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Employment

Research Fellow

Research Fellow
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1 Aug 2016 → present

Research outputs

MRSD: a quantitative approach for assessing suitability of RNA-seq in the investigation of mis-splicing in Mendelian disease

Rowlands, C., Taylor, A., Rice, G., Whiffin, N., Hall, H. N., Newman, W., Black, G., kConFab Investigators, O'Keefe, R., Hubbard, S., Douglas, A. G. L., Baralle, D. & Briggs, T., 12 Dec 2021, (Accepted/In press) In: *American Journal of Human Genetics*.

Enhanced cGAS-STING-dependent interferon signaling associated with mutations in ATAD3A

Lepelley, A., Mina, E. D., Nieuwenhove, E. V., Waumans, L., Fraitag, S., Rice, G. I., Dhir, A., Frémond, M-L., Rodero, M. P., Seabra, L., Carter, E., Bodemer, C., Buhas, D., Callewaert, B., Lonlay, P. D., Somer, L. D., Dymont, D. A., Faes, F., Grove, L., Holden, S. & 10 others, Hully, M., Kurian, M. A., McMillan, H. J., Suetens, K., Tynismaa, H., Chhun, S., Wai, T., Wouters, C., Bader-Meunier, B. & Crow, Y. J., 13 Aug 2021, In: *Journal of Experimental Medicine*. 218, 10, e20201560.
DOI: 10.1084/jem.20201560

Differential levels of IFN α subtypes in autoimmunity and viral infection

Bondet, V., Rodero, M. P., Posseme, C., Bost, P., Decalf, J., Haljasmägi, L., Bekaddour, N., Rice, G. I., Upasani, V., Herbeuval, J-P., Reynolds, J. A., Briggs, T. A., Bruce, I. N., Mauri, C., Isenberg, D., Menon, M., Hunt, D., Schwikowski, B., Mariette, X., Pol, S. & 5 others, Rozenberg, F., Cantaert, T., Eric Gottenberg, J., Kisand, K. & Duffy, D., 1 Aug 2021, In: *Cytokine*. 144, 155533.
DOI: 10.1016/j.cyto.2021.155533

Differential Expression of Interferon-Alpha Protein Provides Clues to Tissue Specificity Across Type I Interferonopathies

Lodi, L., Melki, I., Bondet, V., Seabra, L., Rice, G. I., Carter, E., Lepelley, A., Martin-Niçlós, M. J., Al Adba, B., Bader-Meunier, B., Barth, M., Blauwblomme, T., Bodemer, C., Boespflug-Tanguy, O., Dale, R. C., Desguerre, I., Ducrocq, C., Dulieu, F., Dumaine, C., Ellul, P. & 19 others, Hadchouel, A., Hentgen, V., Hié, M., Hully, M., Jeziorski, E., Lévy, R., Mochel, F., Orcesi, S., Passemard, S., Pouletty, M., Quartier, P., Renaldo, F., Seidl, R., Shetty, J., Neven, B., Blanche, S., Duffy, D., Crow, Y. J. & Frémond, M-L., 1 Apr 2021, In: *Journal of clinical immunology*. 41, 3, p. 603-609 7 p.
DOI: 10.1007/s10875-020-00952-x

LACC1 deficiency links juvenile arthritis with autophagy and metabolism in macrophages

Omarjee, O., Mathieu, A-L., Quiniou, G., Moreews, M., Ainouze, M., Frachette, C., Melki, I., Dumaine, C., Gerfaud-Valentin, M., Duquesne, A., Kallinich, T., Tahir Turanli, E., Malcus, C., Viel, S., Pescarmona, R., Georgin-Lavialle, S., Jamilloux, Y., Larbre, J-P., Sarabay, G., Magnotti, F. & 9 others, Rice, G. I., Bleicher, F., Reboulet, J., Merabet, S., Henry, T., Crow, Y. J., Faure, M., Walzer, T. & Belot, A., 1 Mar 2021, In: *The Journal of experimental medicine*. 218, 3, 20201006.
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From diagnosis to prognosis: Revisiting the meaning of muscle ISG15 overexpression in juvenile inflammatory myopathies

Hou, C., Durrleman, C., Periou, B., Barnerias, C., Bodemer, C., Desguerre, I., Quartier, P., Melki, I., Rice, G. I., Rodero, M. P., Charuel, J-L., Relaix, F., Bader-Meunier, B., Authier, F. J. & Gitiaux, C., Jan 2021, In: *Arthritis & rheumatology (Hoboken, N.J.)*.
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Overview of STING-Associated Vasculopathy with Onset in Infancy (SAVI) Among 21 Patients

Frémond, M-L., Hadchouel, A., Berteloot, L., Melki, I., Bresson, V., Barnabei, L., Jeremiah, N., Belot, A., Bondet, V., Brocq, O., Chan, D., Dagher, R., Dubus, J-C., Duffy, D., Feuillet-Soummer, S., Fusaro, M., Gattorno, M., Insalaco, A.,

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cGAS-mediated induction of type I interferon due to inborn errors of histone pre-mRNA processing

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JAK Inhibition in the Aicardi-Goutieres Syndrome

Neven, B., Al Adba, B., Hully, M., Desguerre, I., Pressiat, C., Boddaert, N., Duffy, D., Rice, G. I., Seabra, L., Fremond, M-L., Blanche, S. & Crow, Y. J., 26 Nov 2020, In: *New England Journal Of Medicine*. 383, 22, p. 2190-2193

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Mutations in COPA lead to abnormal trafficking of STING to the Golgi and interferon signaling

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Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum

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Expanding the clinical spectrum of Fowler syndrome: Three siblings with survival into adulthood and systematic review of the literature

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Clinical Reasoning: A 25-year-old woman with recurrent episodes of collapse and loss of consciousness

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Biallelic mutations in NRROS cause an early onset lethal microgliopathy

Smith, C., McColl, B. W., Patir, A., Barrington, J., Armishaw, J., Clarke, A., Eaton, J., Hobbs, V., Mansour, S., Nolan, M., Rice, G. I., Rodero, M. P., Seabra, L., Uggenti, C., Livingston, J. H., Bridges, L. R., Jeffrey, I. J. M. & Crow, Y. J., May 2020, In: *Acta Neuropathologica*. 139, 5, p. 947-951 5 p.

