# Gillian I Rice

## List of Publications by Citations



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|---|---|---|---|
| 115 papers | 9,164 citations | 47 h-index | 95 g-index |
| 132 ext. papers | 11,129 ext. citations | 9.6 avg, IF | 5.3 L-index |

| # | Paper | IF | Citations |
|---|---|---|---|
| 115 | HIV-1 restriction factor SAMHD1 is a deoxynucleoside triphosphate triphosphohydrolase. *Nature*, **2011**, 480, 379-82 | 50.4 | 578 |
| 114 | Mutations in ADAR1 cause Aicardi-Goutières syndrome associated with a type I interferon signature. *Nature Genetics*, **2012**, 44, 1243-8 | 36.3 | 521 |
| 113 | Mutations involved in Aicardi-Goutières syndrome implicate SAMHD1 as regulator of the innate immune response. *Nature Genetics*, **2009**, 41, 829-32 | 36.3 | 507 |
| 112 | Evaluation of angiotensin-converting enzyme (ACE), its homologue ACE2 and neprilysin in angiotensin peptide metabolism. *Biochemical Journal*, **2004**, 383, 45-51 | 3.8 | 462 |
| 111 | Gain-of-function mutations in IFIH1 cause a spectrum of human disease phenotypes associated with upregulated type I interferon signaling. *Nature Genetics*, **2014**, 46, 503-509 | 36.3 | 376 |
| 110 | Characterization of human disease phenotypes associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR, and IFIH1. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 296-312 | 2.5 | 321 |
| 109 | Human intracellular ISG15 prevents interferon-α over-amplification and auto-inflammation. *Nature*, **2015**, 517, 89-93 | 50.4 | 311 |
| 108 | Clinical and molecular phenotype of Aicardi-Goutieres syndrome. *American Journal of Human Genetics*, **2007**, 81, 713-25 | 11 | 310 |
| 107 | Inherited STING-activating mutation underlies a familial inflammatory syndrome with lupus-like manifestations. *Journal of Clinical Investigation*, **2014**, 124, 5516-20 | 15.9 | 294 |
| 106 | Heterozygous mutations in TREX1 cause familial chilblain lupus and dominant Aicardi-Goutieres syndrome. *American Journal of Human Genetics*, **2007**, 80, 811-5 | 11 | 286 |
| 105 | Assessment of interferon-related biomarkers in Aicardi-Goutières syndrome associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, and ADAR: a case-control study. *Lancet Neurology, The*, **2013**, 12, 1159-69 | 24.1 | 267 |
| 104 | SAMHD1 restricts HIV-1 reverse transcription in quiescent CD4(+) T-cells. *Retrovirology*, **2012**, 9, 87 | 3.6 | 254 |
| 103 | Mitochondrial double-stranded RNA triggers antiviral signalling in humans. *Nature*, **2018**, 560, 238-242 | 50.4 | 211 |
| 102 | Detection of interferon alpha protein reveals differential levels and cellular sources in disease. *Journal of Experimental Medicine*, **2017**, 214, 1547-1555 | 16.6 | 192 |
| 101 | Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. *Nature Genetics*, **2012**, 44, 338-42 | 36.3 | 186 |
| 100 | Tartrate-resistant acid phosphatase deficiency causes a bone dysplasia with autoimmunity and a type I interferon expression signature. *Nature Genetics*, **2011**, 43, 127-31 | 36.3 | 173 |
| 99 | Efficacy of the Janus kinase 1/2 inhibitor ruxolitinib in the treatment of vasculopathy associated with TMEM173-activating mutations in 3 children. *Journal of Allergy and Clinical Immunology*, **2016**, 138, 1752-1755 | 11.5 | 141 |

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| 98 | Circulating activities of angiotensin-converting enzyme, its homolog, angiotensin-converting enzyme 2, and neprilysin in a family study. *Hypertension*, **2006**, 48, 914-20 | 8.5 | 135 |
