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List of Publications by Year in descending order

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242 papers	25,641 citations	7069 78 h-index	7718 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.	13.7	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.	9.4	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.	2.0	732
4	Mutations in ADAR1 cause Aicardi-GoutiÃ¨res syndrome associated with a type I interferon signature. Nature Genetics, 2012, 44, 1243-1248.	9.4	712
5	HIV-1 restriction factor SAMHD1 is a deoxynucleoside triphosphate triphosphohydrolase. Nature, 2011, 480, 379-382.	13.7	707
6	Aicardiâ€“GoutiÃ¨res syndrome and the type I interferonopathies. Nature Reviews Immunology, 2015, 15, 429-440.	10.6	705
7	Mutations involved in Aicardi-GoutiÃ¨res syndrome implicate SAMHD1 as regulator of the innate immune response. Nature Genetics, 2009, 41, 829-832.	9.4	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.	9.4	592
9	ASPM is a major determinant of cerebral cortical size. Nature Genetics, 2002, 32, 316-320.	9.4	538
10	The spectrum of SCN1A-related infantile epileptic encephalopathies. Brain, 2007, 130, 843-852.	3.7	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.	2.6	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.	9.4	490
13	The 2017 IUIS Phenotypic Classification for Primary Immunodeficiencies. Journal of Clinical Immunology, 2018, 38, 129-143.	2.0	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.	4.9	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.	0.7	447
16	Inherited STING-activating mutation underlies a familial inflammatory syndrome with lupus-like manifestations. Journal of Clinical Investigation, 2014, 124, 5516-5520.	3.9	435
17	Human intracellular ISG15 prevents interferon-Î±/Î² over-amplification and auto-inflammation. Nature, 2015, 517, 89-93.	13.7	432
18	Mitochondrial double-stranded RNA triggers antiviral signalling in humans. Nature, 2018, 560, 238-242.	13.7	397

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19	Clinical and Molecular Phenotype of Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 81, 713-725.	2.6	375
20	Mutations in the Pericentrin (PCNT) Gene Cause Primordial Dwarfism. Science, 2008, 319, 816-819.	6.0	370
21	Type I interferonâ€“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-GoutiÃ¨res 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 PIK3CAâ€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-Î² 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. American Journal of Human Genetics, 2018, 102, 175-187.	2.6	204
38	A Specific IFIH1 Gain-of-Function Mutation Causes Singleton-Merten Syndrome. American Journal of Human Genetics, 2015, 96, 275-282.	2.6	188
39	Preexisting autoantibodies to type I IFNs underlie critical COVID-19 pneumonia in patients with APS-1. Journal of Experimental Medicine, 2021, 218, .	4.2	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.	2.6	171
41	The SKIV2L RNA exosome limits activation of the RIG-I-like receptors. Nature Immunology, 2014, 15, 839-845.	7.0	170
42	Type I interferon-mediated autoinflammation due to DNase II deficiency. Nature Communications, 2017, 8, 2176.	5.8	164
43	The type I interferonopathies: 10 years on. Nature Reviews Immunology, 2022, 22, 471-483.	10.6	164
44	Assessment of Type I Interferon Signaling in Pediatric Inflammatory Disease. Journal of Clinical Immunology, 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. American Journal of Human Genetics, 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. Science Immunology, 2021, 6, .	5.6	161
47	Type I interferonopathies: Mendelian type I interferon up-regulation. Current Opinion in Immunology, 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. Journal of Allergy and Clinical Immunology, 2017, 140, 543-552.e5.	1.5	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.	2.0	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.	2.6	151
52	Life-threatening influenza pneumonitis in a child with inherited IRF9 deficiency. Journal of Experimental Medicine, 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. European Journal of Human Genetics, 2012, 20, 381-388.	1.4	142
