Gillian I Rice

List of Publications by Year in Descending Order

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	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
114	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
113	Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum. *American Journal of Medical Genetics, Part A*, **2021**, 185, 15-25	2.5	5
112	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
111	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
110	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
109	LACC1 deficiency links juvenile arthritis with autophagy and metabolism in macrophages. *Journal of Experimental Medicine*, **2021**, 218,	16.6	3
108	Differential levels of IFN-β subtypes in autoimmunity and viral infection. *Cytokine*, **2021**, 144, 155533	4	5
107	Enhanced cGAS-STING-dependent interferon signaling associated with mutations in ATAD3A. *Journal of Experimental Medicine*, **2021**, 218,	16.6	8
106	Clinical Reasoning: A 25-year-old woman with recurrent episodes of collapse and loss of consciousness. *Neurology*, **2020**, 94, 994-999	6.5	1
105	Biallelic mutations in NRROS cause an early onset lethal microgliopathy. *Acta Neuropathologica*, **2020**, 139, 947-951	14.3	6
104	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
103	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
102	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	
101	Use of ruxolitinib in COPA syndrome manifesting as life-threatening alveolar haemorrhage. *Thorax*, **2020**, 75, 92-95	7.3	17
100	Biallelic Mutations in MTPAP Associated with a Lethal Encephalopathy. *Neuropediatrics*, **2020**, 51, 178-184	1.6	3
99	Anti-MDA5 juvenile idiopathic inflammatory myopathy: a specific subgroup defined by differentially enhanced interferon-β signalling. *Rheumatology*, **2020**, 59, 1927-1937	3.9	12

98	PSMB10, the last immunoproteasome gene missing for PRAAS. *Journal of Allergy and Clinical Immunology*, **2020**, 145, 1015-1017.e6	11.5	20
97	Genetic and phenotypic spectrum associated with IFIH1 gain-of-function. *Human Mutation*, **2020**, 41, 837-849	4.7	31
96	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
95	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
94	JAK Inhibition in the Aicardi-Goutières Syndrome. *New England Journal of Medicine*, **2020**, 383, 2190-2191	59.2	8
93	Mutations in COPA lead to abnormal trafficking of STING to the Golgi and interferon signaling. *Journal of Experimental Medicine*, **2020**, 217,	16.6	47
92	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
91	Bloom syndrome protein restrains innate immune sensing of micronuclei by cGAS. *Journal of Experimental Medicine*, **2019**, 216, 1199-1213	16.6	51
90	Comment on: SAberrant tRNA processing causes an autoinflammatory syndrome responsive to TNF inhibitorsSby Giannelou : mutations in result in a constitutive activation of type I interferon signalling. *Annals of the Rheumatic Diseases*, **2019**, 78, e86	2.4	7
89	RNASEH2B Related Adult-Onset Interferonopathy. *Journal of Clinical Immunology*, **2019**, 39, 620-622	5.7	3
88	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
87	Severe type I interferonopathy and unrestrained interferon signaling due to a homozygous germline mutation in. *Science Immunology*, **2019**, 4,	28	38
86	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
85	An open-label trial of JAK 1/2 blockade in progressive -associated neuroinflammation. *Neurology*, **2018**, 90, 289-291	6.5	41
84	Comprehensive molecular screening strategy of OCLN in band-like calcification with simplified gyration and polymicrogyria. *Clinical Genetics*, **2018**, 93, 228-234	4	8
83	Mitochondrial double-stranded RNA triggers antiviral signalling in humans. *Nature*, **2018**, 560, 238-242	50.4	211
82	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
81	JAK 1/2 Blockade in MDA5 Gain-of-Function. *Journal of Clinical Immunology*, **2018**, 38, 844-846	5.7	17

