
abnormality. Their results identified significant differences between the two groups with the 74% of the traditional group having non-mosaic 45X karyotype and 54% of the incidental group having mosaic 45X/46XX karyotype. Results showed that the girls who were diagnosed incidentally had a “milder” form of TS than those diagnosed clinically. TS has been described largely by the non-mosaic Turner population and therefore Gunther et al. (2004) suggest that the phenotype typically associated with TS karyotype is exaggerated hence for some girls the information does not fit their individual profile.

7.5.2.2 Minimising the social consequences of the diagnosis

When chronic illness is perceived negatively there is the potential for individuals to experience stigma. Goffman (1963 reprinted in 1990) proposed in his seminal work on stigma that society categorises individuals on the basis of normative expectations. An individual can be stigmatised by physical characteristics and behaviours or by membership of ‘marginal’ social groups. If TS is viewed negatively then there is the potential for stigmatisation to occur. The social construction of stigma relies on aspects of individual functioning that are valued by society to be missing. Stigma is therefore considered to be located in the individual. Girls with TS may or may not have visible characteristics relating to their physical appearance and behaviour and sometimes when they do, they are subtle and only discerned by those who have a good knowledge of the condition. The fewer the visible aspects the more of a choice there is to disclose information to others. Goffman (1990) describes the disparity between an individual’s actual social identity and their virtual one as being discredited or discreditable. Discredited is the term applied to those who have been judged and marginalised; discreditable when an individual has not yet revealed their full identity and therefore have not yet been discredited. In these cases the pressure is on managing information

“To display or not to display; to tell or not to tell; to let on or not to let on to lie or not to lie; and in each case, to whom, how, when and where” (p57)

Decisions to reveal and/or conceal the diagnosis involved a complex mix of competing motivations for girls and their parents and are discussed in the next section.

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Parents were anxious that their daughters would be bullied and would not make friends in line with potential social difficulties identified in the TS literature. School environments and the potential for bullying appeared to be a common source of anxiety although only two out of fifteen families reported any direct experience of being bullied. One of those two was related to being small rather than directly to TS. This fear is an example of felt stigma, “fear of an undesired differentness” (Goffman 1990) rather than enacted stigma which is the description given to the experience of direct discrimination.

To counter any potential social problems a strategy identified by several parents was on taking action to improve their daughter’s peer relationships and social skills by encouraging activities outside the home and engaging their daughters in multiple activities. This strategy and subsequent social engagement is not reflected in earlier studies with women with TS. These women are described as being socially anxious with poor communication skills resulting in increased dependency and living at home with parents. This type of social exclusion is identified by Goffman (1990) as a consequence of being discredited. However, although developing intimate partnerships and becoming independent was articulated as an area of concern by some parents in this study, the majority talked of their daughter’s leaving home and having independent lives. This suggests a generational divide in the literature which does not necessarily reflect the experiences of girls more recently diagnosed with TS, who may have benefited from changes in treatment such as early administration of GH, pubertal induction in line with peer development and a strong support group (TSSS).

The act of normalising the social activities of girls allies them with normal girls and counteracts the potential to be viewed as different. However, there is a risk of being identified as different if the girls are not able to participate fully or achieve the necessary skill level comparable with peers. Parents explained that they selected activities in two ways, firstly in response to their daughter’s interest and secondly activities that their daughter was good at. Where the activity proved to be difficult the girls stopped attending and tried something else. In younger girls the parents were also able to manipulate friendship groups for their daughters, placing them in activities where the opportunity to socialise was possible.
Recognising that girls with TS may find social situations difficult a key aim of the TSSS was to provide social opportunities. However, accessing TSSS was dependent upon parental decision to do so and there seemed to be a tension for some families between acknowledging their daughter as a girl with TS and stepping into a community of girls with TS. Girls who had experienced being with others at the TSSS events reported the experience positively. Three mothers (Case B, H, L) stated they thought it would be useful for their daughters to talk to other girls with TS or access a website where they could communicate with others who could share the experience of having TS, to promote a sense of belonging with other girls with TS. However, for some parents mixing with other girls with TS was perceived negatively and parents preferred their daughter’s to socialise with non-TS girls. Goffman (1990) explained that friendships with “devalued people” reinforces stigmatised identities because it illustrates an inability to integrate with normal individuals, which in turn reinforces the stigmatised identity rather than developing normal social relationships.

7.5.3 Reconciling TS as part of girl’s identity

The previous section introduces the notion of stigma associated with social identity of being a girl with TS. This section considers how normalising helped parents and girls reconcile TS as part of their personal identity.

7.5.3.1 Tensions in revealing and concealing information

The risk of being defined by illness may lead to individuals revealing or concealing information. Some parents were concerned about sharing sensitive information with others when their daughter was a baby, recognising that as she grew up their daughter may consider the information to be personal and not want others to know. This motivation was less to do with being secretive and more to do with ownership of information and the child’s right to privacy. Revealing and concealing information is not a clear process or definitive. At the heart of decisions to disclose were issues about how to manage information; what to tell, when to tell, who to tell and how to tell. In some conditions such as Down syndrome there are visible stigmata that are recognised by the general public, which results in immediate disclosure of the diagnosis. However,
in TS there is a degree of choice because visibility varies. Firstly, with the girl’s presenting phenotype and secondly the knowledge of the general public relating to TS.

Stigma is reliant upon the individual displaying differences from the normal population and on those differences being recognised and responded to by others in society. If a girl with TS has mild phenotype then she may be perceived as normal, a concept Goffman refers to as “passing”. It is a central concept of stigma and relates to information management so that the “stigmatised” can pass as a “normal”. For parents of girls with TS there are decisions not only about what to reveal but also how to avoid discreditable qualities; to minimise visible behaviours. However, the limited knowledge of the public about TS means that many of the features and characteristic behaviours may not be noticed or associated with the diagnosis. Nevertheless any visible deviation, physical or behavioural, from perceived societal norms would be stigmatising qualities irrespective of the diagnosis.

There were visible aspects of TS such as short stature automatically disclosed but not attributed to TS, i.e. parents explain that their daughter has a growth problem which is being treated with GH; parents in Case I openly shared information about their daughter’s coeliac disease but did not disclose the primary diagnosis of TS. Omitting information that it is part of the overall diagnosis of TS meant that sensitive information relating to genetics and infertility could be protected and kept private. Many other families disclosed the diagnosis and explained to those who had not heard of the condition that TS fundamentally meant their daughter has a growth problem and difficulties with mathematics, leaving out the more sensitive aspects. A strategy of selective disclosure was seen across these data in relation to sharing information outside the family. It provides a mechanism for negotiating an individual version of TS. If individual components of TS are revealed rather than the diagnostic term, this reduces anxiety about the full consequences of the condition being discovered.

Unlike conditions such as asthma, where it is accepted that some children may be mildly affected and others more severely, it appears with more stigmatised conditions such as epilepsy and TS the individual is assigned an identity rooted in a stereotype
The stereotypical images of women with TS emerged from descriptions of women with TS in the 1950s, which may no longer be wholly relevant to girls currently receiving active medical management of their growth and pubertal development that was unavailable to previous generations.

Another strategy to emerge in relation to controlling information between parents and their daughters was ‘the concept of everything’. This concept changed primarily in response to the girl’s age and stage of development. Goffman (1990) acknowledges the difficulties of revealing and concealing information to children about their illness. The difficulty relates to the child’s ability to understand sensitive and detailed information and about how they may use it. Children are perceived as “unsafe receptacles for information” (Goffman 1990, p71).

TS as a diagnostic label was not in itself reported as invoking stigmatising responses as is reported in literature about epilepsy or autism, largely because the condition is not well publicised and known about. However, the cardinal features of TS, namely short stature and infertility, are recognised as potentially stigmatising conditions in society, which in addition to social difficulties open the girls up to the possibility of stigma from multiple sources. Therefore the dilemma appeared to be how to manage information and situations relating to the component parts of the diagnosis rather than to conceal the diagnosis itself.

7.5.3.2 Managing the health and social needs

Medical management of TS is complicated and involves many body systems in addition to emotional and psychological support of the girls and their parents. Parenting in Goffman’s theory is in the category of “the wise”. These individuals are related to the stigmatised individual in a relationship that wider society treats as one, “they are obliged to share some of the discredit of the stigmatised person” (p43). This is further demonstrated in later discussion relating to infertility and the response of mothers. The second group to be considered within “the wise” category are the doctors and nurses who understand and attend to the needs of those with the stigmatic quality.
These data were dominated by the social meaning attached to this diagnosis, by concern for social functioning, for girls to fit in and be accepted. Many parents and older girls commented on aspects of their lives that demonstrated they were able to function normally in social groups outside the family. Medical management was also discussed within the context of providing positive social outcomes for current and future roles. For example, the administration of GH would enable the girls to become a normal female height; the induction of puberty enables girls to look like and develop secondary sexual characteristics in line with their peer group which would help in forming relationships, treatment options for infertility offers hope of fulfilling future role as mother.

With the exception of treatment associated with the management of co-morbidity there was no separation of the medical from the social outcome. For example, minimal reference was made to the long-term effects of GH or the potential benefits of oestrogen to bone mineralisation or potential hearing loss in early adulthood. The dominant areas of concern were the use of medical treatments and psychological interventions to make the girls appear as normal as possible, which would in turn reduce any potential social difficulties. Concepts of normal and normalising are fundamentally different yet for many girls with TS they are closely entwined. In chronic illness the treatment of the body usually has the consequence of better function which enables individuals to normalise their lifestyle, however, with TS, treatment of the body has the consequence of making the body appear normal, to pass as a girl without TS in order to normalise the girls’ lives. An overriding aim is to reduce social difficulties.

7.5.4 Threats and opportunities to normalising

Most chronic illness deals with physical uncertainty of symptoms or deterioration created by the illness. Although some girls have to manage co-morbidity this seemed overshadowed by two fixed problems: short stature and infertility. These features were of little concern to girls in the pre-oestrogen group where developmentally height was less of an issue than with the post-oestrogen group. For the young girls who knew that infertility may be a problem discussion with parents had focused on doctors helping them to have babies or the possibility of adoption whereas older girls commented on
developing relationships and timing of release of the information to a potential long-term partner. However, for both groups the concerns related to threats to future social roles that could be ameliorated by medical management in the present.

7.5.4.1 Clinic visits

A threat to the normalising process was clinic attendance. The doctor to some extent validated how ‘TS’ or how ‘normal’ the girls were perceived to be. Some parents and girls commented that they did not really think about TS until clinic appointments were due. This highlights the possibility for distancing the condition until there is an associated issue relating to its management. It may be that seeing the same consultant minimises TS through personalising the encounter. In this way the focus remained on the individual girl and there appeared to be less emphasis on the condition, outcomes of examinations and investigations discussed in relation to TS rather than in context of the girl.

Attending specific clinics for girls with TS were significant events in clarifying a girl’s individual profile, especially in the early years following the diagnosis. Parent’s motivation for getting what they needed from clinic visits in terms of information varied. For some these appointments could stimulate discussion of sensitive issues with their daughter, provide opportunities for clarification of information and future planning, and be places to learn more about the condition and extend their own knowledge. For others clinic visits were a straightforward monitoring exercise. Trust and familiarity were valued aspects of the consultation and necessary for free exchange of information between all present. This was important where parents wanted to discuss aspects of management and treatment using information they had gathered from external sources such as TSSS or the internet. Priority for different types of information altered across time for the girls and their parents. There was a degree of uncertainty expressed by parents and girls who were unsure about what would happen at the next clinic appointment. Some of the girls worried about having blood tests. The emphasis on the type of information required changed in line with the needs of parents in parenting their daughters and developmentally for the girls. These changing priorities are depicted in Chapter 5.
7.5.4.2 Management of growth and puberty

The administration of GH is a threat and an opportunity to normalising girls with TS. It is not clear whether HRQOL is improved in childhood and in these interviews the girls were not unduly concerned about their height or having GH. They all understood it would help them grow and older girls quantified this as a female height similar to their mothers’ or as being within the expected height for non-TS women. However, to decline GH and the opportunity of additional growth was not considered an option by any parents interviewed in this study. It was considered to be a positive action. Two sets of parents mentioned being concerned about long-term use but it did not undermine their decision to accept the treatment. It could be suggested that the medical management supports a view of intolerance of difference in society by actively engaging in “curing” visible difference to improve life chances of girls’ fitting in, strengthening the argument that the individual is “defective” in some way. Ultimately to change the physical appearance of their daughter is a way in which parents may perceive they have some control in reducing felt stigma.

