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	Cerebrospinal fluid neopterin as a biomarker of treatment response to Janus kinase inhibition in Aicardiâ€“GoutiÃ¨res syndrome. Developmental Medicine and Child Neurology, 2022, 64, 266-271.	2.1	12
2	Autosomal dominant ADAR c.3019G>A (p.(G1007R)) variant is an important mimic of hereditary spastic paraplegia and cerebral palsy. Brain and Development, 2022, 44, 153-160.	1.1	3
3	The type I interferonopathies: 10 years on. Nature Reviews Immunology, 2022, 22, 471-483.	22.7	164
4	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
5	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
6	A partial form of inherited human USP18 deficiency underlies infection and inflammation. Journal of Experimental Medicine, 2022, 219, .	8.5	28
7	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
8	Type I interferonâ€“related kidney disorders. Kidney International, 2022, 101, 1142-1159.	5.2	21
9	DNASE1L3 deficiency, new phenotypes, and evidence for a transient type I IFN signaling. Journal of Clinical Immunology, 2022, 42, 1310-1320.	3.8	7
10	Delineating the epilepsy phenotype of NRROS-related microgliopathy: A case report and literature review. Seizure: the Journal of the British Epilepsy Association, 2022, 100, 15-20.	2.0	3
11	Cerebral Microangiopathy in Leukoencephalopathy With Cerebral Calcifications and Cysts: A Pathological Description. Journal of Child Neurology, 2021, 36, 133-140.	1.4	3
12	Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum. American Journal of Medical Genetics, Part A, 2021, 185, 15-25.	1.2	15
13	Rheumatoid factor positive polyarticular juvenile idiopathic arthritis associated with a novel COPA mutation. Rheumatology, 2021, 60, e171-e173.	1.9	6
14	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
15	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
16	LACC1 deficiency links juvenile arthritis with autophagy and metabolism in macrophages. Journal of Experimental Medicine, 2021, 218, .	8.5	17
17	STING-Mediated Lung Inflammation and Beyond. Journal of Clinical Immunology, 2021, 41, 501-514.	3.8	48
18	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

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19	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
20	Novel compound heterozygous STN1 variants are associated with Coats Plus syndrome. Molecular Genetics & Genomic Medicine, 2021, 9, e1708.	1.2	3
21	Opsoclonusâ€myoclonus in Aicardiâ€GoutiÃ¨res syndrome. Developmental Medicine and Child Neurology, 2021, 63, 1483-1486.	2.1	4
22	JAK inhibition in the type I interferonopathies. Journal of Allergy and Clinical Immunology, 2021, 148, 991-993.	2.9	19
23	Enhanced cGAS-STINGâ€“dependent interferon signaling associated with mutations in ATAD3A. Journal of Experimental Medicine, 2021, 218, .	8.5	43
24	Erythrocyte-derived mitochondria take to the lupus stage. Cell Metabolism, 2021, 33, 1723-1725.	16.2	3
25	Mitochondrial Nucleic Acid as a Driver of Pathogenic Type I Interferon Induction in Mendelian Disease. Frontiers in Immunology, 2021, 12, 729763.	4.8	2
26	Mitochondrial Nucleic Acid as a Driver of Pathogenic Type I Interferon Induction in Mendelian Disease. Frontiers in Immunology, 2021, 12, 729763.	4.8	18
27	Treatments in Aicardiâ€“GoutiÃ¨res syndrome. Developmental Medicine and Child Neurology, 2020, 62, 42-47.	2.1	70
28	Circulating Interferonâ€Î± Measured With a Highly Sensitive Assay as a Biomarker for Juvenile Inflammatory Myositis Activity: Comment on the Article by Mathian et al. Arthritis and Rheumatology, 2020, 72, 195-197.	5.6	15
29	Inhibition of IFNÎ± secretion in cells from patients with juvenile dermatomyositis under TBK1 inhibitor treatment revealed by single-molecular assay technology. Rheumatology, 2020, 59, 1171-1174.	1.9	5
30	Use of ruxolitinib in COPA syndrome manifesting as life-threatening alveolar haemorrhage. Thorax, 2020, 75, 92-95.	5.6	36