80	Reverse-Transcriptase Inhibitors in the Aicardi-Goutières Syndrome. *New England Journal of Medicine*, **2018**, 379, 2275-7	59.2	61
79	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
78	A child with severe juvenile dermatomyositis treated with ruxolitinib. *Brain*, **2018**, 141, e80	11.2	37
77	Autosomal-dominant early-onset spastic paraparesis with brain calcification due to IFIH1 gain-of-function. *Human Mutation*, **2018**, 39, 1076-1080	4.7	4
76	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
75	Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. *Neuropediatrics*, **2017**, 48, 166-184	1.6	35
74	Detection of interferon alpha protein reveals differential levels and cellular sources in disease. *Journal of Experimental Medicine*, **2017**, 214, 1547-1555	16.6	192
73	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
72	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
71	Assessment of Type I Interferon Signaling in Pediatric Inflammatory Disease. *Journal of Clinical Immunology*, **2017**, 37, 123-132	5.7	94
70	Expression of Cyclic GMP-AMP Synthase in Patients With Systemic Lupus Erythematosus. *Arthritis and Rheumatology*, **2017**, 69, 800-807	9.5	87
69	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
68	Tartrate-Resistant Acid Phosphatase Deficiency in the Predisposition to Systemic Lupus Erythematosus. *Arthritis and Rheumatology*, **2017**, 69, 131-142	9.5	32
67	Type I interferon-mediated autoinflammation due to DNase II deficiency. *Nature Communications*, **2017**, 8, 2176	17.4	111
66	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. *Nature Genetics*, **2016**, 48, 1185-92	36.3	74
65	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
64	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
63	Spondyloenchondrodysplasia Due to Mutations in ACP5: A Comprehensive Survey. *Journal of Clinical Immunology*, **2016**, 36, 220-34	5.7	48

62	ADA2 deficiency: case report of a new phenotype and novel mutation in two sisters. *RMD Open*, **2016**, 2, e000236	5.9	38
61	JAK inhibition in STING-associated interferonopathy. *Annals of the Rheumatic Diseases*, **2016**, 75, e75	2.4	17
60	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
59	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
58	Human disease phenotypes associated with mutations in TREX1. *Journal of Clinical Immunology*, **2015**, 35, 235-43	5.7	108
57	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
56	Human intracellular ISG15 prevents interferon-α over-amplification and auto-inflammation. *Nature*, **2015**, 517, 89-93	50.4	311
55	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
54	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
53	A specific IFIH1 gain-of-function mutation causes Singleton-Merten syndrome. *American Journal of Human Genetics*, **2015**, 96, 275-82	11	134
52	Aicardi-Goutières syndrome harbours abundant systemic and brain-reactive autoantibodies. *Annals of the Rheumatic Diseases*, **2015**, 74, 1931-9	2.4	27
51	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
50	ADAR1 Facilitates HIV-1 Replication in Primary CD4+ T Cells. *PLoS ONE*, **2015**, 10, e0143613	3.7	11
49	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
48	The SKIV2L RNA exosome limits activation of the RIG-I-like receptors. *Nature Immunology*, **2014**, 15, 839-45	19.1	131
47	Inherited STING-activating mutation underlies a familial inflammatory syndrome with lupus-like manifestations. *Journal of Clinical Investigation*, **2014**, 124, 5516-20	15.9	294
46	Leukoencephalopathy with calcifications and cysts: a purely neurological disorder distinct from coats plus. *Neuropediatrics*, **2014**, 45, 175-82	1.6	33
45	Mutations in ADAR1, IFIH1, and RNASEH2B presenting as spastic paraplegia. *Neuropediatrics*, **2014**, 45, 386-93	1.6	53