| 97 | A specific IFIH1 gain-of-function mutation causes Singleton-Merten syndrome. *American Journal of Human Genetics*, **2015**, 96, 275-82 | 11 | 134 |
| 96 | The SKIV2L RNA exosome limits activation of the RIG-I-like receptors. *Nature Immunology*, **2014**, 15, 839-45 | 19.1 | 131 |
| 95 | ACEH/ACE2 is a novel mammalian metallocarboxypeptidase and a homologue of angiotensin-converting enzyme insensitive to ACE inhibitors. *Canadian Journal of Physiology and Pharmacology*, **2002**, 80, 346-53 | 2.4 | 131 |
| 94 | Protein kinase c deficiency causes mendelian systemic lupus erythematosus with B cell-defective apoptosis and hyperproliferation. *Arthritis and Rheumatism*, **2013**, 65, 2161-71 | | 119 |
| 93 | Type I interferon-mediated autoinflammation due to DNase II deficiency. *Nature Communications*, **2017**, 8, 2176 | 17.4 | 111 |
| 92 | Human disease phenotypes associated with mutations in TREX1. *Journal of Clinical Immunology*, **2015**, 35, 235-43 | 5.7 | 108 |
| 91 | Disease-associated mutations identify a novel region in human STING necessary for the control of type I interferon signaling. *Journal of Allergy and Clinical Immunology*, **2017**, 140, 543-552.e5 | 11.5 | 103 |
| 90 | SAMHD1 is a nucleic-acid binding protein that is mislocalized due to aicardi-goutières syndrome-associated mutations. *Human Mutation*, **2012**, 33, 1116-22 | 4.7 | 103 |
| 89 | Neprilysin, obesity and the metabolic syndrome. *International Journal of Obesity*, **2011**, 35, 1031-40 | 5.5 | 102 |
| 88 | Assessment of Type I Interferon Signaling in Pediatric Inflammatory Disease. *Journal of Clinical Immunology*, **2017**, 37, 123-132 | 5.7 | 94 |
| 87 | Expression of Cyclic GMP-AMP Synthase in Patients With Systemic Lupus Erythematosus. *Arthritis and Rheumatology*, **2017**, 69, 800-807 | 9.5 | 87 |
| 86 | A type I interferon signature identifies bilateral striatal necrosis due to mutations in ADAR1. *Journal of Medical Genetics*, **2014**, 51, 76-82 | 5.8 | 85 |
| 85 | Autosomal dominant inheritance of a heterozygous mutation in SAMHD1 causing familial chilblain lupus. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 235-7 | 2.5 | 81 |
| 84 | Intracerebral large artery disease in Aicardi-Goutières syndrome implicates SAMHD1 in vascular homeostasis. *Developmental Medicine and Child Neurology*, **2010**, 52, 725-32 | 3.3 | 80 |
| 83 | Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. *Nature Genetics*, **2016**, 48, 1185-92 | 36.3 | 74 |
| 82 | Stimulator of Interferon Genes-Associated Vasculopathy With Onset in Infancy: A Mimic of Childhood Granulomatosis With Polyangiitis. *JAMA Dermatology*, **2015**, 151, 872-7 | 5.1 | 72 |
| 81 | Cerebroretinal microangiopathy with calcifications and cysts (CRMCC). *American Journal of Medical Genetics, Part A*, **2008**, 146A, 182-90 | 2.5 | 70 |

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| 80 | Identification and characterization of an inborn error of metabolism caused by dihydrofolate reductase deficiency. *American Journal of Human Genetics*, **2011**, 88, 216-25 | 11 | 68 |
| 79 | Mutations in CECR1 associated with a neutrophil signature in peripheral blood. *Pediatric Rheumatology*, **2014**, 12, 44 | 3.5 | 63 |