7.5.5 Summary

There appear to be parallel conversations relating to normalising in TS. There is a medical dialogue about the need to normalise the girls physically through treatment. This includes medical management of any co-morbidity and ill health resulting from TS such as cardiac anomalies and hearing dysfunction. Medical management for TS as in any other condition aims to optimise physical health for the present and the future. Chronic illness usually contains a downward trajectory and this can be seen in TS. There are aspects of the condition that worsen with age. However, a parallel conversation also occurs with specific reference to the impact of the condition on the future roles of the girls. This aspect of normalisation is reflected in Wolfensberger’s idea that where groups in society are at odds with societal expectation of roles they will be perceived as devalued, and these devalued roles are generally considered negative. Choices about their roles as women, particularly roles as partners and mothers are central areas of concern in these data. This conversation is specific and gendered, it has implications for what information is shared and with whom. The concepts of being
normal and of normalising are very different. Normalising is enabled largely through the control of information which is supported by the visibility and invisibility of features of TS and by decisions about the perceived ownership and sensitivity of information.

7.6 Identity

The concept of identity was evident within the information activities. The next section provides a brief review of the main findings to exemplify where identity emerged as a major theme in this study. Illustrated throughout the discussion so far is the sense of ambiguity in what TS meant in general terms and in the interpretation of what TS meant for an individual child. Within this study all parents and girls recognised they had had a diagnosis of TS, the chromosome analysis meant this was unequivocal. As already indicated, for several families there appeared to be little coherence between the chromosomal analysis, the phenotype and the degree to which the term TS was representative of their daughter and her experience of living with TS. This ambiguity reflects a central tenet of symbolic interactionism which is that “truth is tentative” and meanings will change depending upon the context for an individual (Charon 2010). A criticism of symbolic interactionism is that it focuses on this micro level of an individual experience and neglects a macro perspective of society. However, in this study the insight provided by individual accounts has enabled consideration of wider societal issues for girls with TS and their families, when placed within published literature and theories. For example, the impact of potential stigmatising features on future societal roles for women as wives and mothers. A further example being the consideration of different ways in which health information supports the concept of developmentally appropriate education and informed choice in various aspects of their lives.
7.6.1 Brief review of the findings related to identity within the information activities

Data pertinent to gathering and receiving information illustrated that parents sought out information to familiarise themselves with the features and characteristics associated with TS and then compared their daughter’s presenting features to either confirm the diagnosis or explain certain behaviours. Over time their daughter became a source of information as parents observed and interpreted features and characteristics. Siblings became the reference point for development, behaviour and familial characteristics. The more information parents gathered the more comparisons they made with their daughter, using a variety of sources such as written literature, photographs on the TSSS website or personal contact with other girls and families. The girls were primarily dependent upon their parents and the consultant paediatric endocrinologist for information.

Making sense of information was the second area of activity in which parents received a wealth of information in the early diagnostic period. Medical staff investigated and clarified the parameters of the girls’ individual profile, determining which physical features she had, her characteristics and potential abilities. Parents expressed an initial sense of shock at receiving the diagnosis as well as anxiety linked to uncertainty as they came to understand the wide-ranging nature of daughter’s condition. Sadness was specifically expressed in relation to the aspect of infertility. In making sense of information parents were able to develop an understanding of their daughter as a girl with TS. This knowledge base enabled parents to participate in clinical decisions and to take action to encourage social engagement and achievement at school for their daughters.

Parents used information about TS and their daughters to negotiate an individual profile of their daughters’ TS. Parents undertook this activity with themselves, family members and medical staff in order to determine the parameters of their daughter’s TS. The eldest girl interviewed expressed concern about the chromosomal nature of the condition becoming public knowledge and had started to think through the implications of infertility. Other girls had an understanding focused on features that directly affected them rather than the syndrome as a whole. Both parents and the girls illustrated
selectivity in sharing information about chromosomes or infertility; for example, one of the girls had given a talk at school about TS but only included information about growth and having GH injections. Data illustrated a tension, between what information parents should share in order to benefit their daughter in some way, for example to secure appropriate academic support at school and, not sharing information because they did not want their daughter perceived as different from other girls or treated any differently to other children. Table 34 summarises key issues from the findings and identifies three types of identity evidenced from these data.

Table 34: Summary of key findings and associated types of identity

<table>
<thead>
<tr>
<th>Information Activities</th>
<th>Key issues from findings</th>
<th>Types of Identity</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gathering and Receiving</td>
<td>Identifying characteristics and features of TS</td>
<td>Identity of TS</td>
</tr>
<tr>
<td>(Chapter Four)</td>
<td>Observing daughter</td>
<td></td>
</tr>
<tr>
<td>Making Sense of Information</td>
<td>Perceived importance of genetics</td>
<td>Individual identity</td>
</tr>
<tr>
<td>(Chapter Five)</td>
<td>Visibility of TS</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Medical and social divide in TS</td>
<td></td>
</tr>
<tr>
<td>Using and Sharing</td>
<td>Differences between mothers and fathers to probable infertility</td>
<td>Current and future identity</td>
</tr>
<tr>
<td>(Chapter Six)</td>
<td>Optimising future choices</td>
<td></td>
</tr>
</tbody>
</table>

These three types of identity form the subsections that structure this section of the discussion. Within a symbolic interactionist view identity is seen as central to understanding human action (Charon 2001). Developmental needs for information also change as girls grow up and their definition of ‘self’ changes. Guided by a symbolic interactionist view of identity, Stryker (1980) argues part of the ‘self’ is defined by
meanings and expectations associated with roles. Key findings illustrate that meanings of TS to these participants affects all dimensions of ‘self’ (Callero 2003), including the girls’ bodies, their thinking processes, their lifestyles and future within female role expectations.

7.6.2 Identity of TS

An initial identity of TS was assigned to families by the consultant paediatric endocrinologist. Girls were categorised on the basis of chromosomal analysis and identification of commonly associated features and characteristics of TS. At the point of diagnosis official recognition of a difference (that may have previously been perceived by parents or others but not articulated, e.g. Case H) was made. Social settings such as hospital, clinic and support groups that the family will now enter further define their difference, as families associated with TS. Four-monthly appointments at the TS clinic identify a specific group of individuals that will be there and this is the group that girls and their parents are now part of. They are bound together by their genetic make-up. Characteristics and physical features associated with an individual’s genetic make-up are usually referred to positively within families to denote kinship and connection to immediate and wider family members rather than with similarity to a group external to the family. One mother expressed her sadness at noticing her daughter looked like other girls with TS. Her language appeared to assign a specific identity to her daughter, characteristics that were not part of their family, she referred to girls with TS as “they” and “them” and during the interview she enquired “do you see a look of them”. This comment also emphasises the point Goffman makes about the importance of the sense of sight in stigma. However, there also needs to be “perceptibility” and “evidentness”, meaning that the observer needs to recognise the difference and what the implications of that difference might be for social interaction (Goffman 1990).

For some families identifying their daughters with this new social group of girls with TS was not considered to be a positive option. Parents were reluctant for their daughters to meet with other girls purely because they had the same condition. It was perceived as reinforcing their identity as a girl with TS and diminishing them as individuals who could be defined in many other ways.
didn’t want it be Flora with Turner’s syndrome and, and that’s the only thing we see really he wanted her to be known own as Flora ok she had got this thing and we’ll deal with it, but he didn’t want her going on outings just because they were children with Turner’s syndrome really (FP:L44–46)

This statement uses negative language in relation to TS “this thing” and also refers to “going on outings”, which is a colloquial term historically used when referring to the elderly or children with learning disabilities. The use of language here emphasises the symbolic nature of language in the process of developing meanings through communication with others (Charon 2010). However, there are also examples in the findings of parents who are neutral toward the diagnosis on a day-to-day level.

Vigilance by parents of children with chronic illness is an activity frequently cited in the literature. In circumstances where a chronic condition such as asthma or cystic fibrosis threatens their child’s immediate physical health parents need to “anticipate, recognise and respond promptly to changes” (Hayes 2007). However, with regard to physical health there is little urgency on a day-to-day basis for parents of girls with TS to be vigilant about. Their main parental role was related to observing, monitoring and interpreting behaviours and characteristics typical of the condition over a long period of time, from infancy to adolescence. Parental focus was largely on optimising their daughters social functioning. Parents monitored their daughters for responses to treatment; achievement of normal childhood milestones; ability to function socially and their academic performance particularly with mathematics. Subsequent interpretation of these observations contributed to identification of their daughter as a girl with TS. However, these parents also qualified their daughter as being ‘normal’. Vigilance for these parents was less about physical symptoms and more about recognising potential stigmatising threats which varied over time and situations.

7.6.3 Individual Identity

Girls at the point of diagnosis are assigned the status of ‘girl with TS, however, the component parts of the diagnosis may not have been known to the girl possibly due to her age and/or dependence upon the motivation of the parents. Therefore her TS identity is revealed over time. The medical diagnosis is important to categorise the girl for subsequent medical and social support, but she has until this point unwittingly
incorporated aspects of TS as part of her identity in its component parts rather than as a whole.

The genetic nature of the condition and its resulting visible manifestations, e.g. short stature, have in part begin to shape expectations of others potentially defining who the girls are and who they may become through social encounters prior to the diagnosis. Individual identity is the girl’s personal identity, a unique mix of how she (and her parents) manages information about her condition with her life experiences. Goffman (1990) stated that personal identity enables individuals to be differentiated from one another and “can be used to safeguard against potential misrepresentation”. Individual identities of girls in this study were also mediated by their parents and the girls themselves, who controlled information about TS and determined which features and characteristics were applicable to them,

Personal identity pertains not to combinations of social attributes but to the kind of information control the individual can exert (Goffman 1990, p86)

The unfamiliar nature of TS to the general public and the diverse combination of features and characteristics expected of a ‘syndrome’ provided uncertainty that facilitated a fluid identity, one that could be negotiated and renegotiated in different situations and circumstances.

This attitude was reflected in a study undertaken in the USA with mothers, fathers and grandparents of children with Klinefelter or TS and mothers of children with Fragile X syndrome. The aim of the study was to explore how these families interpret a confirmed genetic diagnosis that is associated with a range of possible symptoms which may never be exhibited (Whitmarsh, Davis, Skinner and Bailey 2007). They explain that a syndrome can be diagnosed in the absence of visible signs through genetic testing which is different to the majority of diagnoses where symptoms are assessed clinically prior to assignment of a named condition. Syndromes they contend are “uncertain, varied and vague”. Whitmarsh et al. (2007) reported that the parents of children with these syndromes created a “counter discourse” that expressed doubt about the details or significance of the genetic information; instead they described their children’s achievements and abilities, “resisting the reductionism of a genetic label”. In a diagnosis
of TS there is uncertainty, particularly in young girls, as currently it is not possible to predict their future characteristics.

7.6.3.1 Understanding “My TS”

Identity emerges through feelings and thoughts (Charmaz 2009) and families create meaning through communication. Parents were the primary source of information for girls across the age range in this study. How parents perceived TS and talked about it influenced their daughter’s understanding of the condition and herself particularly when the diagnosis was made early in a girl’s life. Learning experiences that influence the concept of self are described by Goffman (1990) as an individual’s “moral career”, how girls become socialised as a girl with TS. Two phases are involved whereby an individual initially learns societal norms and what it may be like to be stigmatised and secondly, recognition that they possess a stigmatising attribute. Four patterns are noted by Goffman, three of which can be evidenced in these data (Table 35):
### Table 35: Patterns of moral career

<table>
<thead>
<tr>
<th>Pattern of Moral Career</th>
<th>Consequence</th>
<th>Case</th>
</tr>
</thead>
<tbody>
<tr>
<td>Inborn stigma</td>
<td>Socialised into disadvantageous situation incorporating standards which they do not meet, e.g. height, natural puberty and pregnancy, social confidence</td>
<td>A,C,D,E,F,J, K,L,M,N,P</td>
</tr>
</tbody>
</table>
| Capacity of family to provide “protective capsule” | Sustained by selective release of information  
Child sees self as normal  
Anxiety for parents about stigmatising encounters which are avoided until exposed to new situations and/or settings, e.g. school, dating, leaving home/employment | B,C,D,F G, H, I, K, M, N, P |
| Later in life           | Learn they have always been discreditable  
Withholding information  
Finding reason for short stature | D, E, H                  |

As girls grew up, parents tried to identify what was a TS trait and what was their daughter’s normal personality and natural abilities. Cases above that appear in both the inborn stigma and “protective capsule” patterns used different strategies as the girls passed through different development stages. Aspects of TS emerged as the girls grew up and the emphasis for parents and girls shifted from physical needs dominating in infancy and early childhood to social needs dominating in adolescence. This mirrors normal developmental needs of children across their life-course but for girls with TS these aspects are conscious, thought about and subject to analysis and interpretation by parents and medical staff. The moral career for girls with TS also incorporates the
concept of everything whereby parents believe they have told their daughters all the information they need to know about TS. However, these data illustrate a broad range of interpretation as to what this information may include and the depth of information received by the girls. For some the “protective capsule” was a strategy to control information in a rigid manner to deny the full consequences of having TS, whereas for others it provided a safe place to have TS and offer reassurance that they could have TS but still be ‘normal’.

