Gillian I Rice

List of Publications by Year
in descending order

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Version: 2024-02-01

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	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
2	Systemic inflammatory syndrome in children with <scp>FARSA</scp> deficiency. Clinical Genetics, 2022, 101, 552-558.	2.0	7
3	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
4	DNASE1L3 deficiency, new phenotypes, and evidence for a transient type I IFN signaling. Journal of Clinical Immunology, 2022, 42, 1310-1320.	3.8	7
5	Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum. American Journal of Medical Genetics, Part A, 2021, 185, 15-25.	1.2	15
6	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
7	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
8	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
9	LACC1 deficiency links juvenile arthritis with autophagy and metabolism in macrophages. Journal of Experimental Medicine, 2021, 218, .	8.5	17
10	Differential levels of IFNÎ± subtypes in autoimmunity and viral infection. Cytokine, 2021, 144, 155533.	3.2	12
11	Enhanced cGAS-STINGâ€“dependent interferon signaling associated with mutations in ATAD3A. Journal of Experimental Medicine, 2021, 218, .	8.5	43
12	Use of ruxolitinib in COPA syndrome manifesting as life-threatening alveolar haemorrhage. Thorax, 2020, 75, 92-95.	5.6	36
13	Biallelic Mutations in MTPAP Associated with a Lethal Encephalopathy. Neuropediatrics, 2020, 51, 178-184.	0.6	3
14	Anti-MDA5 juvenile idiopathic inflammatory myopathy: a specific subgroup defined by differentially enhanced interferon-Î± signalling. Rheumatology, 2020, 59, 1927-1937.	1.9	26
15	PSMB10, the last immunoproteasome gene missing for PRAAS. Journal of Allergy and Clinical Immunology, 2020, 145, 1015-1017.e6.	2.9	42
16	Genetic and phenotypic spectrum associated with IFIH1 gainâ€ofâ€function. Human Mutation, 2020, 41, 837-849.	2.5	63
17	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
18	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

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19	JAK Inhibition in the Aicardiâ€“GoutiÃ¨res Syndrome. New England Journal of Medicine, 2020, 383, 2190-2193.	27.0	24
20	Mutations in COPA lead to abnormal trafficking of STING to the Golgi and interferon signaling. Journal of Experimental Medicine, 2020, 217, .	8.5	130
21	Clinical Reasoning: A 25-year-old woman with recurrent episodes of collapse and loss of consciousness. Neurology, 2020, 94, 994-999.	1.1	2
22	Biallelic mutations in NRROS cause an early onset lethal microgliopathy. Acta Neuropathologica, 2020, 139, 947-951.	7.7	17
23	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
24	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
25	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
26	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
27	RNASEH2B Related Adult-Onset Interferonopathy. Journal of Clinical Immunology, 2019, 39, 620-622.	3.8	6
28	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
29	E086â€ƒClinical and serological features of increased interferon-alpha activity in an unselected connective tissue disease cohort. Rheumatology, 2019, 58, .	1.9	0
30	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
31	Bloom syndrome protein restrains innate immune sensing of micronuclei by cGAS. Journal of Experimental Medicine, 2019, 216, 1199-1213.	8.5	75
32	OP0107â€…HETEROZYGOUS MUTATIONS IN COPA ARE ASSOCIATED WITH ENHANCED TYPE I INTERFERON SIGNALLING. , 2019, , .		0
33	Severe type I interferonopathy and unrestrained interferon signaling due to a homozygous germline mutation in STAT2. Science Immunology, 2019, 4, .	11.9	80
34	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
35	An open-label trial of JAK 1/2 blockade in progressive IFIH1 -associated neuroinflammation. Neurology, 2018, 90, 289-291.	1.1	60
36	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