| 78 | Familial Aicardi-Goutières syndrome due to SAMHD1 mutations is associated with chronic arthropathy and contractures. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 938-42 | 2.5 | 61 |
| 77 | Reverse-Transcriptase Inhibitors in the Aicardi-Goutières Syndrome. *New England Journal of Medicine*, **2018**, 379, 2275-7 | 59.2 | 61 |
| 76 | Therapies in Aicardi-Goutières syndrome. *Clinical and Experimental Immunology*, **2014**, 175, 1-8 | 6.2 | 60 |
| 75 | Unusual cutaneous features associated with a heterozygous gain-of-function mutation in IFIH1: overlap between Aicardi-Goutières and Singleton-Merten syndromes. *British Journal of Dermatology*, **2015**, 173, 1505-13 | 4 | 55 |
| 74 | Mutations in ADAR1, IFIH1, and RNASEH2B presenting as spastic paraplegia. *Neuropediatrics*, **2014**, 45, 386-93 | 1.6 | 53 |
| 73 | PRKDC mutations associated with immunodeficiency, granuloma, and autoimmune regulator-dependent autoimmunity. *Journal of Allergy and Clinical Immunology*, **2015**, 135, 1578-88.e5 | 11.5 | 52 |
| 72 | cGAS-mediated induction of type I interferon due to inborn errors of histone pre-mRNA processing. *Nature Genetics*, **2020**, 52, 1364-1372 | 36.3 | 52 |
| 71 | Bloom syndrome protein restrains innate immune sensing of micronuclei by cGAS. *Journal of Experimental Medicine*, **2019**, 216, 1199-1213 | 16.6 | 51 |
| 70 | Spondyloenchondrodysplasia Due to Mutations in ACP5: A Comprehensive Survey. *Journal of Clinical Immunology*, **2016**, 36, 220-34 | 5.7 | 48 |
| 69 | Mutations in COPA lead to abnormal trafficking of STING to the Golgi and interferon signaling. *Journal of Experimental Medicine*, **2020**, 217, | 16.6 | 47 |
| 68 | The paraoxonase Gln-Arg 192 polymorphism in subjects with ischaemic heart disease. *Coronary Artery Disease*, **1997**, 8, 677-82 | 1.4 | 44 |
| 67 | A functional XPNPEP2 promoter haplotype leads to reduced plasma aminopeptidase P and increased risk of ACE inhibitor-induced angioedema. *Human Mutation*, **2011**, 32, 1326-31 | 4.7 | 42 |
| 66 | Band-like intracranial calcification with simplified gyration and polymicrogyria: a distinct "pseudo-TORCH" phenotype. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 3173-80 | 2.5 | 42 |
| 65 | An open-label trial of JAK 1/2 blockade in progressive -associated neuroinflammation. *Neurology*, **2018**, 90, 289-291 | 6.5 | 41 |
| 64 | Cutaneous histopathological findings of Aicardi-Goutières syndrome, overlap with chilblain lupus. *Journal of Cutaneous Pathology*, **2008**, 35, 774-8 | 1.7 | 39 |
| 63 | ADA2 deficiency: case report of a new phenotype and novel mutation in two sisters. *RMD Open*, **2016**, 2, e000236 | 5.9 | 38 |

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| 62 | Severe type I interferonopathy and unrestrained interferon signaling due to a homozygous germline mutation in. *Science Immunology*, **2019**, 4, | 28 | 38 |
| 61 | A child with severe juvenile dermatomyositis treated with ruxolitinib. *Brain*, **2018**, 141, e80 | 11.2 | 37 |
| 60 | Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. *Neuropediatrics*, **2017**, 48, 166-184 | 1.6 | 35 |
| 59 | A further example of a distinctive autosomal recessive syndrome comprising neonatal diabetes mellitus, intestinal atresias and gall bladder agenesis. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1713-7 | 2.5 | 34 |
| 58 | Leukoencephalopathy with calcifications and cysts: a purely neurological disorder distinct from coats plus. *Neuropediatrics*, **2014**, 45, 175-82 | 1.6 | 33 |