Some families readily acknowledged their daughter’s diagnosis and were keen that their daughter should experience usual activities with non-TS girls of similar age and development. These parents avoided creating perceived tension with others through selective release of information. Many of these girls had recognisable stigmata only visible to the knowledgeable observer (i.e. those with knowledge of features and characteristics of TS). They expressed a view that openness about the diagnosis enabled it to be contained and discussed in a matter of fact way which reduced any tension associated with exploring the broader aspects of the condition instead it kept the focus on the specific needs of their daughter. Girls and parents in this situation are considered discreditable and therefore the focus is on managing information in social settings. This adaptive strategy is described by Goffman as “covering”.

7.6.3.2 Body Image

There is a presumption that a girl with TS will experience psychological problems related to specific aspects of TS such as poor body image leading to low self-esteem. There is a respected body of knowledge (McQuillan, Greil, White, Casey-Jacob 2003) that illustrates the ‘connectedness’ between the identity of women and their bodies. Any threat to body shape or function will affect their self-identity, perception of function, life chances and experiences (Shilling 2003).

In the current study some parents commented on their daughter’s body in terms of size and shape. Body proportion was an issue for some parents and older girls. Since the 1960s the desired body shape has been thin and slender, however, girls with TS are commonly short with a tendency to put on weight. Shilling (2003) stated that the
influence of body image on self-identity is greater for females than it is for males. Judgements about the girls size and shape came from mothers and fathers in these data as they talked about what their children looked like and what they believed others think they look like. Parents made comparisons with the TS population via TSSS and also with their daughter’s siblings and peer group. Mothers talked about how visible features where possible were minimised for example, longer hair styles would disguise their neck, clothes could minimise body disproportion and make up could be used to accentuate favoured features. Fernandes, Papaikonomon and Nieuwoudt (2006) states “centrality of the body to feminine identity is predominant”, a sentiment reflected in mothers’ concerns about issues such as girls buying fashionable clothes and being like their friends. Negative body image results in low self-esteem which can undermine mental health and well-being.

The literature review identified (Sutton et al. 2005) the pervasiveness of infertility in women with TS stating that across the lifespan it continued to be a source of sadness and regret. With the exception of one family the over-riding negative aspect reported by mothers was that of infertility. This was the information least shared outside close family and stated as the most difficult issue that parents had to explain to their daughters. It could be anticipated that this reflects the age range and corresponding developmental interests of this group, which are broad; however, the ages of respondents reflecting the differing views are not differentiated in the report of the findings.

7.6.4 Current and Future identity

Current and future identities of parents and their daughters are adjusted following diagnosis. Parents become parents of a girl with a chronic condition. In this instance TS also means parents face the potential future loss of identity as grandparents (Loughlin 2006). For girls with a late diagnosis or for those where disclosure of salient facts happens when they are older (when information is considered to be age-appropriate by parents) their identity changes from their perception of being a normal girl to one of a girl with health and social problems. The probability of infertility also threatens their
future identity as a mother. This information potentially impacts on their roles on a day-to-day level and their view of themselves.

Goffman (1990) considers the control of information from the past to minimise its impact on current acceptance of an individual into a social group. He argues that concealing information in this situation has implications for social and personal identity. Girls with TS are not concealing information from their past that will define them but they with their parents are controlling the information that will shape their futures.

7.6.4.1. Being a girl with TS: self-concept

Developmental changes occur in adolescence relating to self-concept. Helgeson and Novak (2007) examined how adolescents with type 1 diabetes integrated having a chronic illness into their expanding self-concept. They found that adolescent females who defined themselves in terms of their illness were particularly problematic when the illness was perceived in negative terms. Charmaz (1991) identified two approaches, those who incorporate the illness into their self-concepts making it a defining part of who they are and those who ‘contain’ the illness by trying to not let it interfere with their lives. Females were more likely to integrate illness into their identities than males.

These data did not indicate that younger girls in the pre-oestrogen group placed the condition centrally in their lives. Where a condition has immediate physical management requirements, such as diabetes, it presents a dichotomy, on the one hand the condition needs to be central in order to maintain good physical health but on the other, for positive mental health it is better if the condition is not central (Helgeson and Novak 2007). Better control in these situations usually equates to better quality of life as the disease process is stable. However, this also assumes the lifelong condition is perceived negatively by the individual. Data from the older girls does not suggest that girls view the diagnosis of TS as negative per se. They discuss aspects that they need to manage but did not describe TS or themselves negatively. However, they did describe themselves in a way that distanced themselves from some of the features and characteristics in order to present themselves similar to girls who did not have TS.
Parents were sensitive to the concept of low self-esteem and poor self-concept because it is often reported to be a disabling feature of the condition. One set of parents specifically stated they wanted their daughter to be proud of who she is, and TS was part of who she is and was nothing to be ashamed of. Their positive parenting style which focused on raising their daughter’s self-esteem and confidence, had been influenced by experiences at TSSS meetings. They described their observations of women with TS who were “dressed like blokes really, dark clothes, jumpers, low self-esteem was what came across” (NP:L40) therefore “we swore we’d bring her up girly and we have”. They were also concerned by the poor communication skills in older women and again their lack of social confidence:

I mean I make a point of everyday of saying she looks lovely or beautiful or she’s the best girl in the world, all these little things and she she believes that and it’s important she’s confident.(NP:L67)

The experiences of these parents highlight the positive and negative sides of accessing support groups. The father in this family concluded:

I used to come away feeling really sad because the whole weekend was just one big reminder of all the problems Sarah has or will have and sometimes it’s best not to see it like that, just see her as Sarah not as a girl with problems y’know? (NP/R1:L305-307)

Girls with TS and their parents in this study gave different examples about information that they shared providing rationales for selective disclosure (Chapter 6). However, information relating to the cardinal features of TS, short stature and infertility were handled very differently. Information about short stature and the need for GH was widely shared whereas infertility and genetic information was the least shared information.

### 7.6.4.2 Future identity as a woman with TS

These data suggested that there was a difference in the response between mothers and fathers to infertility. This is not to suggest that fathers are not distressed by the information, however, they tended to see it as a future problem, one that involved their daughter in decision-making about whether she wanted to have children and not
something they needed to focus on or worry about whilst she was young. Mothers, however, responded more emotionally using terms such as “suicidal”, “end of the world”, “complete sorrow”. Mother’s talked about being a “proper woman” and the centrality of motherhood to womanhood, and of this being the most devastating part of a TS diagnosis to come to terms with.

Hope was maintained because it was a future event and therefore there was uncertainty in new technologies or their daughter’s development. Some mothers were also concerned about the development of intimate relationships in the future including how and when such information would be disclosed to future partners. For some mothers it was about giving their daughter a choice. This led them to look at options and in one case become politically active to successfully contribute to a change in the law to support future fertility options. Some mothers considered action to mediate the infertility through egg donation or involved the girl’s sisters in thinking about the possibility of egg donation in the future. Others thought through adoption as an option. There is no gender crisis here, there is no question that their daughter is a little girl, all parents interviewed were confident about the gender of their child. The sadness was less related to the identity of womanhood and more directly related to the threat to their daughter’s future prospects of motherhood. This was a significant concern for mothers with daughters of all ages and irrespective of the age at which their daughter was diagnosed.

Ethnographic studies describe infertility in the non-TS population as a source of shame and spoiled identity (McNamara 2002; McQuillan et al. 2003; Crawshaw 1995). Literature (Griel et al. 2010; Fernandes et al. 2006) reports, that motherhood brings with it a certain status, identity and achievement. It is viewed as a natural occurrence and for many is an objective in adult life. Motherhood is reported to be a significant life event across different cultures bringing with it personal fulfilment (Crawshaw 1995). Consequently, it is argued that women who are infertile experience a sense of isolation which affects their relationship with others and they are reported as being ‘disconnected’. In health research illness is often discussed as a form of deviance.
Infertility is viewed as a discreditable attribute resulting in felt stigma (McNamara 2002). It is suggested that women with multiple roles experience less stress because they can reduce stigma through adopting other identities and roles, for example, by pursuing careers. However, other studies suggest that multiple roles do not mitigate distress if motherhood is the main goal that cannot be achieved and particularly in communities and families where motherhood is an expectation of women. In societies where motherhood is expected infertility would not be a hidden feature of TS. It is suggested that biological or social motherhood is better than childlessness, however, Sutton et al. (2005) found that for adult women with TS from ages 20 to 59 years the “inability to bear a biological child” was described as painful and was a lifelong sadness. In non-TS women who are infertile significant implications are identified for emotional functioning which negatively impacts on their gender identity and self-esteem (McNamara 2002, McQuillan et al. 2003). Sutton et al. (2005) TS using DSM IV Structured Clinical Interview and Rosenberg self-esteem scale found much higher levels of lifelong depression and similar levels of self-esteem in girls and women with TS compared with other infertile women.

Stigmatisation can only occur where individuals internalise the ‘infertility label’ negatively. The oldest girl in the study at 16 years stated she found it difficult and all the mothers displayed this aspect of the diagnosis negatively,

You think you’re normal, you can live with being small but then when you find that out its kind of hard to take and I think that’s the same for my parents as well. (DG/L468)

The perception of being normal reflects Goffman’s ‘protective capsule’, where as yet she has not been of an age where infertility would be part of her world and therefore could be concealed.

Positive aspects of childlessness are not easy to find (Pfeffer 1987) and even where women are voluntarily childless they engage in defensive techniques to justify their status often “redefining childlessness as a socially valuable decision, and highly valuing their identity in work” to avoid being perceived as selfish. The published literature
referred to here, relates to women for whom infertility was unexpected and is described by McQuillan et al. (2003) as role blockage.

Mothers in the current study express feelings similar to infertile women, a sort of ‘infertility by proxy’ as they experience the potential loss of motherhood for their daughters which is generally a taken for granted life event. It may be that these mothers are expressing courtesy stigma, a transferable stigma described by Goffman (1990). Some of the mothers interviewed had personally experienced loss or threats to their own motherhood through still birth and miscarriages; and some stated they had difficulty conceiving. All the mothers interviewed experienced motherhood as an integral and key aspect of their identity and reflected the collective view of a pro-motherhood society. Through social encounters they have experienced the status and recognition afforded to mothers (Griel et al. 2010).

Identity is formed through the eyes of others, through social interaction and how we respond to the expectation of other people and the society in which we live (McQuillan et al. 2003). A central tenet of symbolic interactionism is that individuals act toward events based on the meaning they have for them, and these meanings arise from social interaction and modified through interpretation (Blumer 1969). In this study the mothers’ distress at probable infertility in their daughters determined efforts to try to correct or ameliorate it, reflecting their world view that infertility is not a natural or desired state. As such they are unwittingly contributing to the stigmatising of their own daughters. Sutton et al. (2005) argue that “protracted grief” of parents relating to infertility could be damaging in the long-term and interfere with adaptation processes. Mothers expressed hope for the future and explained that they did not think about infertility all the time but only when it was raised in social situations.

7.6.5 Summary

Although Goffman’s language is dated the original model still remains highly respected and dominant (Carnevale 2007). Criticism of the theory includes the helpless role that is attributed to stigmatised individuals and lack of analysis of social structures (Scambler 2006), however, Goffman does focus on the social difficulties for those with stigmatic
characteristics, which is in accord with the dilemmas faced by girls with TS and their parents. The process of stigma provides a way of considering how information is used by society and individuals. These data enable the experiential nature of stigma to be examined from the perspective of girls with TS and their parents.

