Yanick J Crow

List of Publications by Year
in descending order

242 papers	25,641 citations	7096 78 h-index	7745 150 g-index
248 all docs	248 docs citations	248 times ranked	26838 citing authors

#	Article	IF	Citations
1	Large-scale discovery of novel genetic causes of developmental disorders. Nature, 2015, 519, 223-228.	27.8	998
2	Mutations in the gene encoding the 3â€²-5â€² DNA exonuclease TREX1 cause Aicardi-GoutiÃ¨res syndrome at the AGS1 locus. Nature Genetics, 2006, 38, 917-920.	21.4	752
3	International Union of Immunological Societies: 2017 Primary Immunodeficiency Diseases Committee Report on Inborn Errors of Immunity. Journal of Clinical Immunology, 2018, 38, 96-128.	3.8	732
4	Mutations in ADAR1 cause Aicardi-GoutiÃ¨res syndrome associated with a type I interferon signature. Nature Genetics, 2012, 44, 1243-1248.	21.4	712
5	HIV-1 restriction factor SAMHD1 is a deoxynucleoside triphosphate triphosphohydrolase. Nature, 2011, 480, 379-382.	27.8	707
6	Aicardiâ€“GoutiÃ¨res syndrome and the type I interferonopathies. Nature Reviews Immunology, 2015, 15, 429-440.	22.7	705
7	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
8	Mutations in genes encoding ribonuclease H2 subunits cause Aicardi-GoutiÃ¨res syndrome and mimic congenital viral brain infection. Nature Genetics, 2006, 38, 910-916.	21.4	592
9	ASPM is a major determinant of cerebral cortical size. Nature Genetics, 2002, 32, 316-320.	21.4	538
10	The spectrum of SCN1A-related infantile epileptic encephalopathies. Brain, 2007, 130, 843-852.	7.6	501
11	Identification of Microcephalin, a Protein Implicated in Determining the Size of the Human Brain. American Journal of Human Genetics, 2002, 71, 136-142.	6.2	499
12	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
13	The 2017 IUIS Phenotypic Classification for Primary Immunodeficiencies. Journal of Clinical Immunology, 2018, 38, 129-143.	3.8	488
14	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
15	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
16	Inherited STING-activating mutation underlies a familial inflammatory syndrome with lupus-like manifestations. Journal of Clinical Investigation, 2014, 124, 5516-5520.	8.2	435
17	Human intracellular ISG15 prevents interferon-Î±/Î² over-amplification and auto-inflammation. Nature, 2015, 517, 89-93.	27.8	432
18	Mitochondrial double-stranded RNA triggers antiviral signalling in humans. Nature, 2018, 560, 238-242.	27.8	397

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19	Clinical and Molecular Phenotype of Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 81, 713-725.	6.2	375
20	Mutations in the Pericentrin (PCNT) Gene Cause Primordial Dwarfism. Science, 2008, 319, 816-819.	12.6	370
21	Type I interferonâ€“mediated monogenic autoinflammation: The type I interferonopathies, a conceptual overview. Journal of Experimental Medicine, 2016, 213, 2527-2538.	8.5	359
22	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
23	Type I interferonopathies: a novel set of inborn errors of immunity. Annals of the New York Academy of Sciences, 2011, 1238, 91-98.	3.8	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.	6.2	325
25	SAMHD1 restricts HIV-1 reverse transcription in quiescent CD4+T-cells. Retrovirology, 2012, 9, 87.	2.0	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.	6.2	291
27	Detection of interferon alpha protein reveals differential levels and cellular sources in disease. Journal of Experimental Medicine, 2017, 214, 1547-1555.	8.5	288
28	Aicardi-Goutieres syndrome and related phenotypes: linking nucleic acid metabolism with autoimmunity. Human Molecular Genetics, 2009, 18, R130-R136.	2.9	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.	11.1	254
30	Clinical delineation and natural history of the PIK3CAâ€related overgrowth spectrum. American Journal of Medical Genetics, Part A, 2014, 164, 1713-1733.	1.2	249
31	Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. Nature Genetics, 2012, 44, 338-342.	21.4	234
32	Human USP18 deficiency underlies type 1 interferonopathy leading to severe pseudo-TORCH syndrome. Journal of Experimental Medicine, 2016, 213, 1163-1174.	8.5	224
33	tRNA splicing endonuclease mutations cause pontocerebellar hypoplasia. Nature Genetics, 2008, 40, 1113-1118.	21.4	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.	21.4	214
35	ADAMTSL2 mutations in geleophysic dysplasia demonstrate a role for ADAMTS-like proteins in TGF-Î² bioavailability regulation. Nature Genetics, 2008, 40, 1119-1123.	21.4	211
36	SAMHD1 is mutated recurrently in chronic lymphocytic leukemia and is involved in response to DNA damage. Blood, 2014, 123, 1021-1031.	1.4	205