| 57 | Angiotensin-converting enzyme (ACE) gene polymorphisms in patients characterised by coronary angiography. *Human Genetics*, **1997**, 100, 420-5 | 6.3 | 33 |
| 56 | Efficacy of JAK1/2 inhibition in the treatment of chilblain lupus due to TREX1 deficiency. *Annals of the Rheumatic Diseases*, **2019**, 78, 431-433 | 2.4 | 33 |
| 55 | Homozygous N-terminal missense mutation in TRNT1 leads to progressive B-cell immunodeficiency in adulthood. *Journal of Allergy and Clinical Immunology*, **2017**, 139, 360-363.e6 | 11.5 | 32 |
| 54 | Tartrate-Resistant Acid Phosphatase Deficiency in the Predisposition to Systemic Lupus Erythematosus. *Arthritis and Rheumatology*, **2017**, 69, 131-142 | 9.5 | 32 |
| 53 | COL4A1 mutations associated with a characteristic pattern of intracranial calcification. *Neuropediatrics*, **2011**, 42, 227-33 | 1.6 | 32 |
| 52 | Genetic and phenotypic spectrum associated with IFIH1 gain-of-function. *Human Mutation*, **2020**, 41, 837-849 | 4.7 | 31 |
| 51 | Musculoskeletal Disease in MDA5-Related Type I Interferonopathy: A Mendelian Mimic of Jaccoud's Arthropathy. *Arthritis and Rheumatology*, **2017**, 69, 2081-2091 | 9.5 | 30 |
| 50 | A de novo p.Asp18Asn mutation in TREX1 in a patient with Aicardi-Goutières syndrome. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2612-7 | 2.5 | 30 |
| 49 | Aicardi-Goutières syndrome: description of a late onset case. *Developmental Medicine and Child Neurology*, **2008**, 50, 631-4 | 3.3 | 30 |
| 48 | Aicardi-Goutières syndrome harbours abundant systemic and brain-reactive autoantibodies. *Annals of the Rheumatic Diseases*, **2015**, 74, 1931-9 | 2.4 | 27 |
| 47 | Characterization of samhd1 morphant zebrafish recapitulates features of the human type I interferonopathy Aicardi-Goutières syndrome. *Journal of Immunology*, **2015**, 194, 2819-25 | 5.3 | 26 |
| 46 | Chilblains as a diagnostic sign of aicardi-goutières syndrome. *Neuropediatrics*, **2010**, 41, 18-23 | 1.6 | 26 |
| 45 | Identification of Novel Polymorphisms within the Protein Z Gene, Haplotype Distribution and Linkage Analysis. *Thrombosis and Haemostasis*, **2001**, 85, 1123-1124 | 7 | 26 |

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| 44 | Systemic lupus erythematosus due to C1q deficiency with progressive encephalopathy, intracranial calcification and acquired moyamoya cerebral vasculopathy. *Lupus*, **2013**, 22, 639-43 | 2.6 | 24 |
| 43 | Two further cases of spondyloenchondrodysplasia (SPENCD) with immune dysregulation. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2810-5 | 2.5 | 24 |
| 42 | Clinical, radiological and possible pathological overlap of cystic leukoencephalopathy without megalencephaly and Aicardi-Goutières syndrome. *European Journal of Paediatric Neurology*, **2016**, 20, 604-10 | 3.8 | 24 |
| 41 | Aicardi-Goutières syndrome presenting atypically as a sub-acute leukoencephalopathy. *European Journal of Paediatric Neurology*, **2008**, 12, 408-11 | 3.8 | 23 |
| 40 | Angiotensin converting enzyme and angiotensin II type 1-receptor gene polymorphisms and risk of ischaemic heart disease. *Cardiovascular Research*, **1999**, 41, 746-53 | 9.9 | 22 |
| 39 | FVIII Coagulant Activity and Antigen in Subjects with Ischaemic Heart Disease. *Thrombosis and Haemostasis*, **1998**, 80, 757-762 | 7 | 21 |
