Yanick J Crow

List of Publications by Year in Descending Order

231 papers	18,838 citations	70 h-index	134 g-index
248 ext. papers	22,897 ext. citations	9.7 avg, IF	6.93 L-index

#	Paper	IF	Citations
231	The 2021 EULAR and ACR points to consider for diagnosis and management of autoinflammatory type I interferonopathies: CANDLE/PRAAS, SAVI and AGS.. *Annals of the Rheumatic Diseases*, **2022**,	2.4	0
230	Mutations in RNU7-1 Weaken Secondary RNA Structure, Induce MCP-1 and CXCL10 in CSF, and Result in Aicardi-Goutières Syndrome with Severe End-Organ Involvement.. *Journal of Clinical Immunology*, **2022**, 1	5.7	1
229	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.. *Arthritis and Rheumatology*, **2022**,	9.5	2
228	Mitochondrial Nucleic Acid as a Driver of Pathogenic Type I Interferon Induction in Mendelian Disease. *Frontiers in Immunology*, **2021**, 12, 729763	8.4	4
227	Mitochondrial Nucleic Acid as a Driver of Pathogenic Type I Interferon Induction in Mendelian Disease. *Frontiers in Immunology*, **2021**, 12, 729763	8.4	2
226	Autosomal dominant ADAR c.3019G>A (p.(G1007R)) variant is an important mimic of hereditary spastic paraplegia and cerebral palsy. *Brain and Development*, **2021**,	2.2	0
225	The type I interferonopathies: 10 years on. *Nature Reviews Immunology*, **2021**,	36.5	20
224	Inflammatory profiles across the spectrum of disease reveal a distinct role for GM-CSF in severe COVID-19. *Science Immunology*, **2021**, 6,	28	82
223	Preexisting autoantibodies to type I IFNs underlie critical COVID-19 pneumonia in patients with APS-1. *Journal of Experimental Medicine*, **2021**, 218,	16.6	79
222	Novel compound heterozygous STN1 variants are associated with Coats Plus syndrome. *Molecular Genetics & Genomic Medicine*, **2021**, e1708	2.3	
221	Opsoclonus-myoclonus in Aicardi-Goutières syndrome. *Developmental Medicine and Child Neurology* , **2021**, 63, 1483-1486	3.3	0
220	Cerebral Microangiopathy in Leukoencephalopathy With Cerebral Calcifications and Cysts: A Pathological Description. *Journal of Child Neurology*, **2021**, 36, 133-140	2.5	0
219	Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum. *American Journal of Medical Genetics, Part A*, **2021**, 185, 15-25	2.5	5
218	Rheumatoid factor positive polyarticular juvenile idiopathic arthritis associated with a novel COPA mutation. *Rheumatology*, **2021**, 60, e171-e173	3.9	1
217	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	5.4	19
216	Differential Expression of Interferon-Alpha Protein Provides Clues to Tissue Specificity Across Type I Interferonopathies. *Journal of Clinical Immunology*, **2021**, 41, 603-609	5.7	7
215	LACC1 deficiency links juvenile arthritis with autophagy and metabolism in macrophages. *Journal of Experimental Medicine*, **2021**, 218,	16.6	3

214	STING-Mediated Lung Inflammation and Beyond. *Journal of Clinical Immunology*, **2021**, 41, 501-514	5.7	14
213	Cerebrospinal fluid neopterin as a biomarker of treatment response to Janus kinase inhibition in Aicardi-Goutières syndrome. *Developmental Medicine and Child Neurology*, **2021**,	3.3	2
212	Enhanced cGAS-STING-dependent interferon signaling associated with mutations in ATAD3A. *Journal of Experimental Medicine*, **2021**, 218,	16.6	8
211	Erythrocyte-derived mitochondria take to the lupus stage. *Cell Metabolism*, **2021**, 33, 1723-1725	24.6	1
210	Clinical Reasoning: A 25-year-old woman with recurrent episodes of collapse and loss of consciousness. *Neurology*, **2020**, 94, 994-999	6.5	1
209	Cardiac valve involvement in -related type I interferonopathy. *Journal of Medical Genetics*, **2020**, 57, 475-478	5.8	6
