

# Yanick J Crow

## List of Publications by Year in descending order

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242  
papers

25,641  
citations

8208

78  
h-index

8878

150  
g-index

248  
all docs

248  
docs citations

248  
times ranked

28846  
citing authors

#	ARTICLE	IF	CITATIONS
1	Large-scale discovery of novel genetic causes of developmental disorders. <i>Nature</i> , 2015, 519, 223-228.	13.7	998
2	Mutations in the gene encoding the 3'→5' DNA exonuclease TREX1 cause Aicardi-Goutières syndrome at the AGS1 locus. <i>Nature Genetics</i> , 2006, 38, 917-920.	9.4	752
3	International Union of Immunological Societies: 2017 Primary Immunodeficiency Diseases Committee Report on Inborn Errors of Immunity. <i>Journal of Clinical Immunology</i> , 2018, 38, 96-128.	2.0	732
4	Mutations in ADAR1 cause Aicardi-Goutières syndrome associated with a type I interferon signature. <i>Nature Genetics</i> , 2012, 44, 1243-1248.	9.4	712
5	HIV-1 restriction factor SAMHD1 is a deoxynucleoside triphosphate triphosphohydrolase. <i>Nature</i> , 2011, 480, 379-382.	13.7	707
6	Aicardi-Goutières syndrome and the type I interferonopathies. <i>Nature Reviews Immunology</i> , 2015, 15, 429-440.	10.6	705
7	Mutations involved in Aicardi-Goutières syndrome implicate SAMHD1 as regulator of the innate immune response. <i>Nature Genetics</i> , 2009, 41, 829-832.	9.4	610
8	Mutations in genes encoding ribonuclease H2 subunits cause Aicardi-Goutières syndrome and mimic congenital viral brain infection. <i>Nature Genetics</i> , 2006, 38, 910-916.	9.4	592
9	ASPM is a major determinant of cerebral cortical size. <i>Nature Genetics</i> , 2002, 32, 316-320.	9.4	538
10	The spectrum of SCN1A-related infantile epileptic encephalopathies. <i>Brain</i> , 2007, 130, 843-852.	3.7	501
11	Identification of Microcephalin, a Protein Implicated in Determining the Size of the Human Brain. <i>American Journal of Human Genetics</i> , 2002, 71, 136-142.	2.6	499
12	Gain-of-function mutations in IFIH1 cause a spectrum of human disease phenotypes associated with upregulated type I interferon signaling. <i>Nature Genetics</i> , 2014, 46, 503-509.	9.4	490
13	The 2017 IUIS Phenotypic Classification for Primary Immunodeficiencies. <i>Journal of Clinical Immunology</i> , 2018, 38, 129-143.	2.0	488
14	Assessment of interferon-related biomarkers in Aicardi-Goutières syndrome associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, and ADAR: a case-control study. <i>Lancet Neurology</i> , The, 2013, 12, 1159-1169.	4.9	473
15	Characterization of human disease phenotypes associated with mutations in <i>TREX1</i> , <i>RNASEH2A</i> , <i>RNASEH2B</i> , <i>RNASEH2C</i> , <i>SAMHD1</i> , <i>ADAR</i> , and <i>IFIH1</i> . <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 296-312.	0.7	447
16	Inherited STING-activating mutation underlies a familial inflammatory syndrome with lupus-like manifestations. <i>Journal of Clinical Investigation</i> , 2014, 124, 5516-5520.	3.9	435
17	Human intracellular ISG15 prevents interferon- $\beta$ over-amplification and auto-inflammation. <i>Nature</i> , 2015, 517, 89-93.	13.7	432
18	Mitochondrial double-stranded RNA triggers antiviral signalling in humans. <i>Nature</i> , 2018, 560, 238-242.	13.7	397