31	Neuromyelitis optica in patients with increased interferon alpha concentrations. Lancet Neurology, The, 2020, 19, 31-33.	10.2	14
32	Anti-MDA5 juvenile idiopathic inflammatory myopathy: a specific subgroup defined by differentially enhanced interferon-Î± signalling. Rheumatology, 2020, 59, 1927-1937.	1.9	26
33	PSMB10, the last immunoproteasome gene missing for PRAAS. Journal of Allergy and Clinical Immunology, 2020, 145, 1015-1017.e6.	2.9	42
34	Genetic and phenotypic spectrum associated with IFIH1 gainâ€ofâ€function. Human Mutation, 2020, 41, 837-849.	2.5	63
35	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
36	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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37	JAK Inhibition in the Aicardiâ€“GoutiÃ¨res Syndrome. New England Journal of Medicine, 2020, 383, 2190-2193.	27.0	24
38	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
39	Apparent Radiological Improvement in an Infant With Labrune Syndrome Treated With Bevacizumab. Pediatric Neurology, 2020, 112, 53-55.	2.1	7
40	Mutations in COPA lead to abnormal trafficking of STING to the Golgi and interferon signaling. Journal of Experimental Medicine, 2020, 217, .	8.5	130
41	Magnetic resonance imaging pattern recognition in childhood bilateral basal ganglia disorders. Brain Communications, 2020, 2, fcaa178.	3.3	17
42	Clinical Reasoning: A 25-year-old woman with recurrent episodes of collapse and loss of consciousness. Neurology, 2020, 94, 994-999.	1.1	2
43	Cardiac valve involvement in ADAR-related type I interferonopathy. Journal of Medical Genetics, 2020, 57, 475-478.	3.2	19
44	Catatonia in a patient with Aicardi-GoutiÃ¨res syndrome efficiently treated with immunoadsorption. Schizophrenia Research, 2020, 222, 484-486.	2.0	6
45	Mendelian disorders of immunity related to an upregulation of type I interferon. , 2020, , 751-772.		2
46	An Indian child with Coats plus syndrome due to mutations in <scp>STN1</scp>. American Journal of Medical Genetics, Part A, 2020, 182, 2139-2144.	1.2	9
47	Biallelic mutations in NRROS cause an early onset lethal microgliopathy. Acta Neuropathologica, 2020, 139, 947-951.	7.7	17
48	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
49	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
50	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
51	COPA Syndrome as a Cause of Lupus Nephritis. Kidney International Reports, 2019, 4, 1187-1189.	0.8	19
52	Leukoencephalopathy, Intracranial Calcifications, Cysts, and SNORD118 Mutation (Labrune Syndrome) with Obstructive Hydrocephalus. World Neurosurgery, 2019, 125, 271-272.	1.3	15
53	Bloom syndrome protein restrains innate immune sensing of micronuclei by cGAS. Journal of Experimental Medicine, 2019, 216, 1199-1213.	8.5	75
54	Severe type I interferonopathy and unrestrained interferon signaling due to a homozygous germline mutation in STAT2. Science Immunology, 2019, 4, .	11.9	80

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55	Understanding the evolving phenotype of vascular complications in telomere biology disorders. Angiogenesis, 2019, 22, 95-102.	7.2	45
56	DDX58 and Classic Singleton-Merten Syndrome. Journal of Clinical Immunology, 2019, 39, 75-80.	3.8	37
57	Self-Awareness: Nucleic Acidâ€“Driven Inflammation and the Type I Interferonopathies. Annual Review of Immunology, 2019, 37, 247-267.	21.8	111
58	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
59	Familial Blau syndrome:First molecularly confirmed report from India. Indian Journal of Ophthalmology, 2019, 67, 165.	1.1	6
60	An open-label trial of JAK 1/2 blockade in progressive IFIH1 -associated neuroinflammation. Neurology, 2018, 90, 289-291.	1.1	60
61	Sort Your Self Out!. Cell, 2018, 172, 640-642.	28.9	5
62	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	6.2	204
63	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
64	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