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37	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	6.2	204
38	A Specific IFIH1 Gain-of-Function Mutation Causes Singleton-Merten Syndrome. American Journal of Human Genetics, 2015, 96, 275-282.	6.2	188
39	Preexisting autoantibodies to type I IFNs underlie critical COVID-19 pneumonia in patients with APS-1. Journal of Experimental Medicine, 2021, 218, .	8.5	185
40	Mutations in PIEZO2 Cause Gordon Syndrome, Marden-Walker Syndrome, and Distal Arthrogryposis Type 5. American Journal of Human Genetics, 2014, 94, 734-744.	6.2	171
41	The SKIV2L RNA exosome limits activation of the RIG-I-like receptors. Nature Immunology, 2014, 15, 839-845.	14.5	170
42	Type I interferon-mediated autoinflammation due to DNase II deficiency. Nature Communications, 2017, 8, 2176.	12.8	164
43	The type I interferonopathies: 10 years on. Nature Reviews Immunology, 2022, 22, 471-483.	22.7	164
44	Assessment of Type I Interferon Signaling in Pediatric Inflammatory Disease. Journal of Clinical Immunology, 2017, 37, 123-132.	3.8	163
45	Brown-Vialetto-Van Laere Syndrome, a Ponto-Bulbar Palsy with Deafness, Is Caused by Mutations in C20orf54. American Journal of Human Genetics, 2010, 86, 485-489.	6.2	161
46	Inflammatory profiles across the spectrum of disease reveal a distinct role for GM-CSF in severe COVID-19. Science Immunology, 2021, 6, .	11.9	161
47	Type I interferonopathies: Mendelian type I interferon up-regulation. Current Opinion in Immunology, 2015, 32, 7-12.	5.5	160
48	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
49	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
50	Human Disease Phenotypes Associated With Mutations in TREX1. Journal of Clinical Immunology, 2015, 35, 235-243.	3.8	154
51	Mutation of the Variant Î±-Tubulin TUBA8 Results in Polymicrogyria with Optic Nerve Hypoplasia. American Journal of Human Genetics, 2009, 85, 737-744.	6.2	151
52	Life-threatening influenza pneumonitis in a child with inherited IRF9 deficiency. Journal of Experimental Medicine, 2018, 215, 2567-2585.	8.5	146
53	How genetically heterogeneous is Kabuki syndrome?: MLL2 testing in 116 patients, review and analyses of mutation and phenotypic spectrum. European Journal of Human Genetics, 2012, 20, 381-388.	2.8	142
54	SAMHD1-dependent retroviral control and escape in mice. EMBO Journal, 2013, 32, 2454-2462.	7.8	141