| 38 | Brief Report: Vitamin D Deficiency Is Associated With Endothelial Dysfunction and Increases Type I Interferon Gene Expression in a Murine Model of Systemic Lupus Erythematosus. *Arthritis and Rheumatology*, **2016**, 68, 2929-2935 | 9.5 | 20 |
| 37 | PSMB10, the last immunoproteasome gene missing for PRAAS. *Journal of Allergy and Clinical Immunology*, **2020**, 145, 1015-1017.e6 | 11.5 | 20 |
| 36 | MDA5-Associated Neuroinflammation and the Singleton-Merten Syndrome: Two Faces of the Same Type I Interferonopathy Spectrum. *Journal of Interferon and Cytokine Research*, **2017**, 37, 214-219 | 3.5 | 19 |
| 35 | Overview of STING-Associated Vasculopathy with Onset in Infancy (SAVI) Among 21 Patients. *Journal of Allergy and Clinical Immunology: in Practice*, **2021**, 9, 803-818.e11 | 5.4 | 19 |
| 34 | Use of ruxolitinib in COPA syndrome manifesting as life-threatening alveolar haemorrhage. *Thorax*, **2020**, 75, 92-95 | 7.3 | 17 |
| 33 | JAK inhibition in STING-associated interferonopathy. *Annals of the Rheumatic Diseases*, **2016**, 75, e75 | 2.4 | 17 |
| 32 | JAK 1/2 Blockade in MDA5 Gain-of-Function. *Journal of Clinical Immunology*, **2018**, 38, 844-846 | 5.7 | 17 |
| 31 | Contribution of rare and predicted pathogenic gene variants to childhood-onset lupus: a large, genetic panel analysis of British and French cohorts. *Lancet Rheumatology, The*, **2020**, 2, e99-e109 | 14.2 | 16 |
| 30 | Elevation of proinflammatory cytokines in patients with Aicardi-Goutières syndrome. *Neurology*, **2013**, 80, 997-1002 | 6.5 | 15 |
| 29 | Synonymous mutations in RNASEH2A create cryptic splice sites impairing RNase H2 enzyme function in Aicardi-Goutières syndrome. *Human Mutation*, **2013**, 34, 1066-70 | 4.7 | 15 |
| 28 | Basal ganglia calcification in a patient with beta-propeller protein-associated neurodegeneration. *Pediatric Neurology*, **2014**, 51, 843-5 | 2.9 | 14 |
| 27 | Anti-MDA5 juvenile idiopathic inflammatory myopathy: a specific subgroup defined by differentially enhanced interferon-signalling. *Rheumatology*, **2020**, 59, 1927-1937 | 3.9 | 12 |

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| 26 | ADAR1 Facilitates HIV-1 Replication in Primary CD4+ T Cells. *PLoS ONE*, **2015**, 10, e0143613 | 3.7 | 11 |
| 25 | Type I interferon in patients with systemic autoimmune rheumatic disease is associated with haematological abnormalities and specific autoantibody profiles. *Arthritis Research and Therapy*, **2019**, 21, 147 | 5.7 | 8 |
| 24 | Comprehensive molecular screening strategy of OCLN in band-like calcification with simplified gyration and polymicrogyria. *Clinical Genetics*, **2018**, 93, 228-234 | 4 | 8 |
| 23 | Human Endothelial Cell-derived Nuclear Proteins that Recognise Polymorphic DNA Elements in the von Willebrand Factor Gene Promoter Include YY1. *Thrombosis and Haemostasis*, **2001**, 86, 672-679 | 7 | 8 |
| 22 | JAK Inhibition in the Aicardi-Goutières Syndrome. *New England Journal of Medicine*, **2020**, 383, 2190-2191 | 59.2 | 8 |
| 21 | Enhanced cGAS-STING-dependent interferon signaling associated with mutations in ATAD3A. *Journal of Experimental Medicine*, **2021**, 218, | 16.6 | 8 |
| 20 | Comment on: 'Aberrant tRNA processing causes an autoinflammatory syndrome responsive to TNF inhibitors' by Giannelou : mutations in result in a constitutive activation of type I interferon signalling. *Annals of the Rheumatic Diseases*, **2019**, 78, e86 | 2.4 | 7 |