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19	Clinical and Molecular Phenotype of Aicardi-Goutières Syndrome. American Journal of Human Genetics, 2007, 81, 713-725.	2.6	375
20	Mutations in the Pericentrin ( <i>PCNT</i> ) Gene Cause Primordial Dwarfism. Science, 2008, 319, 816-819.	6.0	370
21	Type I interferon-mediated monogenic autoinflammation: The type I interferonopathies, a conceptual overview. Journal of Experimental Medicine, 2016, 213, 2527-2538.	4.2	359
22	Heterozygous Mutations in TREX1 Cause Familial Chilblain Lupus and Dominant Aicardi-Goutières Syndrome. American Journal of Human Genetics, 2007, 80, 811-815.	2.6	339
23	Type I interferonopathies: a novel set of inborn errors of immunity. Annals of the New York Academy of Sciences, 2011, 1238, 91-98.	1.8	337
24	Mutations in the Transmembrane Natriuretic Peptide Receptor NPR-B Impair Skeletal Growth and Cause Acromesomelic Dysplasia, Type Maroteaux. American Journal of Human Genetics, 2004, 75, 27-34.	2.6	325
25	SAMHD1 restricts HIV-1 reverse transcription in quiescent CD4+T-cells. Retrovirology, 2012, 9, 87.	0.9	302
26	Infection-Triggered Familial or Recurrent Cases of Acute Necrotizing Encephalopathy Caused by Mutations in a Component of the Nuclear Pore, RANBP2. American Journal of Human Genetics, 2009, 84, 44-51.	2.6	291
27	Detection of interferon alpha protein reveals differential levels and cellular sources in disease. Journal of Experimental Medicine, 2017, 214, 1547-1555.	4.2	288
28	Aicardi-Goutieres syndrome and related phenotypes: linking nucleic acid metabolism with autoimmunity. Human Molecular Genetics, 2009, 18, R130-R136.	1.4	258
29	Modeling of TREX1-Dependent Autoimmune Disease using Human Stem Cells Highlights L1 Accumulation as a Source of Neuroinflammation. Cell Stem Cell, 2017, 21, 319-331.e8.	5.2	254
30	Clinical delineation and natural history of the <i>PIK3CA</i> -related overgrowth spectrum. American Journal of Medical Genetics, Part A, 2014, 164, 1713-1733.	0.7	249
31	Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. Nature Genetics, 2012, 44, 338-342.	9.4	234
32	Human USP18 deficiency underlies type 1 interferonopathy leading to severe pseudo-TORCH syndrome. Journal of Experimental Medicine, 2016, 213, 1163-1174.	4.2	224
33	tRNA splicing endonuclease mutations cause pontocerebellar hypoplasia. Nature Genetics, 2008, 40, 1113-1118.	9.4	217
34	Tartrate-resistant acid phosphatase deficiency causes a bone dysplasia with autoimmunity and a type I interferon expression signature. Nature Genetics, 2011, 43, 127-131.	9.4	214
35	ADAMTSL2 mutations in geleophysic dysplasia demonstrate a role for ADAMTS-like proteins in TGF- $\beta$ bioavailability regulation. Nature Genetics, 2008, 40, 1119-1123.	9.4	211
36	SAMHD1 is mutated recurrently in chronic lymphocytic leukemia and is involved in response to DNA damage. Blood, 2014, 123, 1021-1031.	0.6	205