54	SAMHD1-dependent retroviral control and escape in mice. EMBO Journal, 2013, 32, 2454-2462.	3.5	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.	1.1	132
56	Mutations in COPA lead to abnormal trafficking of STING to the Golgi and interferon signaling. Journal of Experimental Medicine, 2020, 217, .	4.2	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.	0.3	127
58	Aicardiâ€GoutiÃ¨res syndrome: an important Mendelian mimic of congenital infection. Developmental Medicine and Child Neurology, 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. American Journal of Human Genetics, 2010, 87, 354-364.	2.6	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.	1.4	122
61	Clinical and Mutational Spectrum of Mowatâ€“Wilson Syndrome. European Journal of Medical Genetics, 2005, 48, 97-111.	0.7	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.	1.1	121
63	A type I interferon signature identifies bilateral striatal necrosis due to mutations inADAR1. Journal of Medical Genetics, 2014, 51, 76-82.	1.5	118
64	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature Genetics, 2016, 48, 1185-1192.	9.4	114
65	Atypical Progeroid Syndrome due to Heterozygous Missense LMNA Mutations. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 4971-4983.	1.8	113
66	Self-Awareness: Nucleic Acidâ€“Driven Inflammation and the Type I Interferonopathies. Annual Review of Immunology, 2019, 37, 247-267.	9.5	111
67	Stimulator of Interferon Genesâ€“Associated Vasculopathy With Onset in Infancy. JAMA Dermatology, 2015, 151, 872.	2.0	108
68	Reverse-Transcriptase Inhibitors in the Aicardiâ€“GoutiÃ¨res Syndrome. New England Journal of Medicine, 2018, 379, 2275-2277.	13.9	106
69	cGAS-mediated induction of type I interferon due to inborn errors of histone pre-mRNA processing. Nature Genetics, 2020, 52, 1364-1372.	9.4	105
70	A Fifth Locus for Primary Autosomal Recessive Microcephaly Maps to Chromosome 1q31. American Journal of Human Genetics, 2000, 67, 1578-1580.	2.6	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.	2.0	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.	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 Aicardiâ€“GoutiÃ¨res 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 regulatorâ€“dependent autoimmunity. Journal of Allergy and Clinical Immunology, 2015, 135, 1578-1588.e5.	1.5	84
79	Update and Mutational Analysis ofSLC20A2: 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 STAT2. Science Immunology, 2019, 4, .	5.6	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.	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 â€” 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 Aicardiâ€“GoutiÃ¨res 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 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.	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-GoutiÃ¨res Syndrome: Neuroradiologic Findings and Follow-Up. American Journal of Neuroradiology, 2009, 30, 1971-1976.	1.2	72
92	Mutations in ADAR1, IFIH1, and RNASEH2B Presenting As Spastic Paraplegia. Neuropediatrics, 2014, 45, 386-391.	0.3	72
93	Spondyloenchondrodysplasia Due to Mutations in ACP5: A Comprehensive Survey. Journal of Clinical Immunology, 2016, 36, 220-234.	2.0	71
94	Treatments in Aicardiâ€“GoutiÃ¨res syndrome. Developmental Medicine and Child Neurology, 2020, 62, 42-47.	1.1	70
95	Aicardiâ€“GoutiÃ¨res syndrome. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1629-1635.	1.0	69
96	Recognizable phenotypes associated with intracranial calcification. Developmental Medicine and Child Neurology, 2013, 55, 46-57.	1.1	68
97	Reduced penetrance alleles for Huntington's disease: a multi-centre direct observational study. Journal of Medical Genetics, 2006, 44, e68-e68.	1.5	67
98	Delineation of Late Onset Hypoventilation Associated with Hypothalamic Dysfunction Syndrome. Pediatric Research, 2008, 64, 689-694.	1.1	63
99	Genetic and phenotypic spectrum associated with IFIH1 gainâ€ofâ€function. Human Mutation, 2020, 41, 837-849.	1.1	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.	1.5	62
101	Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. Neuropediatrics, 2017, 48, 166-184.	0.3	62
