Gillian I Rice

List of Publications by Year
in descending order

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123 papers	12,766 citations	34105 52 h-index	25787 108 g-index
132 all docs	132 docs citations	132 times ranked	13592 citing authors

#	Article	IF	Citations
1	Mutations in ADAR1 cause Aicardi-GoutiÃ¨res syndrome associated with a type I interferon signature. Nature Genetics, 2012, 44, 1243-1248.	21.4	712
2	HIV-1 restriction factor SAMHD1 is a deoxynucleoside triphosphate triphosphohydrolase. Nature, 2011, 480, 379-382.	27.8	707
3	Mutations involved in Aicardi-GoutiÃ¨res syndrome implicate SAMHD1 as regulator of the innate immune response. Nature Genetics, 2009, 41, 829-832.	21.4	610
4	Evaluation of angiotensin-converting enzyme (ACE), its homologue ACE2 and neprilysin in angiotensin peptide metabolism. Biochemical Journal, 2004, 383, 45-51.	3.7	539
5	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.	21.4	490
6	Assessment of interferon-related biomarkers in Aicardi-GoutiÃ¨res syndrome associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, and ADAR: a case-control study. Lancet Neurology, The, 2013, 12, 1159-1169.	10.2	473
7	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, 167, 296-312.	1.2	447
8	Inherited STING-activating mutation underlies a familial inflammatory syndrome with lupus-like manifestations. Journal of Clinical Investigation, 2014, 124, 5516-5520.	8.2	435
9	Human intracellular ISG15 prevents interferon-Î±/Î² over-amplification and auto-inflammation. Nature, 2015, 517, 89-93.	27.8	432
10	Mitochondrial double-stranded RNA triggers antiviral signalling in humans. Nature, 2018, 560, 238-242.	27.8	397
11	Clinical and Molecular Phenotype of Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 81, 713-725.	6.2	375
12	Heterozygous Mutations in TREX1 Cause Familial Chilblain Lupus and Dominant Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 80, 811-815.	6.2	339
13	SAMHD1 restricts HIV-1 reverse transcription in quiescent CD4+T-cells. Retrovirology, 2012, 9, 87.	2.0	302
14	Detection of interferon alpha protein reveals differential levels and cellular sources in disease. Journal of Experimental Medicine, 2017, 214, 1547-1555.	8.5	288
15	Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. Nature Genetics, 2012, 44, 338-342.	21.4	234
16	Tartrate-resistant acid phosphatase deficiency causes a bone dysplasia with autoimmunity and a type I interferon expression signature. Nature Genetics, 2011, 43, 127-131.	21.4	214
17	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.	2.9	192
18	A Specific IFIH1 Gain-of-Function Mutation Causes Singleton-Merten Syndrome. American Journal of Human Genetics, 2015, 96, 275-282.	6.2	188