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37	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. <i>American Journal of Human Genetics</i> , 2018, 102, 175-187.	2.6	204
38	A Specific IFIH1 Gain-of-Function Mutation Causes Singleton-Merten Syndrome. <i>American Journal of Human Genetics</i> , 2015, 96, 275-282.	2.6	188
39	Preexisting autoantibodies to type I IFNs underlie critical COVID-19 pneumonia in patients with APS-1. <i>Journal of Experimental Medicine</i> , 2021, 218, .	4.2	185
40	Mutations in PIEZO2 Cause Gordon Syndrome, Marden-Walker Syndrome, and Distal Arthrogryposis Type 5. <i>American Journal of Human Genetics</i> , 2014, 94, 734-744.	2.6	171
41	The SKIV2L RNA exosome limits activation of the RIG-I-like receptors. <i>Nature Immunology</i> , 2014, 15, 839-845.	7.0	170
42	Type I interferon-mediated autoinflammation due to DNase II deficiency. <i>Nature Communications</i> , 2017, 8, 2176.	5.8	164
43	The type I interferonopathies: 10 years on. <i>Nature Reviews Immunology</i> , 2022, 22, 471-483.	10.6	164
44	Assessment of Type I Interferon Signaling in Pediatric Inflammatory Disease. <i>Journal of Clinical Immunology</i> , 2017, 37, 123-132.	2.0	163
45	Brown-Vialetto-Van Laere Syndrome, a Ponto-Bulbar Palsy with Deafness, Is Caused by Mutations in C20orf54. <i>American Journal of Human Genetics</i> , 2010, 86, 485-489.	2.6	161
46	Inflammatory profiles across the spectrum of disease reveal a distinct role for GM-CSF in severe COVID-19. <i>Science Immunology</i> , 2021, 6, .	5.6	161
47	Type I interferonopathies: Mendelian type I interferon up-regulation. <i>Current Opinion in Immunology</i> , 2015, 32, 7-12.	2.4	160
48	Disease-associated mutations identify a novel region in human STING necessary for the control of type I interferon signaling. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 140, 543-552.e5.	1.5	159
49	Protein Kinase C $\delta$ Deficiency Causes Mendelian Systemic Lupus Erythematosus With B Cell-Defective Apoptosis and Hyperproliferation. <i>Arthritis and Rheumatism</i> , 2013, 65, 2161-2171.	6.7	155
50	Human Disease Phenotypes Associated With Mutations in TREX1. <i>Journal of Clinical Immunology</i> , 2015, 35, 235-243.	2.0	154
51	Mutation of the Variant $\alpha$ -Tubulin TUBA8 Results in Polymicrogyria with Optic Nerve Hypoplasia. <i>American Journal of Human Genetics</i> , 2009, 85, 737-744.	2.6	151
52	Life-threatening influenza pneumonitis in a child with inherited IRF9 deficiency. <i>Journal of Experimental Medicine</i> , 2018, 215, 2567-2585.	4.2	146
53	How genetically heterogeneous is Kabuki syndrome?: MLL2 testing in 116 patients, review and analyses of mutation and phenotypic spectrum. <i>European Journal of Human Genetics</i> , 2012, 20, 381-388.	1.4	142
54	SAMHD1-dependent retroviral control and escape in mice. <i>EMBO Journal</i> , 2013, 32, 2454-2462.	3.5	141