102	An open-label trial of JAK 1/2 blockade in progressive IFIH1 -associated neuroinflammation. Neurology, 2018, 90, 289-291.	1.5	60
103	Mutations in MFSD8/CLN7 are a frequent cause of variant-late infantile neuronal ceroid lipofuscinosis. Human Mutation, 2009, 30, E530-E540.	1.1	59
104	Neuroradiologic patterns and novel imaging findings in Aicardi-GoutiÃ¨res syndrome. Neurology, 2016, 86, 28-35.	1.5	59
105	A child with severe juvenile dermatomyositis treated with ruxolitinib. Brain, 2018, 141, e80-e80.	3.7	58
106	Paediatric stroke: genetic insights into disease mechanisms and treatment targets. Lancet Neurology, The, 2011, 10, 264-274.	4.9	57
107	Genetic syndromes mimic congenital infections. Journal of Pediatrics, 2005, 146, 701-705.	0.9	54
108	Mosaic structural variation in children with developmental disorders. Human Molecular Genetics, 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. Annals of the Rheumatic Diseases, 2019, 78, 431-433.	0.5	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.	1.4	50
111	STING-Mediated Lung Inflammation and Beyond. Journal of Clinical Immunology, 2021, 41, 501-514.	2.0	48
112	Cutaneous histopathological findings of Aicardiâ€“GoutiÃ¨res syndrome, overlap with chilblain lupus. Journal of Cutaneous Pathology, 2008, 35, 774-778.	0.7	47
113	Tartrateâ€Resistant Acid Phosphatase Deficiency in the Predisposition to Systemic Lupus Erythematosus. Arthritis and Rheumatology, 2017, 69, 131-142.	2.9	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.	0.7	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.	0.6	45
116	Variable Phenotype Including Leigh Syndrome with a 9185T>C Mutation in the MTATP6 Gene. Neuropediatrics, 2007, 38, 313-316.	0.3	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.	1.5	45
118	Understanding the evolving phenotype of vascular complications in telomere biology disorders. Angiogenesis, 2019, 22, 95-102.	3.7	45
119	Musculoskeletal Disease in MDA5â€Related Type I Interferonopathy: A Mendelian Mimic of Jaccoud's Arthropathy. Arthritis and Rheumatology, 2017, 69, 2081-2091.	2.9	44
120	Enhanced cGAS-STINGâ€“dependent interferon signaling associated with mutations in ATAD3A. Journal of Experimental Medicine, 2021, 218, .	4.2	43
121	PSMB10, the last immunoproteasome gene missing for PRAAS. Journal of Allergy and Clinical Immunology, 2020, 145, 1015-1017.e6.	1.5	42
122	Leukoencephalopathy with Calcifications and Cysts: A Purely Neurological Disorder Distinct from Coats Plus. Neuropediatrics, 2014, 45, 175-182.	0.3	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.	1.5	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.	0.7	38
125	Expanding the clinical spectrum of SLC29A3 gene defects. European Journal of Medical Genetics, 2010, 53, 309-313.	0.7	38
126	COL4A1 Mutations Associated with a Characteristic Pattern of Intracranial Calcification. Neuropediatrics, 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. Lancet Rheumatology, The, 2020, 2, e99-e109.	2.2	38
128	DDX58 and Classic Singleton-Merten Syndrome. Journal of Clinical Immunology, 2019, 39, 75-80.	2.0	37
129	Diagnosing fetal alcohol syndrome: new insights from newer genetic technologies. Archives of Disease in Childhood, 2012, 97, 812-817.	1.0	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.	0.4	36
131	Use of ruxolitinib in COPA syndrome manifesting as life-threatening alveolar haemorrhage. Thorax, 2020, 75, 92-95.	2.7	36
132	Aicardiâ€GoutiÃ¨res syndrome: description of a late onset case. Developmental Medicine and Child Neurology, 2008, 50, 631-634.	1.1	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.	0.7	35
134	Aicardiâ€“GoutiÃ¨res syndrome harbours abundant systemic and brain-reactive autoantibodies. Annals of the Rheumatic Diseases, 2015, 74, 1931-1939.	0.5	35
135	A second locus for Aicardi-Goutieres syndrome at chromosome 13q14-21. Journal of Medical Genetics, 2005, 43, 444-450.	1.5	33
136	Early-Onset Aicardi-GoutiÃ¨res Syndrome. Journal of Child Neurology, 2015, 30, 1343-1348.	0.7	33
137	Chilblains as a Diagnostic Sign of Aicardi-GoutiÃ¨res Syndrome. Neuropediatrics, 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. Frontiers in Immunology, 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. Annals of the Rheumatic Diseases, 2022, 81, 601-613.	0.5	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.	0.7	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.	3.3	29