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19	The SKIV2L RNA exosome limits activation of the RIG-I-like receptors. Nature Immunology, 2014, 15, 839-845.	14.5	170
20	Circulating Activities of Angiotensin-Converting Enzyme, Its Homolog, Angiotensin-Converting Enzyme 2, and Neprilysin in a Family Study. Hypertension, 2006, 48, 914-920.	2.7	167
21	Type I interferon-mediated autoinflammation due to DNase II deficiency. Nature Communications, 2017, 8, 2176.	12.8	164
22	Assessment of Type I Interferon Signaling in Pediatric Inflammatory Disease. Journal of Clinical Immunology, 2017, 37, 123-132.	3.8	163
23	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.	2.9	159
24	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, 346-353.	1.4	156
25	Protein Kinase CÎ´ Deficiency Causes Mendelian Systemic Lupus Erythematosus With B Cellâ€Defective Apoptosis and Hyperproliferation. Arthritis and Rheumatism, 2013, 65, 2161-2171.	6.7	155
26	Human Disease Phenotypes Associated With Mutations in TREX1. Journal of Clinical Immunology, 2015, 35, 235-243.	3.8	154
27	Neprilysin, obesity and the metabolic syndrome. International Journal of Obesity, 2011, 35, 1031-1040.	3.4	137
28	Mutations in COPA lead to abnormal trafficking of STING to the Golgi and interferon signaling. Journal of Experimental Medicine, 2020, 217, .	8.5	130
29	Expression of Cyclic GMPâ€AMP Synthase in Patients With Systemic Lupus Erythematosus. Arthritis and Rheumatology, 2017, 69, 800-807.	5.6	129
30	SAMHD1 is a nucleic-acid binding protein that is mislocalized due to aicardi-goutiÃ¨res syndrome-associated mutations. Human Mutation, 2012, 33, 1116-1122.	2.5	121
31	A type I interferon signature identifies bilateral striatal necrosis due to mutations inADAR1. Journal of Medical Genetics, 2014, 51, 76-82.	3.2	118
32	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature Genetics, 2016, 48, 1185-1192.	21.4	114
33	Stimulator of Interferon Genesâ€“Associated Vasculopathy With Onset in Infancy. JAMA Dermatology, 2015, 151, 872.	4.1	108
34	Reverse-Transcriptase Inhibitors in the Aicardiâ€“GoutiÃ¨res Syndrome. New England Journal of Medicine, 2018, 379, 2275-2277.	27.0	106
35	cGAS-mediated induction of type I interferon due to inborn errors of histone pre-mRNA processing. Nature Genetics, 2020, 52, 1364-1372.	21.4	105
36	A functional XPNPEP2 promoter haplotype leads to reduced plasma aminopeptidase P and increased risk of ACE inhibitor-induced angioedema. Human Mutation, 2011, 32, 1326-1331.	2.5	104

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37	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.	3.8	98
38	Autosomal dominant inheritance of a heterozygous mutation in SAMHD1 causing familial chilblain lupus. American Journal of Medical Genetics, Part A, 2011, 155, 235-237.	1.2	97
39	Identification and Characterization of an Inborn Error of Metabolism Caused by Dihydrofolate Reductase Deficiency. American Journal of Human Genetics, 2011, 88, 216-225.	6.2	90
40	Intracerebral large artery disease in Aicardiâ€“GoutiÃ¨res syndrome implicates SAMHD1 in vascular homeostasis. Developmental Medicine and Child Neurology, 2010, 52, 725-732.	2.1	89
41	Mutations in CECR1 associated with a neutrophil signature in peripheral blood. Pediatric Rheumatology, 2014, 12, 44.	2.1	88
42	Cerebroretinal microangiopathy with calcifications and cysts (CRMCC). American Journal of Medical Genetics, Part A, 2008, 146A, 182-190.	1.2	87
43	PRKDC mutations associated with immunodeficiency, granuloma, and autoimmune regulatorâ€“dependent autoimmunity. Journal of Allergy and Clinical Immunology, 2015, 135, 1578-1588.e5.	2.9	84
44	Severe type I interferonopathy and unrestrained interferon signaling due to a homozygous germline mutation in STAT2. Science Immunology, 2019, 4, .	11.9	80
45	Unusual cutaneous features associated with a heterozygous gain-of-function mutation inIFIH1: overlap between Aicardi-GoutiÃ¨res and Singleton-Merten syndromes. British Journal of Dermatology, 2015, 173, 1505-1513.	1.5	76
46	Bloom syndrome protein restrains innate immune sensing of micronuclei by cGAS. Journal of Experimental Medicine, 2019, 216, 1199-1213.	8.5	75
47	Therapies in Aicardiâ€“GoutiÃ¨res syndrome. Clinical and Experimental Immunology, 2013, 175, 1-8.	2.6	74
48	Familial Aicardiâ€“GoutiÃ¨res syndrome due to SAMHD1 mutations is associated with chronic arthropathy and contractures. American Journal of Medical Genetics, Part A, 2010, 152A, 938-942.	1.2	73
49	Mutations in ADAR1, IFIH1, and RNASEH2B Presenting As Spastic Paraplegia. Neuropediatrics, 2014, 45, 386-391.	0.6	72
50	Spondyloenchondrodysplasia Due to Mutations in ACP5: A Comprehensive Survey. Journal of Clinical Immunology, 2016, 36, 220-234.	3.8	71
51	Genetic and phenotypic spectrum associated with IFIH1 gainâ€ofâ€function. Human Mutation, 2020, 41, 837-849.	2.5	63
52	Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. Neuropediatrics, 2017, 48, 166-184.	0.6	62
53	An open-label trial of JAK 1/2 blockade in progressive IFIH1 -associated neuroinflammation. Neurology, 2018, 90, 289-291.	1.1	60
54	A child with severe juvenile dermatomyositis treated with ruxolitinib. Brain, 2018, 141, e80-e80.	7.6	58