Fathers appeared to be protecting their daughter’s current identity as a normal little girl where as mothers were protecting her future social roles as a wife, and importantly mother. Both parents were committed to optimising their daughter’s academic achievements with a view to developing a future role contributing to society. Improving their daughter’s social functioning would improve her life chances through forming future relationships and again both parents were supportive of this.

The knowledge that girls may have difficulties in social situations and future roles may be limited heightened parental fears of their daughter becoming socially isolated and this appeared to be a dominant and constant concern.

7.7 Strengths and Limitations

The chapter begins with a reflexive discussion of the strengths and limitations of this study with specific reference to methodology, methods and analysis. Credibility and trustworthiness of qualitative research can be illustrated through examination of processes that underpinned decisions taken within the study. Huberman and Miles (2002) clarify the fundamental need for researchers to undertake this course of action:

As observers and interpreters of the world, we are inextricably part of it; we cannot step outside our own experience to obtain some observer-independent account of what we experience. Thus it is always possible for there to be different equally valid accounts from different perspectives. (p41)

Reflexivity is integrated into the following discussion because it enables comment on my behaviour throughout the research process, my observations, and subsequent judgements and interpretations ultimately made (Holliday 2002).
7.7.1 Methodology

Consideration of the benefits of specific aspects of this research study adds to material presented in Chapter Three in making explicit the approach taken. Prior to the commencement of this study clinical colleagues had stated there was no recognised structured process to determine the information needs of girls with TS and their parents or any evaluation of the usefulness and effectiveness of information shared with these families. Lack of previous research into the information needs of this patient group led to the decision to explore the subject area broadly.

Undertaking a qualitative exploratory study into the perceptions of girls and their parents enabled information needs of this patient group to be uncovered. The selection of symbolic interactionism as the philosophical approach to this research is appropriate in addressing the research questions. Insight into the perspective of the participants, *their* information needs and associated priorities was made known. The exploratory nature of this study was well served by a flexible qualitative approach which enabled subtle differences, similarities and the complexity of individual experiences in relation to information needs to be accessed (Smith & Bekker 2011). Social encounters are central to any exchange of information and as such symbolic interactionism proved to be a useful strategy to explore meanings derived through social interaction in different settings such as home, hospital and school.

Analysis informed from a symbolic interactionist perspective considered the diagnosis of TS not to be deterministic but rather a term open to individual interpretation (Charon 2010). Such an approach supported exploration of individual experiences and variations across developmental ages, time and context.

A limitation of symbolic interactionism identified in the literature is that meanings are individual and subject to constant change as they are redefined, in different contexts. However within children’s health care this is an accepted part of clinical practice. Irrespective of a child’s health problem, the need for information will change as they grow and develop physically, cognitively, socially and emotionally. Therefore rather than a limitation in this study, symbolic interactionist approach guides the analysis to
consider how girls and parents constructed meanings of TS at different ages and stages of their lives, to explore what contributed to the creation of their meanings as part of their life course. The Framework approach enabled material to be drawn out of the interviews and meanings of TS to be found. Personal interpretations of what TS meant to them, the impact it had for their information needs as individuals and as families, and activities they engaged in to meet their information need were defined.

Overall, a strength of this study was the qualitative methodology which successfully revealed new findings about the information needs of girls with TS and their parents. Participants were engaged and parents and older girls stated that they welcomed an opportunity to discuss information needs. They recognised it as a valuable topic for research that could improve this area of clinical practice for families.

7.7.2 Methods

7.7.2.1 Sample

The small sample size can be seen as a limitation of this study, however, the intention was not to generalise findings but to gain insight into the information needs of a specific group. Participants represented a range of demographic and social characteristics, providing diverse experiences of TS. All participants were English speaking.

Participants were recruited from a tertiary TS clinic therefore it should be noted that they are geographically from one region of the UK. As volunteers they are likely to be a motivated group and their perceptions of TS may differ to those who did not participate.

7.7.2.2 Interviewing parents

Relationships between the researcher and researched will affect the interview and meanings created within it (Hall and Callery 2001). Prior to the commencement of the interview some parents (Case C,F,H) expressed anxiety about whether they would have answers to my questions. The topic guide proved useful in allaying this anxiety as it identified to parents broad issues for discussion rather than a set of precise questions. The topic guide also acted as an aide memoire for parents and me, providing flexibility for interview topics to develop whilst maintaining focus on the central topics. It enabled
participants to link ideas and return to previous points with minimal prompting, which was helpful in addressing the power differential between researcher and participant. It felt reciprocal; participants were active providers of data.

The interviews revealed some differences between the fathers and mothers with regard to their preferences for content, format and timing of disclosure of information to their daughters (Case A,B,D,F,H,K). Nine of the interviews were with both parents, however, mothers interviewed alone also commented on differences in approach to information needs between themselves and fathers, suggesting that in these families mothers and fathers had different priorities about the information they wanted and different ideas about the way to use it. Interviewing parents together limited exploration of their individual information needs although it was through interviewing parents together that the differences were identified.

7.7.2.3 Interviewing girls with TS

When transcribing data from these interviews I found aspects of my own behaviour influenced the data collected in different ways. One was my hesitation in probing their responses, particularly the younger girls. I was uncomfortable in following up responses from the girls when their body language or one word response indicated they did not want to answer or elaborate any further, for example, by avoiding eye contact, giggling nervously or shaking their head. In these situations I tended to move on to another point quickly to ease their discomfort, resulting in some superficial data. This issue was referred to in Chapter Two with reference to work by Cree et al. (2002) that highlighted in their work that researchers backed off when they sensed a child did not want to continue to talk about a sensitive topic area. Girls with TS are reported as finding social situations difficult. In response to this as explained in Chapter Two I had developed techniques for the girls to use during the interviews such as traffic light system to control questioning, drawing pictures to illustrate understanding and an ‘About Me’ booklet to avoid direct questioning. These proved useful in developing dialogue with many of the girls. However, there were some exceptions and in retrospect further exploration with parents at the time of recruitment regarding their daughters’ usual
response in one to one situations would have enabled further detailed and individual planning for the interview.

7.7.3 Analysis

Atypical experiences within information exchange and subsequent perceived information needs were evident in the analysis highlighting variation across the cases. Atypical cases developed the analysis and discussion. Negative evidence and contradictory evidence highlighted alternative interpretations broadening the meaning of understandings of TS across different cases (Barbour 2001). Each view is treated as having equal validity and used to stimulate data analysis (Miles and Huberman 1994; Bloor 1997). The constant comparative method employed in the analysis enabled such differences and similarities to be sought across and within the cases, for example, four of the fifteen cases provided insight into specific circumstances:

- Case N was the only family interviewed where the diagnosis was made prenatally.
- Case E was the only family interviewed where there was a late diagnosis in adolescence.
- Case H illustrated how a mother strictly controlled information and was reluctant to allow public disclosure of the diagnosis.
- Case J was the only family interviewed with a daughter who had learning disability and significant co-morbidity.

The amount and variation of data collected in this exploratory study was considered adequate after fifteen families had been interviewed. The later interviews were not generating any new ideas to add to the analysis, “appropriateness refers to selection of information according to theoretical needs of the study and emerging model” (Morse 1995). The topic guide was altered in response to new situations or content introduction during transcription of interview data. This enabled confirmation of ideas as data collection worked towards “saturation”.

Strategies and decisions were made to increase transparency and accountability of the research procedure. Documentation of the development of the themes within this
research provides evidence of the steps followed explaining processes used and from where the major themes emerged. Supporting documents are available for scrutiny including recordings of the interviews, interview transcripts and field notes; notes made about the analytical process, tables and charts (Robson 2002; Silverman 2006).

“Verification of the study with secondary informants” (Morse 1996) is a process which suggests presenting data back to the informants for them to confirm validity of the study. However, the value of this is disputed within the literature. Transcripts were made available to the participants, however, a limitation in this part-time study was the length of time between the interview and availability of the transcript to participants. No additional comments were made by girls or their parents.

In supervision meetings, supervisors examined excerpts of coded transcript and through detailed discussion would contribute and question the development of categories and themes. This process facilitated further inductive inquiry and challenged the process of linking data. Discussion with the supervisory team was therefore invaluable in bringing new insights and questions to these data.

7.7.4 Real World Validation

Morse states:

If you have developed your concepts well they should be recognisable in other places, in other groups and in other situations (p148)

The panel described at the beginning of Chapter Three supported this research at different stages. They were important in the planning and preparation of material such as the topic guides and very useful in questioning and justifying interpretations made during analysis prompting different questions. External validation can increase confidence in findings (Pyett 2003) and in addition to ongoing review and questioning by supervisors, this study has been presented to different audiences for different purposes throughout its development and implementation:

• A seminar presentation to provide an overview of the study was delivered to fellow students which helped to formulate the approach taken.

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• A poster presentation of the key methodological issues was presented to departmental research staff and students. This was assessed by a supervisor.
• A paper presenting early findings was delivered at the British Society of Paediatric Endocrinology and Diabetes Conference (2010). The paper was well received by experienced nursing and medical staff, some of whom commented that the emergent themes had relevance to their experiences with families with TS.
• The TSSS invited me to present the early findings at their annual conference in June 2010. The experience was a significant point in the research process for me as many parents, some of whom had contributed directly to the study, made considerable comment on how it resonated with their own experiences. Some parents wrote comments which can be found in Appendix 10. Parents also offered different perspectives and interpretations to my own which added to the questioning of data during analysis. I have been invited to return on completion of the thesis.

7.8 Summary
Symbolic interactionism is considered as primarily a philosophical framework. However, Charon (2010) argues “it is an exciting and useful perspective to understanding human social life” (p184). TS affects all aspects of girls’ lives in their present and has implications for their future roles and lifestyles. The discussion has focused on interactions between girls with TS and those around them and how these interactions have shaped the interpretation of what having TS means to them. The influence of symbolic interactionism has been to develop deeper insight into the participants’ meanings and understanding of TS. Focusing on their personal situations has reframed the understanding of the information needs of girls with TS and their parents, offering their perspective. The individual meanings of TS are unique to each girl and parent but there were some shared perceptions found in social contexts such as experiences in school, responses to probable infertility.

Symbolic interactionism and the use of the Framework approach has proved to be a beneficial combination to illuminate the meanings girls and their parents developed in relation to a diagnosis of TS (Charon 2010) and resulting need for information.
CHAPTER EIGHT: CONCLUSIONS AND RECOMMENDATIONS

8.0 Introduction

This research set out to explore the perceived information needs of girls with TS and their parents:

- To identify what girls aged 8–16 years old know and understand about TS
- To explore their experiences of living with TS
- To discover what parents and girls with TS perceive their information needs to be
- To investigate sources of information and support, and their usefulness

The findings identified that there are unmet information needs for these girls and their parents. The problem relating to information needs is not about a lack of information but how it is gathered, made sense of and shared. There is a significant amount of good quality information produced by the TSSS readily available to girls and their parents.

However, their need for information is individual and varies considerably. The findings revealed that the information needs of each family interviewed are a complex phenomenon. The age at diagnosis, particular karyotype, variation in phenotype, decisions about timing and content of information to be shared across developmental stages, in addition to the inherent complexity of the subject areas contributed to the difficulties girls with TS and their parents face in the process of individualising information and developing their understanding of TS.

Exploring the experiences of girls and their parents exposed diversity in how families conceptualised TS. There were similarities of experiences across the cases, but the variation in presentation of TS meant that families also provided insights into their personal meanings. Across the families information needs are addressed by engagement in three major information activities: gathering and receiving, making sense of, and using and sharing.
8.1 The meaning of TS

The meaning of TS to each of the girls and their parents was important because it directly affected their information needs. Parents’ meanings of TS were shaped through interaction with clinical and lay experts, publications, parenting and the response of others to their daughters in a variety of settings including at home, school, in organised activities, and TSSS events. For girls the meanings of TS were also informed through interaction with their parents and other family members. Their understandings were modified over time as information was gathered and received information, made sense of and subsequently used. As they developed their understanding, three major themes are evident from the data: uncertainty, identity and normalising. These themes are central in forming the meaning of TS held by these parents and girls. Key points are summarised below.

8.1.1 Uncertainty

Uncertainty was identified about the extent to which TS matched the child’s phenotype. Although a large amount of information could be gathered, there could be uncertainty about how the general information applied to an individual child. Making sense of information raised additional issues of complexity for the parents as their daughter grew and they tried to decipher what were normal developmental changes and what they considered to be manifestations of TS. Physical stigmata were unchanged, however, behavioural aspects of the condition appeared to change over time as she progressed through new developmental stages. Uncertainty could also arise in using information, including whether what would be appropriate information to share with their child at any one time; uncertainty about how to share information and uncertainty about their daughter’s understanding of information.