143	Systemic lupus erythematosus due to C1q deficiency with progressive encephalopathy, intracranial calcification and acquired moyamoya cerebral vasculopathy. Lupus, 2013, 22, 639-643.	0.8	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.	0.7	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.	0.7	28
146	A partial form of inherited human USP18 deficiency underlies infection and inflammation. Journal of Experimental Medicine, 2022, 219, .	4.2	28
147	Aicardiâ€“GoutiÃ¨res syndrome presenting atypically as a sub-acute leukoencephalopathy. European Journal of Paediatric Neurology, 2008, 12, 408-411.	0.7	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.	0.9	26
150	Treatment of Leukoencephalopathy With Calcifications and Cysts With Bevacizumab. Pediatric Neurology, 2017, 71, 56-59.	1.0	24
151	JAK 1/2 Blockade in MDA5 Gain-of-Function. Journal of Clinical Immunology, 2018, 38, 844-846.	2.0	24
152	JAK Inhibition in the Aicardiâ€“GoutiÃ¨res Syndrome. New England Journal of Medicine, 2020, 383, 2190-2193.	13.9	24
153	Elevation of proinflammatory cytokines in patients with Aicardi-GoutiÃ¨res syndrome. Neurology, 2013, 80, 997-1002.	1.5	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.	2.9	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.	0.6	22
156	Neuromyelitis optica in a child with Aicardi-GoutiÃ^res syndrome. Neurology, 2015, 85, 381-383.	1.5	22
157	JAK inhibition in STING-associated interferonopathy. Annals of the Rheumatic Diseases, 2016, 75, e75-e75.	0.5	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.	2.9	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.	0.5	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.	2.0	21
161	Type I interferonâ€“related kidney disorders. Kidney International, 2022, 101, 1142-1159.	2.6	21
162	Clinical phenotype associated with homozygosity forÂaÂHOXD13 7-residue polyalanine tract expansion. European Journal of Medical Genetics, 2006, 49, 396-401.	0.7	19

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163	COPA Syndrome as a Cause of Lupus Nephritis. Kidney International Reports, 2019, 4, 1187-1189.	0.4	19
164	Cardiac valve involvement in ADAR-related type I interferonopathy. Journal of Medical Genetics, 2020, 57, 475-478.	1.5	19
165	JAK inhibition in the type I interferonopathies. Journal of Allergy and Clinical Immunology, 2021, 148, 991-993.	1.5	19
166	Natural history of cardiac involvement in geleophysic dysplasia. , 2005, 132A, 320-323.		18
167	Chromosome 1q42 deletion and agenesis of the corpus callosum. American Journal of Medical Genetics, Part A, 2005, 138A, 68-69.	0.7	18
168	Neurological presentation of Griscelli syndrome: Obstructive hydrocephalus without haematological abnormalities or organomegaly. Brain and Development, 2007, 29, 247-250.	0.6	18
169	Novel monogenic diseases causing human autoimmunity. Current Opinion in Immunology, 2015, 37, 1-5.	2.4	18
170	Mitochondrial Nucleic Acid as a Driver of Pathogenic Type I Interferon Induction in Mendelian Disease. Frontiers in Immunology, 2021, 12, 729763.	2.2	18
171	Basal Ganglia Calcification in a Patient With Beta-Propeller Protein-Associated Neurodegeneration. Pediatric Neurology, 2014, 51, 843-845.	1.0	17
172	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.	2.6	17
173	Magnetic resonance imaging pattern recognition in childhood bilateral basal ganglia disorders. Brain Communications, 2020, 2, fcaa178.	1.5	17
174	Biallelic mutations in NRROS cause an early onset lethal microgliopathy. Acta Neuropathologica, 2020, 139, 947-951.	3.9	17
175	LACC1 deficiency links juvenile arthritis with autophagy and metabolism in macrophages. Journal of Experimental Medicine, 2021, 218, .	4.2	17
176	Spondylocostal dysostosis associated with a 46,XX,+15,dic(6;15)(q25;qll.2) translocation. Clinical Dysmorphology, 1997, 6, 347-350.	0.1	16
177	The neonatal form of Aicardi-GoutiÃ¨res syndrome masquerading as congenital infection. Early Human Development, 2008, 84, 783-785.	0.8	16
178	Synonymous Mutations inRNASEH2ACreate Cryptic Splice Sites Impairing RNase H2 Enzyme Function in Aicardi-GoutiÃ¨res Syndrome. Human Mutation, 2013, 34, 1066-1070.	1.1	16