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55	Efficacy of JAK1/2 inhibition in the treatment of chilblain lupus due to TREX1 deficiency. Annals of the Rheumatic Diseases, 2019, 78, 431-433.	0.9	53
56	The paraoxonase Gln-Arg 192 polymorphism in subjects with ischaemic heart disease. Coronary Artery Disease, 1997, 8, 677-682.	0.7	52
57	Cutaneous histopathological findings of Aicardiâ€“GoutiÃ¨res syndrome, overlap with chilblain lupus. Journal of Cutaneous Pathology, 2008, 35, 774-778.	1.3	47
58	Tartrateâ€Resistant Acid Phosphatase Deficiency in the Predisposition to Systemic Lupus Erythematosus. Arthritis and Rheumatology, 2017, 69, 131-142.	5.6	47
59	ADA2 deficiency: case report of a new phenotype and novel mutation in two sisters. RMD Open, 2016, 2, e000236.	3.8	47
60	Bandâ€like intracranial calcification with simplified gyration and polymicrogyria: A distinct â€œpseudoâ€TORCHâ€•phenotype. American Journal of Medical Genetics, Part A, 2008, 146A, 3173-3180.	1.2	46
61	Musculoskeletal Disease in MDA5â€Related Type I Interferonopathy: A Mendelian Mimic of Jaccoud's Arthropathy. Arthritis and Rheumatology, 2017, 69, 2081-2091.	5.6	44
62	Enhanced cGAS-STINGâ€“dependent interferon signaling associated with mutations in ATAD3A. Journal of Experimental Medicine, 2021, 218, .	8.5	43
63	PSMB10, the last immunoproteasome gene missing for PRAAS. Journal of Allergy and Clinical Immunology, 2020, 145, 1015-1017.e6.	2.9	42
64	Leukoencephalopathy with Calcifications and Cysts: A Purely Neurological Disorder Distinct from Coats Plus. Neuropediatrics, 2014, 45, 175-182.	0.6	41
65	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.	2.9	41
66	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, 1713-1717.	1.2	38
67	COL4A1 Mutations Associated with a Characteristic Pattern of Intracranial Calcification. Neuropediatrics, 2011, 42, 227-233.	0.6	38
68	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.	3.9	38
69	Characterization of samhd1 Morphant Zebrafish Recapitulates Features of the Human Type I Interferonopathy Aicardi-GoutiÃ¨res Syndrome. Journal of Immunology, 2015, 194, 2819-2825.	0.8	36
70	Use of ruxolitinib in COPA syndrome manifesting as life-threatening alveolar haemorrhage. Thorax, 2020, 75, 92-95.	5.6	36
71	Aicardiâ€GoutiÃ¨res syndrome: description of a late onset case. Developmental Medicine and Child Neurology, 2008, 50, 631-634.	2.1	35
72	A de novo p.Asp18Asn mutation in TREX1 in a patient with Aicardiâ€“GoutiÃ¨res syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 2612-2617.	1.2	35