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55	Intracranial calcification in childhood: a review of aetiologies and recognizable phenotypes. <i>Developmental Medicine and Child Neurology</i> , 2014, 56, 612-626.	1.1	132
56	Mutations in <i>COPA</i> lead to abnormal trafficking of STING to the Golgi and interferon signaling. <i>Journal of Experimental Medicine</i> , 2020, 217, .	4.2	130
57	Neurologic Phenotypes Associated with Mutations in <i>TREX1</i> , <i>RNASEH2A</i> , <i>RNASEH2B</i> , <i>RNASEH2C</i> , <i>SAMHD1</i> , <i>ADAR1</i> , and <i>IFIH1</i> : Aicardi-Goutières Syndrome and Beyond. <i>Neuropediatrics</i> , 2016, 47, 355-360.	0.3	127
58	Aicardi-Goutières syndrome: an important Mendelian mimic of congenital infection. <i>Developmental Medicine and Child Neurology</i> , 2008, 50, 410-416.	1.1	125
59	Recessive Mutations in the Gene Encoding the Tight Junction Protein Occludin Cause Band-like Calcification with Simplified Gyration and Polymicrogyria. <i>American Journal of Human Genetics</i> , 2010, 87, 354-364.	2.6	123
60	Mutations in the palmitoyl-protein thioesterase gene ( <i>PPT</i> ; <i>CLN1</i> ) causing juvenile neuronal ceroid lipofuscinosis with granular osmiophilic deposits [published erratum appears in <i>Hum Mol Genet</i> 1998 Apr;7(4):765]. <i>Human Molecular Genetics</i> , 1998, 7, 291-297.	1.4	122
61	Clinical and Mutational Spectrum of Mowat-Wilson Syndrome. <i>European Journal of Medical Genetics</i> , 2005, 48, 97-111.	0.7	121
62	<i>SAMHD1</i> is a nucleic-acid binding protein that is mislocalized due to aicardi-goutières syndrome-associated mutations. <i>Human Mutation</i> , 2012, 33, 1116-1122.	1.1	121
63	A type I interferon signature identifies bilateral striatal necrosis due to mutations in <i>ADAR1</i> . <i>Journal of Medical Genetics</i> , 2014, 51, 76-82.	1.5	118
64	Mutations in <i>SNORD118</i> cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. <i>Nature Genetics</i> , 2016, 48, 1185-1192.	9.4	114
65	Atypical Progeroid Syndrome due to Heterozygous Missense <i>LMNA</i> Mutations. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2009, 94, 4971-4983.	1.8	113
66	Self-Awareness: Nucleic Acid-Driven Inflammation and the Type I Interferonopathies. <i>Annual Review of Immunology</i> , 2019, 37, 247-267.	9.5	111
67	Stimulator of Interferon Genes-Associated Vasculopathy With Onset in Infancy. <i>JAMA Dermatology</i> , 2015, 151, 872.	2.0	108
68	Reverse-Transcriptase Inhibitors in the Aicardi-Goutières Syndrome. <i>New England Journal of Medicine</i> , 2018, 379, 2275-2277.	13.9	106
69	cGAS-mediated induction of type I interferon due to inborn errors of histone pre-mRNA processing. <i>Nature Genetics</i> , 2020, 52, 1364-1372.	9.4	105
70	A Fifth Locus for Primary Autosomal Recessive Microcephaly Maps to Chromosome 1q31. <i>American Journal of Human Genetics</i> , 2000, 67, 1578-1580.	2.6	101
71	Overview of STING-Associated Vasculopathy with Onset in Infancy (SAVI) Among 21 Patients. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2021, 9, 803-818.e11.	2.0	98
72	Autosomal dominant inheritance of a heterozygous mutation in <i>SAMHD1</i> causing familial chilblain lupus. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 235-237.	0.7	97

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73	Degos Disease. American Journal of Clinical Pathology, 2011, 135, 599-610.	0.4	91
74	Identification and Characterization of an Inborn Error of Metabolism Caused by Dihydrofolate Reductase Deficiency. American Journal of Human Genetics, 2011, 88, 216-225.	2.6	90
75	Intracerebral large artery disease in Aicardi-Goutières syndrome implicates SAMHD1 in vascular homeostasis. Developmental Medicine and Child Neurology, 2010, 52, 725-732.	1.1	89
76	Mutations in CECR1 associated with a neutrophil signature in peripheral blood. Pediatric Rheumatology, 2014, 12, 44.	0.9	88
77	Cerebroretinal microangiopathy with calcifications and cysts (CRMCC). American Journal of Medical Genetics, Part A, 2008, 146A, 182-190.	0.7	87
78	PRKDC mutations associated with immunodeficiency, granuloma, and autoimmune regulator-dependent autoimmunity. Journal of Allergy and Clinical Immunology, 2015, 135, 1578-1588.e5.	1.5	84
79	Update and Mutational Analysis of <i>SLC20A2</i> : A Major Cause of Primary Familial Brain Calcification. Human Mutation, 2015, 36, 489-495.	1.1	80
80	Severe type I interferonopathy and unrestrained interferon signaling due to a homozygous germline mutation in <i>STAT2</i> . Science Immunology, 2019, 4, .	5.6	80
81	Aicardi-Goutières Syndrome Displays Genetic Heterogeneity with One Locus (AGS1) on Chromosome 3p21. American Journal of Human Genetics, 2000, 67, 213-221.	2.6	77
82	Coats' Plus: A Progressive Familial Syndrome of Bilateral Coats' Disease, Characteristic Cerebral Calcification, Leukoencephalopathy, Slow Pre- and Post-Natal Linear Growth and Defects of Bone Marrow and Integument. Neuropediatrics, 2004, 35, 10-19.	0.3	77
83	STING-Associated Vasculopathy with Onset in Infancy – A New Interferonopathy. New England Journal of Medicine, 2014, 371, 568-571.	13.9	77
84	A POT1 mutation implicates defective telomere end fill-in and telomere truncations in Coats plus. Genes and Development, 2016, 30, 812-826.	2.7	77
85	Bloom syndrome protein restrains innate immune sensing of micronuclei by cGAS. Journal of Experimental Medicine, 2019, 216, 1199-1213.	4.2	75
86	8p23.1 duplication syndrome; a novel genomic condition with unexpected complexity revealed by array CGH. European Journal of Human Genetics, 2008, 16, 18-27.	1.4	74
87	Therapies in Aicardi-Goutières syndrome. Clinical and Experimental Immunology, 2013, 175, 1-8.	1.1	74
88	Severe combined immunodeficiency in stimulator of interferon genes (STING) V154M/wild-type mice. Journal of Allergy and Clinical Immunology, 2019, 143, 712-725.e5.	1.5	74
89	Familial Aicardi-Goutières syndrome due to <i>SAMHD1</i> mutations is associated with chronic arthropathy and contractures. American Journal of Medical Genetics, Part A, 2010, 152A, 938-942.	0.7	73
90	Severe childhood SMA and axonal CMT due to anticodon binding domain mutations in the GARS gene. Neurology, 2006, 67, 1710-1712.	1.5	72