| 19 | Analysis of U8 snoRNA Variants in Zebrafish Reveals How Bi-allelic Variants Cause Leukoencephalopathy with Calcifications and Cysts. *American Journal of Human Genetics*, **2020**, 106, 694-706 | 11 | 7 |
| 18 | Differential Expression of Interferon-Alpha Protein Provides Clues to Tissue Specificity Across Type I Interferonopathies. *Journal of Clinical Immunology*, **2021**, 41, 603-609 | 5.7 | 7 |
| 17 | Biallelic mutations in NRROS cause an early onset lethal microgliopathy. *Acta Neuropathologica*, **2020**, 139, 947-951 | 14.3 | 6 |
| 16 | Sine causa tetraparesis: A pilot study on its possible relationship with interferon signature analysis and Aicardi Goutières syndrome related genes analysis. *Medicine (United States)*, **2018**, 97, e13893 | 1.8 | 6 |
| 15 | Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum. *American Journal of Medical Genetics, Part A*, **2021**, 185, 15-25 | 2.5 | 5 |
| 14 | From Diagnosis to Prognosis: Revisiting the Meaning of Muscle ISG15 Overexpression in Juvenile Inflammatory Myopathies. *Arthritis and Rheumatology*, **2021**, 73, 1044-1052 | 9.5 | 5 |
| 13 | Differential levels of IFN subtypes in autoimmunity and viral infection. *Cytokine*, **2021**, 144, 155533 | 4 | 5 |
| 12 | Complexity in unclassified auto-inflammatory disease: a case report illustrating the potential for disease arising from the allelic burden of multiple variants. *Pediatric Rheumatology*, **2019**, 17, 70 | 3.5 | 4 |
| 11 | Combination of exome sequencing and immune testing confirms Aicardi-Goutières syndrome type 5 in a challenging pediatric neurology case. *Journal of Physical Education and Sports Management*, **2018**, 4, | 2.8 | 4 |
| 10 | Autosomal-dominant early-onset spastic paraparesis with brain calcification due to IFIH1 gain-of-function. *Human Mutation*, **2018**, 39, 1076-1080 | 4.7 | 4 |
| 9 | Genetic polymorphism in C3 is associated with progression in chronic kidney disease (CKD) patients with IgA nephropathy but not in other causes of CKD. *PLoS ONE*, **2020**, 15, e0228101 | 3.7 | 3 |

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| 8 | RNASEH2B Related Adult-Onset Interferonopathy. *Journal of Clinical Immunology*, **2019**, 39, 620-622 | 5.7 | 3 |
| 7 | Biallelic Mutations in MTPAP Associated with a Lethal Encephalopathy. *Neuropediatrics*, **2020**, 51, 178-184 | 1.6 | 3 |
| 6 | LACC1 deficiency links juvenile arthritis with autophagy and metabolism in macrophages. *Journal of Experimental Medicine*, **2021**, 218, | 16.6 | 3 |
| 5 | Aicardi-Goutières syndrome presenting with haematemesis in infancy. *Acta Paediatrica, International Journal of Paediatrics*, **2009**, 98, 2005-8 | 3.1 | 2 |
| 4 | MRSD: A quantitative approach for assessing suitability of RNA-seq in the investigation of mis-splicing in Mendelian disease.. *American Journal of Human Genetics*, **2022**, | 11 | 2 |
| 3 | Clinical Reasoning: A 25-year-old woman with recurrent episodes of collapse and loss of consciousness. *Neurology*, **2020**, 94, 994-999 | 6.5 | 1 |
| 2 | Mutations in RNU7-1 Weaken Secondary RNA Structure, Induce MCP-1 and CXCL10 in CSF, and Result in Aicardi-Goutières Syndrome with Severe End-Organ Involvement.. *Journal of Clinical Immunology*, **2022**, 1 | 5.7 | 1 |
| 1 | Expanding the clinical spectrum of Fowler syndrome: Three siblings with survival into adulthood and systematic review of the literature. *Clinical Genetics*, **2020**, 98, 423-432 | 4 | |