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73	Aicardiâ€“GoutiÃ¨res syndrome harbours abundant systemic and brain-reactive autoantibodies. Annals of the Rheumatic Diseases, 2015, 74, 1931-1939.	0.9	35
74	Angiotensin-converting enzyme (ACE) gene polymorphisms in patients characterised by coronary angiography. Human Genetics, 1997, 100, 420-425.	3.8	34
75	Chilblains as a Diagnostic Sign of Aicardi-GoutiÃ¨res Syndrome. Neuropediatrics, 2010, 41, 18-23.	0.6	32
76	Identification of Novel Polymorphisms within the Protein Z Gene, Haplotype Distribution and Linkage Analysis. Thrombosis and Haemostasis, 2001, 85, 1123-1124.	3.4	31
77	Two further cases of spondyloenchondrodysplasia (SPENCD) with immune dysregulation. American Journal of Medical Genetics, Part A, 2008, 146A, 2810-2815.	1.2	30
78	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.	5.6	30
79	Systemic lupus erythematosus due to C1q deficiency with progressive encephalopathy, intracranial calcification and acquired moyamoya cerebral vasculopathy. Lupus, 2013, 22, 639-643.	1.6	29
80	Clinical, radiological and possible pathological overlap of cystic leukoencephalopathy without megalencephaly and Aicardi-GoutiÃ¨res syndrome. European Journal of Paediatric Neurology, 2016, 20, 604-610.	1.6	29
81	Angiotensin converting enzyme and angiotensin II type 1-receptor gene polymorphisms and risk of ischaemic heart disease. Cardiovascular Research, 1999, 41, 746-753.	3.8	28
82	Aicardiâ€“GoutiÃ¨res syndrome presenting atypically as a sub-acute leukoencephalopathy. European Journal of Paediatric Neurology, 2008, 12, 408-411.	1.6	27
83	Anti-MDA5 juvenile idiopathic inflammatory myopathy: a specific subgroup defined by differentially enhanced interferon-Î± signalling. Rheumatology, 2020, 59, 1927-1937.	1.9	26
84	FVIII Coagulant Activity and Antigen in Subjects with Ischaemic Heart Disease. Thrombosis and Haemostasis, 1998, 80, 757-762.	3.4	24
85	JAK 1/2 Blockade in MDA5 Gain-of-Function. Journal of Clinical Immunology, 2018, 38, 844-846.	3.8	24
86	JAK Inhibition in the Aicardiâ€“GoutiÃ¨res Syndrome. New England Journal of Medicine, 2020, 383, 2190-2193.	27.0	24
87	Elevation of proinflammatory cytokines in patients with Aicardi-GoutiÃ¨res syndrome. Neurology, 2013, 80, 997-1002.	1.1	23
88	JAK inhibition in STING-associated interferonopathy. Annals of the Rheumatic Diseases, 2016, 75, e75-e75.	0.9	22
89	MDA5-Associated Neuroinflammation and the Singletonâ€“Merten Syndrome: Two Faces of the Same Type I Interferonopathy Spectrum. Journal of Interferon and Cytokine Research, 2017, 37, 214-219.	1.2	21
90	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.	3.5	20