8.1.2 Normalising

The concerns about identity brought about a desire to normalise the child. Therefore the social implications of TS dominated and overshadowed information about long-term medical implications of TS. The only exception to this was where one girl had significant associated co-morbidity that required daily vigilance by parents in managing
her physical health. Girls’ experiences of TS did not generally include daily activities in managing symptoms as is the case in many other chronic illnesses. The central focus of the medical consultation for the majority of families was the normalising of their bodies through monitoring and managing growth and puberty in line with their peers.

8.1.3 Identity

Uncertainty about the meaning of the diagnosis of TS raised concerns about identity for the girls. Sources which included comprehensive information about current knowledge of TS and associated issues, appears to describe a group identity, however, the meaning and interpretation of TS by each girl and her parents was formed through their unique and individual experience. Identity as a girl with TS was further developed through social encounters between girls (TS and non-TS), parents, health professionals and others. Girls shared information with close friends because TS was part of who they were. The information shared was reflective of their age. Older girls were more selective about what they perceived as sensitive information including the chromosomal nature of the condition and probable infertility.

Social vulnerability is a component of TS reported in the patient information literature. There was anxiety expressed by parents relating to their daughters being identified as different and becoming socially marginalised by their peers as they grew up. Some parents stated this was one of the worst aspects of the condition.

8.2 Implications for clinical practice

8.2.1 Information needs: on-going review

A diagnosis of TS involved the interpretations of the karyotype, phenotype and stereotypical views of girls with TS which provided families with an uncertain picture about the meaning of the condition for individual girls. This was particularly true for parents of girls diagnosed at a young age who had not yet developed or displayed TS traits. These findings highlight the importance of revisiting information at each developmental stage with regular and structured review within clinic visits. Information sharing should be considered as a social interaction to promote openness with girls as
well as with their parents. Parents’ uncertainties about how to share information with their daughters indicate the need for a partnership approach with health professionals to provide support for sharing complex and/or sensitive information with their daughters. The challenge is how to share the extensive and personalised information that health professionals have access to about individual girls. Parents need advice about how to share this information with their daughters. This point is returned to in Subsection 8.2.3 in relation to the route map. The experiences of the families interviewed did not suggest a structured planned approach to teaching and assessment of understanding about TS. A structured planned approach could have potential to promote girls’ understanding and awareness of their bodies and associated treatment, particularly if the focus was on their experience of living with TS and fostering their sense of self.

Raising issues with parents and assessing their understanding of aspects of TS could contribute to reliable information being provided to the girls over a prolonged period of time. The subjective perceptions of parents about the meaning of TS to their daughters and the girls’ views of themselves influence the information they perceive as valuable and so should be explored by health professionals.

The motivations of parents and girls toward information management can also be important to recognise in order to help them share and make sense of information. The broad range of responses across the cases suggests assumptions should not be made about what mothers, fathers and girls need to know and how they wish to receive information. Selective disclosure of information was one way in which identity was controlled. Deferring disclosure of information may be developmentally appropriate but it may also indicate avoidance or denial of an issue. Regular review of information by a familiar health professional could help to identify on-going reluctance to disclose information. Parents could be supported by identification of their own information needs and explicit negotiation about priorities for the timing of sharing information with girls.
8.2.2 Information needs: the role of the PENS

It appears that the role of the PENS differs significantly from the interpretation of the role of clinical nurse specialists working with families of children with other long-term conditions and indeed with children who have other endocrine problems. There may be potential for PENS to play a larger role in supporting and mediating information. All families have regular clinic appointments, therefore access to the PENS is possible. Earlier discussion noted the PENS role was limited to administration of GH. Mishel’s uncertainty illness theory states that “credible sources” are able to influence the subjective experience of uncertainty and Tluczek et al. (2010) identified these sources to be health professionals who were able to interpret information, provide meaning of the condition and explain management treatments and outcomes. It appears that PENS are not perceived as “credible sources” for wider TS issues.

Families valued their 4-6-monthly appointments with the paediatric endocrinologist and if they were unable to see them at each visit they could feel dissatisfaction. It may not be practical to rely on the endocrinologists to this extent because of the pressures on their time. Sharing responsibility for meeting family information needs with PENS in a team approach could be one way of addressing families’ need for continuity and familiarity. This could also provide opportunities to prepare girls for changes in medical management prior to an appointment with the consultant. PENS could also be involved in contributing to the physical assessment of pubertal development. This could enable families’ expectations to be managed and to accept that they would not see a named consultant every time thereby reducing their dependency on the endocrinologist.

8.2.3 Information needs: a route map of issues and resources

A practical suggestion made by two fathers to help with sharing of information was the development of a generic route map. They proposed it should span the ages and stages of development to provide an overview of the ages at which key interventions may occur and indicate what information they should be discussing at a particular time with their daughter. Specific issues could be selected for discussion in clinic. The issues could be selected ahead of time prompted by the map, to enable relevant resources to be
acquired. The age at diagnosis should be a separate element that outlined all initial investigations which would be carried out irrespective of the age of the girl. Following this the family would refer to the generic route map.

Information needs were not just linked to the girl’s immediate medical management and several mothers stated they wanted to know everything. A route map may fulfil their initial need for this overview without bombarding them with large quantities of detailed information. As already reported findings in this research have indicated concern at the depth and breadth of understanding parents and girls have regarding all aspects of TS, it would be beneficial to provide guidance about an appropriate level of information relating to developmental understanding of girls at different ages and at different points in the trajectory of TS.

This strategy could actively engage parents to help them manage the tensions that were uncovered in the findings. These include:

- Tension between revealing and concealing information.
- Tension between wanting their daughter treated the same as other children and yet wanting her to be given special consideration because of TS.
- Tension in managing “nearly normal” persona particularly at school where some teachers found the discrepancies in behaviour difficult to understand and manage.
- Tension between being the same as other girls with TS and having unique and individual issues to manage.

Figure 16 provides an overview of information points, key information and reliable sources identified by participants in the findings of this study. It serves as a starting point for developing the concept of a route-map as suggested by the fathers. Information is linked to three sections reflecting the information trajectory described by parents. Firstly, getting to a diagnosis; secondly confirmation and clarification of the diagnosis for their individual daughter and finally on-going management. In the discussion issues were raised relating to the development of familiarity with health professionals reducing reliance on the paediatric endocrinologist; a need to balance current and long-term health needs; the role of professional and lay experts in providing on-going information.
as the girls’ needs change as they grow and develop. Parents interviewed talked of finding information produced by TSSS as providing useful practical tips for parenting a girl with TS. The plan introduces a team approach to on-going management and identifies the PENS increased input to areas such as preparation and monitoring the induction of puberty. Detailed information plans could also be developed and used as an on-going teaching tool for the endocrine team to develop and revisit information over time. This would be particularly useful for girls diagnosed early in their lives when parents received all information and some of which, as parents in this study reported, was not revisited.
*Figure 16: an overview of key information and sources identified in these data*

**Professional Source of Information**
1. Paediatrician/Obstetrician
2. Paediatric endocrinologist
3. Heart specialist
4. Renal specialist
5. Ears, Nose and Throat specialist
6. Geneticist
7. PENS
8. Clinic nurses
9. Clinical psychologist

**Lay Experts**
- Practical parenting information
  - www.tsss.org.uk or www.childgrowthfoundation.org/Turners_Syndrome.htm
  - TSSS factsheets: feeding; speech and language; induction of puberty; tiredness; spatial awareness; foot care; genetics; disclosure
  - Pharmaceutical booklets about GH available from clinic
  - TSSS: Survive and Succeed at School: guidance for teachers
  - Resources used at School to help to talk about pubertal development

**Getting to a diagnosis**
- TS is individual to each girl, it is complex and can affect many systems of the body. The paediatric endocrinologist will assess these systems (see above) for any abnormality including her heart, kidneys, ears, bones and chromosomes (karyotype). A physical examination will identify the specific features of TS that your daughter has. This is known as her phenotype. Together you will explore information you need to develop your daughter’s understanding of what TS means to her.

**Confirming the diagnosis and clarifying individual**
- Clinical examination by endocrinologist

**Ongoing Management**
- Individual nature of TS means highly individualised management plans are developed
- Parents and girls will develop new skills as girls may have daily GH injections from age 4
- Growing up and changes to girls’ bodies can be discussed from age 9 in readiness for taking tablets to induce puberty
- Induction of puberty is individually negotiated often to coincide with peers. Can be from age 11
- Teachers need information at all stages to help girls to socialise and to achieve her academic potential
- Some girls need some specialised help to develop strategies to manage their feelings as they grow up
- TS is a lifelong condition. At clinic information about long-term health implications is as important as managing more immediate aspects
- General information relating to women not having their own babies can be introduced from an early age and become more focused as girls get older.
8.2.4 Information needs: recognising the social implications for individual girls.

Roles such as being a girl, a wife, a mother, a grandparent, a friend are taken for granted by the general population. These roles are expected yet not consciously thought. This study suggests that TS presents a potential threat to these familiar social roles and as such is at the heart of how and why information about TS is controlled by parents and girls. These roles primarily reflect familiar social relationships and traditional views of female function and are created through interaction with others.

Whether girls were seen to have TS by others depended upon their visibility. Visibility only occurred if the perception of others was that the girl appeared different in looks or through her behaviour, however, this only identified the girls being different and not necessarily as having TS. For this to occur the observer needs to be able to recognise the features and characteristics of TS. Therefore even where a difference is noted, TS as a label for the perceived difference is not always applied. Girls who are seen by others as not physically or behaviourally different can pass for normal. However, there are hidden differences that may impede achievement of female social roles particularly associated with puberty, sexual partnerships, and pregnancy. Therefore parents made decisions about what information could be omitted or concealed in childhood that would later be made known to the girls as they grow and develop.

Conflicting attitudes to raising the public profile of TS and with it the visibility of girls with TS were evident in this study. Visibility of TS was interpreted as an opportunity and a threat to the identity and functioning of girls in social roles. Opportunities included possibilities of improving academic and pastoral support for girls in school, improving knowledge of health professionals, streamlining and accessing services for girls with significant learning difficulties. Threats were identified as misinterpretation and misapplication of the broad features and characteristics to individual girls. The identity of girls with TS resulted from the interpretation of information which was amalgamated with individual experiences of what it meant to have TS, resulting in an individual concept of what it means to be a girl with TS. For some girls TS was a fundamental part of who they were and for others it was not a central part of their everyday lives.
The findings in this study indicate that the felt stigma was a source of distress for parents. Parents could be encouraged to reduce comparisons with other group and helped to recognise stigma as a more general feature of society with more positive views of TS shared during consultations. This is not to deny that some girls may have problems with bullying or difficulty in making friends but it is not an issue for all girls with TS.

8.2.5 Information needs: long-term medical implications of having TS

The genetic and lifelong nature of TS requires an approach to teaching and learning about the condition that incorporates on-going assessment of understanding of future health risks. There was for some parents an anxiety and sadness about sharing some of the more sensitive aspects of TS or exposing their daughters to aspects of the condition that did not currently affect them but were reported in the literature. However, there is a danger in minimising these potential future health needs, which may become overlooked despite having long-term implications for the health of girls with TS. Girls also need to understand the full implications of their diagnosis. Availability of developmentally appropriate rationales for the girls to explain investigations, monitoring and treatment could start to facilitate discussion of the need for long-term healthcare. Meeting the information needs of girls should not only be a responsibility of parents but a shared responsibility with health professionals.

8.3 Recommendations for future research

As stated at the outset no research with a specific focus on information needs of girls with TS and their parents was found in the literature search therefore research into this area is beneficial. To date provision of information has primarily focused on content, i.e. facts about TS reflecting common elements of information relating generally to girls with TS. The resources provided by TSSS are excellent and provide comprehensive material that has proved to be beneficial to families who accessed them. Tailoring of general information to an individual girl’s circumstance was valued by families in this study. Such information facilitated development of close relationships based on trust and familiarity with key medical staff. It can be concluded that the need
for facts and figures about TS is not in question. It is suggested that the widespread concern associated with perceived information needs was associated with when and how to share information with the girls across childhood and adolescence. The age at diagnosis also appeared to be significant in application of information to an individual girl. The dynamics of sharing information and using information in the best interests of girls directs the first suggestion for further research:

1. To develop and evaluate a route map for information sharing with girls with TS and their parents across a larger TS population to add to understanding of the issues and strategies parents and practitioners engage in at different developmental stages and ages at diagnosis.