208	Catatonia in a patient with Aicardi-Goutières syndrome efficiently treated with immunoadsorption. *Schizophrenia Research*, **2020**, 222, 484-486	3.6	4
207	Mendelian disorders of immunity related to an upregulation of type I interferon **2020**, 751-772		1
206	An Indian child with Coats plus syndrome due to mutations in STN1. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2139-2144	2.5	4
205	Biallelic mutations in NRROS cause an early onset lethal microgliopathy. *Acta Neuropathologica*, **2020**, 139, 947-951	14.3	6
204	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	14.2	16
203	Use of ruxolitinib in COPA syndrome manifesting as life-threatening alveolar haemorrhage. *Thorax*, **2020**, 75, 92-95	7.3	17
202	Neuromyelitis optica in patients with increased interferon alpha concentrations. *Lancet Neurology, The*, **2020**, 19, 31-33	24.1	8
201	Anti-MDA5 juvenile idiopathic inflammatory myopathy: a specific subgroup defined by differentially enhanced interferon-signalling. *Rheumatology*, **2020**, 59, 1927-1937	3.9	12
200	PSMB10, the last immunoproteasome gene missing for PRAAS. *Journal of Allergy and Clinical Immunology*, **2020**, 145, 1015-1017.e6	11.5	20
199	Genetic and phenotypic spectrum associated with IFIH1 gain-of-function. *Human Mutation*, **2020**, 41, 837-849	4.7	31
198	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	11	7
197	cGAS-mediated induction of type I interferon due to inborn errors of histone pre-mRNA processing. *Nature Genetics*, **2020**, 52, 1364-1372	36.3	52

196	JAK Inhibition in the Aicardi-Goutières Syndrome. *New England Journal of Medicine*, **2020**, 383, 2190-2191	59.2	8
195	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	8.4	4
194	Apparent Radiological Improvement in an Infant With Labrune Syndrome Treated With Bevacizumab. *Pediatric Neurology*, **2020**, 112, 53-55	2.9	1
193	Mutations in COPA lead to abnormal trafficking of STING to the Golgi and interferon signaling. *Journal of Experimental Medicine*, **2020**, 217,	16.6	47
192	Magnetic resonance imaging pattern recognition in childhood bilateral basal ganglia disorders. *Brain Communications*, **2020**, 2, fcaa178	4.5	8
191	Treatments in Aicardi-Goutières syndrome. *Developmental Medicine and Child Neurology*, **2020**, 62, 42-47	3.3	32
190	Circulating Interferon-α 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	9.5	6
189	Leukoencephalopathy, Intracranial Calcifications, Cysts, and SNORD118 Mutation (Labrune Syndrome) with Obstructive Hydrocephalus. *World Neurosurgery*, **2019**, 125, 271-272	2.1	8
188	Bloom syndrome protein restrains innate immune sensing of micronuclei by cGAS. *Journal of Experimental Medicine*, **2019**, 216, 1199-1213	16.6	51
187	Comment on: Aberrant tRNA processing causes an autoinflammatory syndrome responsive to TNF inhibitors by Giannelou : mutations in result in a constitutive activation of type I interferon signalling. *Annals of the Rheumatic Diseases*, **2019**, 78, e86	2.4	7
186	Severe combined immunodeficiency in stimulator of interferon genes (STING) V154M/wild-type mice. *Journal of Allergy and Clinical Immunology*, **2019**, 143, 712-725.e5	11.5	44
185	COPA Syndrome as a Cause of Lupus Nephritis. *Kidney International Reports*, **2019**, 4, 1187-1189	4.1	8
184	Familial Blau syndrome:First molecularly confirmed report from India. *Indian Journal of Ophthalmology*, **2019**, 67, 165-167	1.6	3
183	Severe type I interferonopathy and unrestrained interferon signaling due to a homozygous germline mutation in. *Science Immunology*, **2019**, 4,	28	38