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55	Intracranial calcification in childhood: a review of aetiologies and recognizable phenotypes. Developmental Medicine and Child Neurology, 2014, 56, 612-626.	2.1	132
56	Mutations in COPA lead to abnormal trafficking of STING to the Golgi and interferon signaling. Journal of Experimental Medicine, 2020, 217, .	8.5	130
57	Neurologic Phenotypes Associated with Mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR1, and IFIH1: Aicardiâ€“GoutiÃ¨res Syndrome and Beyond. Neuropediatrics, 2016, 47, 355-360.	0.6	127
58	Aicardiâ€GoutiÃ¨res syndrome: an important Mendelian mimic of congenital infection. Developmental Medicine and Child Neurology, 2008, 50, 410-416.	2.1	125
59	Recessive Mutations in the Gene Encoding the Tight Junction Protein Occludin Cause Band-like Calcification with Simplified Gyration and Polymicrogyria. American Journal of Human Genetics, 2010, 87, 354-364.	6.2	123
60	Mutations in the palmitoyl-protein thioesterase gene (PPT; CLN1) causing juvenile neuronal ceroid lipofuscinosis with granular osmiophilic deposits [published erratum appears in Hum Mol Genet 1998 Apr;7(4):765]. Human Molecular Genetics, 1998, 7, 291-297.	2.9	122
61	Clinical and Mutational Spectrum of Mowatâ€“Wilson Syndrome. European Journal of Medical Genetics, 2005, 48, 97-111.	1.3	121
62	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
63	A type I interferon signature identifies bilateral striatal necrosis due to mutations inADAR1. Journal of Medical Genetics, 2014, 51, 76-82.	3.2	118
64	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature Genetics, 2016, 48, 1185-1192.	21.4	114
65	Atypical Progeroid Syndrome due to Heterozygous Missense LMNA Mutations. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 4971-4983.	3.6	113
66	Self-Awareness: Nucleic Acidâ€“Driven Inflammation and the Type I Interferonopathies. Annual Review of Immunology, 2019, 37, 247-267.	21.8	111
67	Stimulator of Interferon Genesâ€“Associated Vasculopathy With Onset in Infancy. JAMA Dermatology, 2015, 151, 872.	4.1	108
68	Reverse-Transcriptase Inhibitors in the Aicardiâ€“GoutiÃ¨res Syndrome. New England Journal of Medicine, 2018, 379, 2275-2277.	27.0	106
69	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
70	A Fifth Locus for Primary Autosomal Recessive Microcephaly Maps to Chromosome 1q31. American Journal of Human Genetics, 2000, 67, 1578-1580.	6.2	101
71	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
72	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

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73	Degos Disease. American Journal of Clinical Pathology, 2011, 135, 599-610.	0.7	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.	6.2	90
75	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
76	Mutations in CECR1 associated with a neutrophil signature in peripheral blood. Pediatric Rheumatology, 2014, 12, 44.	2.1	88
77	Cerebroretinal microangiopathy with calcifications and cysts (CRMCC). American Journal of Medical Genetics, Part A, 2008, 146A, 182-190.	1.2	87
78	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
79	Update and Mutational Analysis ofSLC20A2: A Major Cause of Primary Familial Brain Calcification. Human Mutation, 2015, 36, 489-495.	2.5	80
80	Severe type I interferonopathy and unrestrained interferon signaling due to a homozygous germline mutation in STAT2. Science Immunology, 2019, 4, .	11.9	80
81	Aicardi-GoutiÃ¨res Syndrome Displays Genetic Heterogeneity with One Locus (AGS1) on Chromosome 3p21. American Journal of Human Genetics, 2000, 67, 213-221.	6.2	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.	0.6	77
83	STING-Associated Vasculopathy with Onset in Infancy â€” A New Interferonopathy. New England Journal of Medicine, 2014, 371, 568-571.	27.0	77
84	A POT1 mutation implicates defective telomere end fill-in and telomere truncations in Coats plus. Genes and Development, 2016, 30, 812-826.	5.9	77
85	Bloom syndrome protein restrains innate immune sensing of micronuclei by cGAS. Journal of Experimental Medicine, 2019, 216, 1199-1213.	8.5	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.	2.8	74
87	Therapies in Aicardiâ€“GoutiÃ¨res syndrome. Clinical and Experimental Immunology, 2013, 175, 1-8.	2.6	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.	2.9	74
89	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
90	Severe childhood SMA and axonal CMT due to anticodon binding domain mutations in the GARS gene. Neurology, 2006, 67, 1710-1712.	1.1	72