Improving knowledge of CYP and their families in relation to their health needs sits within the remit of children’s nursing. Where medical conditions are complex, on-going and life-limiting, the role of the clinical nurse specialist is highlighted as central in facilitating coping and adaptation to the condition. Education and effective communication with families and children over time enables and promotes self-care management. Effective therapeutic relationships are at the heart of these activities. In this study the PENS role did not reflect a pattern of engagement seen across a range of other clinical nurse specialists working with children with long-term conditions. They were identified as having a very specific role in teaching and supporting the administration of GH. An on-going therapeutic relationship was not described. Parents were able to seek information about what they needed to know but the difficulty reported was in sharing sensitive information and judging the depth and timing of information shared with their daughter. This study was undertaken in a single tertiary centre and therefore these conclusions are specific to one way of working. This leads to the second area for further research:

2. To undertake a national review of the role of PENS in meeting the information needs of girls with TS and their parents

Within this study was the notion of the girls not being easily perceived as patients. This orientation to wellness and being normal may have implications on service delivery and the perception of nurses only needing to be involved in current management of medical aspects of the condition. This potentially negates the need for proactive involvement in
the information needs for girls and their parents, which may impact negatively on their understanding of the longer-term health issues. In these data, with the exception of infertility, there was limited reference by families to other long-term implications of TS. Women with TS need on-going follow up throughout their adult life, for example, they are known to be susceptible to osteoporosis, cardiovascular disease, hypertension associated with renal disorders, type 2 diabetes, thyroid disease; and sensorineural hearing loss (TSSS 2008). To increase awareness of this potential gap in information provision it is necessary to:

3. Examine the knowledge and understanding of the long-term implications of TS amongst girls with TS and their parents.

This study highlighted potential difficulties experienced by girls with TS and their parents in accessing accurate and relevant information. Two main areas were revealed as dominant themes in this study and are worthy of further research. One is the role of parents as primary information providers particularly when diagnosis was confirmed early in the girls’ life. The other is the need to develop a greater understanding of the loss experienced by mothers and fathers relating to probable infertility of their daughter. In the literature review, a study by Beresford (2000) was identified that recognised parents as the main source of information for children with chronic illness or disability. Beresford advocated that parents require a good understanding of the child’s condition, how it is treated and managed and the long-term implications for health and well-being. The current study did not set out to test parental knowledge but rather to develop an understanding of information needs from their perspective, however, there was some evidence of misunderstandings around the induction of puberty and genetic information. Parents also referred to the complexity and difficult nature of some information to which they were exposed. Clearer understanding of these needs could lead to development of specific learning material to facilitate parental understanding, thereby improving knowledge and understanding of girls about TS.

4. To determine parental knowledge and understanding of key components of TS and ascertain common areas of difficulty
Stress and anxiety are known to impede the information process. Mothers and fathers reported in this study that their daughter’s probable infertility was a cause of distress to them and in a small number of families an on-going sadness relating to this issue pervaded the interview. In one family this issue had prevented the sharing of information of the diagnosis with others and in another case the mother described the tension associated with fear of unanticipated disclosure. There appeared to be different responses and strategies for managing this loss across families and genders suggesting a complex mix of cognitive, social, emotional; and cultural issues. Sutton et al. (2005) identified infertility as a major cause of distress from childhood to mature adulthood in their sample of 97 participants with TS. They concluded that “protracted grief” of parents could be damaging in the long-term and interfere with adaptation processes. In this study the potential loss of motherhood as a social role and accepted identity for women emerged as an important aspect of the families’ experiences of having TS. Examining the meaning of probable infertility to these families, including the range of responses and strategies used in addressing it would be of value to practitioners and families.

5. Explore the impact of probable infertility of a daughter with TS on parents including their responses and mechanisms for managing information

The final area would be to conduct further research with girls and women who have been treated with GH and had puberty induced. There is a generational divide described by some families that influences their parenting style and perception of girls with TS and it is based on observations or research reporting their views of TS. More recent experiences of girls living with TS following medical management of short stature and pubertal development are not well reported and thus research in this area is required:

6. Examination of the similarities and differences in the social experiences of untreated women and women who received medical management of short stature and puberty.
8.4 Conclusion

Information needs reflect a complex interplay of factors including karyotype, phenotype, neurocognitive and behaviour profile, stereotype, medical profile, developmental issues, social functioning, attitude to the diagnosis, and motivation for using information. Information needs were also affected by the perceived impact of these factors on current and future social roles of girls with TS. Discrete perceptions provided various discourses about what it is to be a girl with TS, or parent of a daughter with TS. Through the personal accounts of the girls and their parents participating in this study a broad range of socially defined experiences have been uncovered, providing insights into how these affect their individually perceived information needs.

A clear understanding of the frames of reference for TS, shaped by the girls and their parents, forms the foundation for developing individual information strategies for families, which should include guidance as well as direct involvement on what information to share, when it should be shared and how it can be shared. The strategy to manage information should take account of timing, depth and breadth of information in addition to useful methods parents can use to engage their daughters in learning about the meaning of TS to them.
REFERENCE LIST


Williams B., Corlett J., Dowell J.S., Coyle J., Mukhopadhyay (2009) ”I’ve never not had it so I don’t know what it’s like to”: non-difference and biological
disruption among children and young people with cystic fibrosis. *Qualitative Health Research* 19,(10), 1143–1455.


APPENDIX 1-3

The Information leaflets were produced as A5 size booklets for the girls aged 8-11 and 12-16. For ease of presentation the information is presented here as an A4 document.

All contact details have been removed to protect the anonymity of the participants.
Appendix 1: Information for girls 12-16 years

This is an invitation to take part in a study about the information needs of girls with Turner syndrome and their parents.

Before you agree to take part it is important to know more about the project and why it is being done.

Please read this information and talk to other people about it if you want to.

Thank you for spending your time reading this leaflet

What is this project about?
This project is about working with girls like yourself with Turner syndrome and your parents to discover the sorts of information you need, and the best ways of finding out that information.

About me
My name is Jacquie Collin. I am a Children’s Nurse and I teach nursing students at Keele University in Staffordshire.

Currently I am doing a PhD degree at Manchester University.

Your chance to have a say …

This project gives you the chance to let doctors, nurses and other professionals know the sorts of information children and young people need as they grow up with Turner syndrome. It will also let them know useful ways of giving information or helping children and young people to find out things for themselves.

Who else is involved?

I will be talking to 20 girls between the ages of 8 years and 16 years and their parents.
**Why me?**

Your doctor has suggested that you may like to join in this project. I am inviting you to take part because you are a young person who has Turner syndrome. I am interested to talk to you about how you find things out and what you think has been helpful in learning about Turner syndrome.

**What would I have to do?**

If you are interested in joining the project I will visit you at home so that you can ask me any questions about the project. It will also give us an opportunity to get to know each other before we start the interview.

Taking part involves talking about Turner syndrome to me. I would like to hear what you think it is like living with Turner syndrome, how you find things out and what you think are helpful ways of getting information.

If you don’t mind I would like to tape record the interview and may make some notes. You can turn the tape recorder off at any time during the interview.

**Is it private and confidential?**

Everything we talk about will be confidential. I will not tell anyone else what you say and whatever I read will be private. Only if you share information that raises concern for your health or your safety will we need to tell someone else what we have discussed. We would need to find someone to help us to resolve the problem. It may be your parents or [Name Removed] Nurse specialist, or your doctor. We would decide together who to tell to ensure that you were helped.

When I write a report about the project I will make sure that the children and young people who take part cannot be identified. I will not use your name.

**What if I don't want to take part?**

That's OK.

I only want young people to take part if they really want to. Nobody will be angry or upset if you don't want to take part.

If you decide to take part and then change your mind, that's OK too.

**Yes, I do want to take part**
You or your parent(s) or the person who looks after you needs to complete the contact form and send it back to me. When I've heard from you, I'll get in touch to arrange to visit you.

Any questions?

You and your parent(s) or the person who looks after you can get in touch with me or my supervisors and we will answer your questions. Contact details are on the next page.

Thank you
Hello

My name is Jacquie Collin and I am a Children's Nurse. I would like to tell you about a project I am doing with girls with Turner syndrome and their parents.

Before you can make up your mind about helping you will need to understand all about the project. Please read this leaflet to find out more.

What’s the project about?
I am interested in hearing what you think it is like to have Turner syndrome and how you found out all about it.

Who else is helping with the project?
I will be talking to 20 girls between the ages of 8 years and 16 years and their parents.

Where will you talk to me?
I will come to your home at a time agreed with you, your parent(s) or the person who looks after you. I will talk to you privately but if you want someone else to come too that’s OK.
How will we remember what we say?

I will listen carefully and make notes. I will also tape record what we say if that is OK with you. You can hold the tape recorder and turn it off at any time during our talk.

Is it an important project? Yes!
It is important to hear from girls with Turner syndrome and their parents.

This project will give you the chance to let doctors, nurses and others know the sorts of information children need as they grow up with Turner syndrome. I will make a list of all the things that you and the other girls tell me are important and send it to girls in other parts of the country who also have Turner syndrome. This is called a questionnaire. All this information will help doctors and nurses understand more about what girls with Turner syndrome say is important to them. It will also help them find out useful ways of giving information or help children to find things out for themselves.

Will everything we talk about be private?
I will write a report about all the things that the girls helping in the project and their parents tell me. This will be printed in a magazine for doctors and nurses but your name will not be used.

If a problem crops up when we are talking we can tell your parent(s) about it or [name removed] Nurse specialist or your doctor.

What if I don't want to help?
That's OK,
Nobody will be cross or upset. I only want you to join in if you really want to.
If you say you want to join in and then change your mind that is OK too. When we are talking together you will have a red card to hold up if you do not want to answer a question or continue talking about something or you can switch off the tape recorder.

If you would like to help, What happens next?
Ask your Mum or Dad or the person who looks after you to complete the contact form and send it back to me. When I've heard from you I'll get in touch and arrange to come and see you.

Thank you for reading this information leaflet.
Any more questions?
You and your Parent(s) or the person who looks after you can email, phone or write to me or my supervisor if you want to ask anything else about the study.
Appendix 3: Information leaflet for parents

INFORMATION LEAFLET FOR PARENTS

Study Title:

Perceived Information Needs of girls with Turner syndrome and their parents.

I am writing to invite you (mother or father or both; guardian) and your daughter to participate in a study about the information needs of girls with Turner syndrome and their parents. I am a Children’s Nurse currently employed by Keele University to teach student nurses. My work on this study is for a PhD degree which I am undertaking at Manchester University.

What is the purpose of the study?

The purpose of this study is to develop clinical practice by understanding more clearly the sorts of information children and young people need as they grow up with Turner syndrome and also the information needs of their parents. I will be interviewing at least 20 girls with Turner syndrome between the ages of 8 and 16 years and their parents.

Why have you and your daughter been chosen?

Your daughter’s doctor has suggested that you and your daughter may wish to participate in the study because your daughter is aged between 8 and 16 years and you have experience of living with Turner syndrome. I have prepared a separate leaflet about the study for your daughter which you may wish to read together.

What would we have to do?

The study will involve me visiting your home, listening to your experiences and discussing how you have developed your understanding of Turner syndrome. I would arrange the visit with you to minimise any inconvenience to you and your family. I would like to carry out the interviews in
private, firstly with you and then with your daughter. I do not have a list of questions but I have identified some topics to guide the conversation with you and your daughter. Each interview would last about one hour and I would tape record our conversation. I may also take some notes. Everything we discuss will be confidential. The information collected will be used to develop a questionnaire that will be sent out nationally to girls with Turner syndrome. Health Professionals recognise the importance of providing information to girls and their parents. This study will help the doctors and nurses understand what information is most important to girls and parents including insight into what ways are considered the most useful.

If you would like to take part please complete the contact form and return it to me. When I have heard from you I will get in touch to arrange a visit. If you agree to participate you and your daughter will be asked to sign a consent form.

**Is it confidential?**

All information collected during the course of the research will be kept confidential. If a problem crops up during our conversation that we need to resolve, we will decide together who best can help us. Your doctor and [name removed] Nurse specialist are aware of this study and its aims. I intend to publish the findings and therefore I will make sure that those taking part are not identified in any way.