182	Understanding the evolving phenotype of vascular complications in telomere biology disorders. *Angiogenesis*, **2019**, 22, 95-102	10.6	29
181	DDX58 and Classic Singleton-Merten Syndrome. *Journal of Clinical Immunology*, **2019**, 39, 75-80	5.7	21
180	Self-Awareness: Nucleic Acid-Driven Inflammation and the Type I Interferonopathies. *Annual Review of Immunology*, **2019**, 37, 247-267	34.7	73
179	Efficacy of JAK1/2 inhibition in the treatment of chilblain lupus due to TREX1 deficiency. *Annals of the Rheumatic Diseases*, **2019**, 78, 431-433	2.4	33

178	An open-label trial of JAK 1/2 blockade in progressive -associated neuroinflammation. *Neurology*, **2018**, 90, 289-291	6.5	41
177	Sort Your Self Out!. *Cell*, **2018**, 172, 640-642	56.2	3
176	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. *American Journal of Human Genetics*, **2018**, 102, 175-187	11	108
175	Comprehensive molecular screening strategy of OCLN in band-like calcification with simplified gyration and polymicrogyria. *Clinical Genetics*, **2018**, 93, 228-234	4	8
174	Mitochondrial double-stranded RNA triggers antiviral signalling in humans. *Nature*, **2018**, 560, 238-242	50.4	211
173	Development and Validation of an Ultrasensitive Single Molecule Array Digital Enzyme-linked Immunosorbent Assay for Human Interferon- *Journal of Visualized Experiments*, **2018**,	1.6	5
172	Life-threatening influenza pneumonitis in a child with inherited IRF9 deficiency. *Journal of Experimental Medicine*, **2018**, 215, 2567-2585	16.6	98
171	Taking the STING out of inflammation. *Nature Reviews Rheumatology*, **2018**, 14, 508-509	8.1	4
170	International Union of Immunological Societies: 2017 Primary Immunodeficiency Diseases Committee Report on Inborn Errors of Immunity. *Journal of Clinical Immunology*, **2018**, 38, 96-128	5.7	510
169	The 2017 IUIS Phenotypic Classification for Primary Immunodeficiencies. *Journal of Clinical Immunology*, **2018**, 38, 129-143	5.7	345
168	JAK 1/2 Blockade in MDA5 Gain-of-Function. *Journal of Clinical Immunology*, **2018**, 38, 844-846	5.7	17
167	Reverse-Transcriptase Inhibitors in the Aicardi-Goutières Syndrome. *New England Journal of Medicine*, **2018**, 379, 2275-7	59.2	61
166	A child with severe juvenile dermatomyositis treated with ruxolitinib. *Brain*, **2018**, 141, e80	11.2	37
165	A Brief Historical Perspective on the Pathological Consequences of Excessive Type I Interferon Exposure In vivo. *Journal of Clinical Immunology*, **2018**, 38, 694-698	5.7	8
164	Autosomal-dominant early-onset spastic paraparesis with brain calcification due to IFIH1 gain-of-function. *Human Mutation*, **2018**, 39, 1076-1080	4.7	4
163	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	11.5	103
162	Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. *Neuropediatrics*, **2017**, 48, 166-184	1.6	35
161	Brief Report: Blockade of TANK-Binding Kinase 1/IKKe Inhibits Mutant Stimulator of Interferon Genes (STING)-Mediated Inflammatory Responses in Human Peripheral Blood Mononuclear Cells. *Arthritis and Rheumatology*, **2017**, 69, 1495-1501	9.5	17

160	Detection of interferon alpha protein reveals differential levels and cellular sources in disease. *Journal of Experimental Medicine*, **2017**, 214, 1547-1555	16.6	192
159	MDA5-Associated Neuroinflammation and the Singleton-Merten Syndrome: Two Faces of the Same Type I Interferonopathy Spectrum. *Journal of Interferon and Cytokine Research*, **2017**, 37, 214-219	3.5	19
158	Polymorphisms in IFIH1: the good and the bad. *Nature Immunology*, **2017**, 18, 708-709	19.1	7