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91	Aicardi-GoutiÃ¨res Syndrome: Neuroradiologic Findings and Follow-Up. American Journal of Neuroradiology, 2009, 30, 1971-1976.	2.4	72
92	Mutations in ADAR1, IFIH1, and RNASEH2B Presenting As Spastic Paraplegia. Neuropediatrics, 2014, 45, 386-391.	0.6	72
93	Spondyloenchondrodysplasia Due to Mutations in ACP5: A Comprehensive Survey. Journal of Clinical Immunology, 2016, 36, 220-234.	3.8	71
94	Treatments in Aicardiâ€“GoutiÃ¨res syndrome. Developmental Medicine and Child Neurology, 2020, 62, 42-47.	2.1	70
95	Aicardiâ€“GoutiÃ¨res syndrome. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1629-1635.	1.8	69
96	Recognizable phenotypes associated with intracranial calcification. Developmental Medicine and Child Neurology, 2013, 55, 46-57.	2.1	68
97	Reduced penetrance alleles for Huntington's disease: a multi-centre direct observational study. Journal of Medical Genetics, 2006, 44, e68-e68.	3.2	67
98	Delineation of Late Onset Hypoventilation Associated with Hypothalamic Dysfunction Syndrome. Pediatric Research, 2008, 64, 689-694.	2.3	63
99	Genetic and phenotypic spectrum associated with IFIH1 gainâ€ofâ€function. Human Mutation, 2020, 41, 837-849.	2.5	63
100	Gross rearrangements of the MECP2 gene are found in both classical and atypical Rett syndrome patients. Journal of Medical Genetics, 2005, 43, 451-456.	3.2	62
101	Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. Neuropediatrics, 2017, 48, 166-184.	0.6	62
102	An open-label trial of JAK 1/2 blockade in progressive IFIH1 -associated neuroinflammation. Neurology, 2018, 90, 289-291.	1.1	60
103	Mutations in MFSD8/CLN7 are a frequent cause of variant-late infantile neuronal ceroid lipofuscinosis. Human Mutation, 2009, 30, E530-E540.	2.5	59
104	Neuroradiologic patterns and novel imaging findings in Aicardi-GoutiÃ¨res syndrome. Neurology, 2016, 86, 28-35.	1.1	59
105	A child with severe juvenile dermatomyositis treated with ruxolitinib. Brain, 2018, 141, e80-e80.	7.6	58
106	Paediatric stroke: genetic insights into disease mechanisms and treatment targets. Lancet Neurology, The, 2011, 10, 264-274.	10.2	57
107	Genetic syndromes mimic congenital infections. Journal of Pediatrics, 2005, 146, 701-705.	1.8	54
108	Mosaic structural variation in children with developmental disorders. Human Molecular Genetics, 2015, 24, 2733-2745.	2.9	54