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Contribution of rare and predicted pathogenic gene variants to childhood-onset lupus: a large, genetic panel analysis of British and French cohorts

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Genetic polymorphism in C3 is associated with progression in chronic kidney disease (CKD) patients with IgA nephropathy but not in other causes of CKD

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Use of ruxolitinib in COPA syndrome manifesting as life-threatening alveolar haemorrhage

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Anti-MDA5 juvenile idiopathic inflammatory myopathy: a specific subgroup defined by differentially enhanced interferon- α signalling

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PSMB10, the last immunoproteasome gene missing for PRAAS

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HETEROZYGOUS MUTATIONS IN COPA ARE ASSOCIATED WITH ENHANCED TYPE I INTERFERON SIGNALLING

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CLINICAL AND SEROLOGICAL FEATURES OF INCREASED INTERFERON-ALPHA ACTIVITY IN AN UNSELECTED CONNECTIVE TISSUE DISEASE COHORT

Reynolds, J. A., Briggs, T. A., Rice, G. I., Darmalinggam, S., Bondet, V., Bruce, E., Khan, M., Haque, S., Chinoy, H., Herrick, A. L., McCarthy, E. M., Zeef, L., Hayes, A., Duffy, D., Parker, B. & Bruce, I. N., Apr 2019, In: *Rheumatology*. 58, p. 194-194

A child with severe juvenile dermatomyositis treated with ruxolitinib (vol 141, pg e80, 2018)

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Bloom syndrome protein restrains innate immune sensing of micronuclei by cGAS

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DOI: 10.1084/jem.20181329

Cardiac valve involvement in ADAR-related type I interferonopathy

Crow, Y., Keshavan, N., Barbet, J. P., Bercu, G., Bondet, V., Boussard, C., Dedieu, N., Duffy, D., Hully, M., Giardini, A., Gitiaux, C., Rice, G. I., Seabra, L., Bader-Meunier, B. & Rahman, S., 2019, In: *Journal of Medical Genetics*.
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Type I interferon in patients with systemic autoimmune rheumatic disease is associated with haematological abnormalities and specific autoantibody profiles

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VARIABLE EXPRESSION IN SAMHD1 - ASSOCIATED FAMILIAL AICARDI-GOUTIERES SYNDROME

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Sine causa tetraparesis: A pilot study on its possible relationship with interferon signature analysis and Aicardi Goutières syndrome related genes analysis

AGS study group, Dec 2018, In: *Medicine (Philadelphia)*. 97, 52, p. e13893
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A child with severe juvenile dermatomyositis treated with ruxolitinib

Aeschlimann, F. A., Frémond, M., Duffy, D., Rice, G. I., Charuel, J., Bondet, V., Saire, E., Neven, B., Bodemer, C., Balu, L., Gitiaux, C., Crow, Y. J. & Bader-meunier, B., 1 Nov 2018, In: *Brain*. 141, 11, p. e80
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JAK 1/2 Blockade in MDA5 Gain-of-Function

McLellan, K. E., Martin, N., Davidson, J. E., Cordeiro, N., Oates, B. D., Neven, B., Rice, G. I. & Crow, Y. J., Nov 2018, In: *Journal of clinical immunology*. 38, 8, p. 844-846
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Haskell, G. T., Mori, M., Powell, C., Amrhein, T. J., Rice, G. I., Bailey, L., Strande, N., Weck, K. E., Evans, J. P., Berg, J. S. & Kishnani, P., 1 Oct 2018, In: *Cold Spring Harbor Molecular Case Studies*. 4, 5, p. a002758
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LUPUS ENGELURE FAMILIAL SUR TROIS GENERATIONS : A PROPOS DE QUATRE CAS

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COPA syndrome restricted to life-threatening alveolar hemorrhages: clinical, pathological, molecular and biological characterization

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Measurement of Interferon Alpha Expression Using Multiple Methodologies Identifies a Signature in a Subgroup of Connective Tissue Disease Patients with Haematological Abnormalities

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Comment on: 'Aberrant tRNA processing causes an autoinflammatory syndrome responsive to TNF inhibitors' by Giannelou et al: mutations in TRNT1 result in a constitutive activation of type I interferon signalling

Frémond, M-L., Melki, I., Kracker, S., Bondet, V., Duffy, D., Rice, G. I., Crow, Y. J. & Bader-Meunier, B., 1 Jun 2018, (E-pub ahead of print) In: *Annals of the rheumatic diseases*.
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An open label trial of JAK 1/2 blockade in progressive IFIH1 associated neuroinflammation

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Autosomal dominant early-onset spastic paraparesis with brain calcification due to IFIH1 gain-of-function

Ruau, L., Rice, G., Cabrol, C., Piard, J., Rodero, M., van Eyk, L., Boucher-Brischoux, E., Maertens de Noordhout, A., Mare, R., Scalais, E., Pauly, F., Debray, F. G., Dobyns, W. B., Uggenti, C., Park, J. W., Hur, S., Livingstone, J. H., Crow, Y. & Van Maldergem, L., 2018, In: *Human Mutation*. 39, 8
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Efficacy of JAK1/2 inhibition in the treatment of chilblain lupus due to TREX1 deficiency

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