65	The 2017 IUIS Phenotypic Classification for Primary Immunodeficiencies. Journal of Clinical Immunology, 2018, 38, 129-143.	3.8	488
66	JAK 1/2 Blockade in MDA5 Gain-of-Function. Journal of Clinical Immunology, 2018, 38, 844-846.	3.8	24
67	Reverse-Transcriptase Inhibitors in the Aicardiâ€“GoutiÃ¨res Syndrome. New England Journal of Medicine, 2018, 379, 2275-2277.	27.0	106
68	A child with severe juvenile dermatomyositis treated with ruxolitinib. Brain, 2018, 141, e80-e80.	7.6	58
69	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
70	Autosomal-dominant early-onset spastic paraparesis with brain calcification due to IFIH1 gain-of-function. Human Mutation, 2018, 39, 1076-1080.	2.5	8
71	Mitochondrial double-stranded RNA triggers antiviral signalling in humans. Nature, 2018, 560, 238-242.	27.8	397
72	Development and Validation of an Ultrasensitive Single Molecule Array Digital Enzyme-linked Immunosorbent Assay for Human Interferon-α. Journal of Visualized Experiments, 2018, , .	0.3	8

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73	Life-threatening influenza pneumonitis in a child with inherited IRF9 deficiency. Journal of Experimental Medicine, 2018, 215, 2567-2585.	8.5	146
74	Taking the STING out of inflammation. Nature Reviews Rheumatology, 2018, 14, 508-509.	8.0	6
75	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
76	Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. Neuropediatrics, 2017, 48, 166-184.	0.6	62
77	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
78	Detection of interferon alpha protein reveals differential levels and cellular sources in disease. Journal of Experimental Medicine, 2017, 214, 1547-1555.	8.5	288
79	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
80	Polymorphisms in IFIH1: the good and the bad. Nature Immunology, 2017, 18, 708-709.	14.5	7
81	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
82	Treatment of Leukoencephalopathy With Calcifications and Cysts With Bevacizumab. Pediatric Neurology, 2017, 71, 56-59.	2.1	24
83	Familial and syndromic lupus share the same phenotype as other early-onset forms of lupus. Joint Bone Spine, 2017, 84, 589-593.	1.6	7
84	Assessment of Type I Interferon Signaling in Pediatric Inflammatory Disease. Journal of Clinical Immunology, 2017, 37, 123-132.	3.8	163
85	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
86	Leukoencephalopathy with calcification and cysts: A cerebral microangiopathy caused by mutations in SNORD118. Journal of the Neurological Sciences, 2017, 372, 443.	0.6	2
87	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
88	Tartrateâ€Resistant Acid Phosphatase Deficiency in the Predisposition to Systemic Lupus Erythematosus. Arthritis and Rheumatology, 2017, 69, 131-142.	5.6	47
89	Type I interferon-mediated autoinflammation due to DNase II deficiency. Nature Communications, 2017, 8, 2176.	12.8	164
90	JAK inhibition in STING-associated interferonopathy. Annals of the Rheumatic Diseases, 2016, 75, e75-e75.	0.9	22

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91	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
92	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature Genetics, 2016, 48, 1185-1192.	21.4	114
93	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
94	Type I interferonâ€“mediated monogenic autoinflammation: The type I interferonopathies, a conceptual overview. Journal of Experimental Medicine, 2016, 213, 2527-2538.	8.5	359
95	Human USP18 deficiency underlies type 1 interferonopathy leading to severe pseudo-TORCH syndrome. Journal of Experimental Medicine, 2016, 213, 1163-1174.	8.5	224
96	Spondyloenchondrodysplasia Due to Mutations in ACP5: A Comprehensive Survey. Journal of Clinical Immunology, 2016, 36, 220-234.	3.8	71
97	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
98	Neuroradiologic patterns and novel imaging findings in Aicardi-GoutiÃ¨res syndrome. Neurology, 2016, 86, 28-35.	1.1	59