**Do we have to take part?**

No. It is up to you and your daughter to decide whether or not to take part. You are both free to withdraw at any time and without giving a reason. Your decisions about this will not affect the standard of care your child receives.

**Thank you for spending time to read this information**

If you have any questions or wish to clarify any aspect of the information in this leaflet please get in touch with me or my supervisors.
Appendix 4: Consent Form

CONSENT FORM

Title of Project: Perceived Information Needs of Girls with Turner Syndrome and their Parents’

Name of Researcher: Jacquie Collin

Please initial box

1. I confirm that I have read and understand the information sheet dated 18.11.06 (version 3) for the above study and have had the opportunity to ask questions.

2. I understand that my participation is voluntary and that I am free to withdraw at any time, without giving any reason, without my child’s medical care or legal rights being affected.

3. I understand that sections of any of my child’s medical notes may be looked at by responsible individuals from the University of Manchester or from regulatory authorities where it is relevant to my child’s participation in this research. I give permission for these individuals to have access to my child’s records.

4. I agree to take part in the above study and for my child to participate if they want to.

Name of Child
Name of Parent Signature Date
Name of Researcher Signature Date
Appendix 5: Assent Form

ASSENT FORM FOR CHILDREN/ YOUNG PERSON

Project title: Perceived information needs of girls with Turner Syndrome and their parents

............................. to circle all they agree with please:

Have you read (or had read to you) about this project? Yes/No
Has somebody else explained this project to you? Yes/No
Do you understand what this project is about? Yes/No
Have you asked all the questions you want? Yes/No
Have you had your questions answered in a way you understand? Yes/No
Do you understand it’s OK to stop taking part at any time? Yes/No
Are you happy to take part? Yes/No

If any answers are ‘no’ or you don’t want to take part, don’t sign your name!

If you do want to take part, please write your name and today’s date

Your name ___________________________
Date ___________________________

Your parent or guardian must write their name here too if they are happy for you to do the project

Print Name ___________________________
Sign ___________________________
Date ___________________________

The researcher who explained this project to you needs to sign too:

Print Name ___________________________
Sign ___________________________

Thank you for your help

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# Appendix 6: Pre-oestrogen: getting to a diagnosis

<table>
<thead>
<tr>
<th>Case</th>
<th>Concerns that triggered referral</th>
<th>Route to Specialist Endocrine Services</th>
<th>Comments</th>
</tr>
</thead>
<tbody>
<tr>
<td>C</td>
<td>Mothers continued concern about her daughter’s persistent swollen feet since birth</td>
<td>At birth Mum and her sister were concerned about the baby’s puffy hands and feet; seen by paediatrician, heart checked, told everything was fine. Midwife in hospital noticed small nails underdeveloped. No follow up. Community midwife reported swollen feet to GP but no response. Midwife advised mum to get a second opinion if the swelling unresolved. At 7 months a different GP is seen, did not know why they were swollen but as mum was worried referred to paediatrician. They seemed to know straight away, took some blood tests and 2 weeks later at 8pm they received a call to ask them to go to the hospital the next day. The chromosome analysis was not straightforward 46XY, paediatrician told parents not to tell family or friends; no further mention of TS. Then went to see Endocrinologist, further tests, “it was Turner’s but it was quite unusual”</td>
<td>“got a phone call at 8pm... they were thinking of TS but because like its strange the chromosomes quite unusual they rang us in the evening and said we must go down to (hospital) the next morning” (Mother) “so the chromosomes are 46XY that was just from a blood test em she said and advised us not to tell any family and friends .. ....we felt terrible as it wasn’t like that as long as it wasn’t anything life-threatening I could cope with anything I didn’t feel it was something to be ashamed of” (Mother) The endocrinologist was “quite different”, made it clear that the parents could tell people it was fine you shouldn’t bring up the child with a secret</td>
</tr>
</tbody>
</table>

Diagnosed aged 8 months
| **G** | Born with ambiguous genitalia at 26 weeks gestation, cared for in neonatal unit. | “When she was first born they thought she was a boy but they soon told us she was a girl”. Seen by Endocrinologist on neonatal unit. In hospital for 3 months before discharge. **Diagnosed soon after birth** | “I knew from the start... doctor told us everything soon after she was born... so as soon as she was born we knew what it was, about Turner’s syndrome, not having children and the height” |
| **H** | Parents’ concerned daughter was smaller than her twin sister. She had grommets inserted following recurrent ear infection. During a follow up consultation with ENT surgeon, Mother asked “now she’s got the grommets will she start growing”. ENT surgeon stated that was a bit of an old wives tale but if parents were concerned about their daughter’s height he would refer them to a Paediatrician. | Mother had always felt there was something wrong, “..I always felt from the word go that there was something – different- not quite right about her”. As she grew up there were lots of things including sternomastoid tumour, poor feeding, not growing or developing as well as twin sister, recurrent ear infections, headaches, therefore initially frequent consultations with HV, GP, and Physio. Later had investigations within private health care for headaches and recurrent ear infections before being referred to paediatrician for growth problem. Paediatrician wrote in letter he was investigating for TS. **Subsequent referral to Endocrinologist** **Diagnosed at 8 years** | Mother states that having seen girls with TS on the internet “I knew before he even, the paediatric guy, confirmed what it was, that she’d got it”.

Mother looked it up on the internet “it’s just her ..and I thought oh God”.

Parents felt that the diagnosis was made early enough for GH to positively impact on her final height.

“some children don’t find out until they get to puberty and then it’s too late to help them with their growth so that was a positive thing”.

| **J** | She was taken to the special care baby unit because she had | It was about a week before one of the doctors spoke to the parents to tell them their **“Everything was normal on the scans ... normal size and everything so when she was”** |
swallowed meconium and had problems with feeding. One of the paediatric registrars (on training circuit from nearby Children’s Hospital) noticed her puffy hands and feet. daughter had TS

The consultant paediatrician gave parents the initial information and then they were referred to the Endocrinologist.

### Diagnosed aged one week

born we didn’t really expect to have a daughter with TS” (Mother)

“just shock initially” (Father)

“(perhaps) some mistake on the scan because she was so particularly small (Mother)

Once parents had seen the Endocrinologist “then things came to light” (Father)

### K

At birth her parents noted she had a swollen right foot which the midwives said was positional and would go down. Mother’s previous pregnancy had resulted in a stillbirth and therefore the Paediatrician was asked to check the baby. German doctor on attachment knew exactly what the baby had and took blood tests.

Found out it was TS when she was five days old and parents were then referred to the Endocrinologist. They had their first consultation when their daughter was ten days old.

### Diagnosed confirmed at five days old

When the results came back “the German doctor was not around and the others did not know much about it.”

“we were pretty shell shocked in those first few days” (Father)

“And it was only through seeing (Endocrinologist) that we felt that somebody who actually knew something was going to put us through hopefully what would be the right process” (Mother).

### L

“She wasn’t putting any weight on, so we said we better get her

Parents took daughter to GP who referred them to local paediatrician. She had some

“It was a big shock” (Mother)
**checked out”** (Mother).

<table>
<thead>
<tr>
<th><strong>M</strong></th>
<th>School nurse raised concerns about her height and asked if she could refer her to Community Paediatrician.</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Mother concerned about daughter’s growth and asked for referral to Paediatricians when she was 3 months old but they did not agree that anything was wrong. No follow up. When she was 9 months old she was in size 3 month clothes and was unwell with lots of ear infections. Mother asked HV to weigh the baby and shared her concerns. No action taken. School nurse raised concerns about daughter’s height and referred her to community paediatrician. Daughter examined by Registrar working with Community Paediatrician who took blood samples. Community paediatrician arranged to meet parents with results and confirmed diagnosis of TS. Subsequently referred to Endocrinologist.</td>
</tr>
</tbody>
</table>
|       | “there were lots of bits and pieces but no one took any notice”  
“believe it or not [Dr] said she’s undersize and underweight and I thought no (sarcasm)”  
“.fair play to the young doctor, I think [Dr] was the registrar, as soon as she (daughter) walked in the room [Dr] knew what they was looking for…”  
Community paediatrician “brilliant in all aspects”  
“.it just tied everything up I wasn’t imagining things there were reasons why she hadn’t grown, the ear infections, problems with her nails, there was a long list”  
“devastation and relief, more devastation at the time…” |

<table>
<thead>
<tr>
<th><strong>N</strong></th>
<th>Anomalies found on routine 19 week scan. Amniocentesis confirmed diagnosis of TS at 24 week</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Obstetrician told parents results of amniocentesis. Referred to consultant paediatrician who gave limited and inaccurate</td>
</tr>
<tr>
<td></td>
<td>“it’s not fatal, I couldn’t speak because at no point had anybody said it could be, it hadn’t entered our</td>
</tr>
</tbody>
</table>
Diagnosis made and referred to endocrinologist.

As soon as their daughter was born the father noticed she had swollen hands and feet and was insistent that the paediatrician should examine her.

Father alerted midwife to baby’s abnormally swollen hands and feet. She said they would drain however father was insistent that a paediatrician should be called. She agreed something was wrong and asked consultant paediatrician to examine baby. Parents told it may be TS. Blood samples sent for chromosomes. Parents advised not to look anything up before the results came back.

“[Dr] didn’t know what she’d look like when she was born and she might not make it to be born, her heart could stop at any point. So I stopped work straight away” (Mother)

“Paediatrician said stop getting upset, she’s a beautiful healthy little girl apart from she’s got TS and I’ll put you in touch with people who will explain why it happened and what may or may not be wrong” (Father)

Initial consultation was “very clinical” (Father) “it’s a bit embarrassing to keep saying I don’t understand what you’re saying to me”

<table>
<thead>
<tr>
<th>gestation.</th>
<th>information.</th>
<th>heads” (Mother)</th>
</tr>
</thead>
<tbody>
<tr>
<td>“you’re having a little girl and she’s got TS. We were made up” (Father)</td>
<td>Paediatrician was well meaning but didn’t have information parents needed.</td>
<td>“My Dad’s a paramedic so we found an old medical book and looked under Turners and there was this list, absolutely worse case scenario, mental retardation we picked up on straight away” (Mother)</td>
</tr>
<tr>
<td>Diagnosed at 24 week gestation</td>
<td>“They said yes it is and I started crying” (Mother)</td>
<td>“Paediatrician said stop getting upset, she’s a beautiful healthy little girl apart from she’s got TS and I’ll put you in touch with people who will explain why it happened and what may or may not be wrong” (Father)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>P</th>
<th>As soon as their daughter was born the father noticed she had swollen hands and feet and was insistent that the paediatrician should examine her.</th>
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</tr>
</thead>
<tbody>
<tr>
<td>Father alerted midwife to baby’s abnormally swollen hands and feet. She said they would drain however father was insistent that a paediatrician should be called. She agreed something was wrong and asked consultant paediatrician to examine baby. Parents told it may be TS. Blood samples sent for chromosomes. Parents advised not to look anything up before the results came back.</td>
<td>“They said yes it is and I started crying” (Mother)</td>
<td>“Paediatrician said stop getting upset, she’s a beautiful healthy little girl apart from she’s got TS and I’ll put you in touch with people who will explain why it happened and what may or may not be wrong” (Father)</td>
</tr>
<tr>
<td>Diagnosis made and referred to endocrinologist.</td>
<td>“Paediatrician said stop getting upset, she’s a beautiful healthy little girl apart from she’s got TS and I’ll put you in touch with people who will explain why it happened and what may or may not be wrong” (Father)</td>
<td>“Initial consultation was “very clinical” (Father) “it’s a bit embarrassing to keep saying I don’t understand what you’re saying to me”</td>
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</table>
## Appendix 7: Post-oestrogen group: getting to a diagnosis