44	Therapies in Aicardi-Goutières syndrome. *Clinical and Experimental Immunology*, **2014**, 175, 1-8	6.2	60
43	Basal ganglia calcification in a patient with beta-propeller protein-associated neurodegeneration. *Pediatric Neurology*, **2014**, 51, 843-5	2.9	14
42	Mutations in CECR1 associated with a neutrophil signature in peripheral blood. *Pediatric Rheumatology*, **2014**, 12, 44	3.5	63
41	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
40	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
39	Elevation of proinflammatory cytokines in patients with Aicardi-Goutières syndrome. *Neurology*, **2013**, 80, 997-1002	6.5	15
38	Protein kinase cδ deficiency causes mendelian systemic lupus erythematosus with B cell-defective apoptosis and hyperproliferation. *Arthritis and Rheumatism*, **2013**, 65, 2161-71		119
37	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
36	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
35	SAMHD1 restricts HIV-1 reverse transcription in quiescent CD4(+) T-cells. *Retrovirology*, **2012**, 9, 87	3.6	254
34	Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. *Nature Genetics*, **2012**, 44, 338-42	36.3	186
33	Mutations in ADAR1 cause Aicardi-Goutières syndrome associated with a type I interferon signature. *Nature Genetics*, **2012**, 44, 1243-8	36.3	521
32	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
31	HIV-1 restriction factor SAMHD1 is a deoxynucleoside triphosphate triphosphohydrolase. *Nature*, **2011**, 480, 379-82	50.4	578
30	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
29	Neprilysin, obesity and the metabolic syndrome. *International Journal of Obesity*, **2011**, 35, 1031-40	5.5	102
28	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
27	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

26	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
25	COL4A1 mutations associated with a characteristic pattern of intracranial calcification. *Neuropediatrics*, **2011**, 42, 227-33	1.6	32
24	Chilblains as a diagnostic sign of aicardi-goutières syndrome. *Neuropediatrics*, **2010**, 41, 18-23	1.6	26
23	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
22	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
21	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
20	Aicardi-Goutières syndrome presenting with haematemesis in infancy. *Acta Paediatrica, International Journal of Paediatrics*, **2009**, 98, 2005-8	3.1	2
19	Mutations involved in Aicardi-Goutières syndrome implicate SAMHD1 as regulator of the innate immune response. *Nature Genetics*, **2009**, 41, 829-32	36.3	507
18	Aicardi-Goutières syndrome: description of a late onset case. *Developmental Medicine and Child Neurology*, **2008**, 50, 631-4	3.3	30
17	Aicardi-Goutières syndrome presenting atypically as a sub-acute leukoencephalopathy. *European Journal of Paediatric Neurology*, **2008**, 12, 408-11	3.8	23
16	Cerebroretinal microangiopathy with calcifications and cysts (CRMCC). *American Journal of Medical Genetics, Part A*, **2008**, 146A, 182-90	2.5	70
15	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
14	Two further cases of spondyloenchondrodysplasia (SPENCD) with immune dysregulation. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2810-5	2.5	24
13	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
12	Cutaneous histopathological findings of Aicardi-Goutières syndrome, overlap with chilblain lupus. *Journal of Cutaneous Pathology*, **2008**, 35, 774-8	1.7	39
11	Heterozygous mutations in TREX1 cause familial chilblain lupus and dominant Aicardi-Goutieres syndrome. *American Journal of Human Genetics*, **2007**, 80, 811-5	11	286
10	Clinical and molecular phenotype of Aicardi-Goutieres syndrome. *American Journal of Human Genetics*, **2007**, 81, 713-25	11	310
9	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

8	Evaluation of angiotensin-converting enzyme (ACE), its homologue ACE2 and neprilysin in angiotensin peptide metabolism. *Biochemical Journal*, **2004**, 383, 45-51	3.8	462
7	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
6	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
5	Identification of Novel Polymorphisms within the Protein Z Gene, Haplotype Distribution and Linkage Analysis. *Thrombosis and Haemostasis*, **2001**, 85, 1123-1124	7	26
4	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
3	FVIII Coagulant Activity and Antigen in Subjects with Ischaemic Heart Disease. *Thrombosis and Haemostasis*, **1998**, 80, 757-762	7	21
2	Angiotensin-converting enzyme (ACE) gene polymorphisms in patients characterised by coronary angiography. *Human Genetics*, **1997**, 100, 420-5	6.3	33
1	The paraoxonase Gln-Arg 192 polymorphism in subjects with ischaemic heart disease. *Coronary Artery Disease*, **1997**, 8, 677-82	1.4	44