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37	Sine causa tetraparesis. Medicine (United States), 2018, 97, e13893.	1.0	9
38	JAK 1/2 Blockade in MDA5 Gain-of-Function. Journal of Clinical Immunology, 2018, 38, 844-846.	3.8	24
39	Reverse-Transcriptase Inhibitors in the Aicardiâ€“GoutiÃ¨res Syndrome. New England Journal of Medicine, 2018, 379, 2275-2277.	27.0	106
40	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
41	A child with severe juvenile dermatomyositis treated with ruxolitinib. Brain, 2018, 141, e80-e80.	7.6	58
42	Autosomal-dominant early-onset spastic paraparesis with brain calcification due to IFIH1 gain-of-function. Human Mutation, 2018, 39, 1076-1080.	2.5	8
43	Mitochondrial double-stranded RNA triggers antiviral signalling in humans. Nature, 2018, 560, 238-242.	27.8	397
44	COPA syndrome restricted to life-threatening alveolar hemorrhages: clinical, pathological, molecular and biological characterization. , 2018, , .		1
45	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
46	Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. Neuropediatrics, 2017, 48, 166-184.	0.6	62
47	Detection of interferon alpha protein reveals differential levels and cellular sources in disease. Journal of Experimental Medicine, 2017, 214, 1547-1555.	8.5	288
48	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
49	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
50	Assessment of Type I Interferon Signaling in Pediatric Inflammatory Disease. Journal of Clinical Immunology, 2017, 37, 123-132.	3.8	163
51	Expression of Cyclic GMPâ€AMP Synthase in Patients With Systemic Lupus Erythematosus. Arthritis and Rheumatology, 2017, 69, 800-807.	5.6	129
52	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
53	Tartrateâ€Resistant Acid Phosphatase Deficiency in the Predisposition to Systemic Lupus Erythematosus. Arthritis and Rheumatology, 2017, 69, 131-142.	5.6	47
54	Type I interferon-mediated autoinflammation due to DNase II deficiency. Nature Communications, 2017, 8, 2176.	12.8	164

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55	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
56	JAK inhibition in STING-associated interferonopathy. Annals of the Rheumatic Diseases, 2016, 75, e75-e75.	0.9	22
57	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
58	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature Genetics, 2016, 48, 1185-1192.	21.4	114
59	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
60	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
61	Spondyloenchondrodysplasia Due to Mutations in ACP5: A Comprehensive Survey. Journal of Clinical Immunology, 2016, 36, 220-234.	3.8	71
62	ADA2 deficiency: case report of a new phenotype and novel mutation in two sisters. RMD Open, 2016, 2, e000236.	3.8	47
63	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
64	Stimulator of Interferon Genesâ€“Associated Vasculopathy With Onset in Infancy. JAMA Dermatology, 2015, 151, 872.	4.1	108
65	A Specific IFIH1 Gain-of-Function Mutation Causes Singleton-Merten Syndrome. American Journal of Human Genetics, 2015, 96, 275-282.	6.2	188
66	Aicardiâ€“GoutiÃ¨res syndrome harbours abundant systemic and brain-reactive autoantibodies. Annals of the Rheumatic Diseases, 2015, 74, 1931-1939.	0.9	35
67	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
68	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
69	Human Disease Phenotypes Associated With Mutations in TREX1. Journal of Clinical Immunology, 2015, 35, 235-243.	3.8	154
70	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
71	Human intracellular ISG15 prevents interferon-Î±/Î² over-amplification and auto-inflammation. Nature, 2015, 517, 89-93.	27.8	432
72	ADAR1 Facilitates HIV-1 Replication in Primary CD4+ T Cells. PLoS ONE, 2015, 10, e0143613.	2.5	16

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73	Inherited STING-activating mutation underlies a familial inflammatory syndrome with lupus-like manifestations. Journal of Clinical Investigation, 2014, 124, 5516-5520.	8.2	435
74	Leukoencephalopathy with Calcifications and Cysts: A Purely Neurological Disorder Distinct from Coats Plus. Neuropediatrics, 2014, 45, 175-182.	0.6	41
75	Mutations in ADAR1, IFIH1, and RNASEH2B Presenting As Spastic Paraplegia. Neuropediatrics, 2014, 45, 386-391.	0.6	72
76	Basal Ganglia Calcification in a Patient With Beta-Propeller Protein-Associated Neurodegeneration. Pediatric Neurology, 2014, 51, 843-845.	2.1	17
77	Mutations in CECR1 associated with a neutrophil signature in peripheral blood. Pediatric Rheumatology, 2014, 12, 44.	2.1	88
78	PRKDC mutations associated with immunodeficiency, granuloma and aire-dependent autoimmunity. Pediatric Rheumatology, 2014, 12, .	2.1	1
79	A type I interferon signature identifies bilateral striatal necrosis due to mutations inADAR1. Journal of Medical Genetics, 2014, 51, 76-82.	3.2	118
80	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
81	The SKIV2L RNA exosome limits activation of the RIG-I-like receptors. Nature Immunology, 2014, 15, 839-845.	14.5	170
82	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
83	Elevation of proinflammatory cytokines in patients with Aicardi-GoutiÃ¨res syndrome. Neurology, 2013, 80, 997-1002.	1.1	23
84	Protein Kinase CÎ´ Deficiency Causes Mendelian Systemic Lupus Erythematosus With B Cellâ€Đefective Apoptosis and Hyperproliferation. Arthritis and Rheumatism, 2013, 65, 2161-2171.	6.7	155
85	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
86	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
87	Therapies in Aicardiâ€“GoutiÃ¨res syndrome. Clinical and Experimental Immunology, 2013, 175, 1-8.	2.6	74
88	SAMHD1 restricts HIV-1 reverse transcription in quiescent CD4+T-cells. Retrovirology, 2012, 9, 87.	2.0	302
89	Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. Nature Genetics, 2012, 44, 338-342.	21.4	234
90	Mutations in ADAR1 cause Aicardi-GoutiÃ¨res syndrome associated with a type I interferon signature. Nature Genetics, 2012, 44, 1243-1248.	21.4	712