179	Differential Expression of Interferon-Alpha Protein Provides Clues to Tissue Specificity Across Type I Interferonopathies. Journal of Clinical Immunology, 2021, 41, 603-609.	2.0	16
180	ADAR1 Facilitates HIV-1 Replication in Primary CD4+ T Cells. PLoS ONE, 2015, 10, e0143613.	1.1	16

#	Article	IF	Citations
181	Leukoencephalopathy, Intracranial Calcifications, Cysts, and SNORD118 Mutation (Labrune Syndrome) with Obstructive Hydrocephalus. World Neurosurgery, 2019, 125, 271-272.	0.7	15
182	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.	2.9	15
183	Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum. American Journal of Medical Genetics, Part A, 2021, 185, 15-25.	0.7	15
184	Intracranial calcification in early infantile Krabbe disease: nothing new under the sun. Developmental Medicine and Child Neurology, 2012, 54, 376-379.	1.1	14
185	The eukaryotic elongation factor eEF1A1 interacts with SAMHD1. Biochemical Journal, 2015, 466, 69-76.	1.7	14
186	Neuromyelitis optica in patients with increased interferon alpha concentrations. Lancet Neurology, The, 2020, 19, 31-33.	4.9	14
187	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.	0.5	12
188	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.	1.1	12
189	Elevated Interferon-Alpha in Fetal Blood in the Prenatal Diagnosis of Aicardi-GoutiÃ¨res Syndrome. Fetal Diagnosis and Therapy, 2006, 21, 153-155.	0.6	11
190	Treatment of Gastrointestinal Bleeding in a Probable Case of Cerebroretinal Microangiopathy with Calcifications and Cysts. Molecular Syndromology, 2010, 1, 159-162.	0.3	11
191	The Molecular Basis of GROD-Storing Neuronal Ceroid Lipofuscinoses in Scotland. Molecular Genetics and Metabolism, 1999, 66, 245-247.	0.5	10
192	Mental retardation, keratoconus, febrile seizures and sinoatrial block: a previously undescribed autosomal recessive disorder. Clinical Genetics, 2005, 67, 448-449.	1.0	10
193	Mosaic Tetrasomy 9p: A Mendelian Condition Associated With Pediatric-Onset Overlap Myositis. Pediatrics, 2015, 136, e544-e547.	1.0	10
194	Infantile neurological Degos disease. European Journal of Paediatric Neurology, 2011, 15, 167-170.	0.7	9
195	Comprehensive molecular screening strategy of <scp>OCLN</scp> in bandâ€like calcification with simplified gyration and polymicrogyria. Clinical Genetics, 2018, 93, 228-234.	1.0	9
196	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.	0.7	9
197	Autosomal-dominant early-onset spastic paraparesis with brain calcification due to IFIH1 gain-of-function. Human Mutation, 2018, 39, 1076-1080.	1.1	8
198	Development and Validation of an Ultrasensitive Single Molecule Array Digital Enzyme-linked Immunosorbent Assay for Human Interferon-α. Journal of Visualized Experiments, 2018, , .	0.2	8

#	Article	IF	Citations
199	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.	2.0	8
200	Polymorphisms in IFIH1: the good and the bad. Nature Immunology, 2017, 18, 708-709.	7.0	7
201	Familial and syndromic lupus share the same phenotype as other early-onset forms of lupus. Joint Bone Spine, 2017, 84, 589-593.	0.8	7
202	Apparent Radiological Improvement in an Infant With Labrune Syndrome Treated With Bevacizumab. Pediatric Neurology, 2020, 112, 53-55.	1.0	7
203	DNASE1L3 deficiency, new phenotypes, and evidence for a transient type I IFN signaling. Journal of Clinical Immunology, 2022, 42, 1310-1320.	2.0	7
204	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.	3.3	6
205	Taking the STING out of inflammation. Nature Reviews Rheumatology, 2018, 14, 508-509.	3.5	6
206	Catatonia in a patient with Aicardi-GoutiÃ¨res syndrome efficiently treated with immunoadsorption. Schizophrenia Research, 2020, 222, 484-486.	1.1	6
207	Rheumatoid factor positive polyarticular juvenile idiopathic arthritis associated with a novel COPA mutation. Rheumatology, 2021, 60, e171-e173.	0.9	6
208	Familial Blau syndrome:First molecularly confirmed report from India. Indian Journal of Ophthalmology, 2019, 67, 165.	0.5	6
209	Maternal serum alpha-fetoprotein levels in congenital nephrosis. , 1997, 17, 1089-1089.		5
210	The genetics of Aicardi-GoutiÃ¨res syndrome. European Journal of Paediatric Neurology, 2002, 6, A33-A35.	0.7	5