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91	Aicardi-Goutières Syndrome: Neuroradiologic Findings and Follow-Up. <i>American Journal of Neuroradiology</i> , 2009, 30, 1971-1976.	1.2	72
92	Mutations in ADAR1, IFIH1, and RNASEH2B Presenting As Spastic Paraplegia. <i>Neuropediatrics</i> , 2014, 45, 386-391.	0.3	72
93	Spondyloenchondrodysplasia Due to Mutations in ACP5: A Comprehensive Survey. <i>Journal of Clinical Immunology</i> , 2016, 36, 220-234.	2.0	71
94	Treatments in Aicardi-Goutières syndrome. <i>Developmental Medicine and Child Neurology</i> , 2020, 62, 42-47.	1.1	70
95	Aicardi-Goutières syndrome. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2013, 113, 1629-1635.	1.0	69
96	Recognizable phenotypes associated with intracranial calcification. <i>Developmental Medicine and Child Neurology</i> , 2013, 55, 46-57.	1.1	68
97	Reduced penetrance alleles for Huntington's disease: a multi-centre direct observational study. <i>Journal of Medical Genetics</i> , 2006, 44, e68-e68.	1.5	67
98	Delineation of Late Onset Hypoventilation Associated with Hypothalamic Dysfunction Syndrome. <i>Pediatric Research</i> , 2008, 64, 689-694.	1.1	63
99	Genetic and phenotypic spectrum associated with IFIH1 gain-of-function. <i>Human Mutation</i> , 2020, 41, 837-849.	1.1	63
100	Gross rearrangements of the MECP2 gene are found in both classical and atypical Rett syndrome patients. <i>Journal of Medical Genetics</i> , 2005, 43, 451-456.	1.5	62
101	Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. <i>Neuropediatrics</i> , 2017, 48, 166-184.	0.3	62
102	An open-label trial of JAK 1/2 blockade in progressive IFIH1-associated neuroinflammation. <i>Neurology</i> , 2018, 90, 289-291.	1.5	60
103	Mutations in MFSD8/CLN7 are a frequent cause of variant-late infantile neuronal ceroid lipofuscinosis. <i>Human Mutation</i> , 2009, 30, E530-E540.	1.1	59
104	Neuroradiologic patterns and novel imaging findings in Aicardi-Goutières syndrome. <i>Neurology</i> , 2016, 86, 28-35.	1.5	59
105	A child with severe juvenile dermatomyositis treated with ruxolitinib. <i>Brain</i> , 2018, 141, e80-e80.	3.7	58
106	Paediatric stroke: genetic insights into disease mechanisms and treatment targets. <i>Lancet Neurology</i> , 2011, 10, 264-274.	4.9	57
107	Genetic syndromes mimic congenital infections. <i>Journal of Pediatrics</i> , 2005, 146, 701-705.	0.9	54
108	Mosaic structural variation in children with developmental disorders. <i>Human Molecular Genetics</i> , 2015, 24, 2733-2745.	1.4	54