157	Musculoskeletal Disease in MDA5-Related Type I Interferonopathy: A Mendelian Mimic of Jaccoud's Arthropathy. *Arthritis and Rheumatology*, **2017**, 69, 2081-2091	9.5	30
156	Treatment of Leukoencephalopathy With Calcifications and Cysts With Bevacizumab. *Pediatric Neurology*, **2017**, 71, 56-59	2.9	14
155	Familial and syndromic lupus share the same phenotype as other early-onset forms of lupus. *Joint Bone Spine*, **2017**, 84, 589-593	2.9	4
154	Assessment of Type I Interferon Signaling in Pediatric Inflammatory Disease. *Journal of Clinical Immunology*, **2017**, 37, 123-132	5.7	94
153	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	18	158
152	Leukoencephalopathy with calcification and cysts: A cerebral microangiopathy caused by mutations in SNORD118. *Journal of the Neurological Sciences*, **2017**, 372, 443	3.2	1
151	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	11.5	32
150	Tartrate-Resistant Acid Phosphatase Deficiency in the Predisposition to Systemic Lupus Erythematosus. *Arthritis and Rheumatology*, **2017**, 69, 131-142	9.5	32
149	Type I interferon-mediated autoinflammation due to DNase II deficiency. *Nature Communications*, **2017**, 8, 2176	17.4	111
148	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. *Nature Genetics*, **2016**, 48, 1185-92	36.3	74
147	Neurologic Phenotypes Associated with Mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR1, and IFIH1: Aicardi-Goutières Syndrome and Beyond. *Neuropediatrics*, **2016**, 47, 355-360	1.6	81
146	Type I interferon-mediated monogenic autoinflammation: The type I interferonopathies, a conceptual overview. *Journal of Experimental Medicine*, **2016**, 213, 2527-2538	16.6	243
145	Human USP18 deficiency underlies type 1 interferonopathy leading to severe pseudo-TORCH syndrome. *Journal of Experimental Medicine*, **2016**, 213, 1163-74	16.6	154
144	Spondyloenchondrodysplasia Due to Mutations in ACP5: A Comprehensive Survey. *Journal of Clinical Immunology*, **2016**, 36, 220-34	5.7	48
143	A POT1 mutation implicates defective telomere end fill-in and telomere truncations in Coats plus. *Genes and Development*, **2016**, 30, 812-26	12.6	50

142	Neuroradiologic patterns and novel imaging findings in Aicardi-Goutières syndrome. *Neurology*, **2016**, 86, 28-35	6.5	44
141	JAK inhibition in STING-associated interferonopathy. *Annals of the Rheumatic Diseases*, **2016**, 75, e75	2.4	17
140	Clinical, radiological and possible pathological overlap of cystic leukoencephalopathy without megalencephaly and Aicardi-Goutières syndrome. *European Journal of Paediatric Neurology*, **2016**, 20, 604-10	3.8	24
139	Update and Mutational Analysis of SLC20A2: A Major Cause of Primary Familial Brain Calcification. *Human Mutation*, **2015**, 36, 489-95	4.7	54
138	The eukaryotic elongation factor eEF1A1 interacts with SAMHD1. *Biochemical Journal*, **2015**, 466, 69-76	3.8	11
137	Characterization of samhd1 morphant zebrafish recapitulates features of the human type I interferonopathy Aicardi-Goutières syndrome. *Journal of Immunology*, **2015**, 194, 2819-25	5.3	26
136	Human disease phenotypes associated with mutations in TREX1. *Journal of Clinical Immunology*, **2015**, 35, 235-43	5.7	108
135	PRKDC mutations associated with immunodeficiency, granuloma, and autoimmune regulator-dependent autoimmunity. *Journal of Allergy and Clinical Immunology*, **2015**, 135, 1578-88.e5	11.5	52
134	Neuromyelitis optica in a child with Aicardi-Goutières syndrome. *Neurology*, **2015**, 85, 381-3	6.5	17
133	Mosaic Tetrasomy 9p: A Mendelian Condition Associated With Pediatric-Onset Overlap Myositis. *Pediatrics*, **2015**, 136, e544-7	7.4	10