99	Stimulator of Interferon Genesâ€“Associated Vasculopathy With Onset in Infancy. JAMA Dermatology, 2015, 151, 872.	4.1	108
100	Aicardiâ€“GoutiÃ¨res syndrome and the type I interferonopathies. Nature Reviews Immunology, 2015, 15, 429-440.	22.7	705
101	Early-Onset Aicardi-GoutiÃ¨res Syndrome. Journal of Child Neurology, 2015, 30, 1343-1348.	1.4	33
102	A Specific IFIH1 Gain-of-Function Mutation Causes Singleton-Merten Syndrome. American Journal of Human Genetics, 2015, 96, 275-282.	6.2	188
103	Mosaic structural variation in children with developmental disorders. Human Molecular Genetics, 2015, 24, 2733-2745.	2.9	54
104	Aicardiâ€“GoutiÃ¨res syndrome harbours abundant systemic and brain-reactive autoantibodies. Annals of the Rheumatic Diseases, 2015, 74, 1931-1939.	0.9	35
105	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
106	Update and Mutational Analysis ofSLC20A2: A Major Cause of Primary Familial Brain Calcification. Human Mutation, 2015, 36, 489-495.	2.5	80
107	The eukaryotic elongation factor eEF1A1 interacts with SAMHD1. Biochemical Journal, 2015, 466, 69-76.	3.7	14
108	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

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109	Human Disease Phenotypes Associated With Mutations in TREX1. Journal of Clinical Immunology, 2015, 35, 235-243.	3.8	154
110	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
111	Neuromyelitis optica in a child with Aicardi-GoutiÃˆres syndrome. Neurology, 2015, 85, 381-383.	1.1	22
112	Mosaic Tetrasomy 9p: A Mendelian Condition Associated With Pediatric-Onset Overlap Myositis. Pediatrics, 2015, 136, e544-e547.	2.1	10
113	Novel monogenic diseases causing human autoimmunity. Current Opinion in Immunology, 2015, 37, 1-5.	5.5	18
114	cGMP-AMP synthase paves the way to autoimmunity. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 12903-12904.	7.1	6
115	Large-scale discovery of novel genetic causes of developmental disorders. Nature, 2015, 519, 223-228.	27.8	998
116	Human intracellular ISG15 prevents interferon-Î±/Î² over-amplification and auto-inflammation. Nature, 2015, 517, 89-93.	27.8	432
117	Type I interferonopathies: Mendelian type I interferon up-regulation. Current Opinion in Immunology, 2015, 32, 7-12.	5.5	160
118	ADAR1 Facilitates HIV-1 Replication in Primary CD4+ T Cells. PLoS ONE, 2015, 10, e0143613.	2.5	16
119	Inherited STING-activating mutation underlies a familial inflammatory syndrome with lupus-like manifestations. Journal of Clinical Investigation, 2014, 124, 5516-5520.	8.2	435
120	STING-Associated Vasculopathy with Onset in Infancy â€” A New Interferonopathy. New England Journal of Medicine, 2014, 371, 568-571.	27.0	77
121	Leukoencephalopathy with Calcifications and Cysts: A Purely Neurological Disorder Distinct from Coats Plus. Neuropediatrics, 2014, 45, 175-182.	0.6	41
122	Mutations in ADAR1, IFIH1, and RNASEH2B Presenting As Spastic Paraplegia. Neuropediatrics, 2014, 45, 386-391.	0.6	72
123	Basal Ganglia Calcification in a Patient With Beta-Propeller Protein-Associated Neurodegeneration. Pediatric Neurology, 2014, 51, 843-845.	2.1	17
124	Mutations in CECR1 associated with a neutrophil signature in peripheral blood. Pediatric Rheumatology, 2014, 12, 44.	2.1	88
125	PRKDC mutations associated with immunodeficiency, granuloma and aire-dependent autoimmunity. Pediatric Rheumatology, 2014, 12, .	2.1	1
126	A type I interferon signature identifies bilateral striatal necrosis due to mutations inADAR1. Journal of Medical Genetics, 2014, 51, 76-82.	3.2	118

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127	Reply. Arthritis and Rheumatology, 2014, 66, 229-230.	5.6	0
128	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
129	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
130	Intracranial calcification in childhood: a review of aetiologies and recognizable phenotypes. Developmental Medicine and Child Neurology, 2014, 56, 612-626.	2.1	132