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109	Efficacy of JAK1/2 inhibition in the treatment of chilblain lupus due to TREX1 deficiency. <i>Annals of the Rheumatic Diseases</i> , 2019, 78, 431-433.	0.5	53
110	Further delineation of the phenotype of severe congenital neutropenia type 4 due to mutations in G6PC3. <i>European Journal of Human Genetics</i> , 2011, 19, 18-22.	1.4	50
111	STING-Mediated Lung Inflammation and Beyond. <i>Journal of Clinical Immunology</i> , 2021, 41, 501-514.	2.0	48
112	Cutaneous histopathological findings of Aicardiâ€“Goutiâ€™res syndrome, overlap with chilblain lupus. <i>Journal of Cutaneous Pathology</i> , 2008, 35, 774-778.	0.7	47
113	Tartrateâ€“Resistant Acid Phosphatase Deficiency in the Predisposition to Systemic Lupus Erythematosus. <i>Arthritis and Rheumatology</i> , 2017, 69, 131-142.	2.9	47
114	Bandâ€“like intracranial calcification with simplified gyration and polymicrogyria: A distinct â€œpseudoâ€“TORCHâ€“phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 3173-3180.	0.7	46
115	Brownâ€“Vialetoâ€“Van Laere syndrome; variability in age at onset and disease progression highlighting the phenotypic overlap with Fazio-Londe disease. <i>Brain and Development</i> , 2005, 27, 443-446.	0.6	45
116	Variable Phenotype Including Leigh Syndrome with a 9185T>C Mutation in the <i>MTATP6</i> Gene. <i>Neuropediatrics</i> , 2007, 38, 313-316.	0.3	45
117	Molecular screening of ADAMTSL2 gene in 33 patients reveals the genetic heterogeneity of geleophysic dysplasia. <i>Journal of Medical Genetics</i> , 2011, 48, 417-421.	1.5	45
118	Understanding the evolving phenotype of vascular complications in telomere biology disorders. <i>Angiogenesis</i> , 2019, 22, 95-102.	3.7	45
119	Musculoskeletal Disease in MDA5â€“Related Type I Interferonopathy: A Mendelian Mimic of Jaccoud's Arthropathy. <i>Arthritis and Rheumatology</i> , 2017, 69, 2081-2091.	2.9	44
120	Enhanced cGAS-STINGâ€“dependent interferon signaling associated with mutations in ATAD3A. <i>Journal of Experimental Medicine</i> , 2021, 218, .	4.2	43
121	PSMB10, the last immunoproteasome gene missing for PRAAS. <i>Journal of Allergy and Clinical Immunology</i> , 2020, 145, 1015-1017.e6.	1.5	42
122	Leukoencephalopathy with Calcifications and Cysts: A Purely Neurological Disorder Distinct from Coats Plus. <i>Neuropediatrics</i> , 2014, 45, 175-182.	0.3	41
123	Homozygous N-terminal missense mutation in TRNT1 leads to progressive B-cell immunodeficiency in adulthood. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 139, 360-363.e6.	1.5	41
124	A further example of a distinctive autosomal recessive syndrome comprising neonatal diabetes mellitus, intestinal atresias and gall bladder agenesis. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1713-1717.	0.7	38
125	Expanding the clinical spectrum of SLC29A3 gene defects. <i>European Journal of Medical Genetics</i> , 2010, 53, 309-313.	0.7	38
126	COL4A1 Mutations Associated with a Characteristic Pattern of Intracranial Calcification. <i>Neuropediatrics</i> , 2011, 42, 227-233.	0.3	38