<table>
<thead>
<tr>
<th>Case</th>
<th>Concerns that triggered referral</th>
<th>Route to Specialist Endocrine Services</th>
<th>Comments</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>“lots of things”, including delayed speech, coordination, and heart problem. When she was about 6 years old paediatrician asked parents if they were concerned about their daughter’s height. They said “yes she’s small” but they had not raised it because they were a small family.</td>
<td>Followed up at the heart clinic and by paediatrician because she had been “very poorly ... she was all bloated at birth”. (Mother) Paediatrician did some blood tests and told parents their daughter had TS. Subsequent referral to Paediatric Endocrinologist.</td>
<td>“once they put it all together by (paediatrician) y’know it was just it was like a weight falling off your shoulders, yes it’s something now, they know what it is and they can do something about it, it all makes sense.... before it was so frustrating we wanted somebody to, why didn’t they pull it all together at the different clinics all the time? (Father) Maybe they’ve not seen, I dunno depends who you get doesn’t it, whether they’ve actually seen it before (Mother)</td>
</tr>
<tr>
<td>B</td>
<td>When she was 5 yrs old the Paediatrician said she wasn’t growing well and he saw her 12 months later. Blood tests were carried out at this appointment.</td>
<td>Followed up from birth with a heart problem; later had speech problems. Paediatrician explained diagnosis, referred them to Endocrinologist following blood tests.</td>
<td>“we didn’t expect anything that night we came back and they explained exactly what it was and from then on we just went to see Dr (Endocrinologist)” (Mother)</td>
</tr>
<tr>
<td>D</td>
<td>“I noticed that from the age of six really, didn’t seem to be growing but</td>
<td>“… and my doctor then referred us to a lady in (local hospital) who took</td>
<td>“it came out of the blue really, it was a big shock.”</td>
</tr>
</tbody>
</table>

Diagnosed at age 7 yrs

Diagnosed at age 6 yrs
<table>
<thead>
<tr>
<th>ID</th>
<th>Description</th>
<th>Source</th>
<th>Diagnosis Age</th>
<th>Notes</th>
</tr>
</thead>
<tbody>
<tr>
<td>E</td>
<td>Games teacher said her coordination was not very good and she seemed ‘backward’ compared to the other girls and suggested her parents had her ‘seen’.</td>
<td>Referred to Paediatrician straight away where tests were carried out and diagnosis made. Referred to Endocrinologist</td>
<td>Diagnosed at age 9 yrs</td>
<td>“several times I’d been to the doctors because she was very small and they always said she was below the centile but she was fine” (Mother)</td>
</tr>
<tr>
<td>F</td>
<td>Community Paediatrician concerned about height at School medical</td>
<td>Referred to Paediatrician at local hospital for tests and told diagnosis. Referred to Paediatric Endocrinologist</td>
<td>Diagnosed at age 13 yrs 8 months</td>
<td>“we didn’t know there was anything wrong” (Mother)</td>
</tr>
<tr>
<td>I</td>
<td>“she just wasn’t growing, we noticed and we were concerned”. “we noticed from the age of 5 yrs</td>
<td>“GP picked up it was Turner syndrome” and then they were referred to the paediatrician at the local hospital who referred them to paediatric endocrinologist where the diagnosis was confirmed.</td>
<td>Diagnosed at age: 8yrs</td>
<td>“all credit goes to our GP” “sent to (Specialist clinic) and from there she was diagnosed with TS” (Father)</td>
</tr>
</tbody>
</table>
Appendix 8: Tips for teachers

10 tips that can be easily applied when teaching a child with Turner syndrome

- Try to ensure that the child is seated so that she is facing the teacher for the majority of the time.

- Make eye contact with the girl whenever possible (This may not be easy for her).

- Use handouts, rather than the blackboard, whenever possible.

- Try to keep background noise to a minimum.

- When giving an instruction start with the girl’s name and finish with her name.

- Give full and detailed instructions and repeat if possible.

- Encourage the use of visual aids and colour to aid memory.

- Set realistic targets and award with lots of praise.

- If not reaching academic targets then praise helpfulness etc.

- Listen to her concerns and treat her fairly and this will help the girl with TS reach her full potential and become a valued member of the class.

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Appendix 9: Real World Validation

Comments from parents following presentation of early findings at TSSS annual conference
The perceived information needs of girls with Turner syndrome and their parents

General Comments

I would welcome any comments you may have about the content of this presentation.

Totally agree with the mums wanting to read every single (book/leaflet/internet) piece of information — when we got the diagnosis I was 28 weeks pregnant and I stayed up all night reading the simple book etc. My husband read a couple of chapters & then stopped.

The 2 hardest things we feel at the moment are — how to tell her/when/in what way (she is only 3 now) and the uncertainties about what we’ll be facing in the future.

The infertility aspect is the most painful of anything by far.
The perceived information needs of girls with Turner syndrome and their parents

General Comments

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I agree with the comments about the depth of sadness about infertility. I felt I could 'cope' with everything else i.e. short stature etc as long as she was healthy but felt the future sadness on her behalf. I felt guilty in a way that I'd had my children so easily and that she wouldn't. I worry about her future relationships and hope desperately that she'll find someone patient and understanding and just feel it an added complication in life.

I am looking into egg collection
and freeze it for her future use in case she decides to use them. I hope this is successful and that I am able to give her that option. In a way it feels like a placation of my own guilt about her condition.

I often explain her condition in scientific terms, talking about chromosomes and risk, but often wonder afterwards whether it was perceived that it's a genetic problem that I have passed onto her.
The perceived information needs of girls with Turner syndrome and their parents

General Comments

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I would really have welcomed some information on when and how to talk about puberty / fertility issues.

I waited and waited and then read an article in the TSSS magazine about infertility came to me and said "Mum, will I be able to have children?"

That was not what I wanted or intended for her to find out, but by hanging on to the information that was less it turned out...
The perceived information needs of girls with Turner syndrome and their parents

General Comments

I would welcome any comments you may have about the content of this presentation.

The information given related a lot to particularly how my husband + I found to deal with info at diagnosis. And also infertility issues. I personally when I knew we didn't have medical issues i.e. heart prob should have found the infertility issue heartbreaking. Knowing she wouldn't experience what I had just experienced to get her. However what I have learnt from your discussion in talking with other parents is that there is more than one way to become a mother + its Charlotte issue to deal with when she so wishes.
The perceived information needs of girls with Turner syndrome and their parents

General Comments

I would welcome any comments you may have about the content of this presentation.

Excellent, would have liked to chat all afternoon. Half I totally agreed with half I didn't - typical T.S.

So much depends on time of diagnosis & general attitude of parents.

My daughter would have liked more continuity with the Drs she was seeing; she turned everything to a positive.
Appendix 10:

Mothers and Fathers’ comments on infertility across all cases
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<tr>
<th>Case/Line.</th>
<th>Mothers</th>
<th>Case/Line</th>
<th>Fathers</th>
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<tr>
<td>A:L408</td>
<td>Yes well but you see I struggle as well with her and babies possibly not being able to have them probably not being able to have them and that’s hard, I don’t know just don’t know how you deal with that one. I mean things can change between now and things, science moves all the time doesn’t it? So there might not be the issues for a girl now as there are when she’d be she’d be considering families but I don’t know</td>
<td>Left interview to take siblings out and was not present when fertility was discussed</td>
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<td>B: L279</td>
<td>I wouldn’t accept it’s black and white as you can’t have kids or whatever cause I remember cause that was always my concern right at the beginning mentioning it to Dr name (endocrinologist) and immediately the thing I thought was can I donate eggs or what can I do? What can we do?</td>
<td>L:287</td>
<td>Well you don’t know what B wants to do. We’ve talked about the fact that not everybody has babies</td>
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<td>L:292</td>
<td>That is your problem it’s not B’s problem cause B not got to that problem yet and your worrying about it ‘cause its nothing do with you at the end of the day it’s B</td>
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<td></td>
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<td>L567</td>
<td>...... you’ve (his wife) got to have a handle on it.</td>
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<td>C: L</td>
<td>Father not interviewed</td>
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<td>I worry, you worry that, I don’t know ‘cos I know that if I couldn’t have children it would be quite a big, even though it’s not the end of the world, it can be the end of the world to somebody can’t it, be quite a big thing … I’d rather adopt although you shouldn’t give your own opinion but donor egg I find quite big, I don’t know it gets so complicated</td>
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<th>D:L174</th>
<th>D:L18</th>
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<td>even though you know that she can’t have children at this moment in time you just don’t know by the time she gets to twenty four what might be available so I think I sort of take everything with a pinch of salt even though deep down probably know but there again she may be a child that doesn’t want to have children and she might decide she doesn’t want a family and who are we, it’s her choice at the end of the day well if she was perfectly normal she may not have had children anyway a she may not have wanted them and she still may not have been able to have them she may have been somebody who constantly miscarriages you just don’t know you?</td>
<td>Yeah, it was a big shock, a shock at the time even more a big shock for you rather than me I think I took it more laid back but what probably distresses us both more than anything is knowing that R can’t have children at a later date, she knows this. Unless she has test tube or fertility treatment er ignoring that she won’t have a child of her own genes.</td>
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<td>E:69</td>
<td>Infertility, it’s always been my main concern. The thought of, if I’d not been able to have children I’d have been suicidal, that was the main thing for me, I mean she may not want children but for me that was the biggest concern</td>
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<td>F:98</td>
<td>they were saying she hadn’t got any ovaries but she can have IVF and things that have come on a lot more now than what they were twenty years ago. Its like she’s had, when she’s ready, it like you don’t know what they can do then. But its just the fact that, its hanging over us really its like she’ll, you should be able, I mean we had problems conceiving her anyway but you should be able to make your mind up you think oh God. But she might decide she doesn’t want any but its just because that choice is taken away from her really, but after that we just thought well at the and of the day they are not turning round and saying she’s only got so long to live and they can do stuff for her.</td>
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<td>F:108</td>
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<td>G: L211-15</td>
<td>Maybe in ten years they will have come up with new technologies to help her become a mother one day because I think as a woman it is very important erm even if she was married and very happy the one thing you would want is a child so I do know, I just hope there will be some way.</td>
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<td>H: L241</td>
<td>Yes and when I first found out I suppose, because that’s the bit that bothers me the most about the Turner syndrome. I thought everything else you can probably go through life and cope with but that’s the bit that’s the hardest….</td>
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<td>L: 245</td>
<td>And knowing how I wanted them so much and IVF wasn’t straightforward you know. To know that’s she’s probably got to go through all that. You know the success rates and all that but then it’s another ten years or so and things improve so…</td>
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<td>L: 249</td>
<td>But that’s my initial feeling of complete sorrow for her really</td>
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<td>I: L</td>
<td><em>Mother agreed with husband’s comments</em></td>
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<td>J:L</td>
<td>This child had learning difficulties which were severe enough for parents to indicate that their daughter would be dependent throughout her life therefore fertility was not raised an issue</td>
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<td>K:L406</td>
<td>In a way its nothing to do with us because whether she wants children or not is her own decision but we want to be able to give her choice if possible</td>
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<td>L:694</td>
<td>My fear was she wasn’t going to be a proper girl because she couldn’t have children she’s physically not a proper girl and then not a proper woman. I thought you know it’s not what monster have I given birth to because that is far too extreme but it was that kind of fear</td>
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<td>L:L38</td>
<td>When her time comes it’ll be really hard for her.</td>
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<td>K:L444</td>
<td>Its options isn’t it, we can sort out the financial side she’ll have money to do things but this [egg donation] maybe her only option in reality ten or fifteen years time there’ll be all sorts of things that have happened things change so much er but if you are sort of taking more options so she had choice further down the line, but still she may not want to have children you never know but it’s up to her.</td>
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L:44  I feel really sorry for her, I mean I’d freeze me eggs for her or have one for her y’know but I’ll be older then so it’s gonna be hard

M:L191  That was a bit of a killer

L199  we did explain to her that the way the medical profession is, she might be able to have children but assisted with maybe the help of her sisters, or the medics and if not she adopts

L474  she said Mum, will I be able to have kids? As a mother, or if anybody has ever gone through it, it was devastation, I just sat there and all I wanted to do was burst into tears and hold her, but, I had to say very calm and collective, well, it’s possible, but, it’s highly unlikely at the moment and I explained about the scan

N:L  I told her a long time ago. I’d always said ‘cos I was originally told I couldn’t have children and then I had S erm

N:L  Some of the issues tend to be more like, I get more involved in the psychological side of things rather than the more feminine issues, you (Wife) deal with those because I would feel
<table>
<thead>
<tr>
<th>P:L865</th>
<th>All my worries are about having kids</th>
<th>L:868</th>
<th>Well what can we do about it now, give her a hug and a cuddle, just make sure she’s happy</th>
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<td>so I’ve always said not everyone can have them and it kept coming up over and over so I did say well actually you probably can’t and she was fine with it</td>
<td>awkward about that</td>
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<td>Int: So issues around puberty, infertility</td>
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<td>R2: Yeah, <em>Wife’s name</em> talk more about that because she understands more than I would about how sensitive that all is.</td>
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