211	Sedaghatian spondylometaphyseal dysplasia with pachygyria and absence of the corpus callosum. American Journal of Medical Genetics, Part A, 2006, 140A, 1854-1858.	0.7	5
212	3C syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 1026-1027.	0.7	5
213	Elevated pterins in cerebral spinal fluidâ€“ biochemical marker of Aicardiâ€“GoutiÃ¨res syndrome. Developmental Medicine and Child Neurology, 2009, 51, 841-842.	1.1	5
214	Sort Your Self Out!. Cell, 2018, 172, 640-642.	13.5	5
215	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.	0.9	5
216	The story of DNase II: A stifled deathâ€wish leads to selfâ€harm. European Journal of Immunology, 2010, 40, 2376-2378.	1.6	4

#	Article	IF	Citations
217	New subtype of familial intracranial calcification in a mother and two children. American Journal of Medical Genetics, Part A, 2010, 152A, 943-946.	0.7	4
218	Opsoclonusâ€myoclonus in Aicardiâ€GoutiÃ¨res syndrome. Developmental Medicine and Child Neurology, 2021, 63, 1483-1486.	1.1	4
219	Cerebral Microangiopathy in Leukoencephalopathy With Cerebral Calcifications and Cysts: A Pathological Description. Journal of Child Neurology, 2021, 36, 133-140.	0.7	3
220	Novel compound heterozygous STN1 variants are associated with Coats Plus syndrome. Molecular Genetics & Genomic Medicine, 2021, 9, e1708.	0.6	3
221	Erythrocyte-derived mitochondria take to the lupus stage. Cell Metabolism, 2021, 33, 1723-1725.	7.2	3
222	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.	0.6	3
223	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.	0.9	3
224	Focal dermal hypoplasia with subependymal heterotopia and hypoplastic corpus callosum. Clinical Dysmorphology, 2007, 16, 59-61.	0.1	2
225	Aicardiâ€“GoutiÃ¨res syndrome presenting with haematemesis in infancy. Acta Paediatrica, International Journal of Paediatrics, 2009, 98, 2005-2008.	0.7	2
226	Newly recognized recessive syndrome characterized by dysmorphic features, hypogonadotropic hypogonadism, severe microcephaly, and sensorineural hearing loss maps to 3p21.3. American Journal of Medical Genetics, Part A, 2011, 155, 2910-2915.	0.7	2
227	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.	0.7	2
228	Leukoencephalopathy with calcification and cysts: A cerebral microangiopathy caused by mutations in SNORD118. Journal of the Neurological Sciences, 2017, 372, 443.	0.3	2
229	Clinical Reasoning: A 25-year-old woman with recurrent episodes of collapse and loss of consciousness. Neurology, 2020, 94, 994-999.	1.5	2
230	Mendelian disorders of immunity related to an upregulation of type I interferon. , 2020, , 751-772.		2
231	Mitochondrial Nucleic Acid as a Driver of Pathogenic Type I Interferon Induction in Mendelian Disease. Frontiers in Immunology, 2021, 12, 729763.	2.2	2
232	A newly recognized, likely autosomal recessive syndrome comprising agammaglobulinemia, microcephaly, craniosynostosis, severe dermatitis, and other features. American Journal of Medical Genetics, Part A, 2006, 140A, 1131-1135.	0.7	1
233	Autoimmunity. Current Opinion in Immunology, 2012, 24, 649-650.	2.4	1
234	PRKDC mutations associated with immunodeficiency, granuloma and aire-dependent autoimmunity. Pediatric Rheumatology, 2014, 12, .	0.9	1

#	Article	IF	Citations
235	Congenital palmar polyonychia with postaxial limb defects may be the same as the ulnar-mammary syndrome. American Journal of Medical Genetics, Part A, 2005, 137A, 233-233.	0.7	0
236	Sequencing revolution. Developmental Medicine and Child Neurology, 2011, 53, 673-674.	1.1	0
237	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, .	0.9	0
238	Severe neonatalâ€onset panniculitis in a female infant with Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 3087-3089.	0.7	0
239	Reply. Arthritis and Rheumatology, 2014, 66, 229-230.	2.9	0
240	A child with severe juvenile dermatomyositis treated with ruxolitinib. Journal of Financial Econometrics, 0, , .	0.8	0
241	SAMHD1, A Putative Tumour Suppressor, Is Recurrently Mutated in Chronic Lymphocytic Leukaemia, and Is Associated with Poor Risk Features. Blood, 2012, 120, 713-713.	0.6	0
242	Mendelian Disorders of Immunity Related to an Upregulation of Type I Interferon. , 2014, , 591-602.		0