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127	Contribution of rare and predicted pathogenic gene variants to childhood-onset lupus: a large, genetic panel analysis of British and French cohorts. <i>Lancet Rheumatology, The</i> , 2020, 2, e99-e109.	2.2	38
128	DDX58 and Classic Singleton-Merten Syndrome. <i>Journal of Clinical Immunology</i> , 2019, 39, 75-80.	2.0	37
129	Diagnosing fetal alcohol syndrome: new insights from newer genetic technologies. <i>Archives of Disease in Childhood</i> , 2012, 97, 812-817.	1.0	36
130	Characterization of <i>samhd1</i> Morphant Zebrafish Recapitulates Features of the Human Type I Interferonopathy Aicardi-Goutières Syndrome. <i>Journal of Immunology</i> , 2015, 194, 2819-2825.	0.4	36
131	Use of ruxolitinib in COPA syndrome manifesting as life-threatening alveolar haemorrhage. <i>Thorax</i> , 2020, 75, 92-95.	2.7	36
132	Aicardi-Goutières syndrome: description of a late onset case. <i>Developmental Medicine and Child Neurology</i> , 2008, 50, 631-634.	1.1	35
133	A de novo p.Asp18Asn mutation in <i>TREX1</i> in a patient with Aicardi-Goutières syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2612-2617.	0.7	35
134	Aicardi-Goutières syndrome harbours abundant systemic and brain-reactive autoantibodies. <i>Annals of the Rheumatic Diseases</i> , 2015, 74, 1931-1939.	0.5	35
135	A second locus for Aicardi-Goutières syndrome at chromosome 13q14-21. <i>Journal of Medical Genetics</i> , 2005, 43, 444-450.	1.5	33
136	Early-Onset Aicardi-Goutières Syndrome. <i>Journal of Child Neurology</i> , 2015, 30, 1343-1348.	0.7	33
137	Chilblains as a Diagnostic Sign of Aicardi-Goutières Syndrome. <i>Neuropediatrics</i> , 2010, 41, 18-23.	0.3	32
138	Adult-Onset ANCA-Associated Vasculitis in SAVI: Extension of the Phenotypic Spectrum, Case Report and Review of the Literature. <i>Frontiers in Immunology</i> , 2020, 11, 575219.	2.2	32
139	The 2021 European Alliance of Associations for Rheumatology/American College of Rheumatology points to consider for diagnosis and management of autoinflammatory type I interferonopathies: CANDLE/PRAAS, SAVI and AGS. <i>Annals of the Rheumatic Diseases</i> , 2022, 81, 601-613.	0.5	31
140	Congenital glaucoma and brain stem atrophy as features of Aicardi-Goutières syndrome. , 2004, 129A, 303-307.		30
141	Two further cases of spondyloenchondrodysplasia (SPENCD) with immune dysregulation. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 2810-2815.	0.7	30
142	Cerebral vasculopathy is a common feature in Aicardi-Goutières syndrome associated with <i>SAMHD1</i> mutations. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2011, 108, E232; author reply E233.	3.3	29
143	Systemic lupus erythematosus due to C1q deficiency with progressive encephalopathy, intracranial calcification and acquired moyamoya cerebral vasculopathy. <i>Lupus</i> , 2013, 22, 639-643.	0.8	29
144	Clinical, radiological and possible pathological overlap of cystic leukoencephalopathy without megalencephaly and Aicardi-Goutières syndrome. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 604-610.	0.7	29

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145	Striking intrafamilial phenotypic variability in Aicardi-Goutières syndrome associated with the recurrent Asian founder mutation in <i>RNASEH2C</i> . <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 338-342.	0.7	28
146	A partial form of inherited human USP18 deficiency underlies infection and inflammation. <i>Journal of Experimental Medicine</i> , 2022, 219, .	4.2	28
147	Aicardi-Goutières syndrome presenting atypically as a sub-acute leukoencephalopathy. <i>European Journal of Paediatric Neurology</i> , 2008, 12, 408-411.	0.7	27
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