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91	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
92	HIV-1 restriction factor SAMHD1 is a deoxynucleoside triphosphate triphosphohydrolase. Nature, 2011, 480, 379-382.	27.8	707
93	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
94	Neprilysin, obesity and the metabolic syndrome. International Journal of Obesity, 2011, 35, 1031-1040.	3.4	137
95	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
96	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
97	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
98	COL4A1 Mutations Associated with a Characteristic Pattern of Intracranial Calcification. Neuropediatrics, 2011, 42, 227-233.	0.6	38
99	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
100	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
101	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
102	Chilblains as a Diagnostic Sign of Aicardi-GoutiÃ¨res Syndrome. Neuropediatrics, 2010, 41, 18-23.	0.6	32
103	Aicardiâ€“GoutiÃ¨res syndrome presenting with haematemesis in infancy. Acta Paediatrica, International Journal of Paediatrics, 2009, 98, 2005-2008.	1.5	2
104	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
105	Cerebroretinal microangiopathy with calcifications and cysts (CRMCC). American Journal of Medical Genetics, Part A, 2008, 146A, 182-190.	1.2	87
106	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
107	Two further cases of spondyloenchondrodysplasia (SPENCD) with immune dysregulation. American Journal of Medical Genetics, Part A, 2008, 146A, 2810-2815.	1.2	30
108	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

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109	Cutaneous histopathological findings of Aicardiâ€“GoutiÃ¨res syndrome, overlap with chilblain lupus. Journal of Cutaneous Pathology, 2008, 35, 774-778.	1.3	47
110	Aicardiâ€GoutiÃ¨res syndrome: description of a late onset case. Developmental Medicine and Child Neurology, 2008, 50, 631-634.	2.1	35
111	Aicardiâ€“GoutiÃ¨res syndrome presenting atypically as a sub-acute leukoencephalopathy. European Journal of Paediatric Neurology, 2008, 12, 408-411.	1.6	27
112	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
113	Clinical and Molecular Phenotype of Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 81, 713-725.	6.2	375
114	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
115	Evaluation of angiotensin-converting enzyme (ACE), its homologue ACE2 and neprilysin in angiotensin peptide metabolism. Biochemical Journal, 2004, 383, 45-51.	3.7	539
116	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
117	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
118	Identification of Novel Polymorphisms within the Protein Z Gene, Haplotype Distribution and Linkage Analysis. Thrombosis and Haemostasis, 2001, 85, 1123-1124.	3.4	31
119	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
120	FVIII Coagulant Activity and Antigen in Subjects with Ischaemic Heart Disease. Thrombosis and Haemostasis, 1998, 80, 757-762.	3.4	24
121	Angiotensin-converting enzyme (ACE) gene polymorphisms in patients characterised by coronary angiography. Human Genetics, 1997, 100, 420-425.	3.8	34
122	The paraoxonase Gln-Arg 192 polymorphism in subjects with ischaemic heart disease. Coronary Artery Disease, 1997, 8, 677-682.	0.7	52
123	A child with severe juvenile dermatomyositis treated with ruxolitinib. Journal of Financial Econometrics, 0, , .	1.5	0