132	Novel monogenic diseases causing human autoimmunity. *Current Opinion in Immunology*, **2015**, 37, 1-5	7.8	12
131	cGMP-AMP synthase paves the way to autoimmunity. *Proceedings of the National Academy of Sciences of the United States of America*, **2015**, 112, 12903-4	11.5	5
130	Large-scale discovery of novel genetic causes of developmental disorders. *Nature*, **2015**, 519, 223-8	50.4	706
129	Human intracellular ISG15 prevents interferon-α over-amplification and auto-inflammation. *Nature*, **2015**, 517, 89-93	50.4	311
128	Type I interferonopathies: mendelian type I interferon up-regulation. *Current Opinion in Immunology*, **2015**, 32, 7-12	7.8	128
127	Stimulator of Interferon Genes-Associated Vasculopathy With Onset in Infancy: A Mimic of Childhood Granulomatosis With Polyangiitis. *JAMA Dermatology*, **2015**, 151, 872-7	5.1	72
126	Aicardi-Goutières syndrome and the type I interferonopathies. *Nature Reviews Immunology*, **2015**, 15, 429-40	36.5	493
125	Early-Onset Aicardi-Goutières Syndrome: Magnetic Resonance Imaging (MRI) Pattern Recognition. *Journal of Child Neurology*, **2015**, 30, 1343-8	2.5	26

124	A specific IFIH1 gain-of-function mutation causes Singleton-Merten syndrome. *American Journal of Human Genetics*, **2015**, 96, 275-82	11	134
123	Mosaic structural variation in children with developmental disorders. *Human Molecular Genetics*, **2015**, 24, 2733-45	5.6	39
122	Aicardi-Goutières syndrome harbours abundant systemic and brain-reactive autoantibodies. *Annals of the Rheumatic Diseases*, **2015**, 74, 1931-9	2.4	27
121	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**, 167A, 296-312	2.5	321
120	ADAR1 Facilitates HIV-1 Replication in Primary CD4+ T Cells. *PLoS ONE*, **2015**, 10, e0143613	3.7	11
119	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	36.3	376
118	Clinical delineation and natural history of the PIK3CA-related overgrowth spectrum. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1713-33	2.5	198
117	Intracranial calcification in childhood: a review of aetiologies and recognizable phenotypes. *Developmental Medicine and Child Neurology*, **2014**, 56, 612-26	3.3	111
116	The SKIV2L RNA exosome limits activation of the RIG-I-like receptors. *Nature Immunology*, **2014**, 15, 839-45	19.1	131
115	Mutations in PIEZO2 cause Gordon syndrome, Marden-Walker syndrome, and distal arthrogryposis type 5. *American Journal of Human Genetics*, **2014**, 94, 734-44	11	124
114	SAMHD1 is mutated recurrently in chronic lymphocytic leukemia and is involved in response to DNA damage. *Blood*, **2014**, 123, 1021-31	2.2	154
113	Inherited STING-activating mutation underlies a familial inflammatory syndrome with lupus-like manifestations. *Journal of Clinical Investigation*, **2014**, 124, 5516-20	15.9	294
112	Leukoencephalopathy with calcifications and cysts: a purely neurological disorder distinct from coats plus. *Neuropediatrics*, **2014**, 45, 175-82	1.6	33
111	Mutations in ADAR1, IFIH1, and RNASEH2B presenting as spastic paraplegia. *Neuropediatrics*, **2014**, 45, 386-93	1.6	53
110	Therapies in Aicardi-Goutières syndrome. *Clinical and Experimental Immunology*, **2014**, 175, 1-8	6.2	60
109	Basal ganglia calcification in a patient with beta-propeller protein-associated neurodegeneration. *Pediatric Neurology*, **2014**, 51, 843-5	2.9	14
108	Mutations in CECR1 associated with a neutrophil signature in peripheral blood. *Pediatric Rheumatology*, **2014**, 12, 44	3.5	63
107	A type I interferon signature identifies bilateral striatal necrosis due to mutations in ADAR1. *Journal of Medical Genetics*, **2014**, 51, 76-82	5.8	85