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91	Basal Ganglia Calcification in a Patient With Beta-Propeller Protein-Associated Neurodegeneration. Pediatric Neurology, 2014, 51, 843-845.	2.1	17
92	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.	6.2	17
93	Biallelic mutations in NRROS cause an early onset lethal microgliopathy. Acta Neuropathologica, 2020, 139, 947-951.	7.7	17
94	LACC1 deficiency links juvenile arthritis with autophagy and metabolism in macrophages. Journal of Experimental Medicine, 2021, 218, .	8.5	17
95	Synonymous Mutations inRNASEH2ACreate Cryptic Splice Sites Impairing RNase H2 Enzyme Function in Aicardi-GoutiÃ¨res Syndrome. Human Mutation, 2013, 34, 1066-1070.	2.5	16
96	Differential Expression of Interferon-Alpha Protein Provides Clues to Tissue Specificity Across Type I Interferonopathies. Journal of Clinical Immunology, 2021, 41, 603-609.	3.8	16
97	ADAR1 Facilitates HIV-1 Replication in Primary CD4+ T Cells. PLoS ONE, 2015, 10, e0143613.	2.5	16
98	Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum. American Journal of Medical Genetics, Part A, 2021, 185, 15-25.	1.2	15
99	From Diagnosis to Prognosis: Revisiting the Meaning of Muscle ISG15 Overexpression in Juvenile Inflammatory Myopathies. Arthritis and Rheumatology, 2021, 73, 1044-1052.	5.6	13
100	Comment on: â€˜Aberrant tRNA processing causes an autoinflammatory syndrome responsive to TNF inhibitorsâ€™ by Giannelou et al: mutations in TRNT1 result in a constitutive activation of type I interferon signalling. Annals of the Rheumatic Diseases, 2019, 78, e86-e86.	0.9	12
101	Differential levels of IFNÎ± subtypes in autoimmunity and viral infection. Cytokine, 2021, 144, 155533.	3.2	12
102	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, 109, 210-222.	6.2	12
103	Comprehensive molecular screening strategy of <scp>OCLN</scp> in bandâ€like calcification with simplified gyration and polymicrogyria. Clinical Genetics, 2018, 93, 228-234.	2.0	9
104	Sine causa tetraparesis. Medicine (United States), 2018, 97, e13893.	1.0	9
105	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.	3.4	8
106	Autosomal-dominant early-onset spastic paraparesis with brain calcification due to IFIH1 gain-of-function. Human Mutation, 2018, 39, 1076-1080.	2.5	8
107	Mutations in RNU7-1 Weaken Secondary RNA Structure, Induce MCP-1 and CXCL10 in CSF, and Result in Aicardi-GoutiÃ¨res Syndrome with Severe End-Organ Involvement. Journal of Clinical Immunology, 2022, 42, 962-974.	3.8	8
108	Systemic inflammatory syndrome in children with <scp>FARSA</scp> deficiency. Clinical Genetics, 2022, 101, 552-558.	2.0	7

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109	DNASE1L3 deficiency, new phenotypes, and evidence for a transient type I IFN signaling. Journal of Clinical Immunology, 2022, 42, 1310-1320.	3.8	7
110	Combination of exome sequencing and immune testing confirms Aicardiâ€“GoutiÃ¨res syndrome type 5 in a challenging pediatric neurology case. Journal of Physical Education and Sports Management, 2018, 4, a002758.	1.2	6
111	RNASEH2B Related Adult-Onset Interferonopathy. Journal of Clinical Immunology, 2019, 39, 620-622.	3.8	6
112	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.	2.1	6
113	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.	2.5	6
114	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.	2.0	4
115	Biallelic Mutations in MTPAP Associated with a Lethal Encephalopathy. Neuropediatrics, 2020, 51, 178-184.	0.6	3
116	Aicardiâ€“GoutiÃ¨res syndrome presenting with haematemesis in infancy. Acta Paediatrica, International Journal of Paediatrics, 2009, 98, 2005-2008.	1.5	2
117	Clinical Reasoning: A 25-year-old woman with recurrent episodes of collapse and loss of consciousness. Neurology, 2020, 94, 994-999.	1.1	2
118	PRKDC mutations associated with immunodeficiency, granuloma and aire-dependent autoimmunity. Pediatric Rheumatology, 2014, 12, .	2.1	1
119	COPA syndrome restricted to life-threatening alveolar hemorrhages: clinical, pathological, molecular and biological characterization. , 2018, , .		1
120	320.â€ƒTYPE 1 INTERFERON EXPRESSION IS ASSOCIATED WITH AUTOANTIBODIES ACROSS SYSTEMIC AUTOIMMUNE DISEASES: RESULTS FROM THE LUPUS EXTENDED AUTOIMMUNE PHENOTYPE STUDY. Rheumatology, 2017, 56, .	1.9	0
121	A child with severe juvenile dermatomyositis treated with ruxolitinib. Journal of Financial Econometrics, 0, , .	1.5	0
122	E086â€ƒClinical and serological features of increased interferon-alpha activity in an unselected connective tissue disease cohort. Rheumatology, 2019, 58, .	1.9	0
123	OP0107â€…HETEROZYGOUS MUTATIONS IN COPA ARE ASSOCIATED WITH ENHANCED TYPE I INTERFERON SIGNALLING. , 2019, , .		0