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109	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
110	Further delineation of the phenotype of severe congenital neutropenia type 4 due to mutations in G6PC3. European Journal of Human Genetics, 2011, 19, 18-22.	2.8	50
111	STING-Mediated Lung Inflammation and Beyond. Journal of Clinical Immunology, 2021, 41, 501-514.	3.8	48
112	Cutaneous histopathological findings of Aicardiâ€“GoutiÃ¨res syndrome, overlap with chilblain lupus. Journal of Cutaneous Pathology, 2008, 35, 774-778.	1.3	47
113	Tartrateâ€Resistant Acid Phosphatase Deficiency in the Predisposition to Systemic Lupus Erythematosus. Arthritis and Rheumatology, 2017, 69, 131-142.	5.6	47
114	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
115	Brownâ€“Vialettoâ€“Van Laere syndrome; variability in age at onset and disease progression highlighting the phenotypic overlap with Fazio-Londe disease. Brain and Development, 2005, 27, 443-446.	1.1	45
116	Variable Phenotype Including Leigh Syndrome with a 9185T>C Mutation in the MTATP6 Gene. Neuropediatrics, 2007, 38, 313-316.	0.6	45
117	Molecular screening of ADAMTSL2 gene in 33 patients reveals the genetic heterogeneity of geleophysic dysplasia. Journal of Medical Genetics, 2011, 48, 417-421.	3.2	45
118	Understanding the evolving phenotype of vascular complications in telomere biology disorders. Angiogenesis, 2019, 22, 95-102.	7.2	45
119	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
120	Enhanced cGAS-STINGâ€“dependent interferon signaling associated with mutations in ATAD3A. Journal of Experimental Medicine, 2021, 218, .	8.5	43
121	PSMB10, the last immunoproteasome gene missing for PRAAS. Journal of Allergy and Clinical Immunology, 2020, 145, 1015-1017.e6.	2.9	42
122	Leukoencephalopathy with Calcifications and Cysts: A Purely Neurological Disorder Distinct from Coats Plus. Neuropediatrics, 2014, 45, 175-182.	0.6	41
123	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
124	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
125	Expanding the clinical spectrum of SLC29A3 gene defects. European Journal of Medical Genetics, 2010, 53, 309-313.	1.3	38
126	COL4A1 Mutations Associated with a Characteristic Pattern of Intracranial Calcification. Neuropediatrics, 2011, 42, 227-233.	0.6	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. Lancet Rheumatology, The, 2020, 2, e99-e109.	3.9	38
128	DDX58 and Classic Singleton-Merten Syndrome. Journal of Clinical Immunology, 2019, 39, 75-80.	3.8	37
129	Diagnosing fetal alcohol syndrome: new insights from newer genetic technologies. Archives of Disease in Childhood, 2012, 97, 812-817.	1.9	36
130	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
131	Use of ruxolitinib in COPA syndrome manifesting as life-threatening alveolar haemorrhage. Thorax, 2020, 75, 92-95.	5.6	36
132	Aicardiâ€GoutiÃ¨res syndrome: description of a late onset case. Developmental Medicine and Child Neurology, 2008, 50, 631-634.	2.1	35
133	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
134	Aicardiâ€“GoutiÃ¨res syndrome harbours abundant systemic and brain-reactive autoantibodies. Annals of the Rheumatic Diseases, 2015, 74, 1931-1939.	0.9	35
135	A second locus for Aicardi-Goutieres syndrome at chromosome 13q14-21. Journal of Medical Genetics, 2005, 43, 444-450.	3.2	33
136	Early-Onset Aicardi-GoutiÃ¨res Syndrome. Journal of Child Neurology, 2015, 30, 1343-1348.	1.4	33
137	Chilblains as a Diagnostic Sign of Aicardi-GoutiÃ¨res Syndrome. Neuropediatrics, 2010, 41, 18-23.	0.6	32
138	Adult-Onset ANCA-Associated Vasculitis in SAVI: Extension of the Phenotypic Spectrum, Case Report and Review of the Literature. Frontiers in Immunology, 2020, 11, 575219.	4.8	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. Annals of the Rheumatic Diseases, 2022, 81, 601-613.	0.9	31
140	Congenital glaucoma and brain stem atrophy as features of Aicardi-GoutiÃ¨res syndrome. , 2004, 129A, 303-307.		30
141	Two further cases of spondyloenchondrodysplasia (SPENCD) with immune dysregulation. American Journal of Medical Genetics, Part A, 2008, 146A, 2810-2815.	1.2	30
142	Cerebral vasculopathy is a common feature in Aicardiâ€“GoutiÃ¨res syndrome associated with SAMHD1 mutations. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, E232; author reply E233.	7.1	29
143	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
144	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