106	Reply: To PMID 23666743. *Arthritis and Rheumatology*, **2014**, 66, 229-30	9.5	
105	Mendelian Disorders of Immunity Related to an Upregulation of Type I Interferon **2014**, 591-602		
104	SAMHD1-dependent retroviral control and escape in mice. *EMBO Journal*, **2013**, 32, 2454-62	13	116
103	Assessment of interferon-related biomarkers in Aicardi-Goutières syndrome associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, and ADAR: a case-control study. *Lancet Neurology, The*, **2013**, 12, 1159-69	24.1	267
102	Exudative retinopathy, cerebral calcifications, duodenal atresia, preaxial polydactyly, micropenis, microcephaly and short stature: a new syndrome?. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1829-32	2.5	2
101	Recognizable phenotypes associated with intracranial calcification. *Developmental Medicine and Child Neurology*, **2013**, 55, 46-57	3.3	53
100	Striking intrafamilial phenotypic variability in Aicardi-Goutières syndrome associated with the recurrent Asian founder mutation in RNASEH2C. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 338-42	2.5	25
99	Elevation of proinflammatory cytokines in patients with Aicardi-Goutières syndrome. *Neurology*, **2013**, 80, 997-1002	6.5	15
98	Aicardi-Goutières syndrome. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2013**, 113, 1629-35	3	55
97	Protein kinase cδ deficiency causes mendelian systemic lupus erythematosus with B cell-defective apoptosis and hyperproliferation. *Arthritis and Rheumatism*, **2013**, 65, 2161-71		119
96	Systemic lupus erythematosus due to C1q deficiency with progressive encephalopathy, intracranial calcification and acquired moyamoya cerebral vasculopathy. *Lupus*, **2013**, 22, 639-43	2.6	24
95	Synonymous mutations in RNASEH2A create cryptic splice sites impairing RNase H2 enzyme function in Aicardi-Goutières syndrome. *Human Mutation*, **2013**, 34, 1066-70	4.7	15
94	Intracranial calcification in early infantile Krabbe disease: nothing new under the sun. *Developmental Medicine and Child Neurology*, **2012**, 54, 376-9	3.3	11
93	SAMHD1 restricts HIV-1 reverse transcription in quiescent CD4(+) T-cells. *Retrovirology*, **2012**, 9, 87	3.6	254
92	Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. *Nature Genetics*, **2012**, 44, 338-42	36.3	186
91	Mutations in ADAR1 cause Aicardi-Goutières syndrome associated with a type I interferon signature. *Nature Genetics*, **2012**, 44, 1243-8	36.3	521
90	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, 381-8	5.3	113
89	SAMHD1 is a nucleic-acid binding protein that is mislocalized due to aicardi-goutières syndrome-associated mutations. *Human Mutation*, **2012**, 33, 1116-22	4.7	103

88	Diagnosing fetal alcohol syndrome: new insights from newer genetic technologies. *Archives of Disease in Childhood*, **2012**, 97, 812-7	2.2	27
87	SAMHD1, A Putative Tumour Suppressor, Is Recurrently Mutated in Chronic Lymphocytic Leukaemia, and Is Associated with Poor Risk Features. *Blood*, **2012**, 120, 713-713	2.2	
86	Degos disease: a C5b-9/interferon-□mediated endotheliopathy syndrome. *American Journal of Clinical Pathology*, **2011**, 135, 599-610	1.9	71
85	Molecular screening of ADAMTSL2 gene in 33 patients reveals the genetic heterogeneity of geleophysic dysplasia. *Journal of Medical Genetics*, **2011**, 48, 417-21	5.8	33
84	HIV-1 restriction factor SAMHD1 is a deoxynucleoside triphosphate triphosphohydrolase. *Nature*, **2011**, 480, 379-82	50.4	578
83	Tartrate-resistant acid phosphatase deficiency causes a bone dysplasia with autoimmunity and a type I interferon expression signature. *Nature Genetics*, **2011**, 43, 127-31	36.3	173