131	The SKIV2L RNA exosome limits activation of the RIG-I-like receptors. Nature Immunology, 2014, 15, 839-845.	14.5	170
132	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
133	SAMHD1 is mutated recurrently in chronic lymphocytic leukemia and is involved in response to DNA damage. Blood, 2014, 123, 1021-1031.	1.4	205
134	Mendelian Disorders of Immunity Related to an Upregulation of Type I Interferon. , 2014, , 591-602.		0
135	SAMHD1-dependent retroviral control and escape in mice. EMBO Journal, 2013, 32, 2454-2462.	7.8	141
136	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
137	Exudative retinopathy, cerebral calcifications, duodenal atresia, preaxial polydactyly, micropenis, microcephaly and short stature: A new syndrome?. American Journal of Medical Genetics, Part A, 2013, 161, 1829-1832.	1.2	2
138	Recognizable phenotypes associated with intracranial calcification. Developmental Medicine and Child Neurology, 2013, 55, 46-57.	2.1	68
139	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
140	Elevation of proinflammatory cytokines in patients with Aicardi-GoutiÃ¨res syndrome. Neurology, 2013, 80, 997-1002.	1.1	23
141	Aicardiâ€“GoutiÃ¨res syndrome. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1629-1635.	1.8	69
142	Protein Kinase CÎ´ Deficiency Causes Mendelian Systemic Lupus Erythematosus With B Cellâ€Ðefective Apoptosis and Hyperproliferation. Arthritis and Rheumatism, 2013, 65, 2161-2171.	6.7	155
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	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

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145	Therapies in Aicardiâ€“GoutiÃ¨res syndrome. Clinical and Experimental Immunology, 2013, 175, 1-8.	2.6	74
146	Diagnosing fetal alcohol syndrome: new insights from newer genetic technologies. Archives of Disease in Childhood, 2012, 97, 812-817.	1.9	36
147	Intracranial calcification in early infantile Krabbe disease: nothing new under the sun. Developmental Medicine and Child Neurology, 2012, 54, 376-379.	2.1	14
148	SAMHD1 restricts HIV-1 reverse transcription in quiescent CD4+T-cells. Retrovirology, 2012, 9, 87.	2.0	302
149	Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. Nature Genetics, 2012, 44, 338-342.	21.4	234
150	Mutations in ADAR1 cause Aicardi-GoutiÃ¨res syndrome associated with a type I interferon signature. Nature Genetics, 2012, 44, 1243-1248.	21.4	712
151	Autoimmunity. Current Opinion in Immunology, 2012, 24, 649-650.	5.5	1
152	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
153	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
154	SAMHD1, A Putative Tumour Suppressor, Is Recurrently Mutated in Chronic Lymphocytic Leukaemia, and Is Associated with Poor Risk Features. Blood, 2012, 120, 713-713.	1.4	0
155	Degos Disease. American Journal of Clinical Pathology, 2011, 135, 599-610.	0.7	91
156	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
157	HIV-1 restriction factor SAMHD1 is a deoxynucleoside triphosphate triphosphohydrolase. Nature, 2011, 480, 379-382.	27.8	707
158	Sequencing revolution. Developmental Medicine and Child Neurology, 2011, 53, 673-674.	2.1	0
159	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
160	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
161	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
162	Paediatric stroke: genetic insights into disease mechanisms and treatment targets. Lancet Neurology, The, 2011, 10, 264-274.	10.2	57

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163	Infantile neurological Degos disease. European Journal of Paediatric Neurology, 2011, 15, 167-170.	1.6	9
164	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
165	Phenotypic variation in familial chilblain lupus (FCL) and Aicardi-GoutiÃ¨res syndrome (AGS) associated with TREX1 mutation in 4 family members. Pediatric Rheumatology, 2011, 9, .	2.1	0
166	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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