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145	Striking intrafamilial phenotypic variability in Aicardiâ€“GoutiÃ¨res syndrome associated with the recurrent Asian founder mutation in RNASEH2C. American Journal of Medical Genetics, Part A, 2013, 161, 338-342.	1.2	28
146	A partial form of inherited human USP18 deficiency underlies infection and inflammation. Journal of Experimental Medicine, 2022, 219, .	8.5	28
147	Aicardiâ€“GoutiÃ¨res syndrome presenting atypically as a sub-acute leukoencephalopathy. European Journal of Paediatric Neurology, 2008, 12, 408-411.	1.6	27
148	Lupus: How much â€œcomplexityâ€•is really (just) genetic heterogeneity?. Arthritis and Rheumatism, 2011, 63, 3661-3664.	6.7	27
149	Anti-MDA5 juvenile idiopathic inflammatory myopathy: a specific subgroup defined by differentially enhanced interferon-Î± signalling. Rheumatology, 2020, 59, 1927-1937.	1.9	26
150	Treatment of Leukoencephalopathy With Calcifications and Cysts With Bevacizumab. Pediatric Neurology, 2017, 71, 56-59.	2.1	24
151	JAK 1/2 Blockade in MDA5 Gain-of-Function. Journal of Clinical Immunology, 2018, 38, 844-846.	3.8	24
152	JAK Inhibition in the Aicardiâ€“GoutiÃ¨res Syndrome. New England Journal of Medicine, 2020, 383, 2190-2193.	27.0	24
153	Elevation of proinflammatory cytokines in patients with Aicardi-GoutiÃ¨res syndrome. Neurology, 2013, 80, 997-1002.	1.1	23
154	The 2021 European Alliance of Associations for Rheumatology/American College of Rheumatology Points to Consider for Diagnosis and Management of Autoinflammatory Type I Interferonopathies: <scp>CANDLE</scp>/<scp>PRAAS</scp>, <scp>SAVI</scp>, and <scp>AGS</scp>. Arthritis and Rheumatology, 2022, 74, 735-751.	5.6	23
155	Blue (or purple) toes: Chilblains or chilblain lupus-like lesions are a manifestation of Aicardiâ€“GoutiÃ¨res syndrome and familial chilblain lupus. Journal of the American Academy of Dermatology, 2009, 61, 727-728.	1.2	22
156	Neuromyelitis optica in a child with Aicardi-GoutiÃ^res syndrome. Neurology, 2015, 85, 381-383.	1.1	22
157	JAK inhibition in STING-associated interferonopathy. Annals of the Rheumatic Diseases, 2016, 75, e75-e75.	0.9	22
158	Brief Report: Blockade of TANKâ€Binding Kinase 1/IKKÉ› Inhibits Mutant Stimulator of Interferon Genes (STING)â€“Mediated Inflammatory Responses in Human Peripheral Blood Mononuclear Cells. Arthritis and Rheumatology, 2017, 69, 1495-1501.	5.6	22
159	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
160	A Brief Historical Perspective on the Pathological Consequences of Excessive Type I Interferon Exposure In vivo. Journal of Clinical Immunology, 2018, 38, 694-698.	3.8	21
161	Type I interferonâ€“related kidney disorders. Kidney International, 2022, 101, 1142-1159.	5.2	21
162	Clinical phenotype associated with homozygosity forÂaÂHOXD13 7-residue polyalanine tract expansion. European Journal of Medical Genetics, 2006, 49, 396-401.	1.3	19

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163	COPA Syndrome as a Cause of Lupus Nephritis. Kidney International Reports, 2019, 4, 1187-1189.	0.8	19
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