82	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	5.3	40
81	Type I interferonopathies: a novel set of inborn errors of immunity. *Annals of the New York Academy of Sciences*, **2011**, 1238, 91-8	6.5	252
80	Paediatric stroke: genetic insights into disease mechanisms and treatment targets. *Lancet Neurology, The*, **2011**, 10, 264-74	24.1	42
79	Infantile neurological Degos disease. *European Journal of Paediatric Neurology*, **2011**, 15, 167-70	3.8	8
78	Identification and characterization of an inborn error of metabolism caused by dihydrofolate reductase deficiency. *American Journal of Human Genetics*, **2011**, 88, 216-25	11	68
77	Phenotypic variation in familial chilblain lupus (FCL) and Aicardi-Goutiᅠᅠes syndrome (AGS) associated with TREX1 mutation in 4 family members. *Pediatric Rheumatology*, **2011**, 9,	3.5	78
76	Autosomal dominant inheritance of a heterozygous mutation in SAMHD1 causing familial chilblain lupus. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 235-7	2.5	81
75	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**, 155A, 2910-5	2.5	2
74	Severe neonatal-onset panniculitis in a female infant with Prader-Willi syndrome. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 3087-9	2.5	
73	Lupus: how much "complexity" is really (just) genetic heterogeneity?. *Arthritis and Rheumatism*, **2011**, 63, 3661-4		19
72	Treatment of Gastrointestinal Bleeding in a Probable Case of Cerebroretinal Microangiopathy with Calcifications and Cysts. *Molecular Syndromology*, **2011**, 1, 159-162	1.5	10
71	Cerebral vasculopathy is a common feature in Aicardi-Goutieres syndrome associated with SAMHD1 mutations. *Proceedings of the National Academy of Sciences of the United States of America*, **2011**, 108, E232; author reply E233	11.5	23

70	COL4A1 mutations associated with a characteristic pattern of intracranial calcification. *Neuropediatrics*, **2011**, 42, 227-33	1.6	32
69	Chilblains as a diagnostic sign of aicardi-goutières syndrome. *Neuropediatrics*, **2010**, 41, 18-23	1.6	26
68	Expanding the clinical spectrum of SLC29A3 gene defects. *European Journal of Medical Genetics*, **2010**, 53, 309-13	2.6	33
67	3C syndrome. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1026-7	2.5	3
66	Brown-Vialetto-Van Laere syndrome, a ponto-bulbar palsy with deafness, is caused by mutations in c20orf54. *American Journal of Human Genetics*, **2010**, 86, 485-9	11	139
65	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, 354-64	11	98
64	The story of DNase II: a stifled death-wish leads to self-harm. *European Journal of Immunology*, **2010** , 40, 2376-8	6.1	3
63	Familial Aicardi-Goutières syndrome due to SAMHD1 mutations is associated with chronic arthropathy and contractures. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 938-42	2.5	61
62	New subtype of familial intracranial calcification in a mother and two children. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 943-6	2.5	4
61	A de novo p.Asp18Asn mutation in TREX1 in a patient with Aicardi-Goutières syndrome. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2612-7	2.5	30
60	Intracerebral large artery disease in Aicardi-Goutières syndrome implicates SAMHD1 in vascular homeostasis. *Developmental Medicine and Child Neurology*, **2010**, 52, 725-32	3.3	80
59	Aicardi-Goutieres syndrome and related phenotypes: linking nucleic acid metabolism with autoimmunity. *Human Molecular Genetics*, **2009**, 18, R130-6	5.6	230
58	Atypical progeroid syndrome due to heterozygous missense LMNA mutations. *Journal of Clinical Endocrinology and Metabolism*, **2009**, 94, 4971-83	5.6	95
57	Aicardi-Goutieres syndrome: neuroradiologic findings and follow-up. *American Journal of Neuroradiology*, **2009**, 30, 1971-6	4.4	59
56	Aicardi-Goutières syndrome presenting with haematemesis in infancy. *Acta Paediatrica, International Journal of Paediatrics*, **2009**, 98, 2005-8	3.1	2
55	Mutations in MFSD8/CLN7 are a frequent cause of variant-late infantile neuronal ceroid lipofuscinosis. *Human Mutation*, **2009**, 30, E530-40	4.7	40
54	Mutations involved in Aicardi-Goutières syndrome implicate SAMHD1 as regulator of the innate immune response. *Nature Genetics*, **2009**, 41, 829-32	36.3	507
53	Elevated pterins in cerebral spinal fluid--biochemical marker of Aicardi-Goutières syndrome. *Developmental Medicine and Child Neurology*, **2009**, 51, 841-2	3.3	3

52	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	11	196
51	Mutation of the variant alpha-tubulin TUBA8 results in polymicrogyria with optic nerve hypoplasia. *American Journal of Human Genetics*, **2009**, 85, 737-44	11	132
50	Blue (or purple) toes: chilblains or chilblain lupus-like lesions are a manifestation of Aicardi-Goutières syndrome and familial chilblain lupus. *Journal of the American Academy of Dermatology*, **2009**, 61, 727-8	4.5	15
49	ADAMTSL2 mutations in geleophysic dysplasia demonstrate a role for ADAMTS-like proteins in TGF-beta bioavailability regulation. *Nature Genetics*, **2008**, 40, 1119-23	36.3	172
48	tRNA splicing endonuclease mutations cause pontocerebellar hypoplasia. *Nature Genetics*, **2008**, 40, 1113-8	36.3	177
47	8p23.1 duplication syndrome; a novel genomic condition with unexpected complexity revealed by array CGH. *European Journal of Human Genetics*, **2008**, 16, 18-27	5.3	66
46	Aicardi-Goutières syndrome: an important Mendelian mimic of congenital infection. *Developmental Medicine and Child Neurology*, **2008**, 50, 410-6	3.3	115
45	Aicardi-Goutières syndrome: description of a late onset case. *Developmental Medicine and Child Neurology*, **2008**, 50, 631-4	3.3	30
44	Aicardi-Goutières syndrome presenting atypically as a sub-acute leukoencephalopathy. *European Journal of Paediatric Neurology*, **2008**, 12, 408-11	3.8	23
43	The neonatal form of Aicardi-Goutières syndrome masquerading as congenital infection. *Early Human Development*, **2008**, 84, 783-5	2.2	12
42	Mutations in the pericentrin (PCNT) gene cause primordial dwarfism. *Science*, **2008**, 319, 816-9	33.3	325
41	Delineation of late onset hypoventilation associated with hypothalamic dysfunction syndrome. *Pediatric Research*, **2008**, 64, 689-94	3.2	54
40	Cerebroretinal microangiopathy with calcifications and cysts (CRMCC). *American Journal of Medical Genetics, Part A*, **2008**, 146A, 182-90	2.5	70
39	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, 1713-7	2.5	34
38	Two further cases of spondyloenchondrodysplasia (SPENCD) with immune dysregulation. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2810-5	2.5	24
37	Band-like intracranial calcification with simplified gyration and polymicrogyria: a distinct "pseudo-TORCH" phenotype. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 3173-80	2.5	42
36	Cutaneous histopathological findings of Aicardi-Goutières syndrome, overlap with chilblain lupus. *Journal of Cutaneous Pathology*, **2008**, 35, 774-8	1.7	39
35	Neurological presentation of Griscelli syndrome: obstructive hydrocephalus without haematological abnormalities or organomegaly. *Brain and Development*, **2007**, 29, 247-50	2.2	13

34	Variable phenotype including Leigh syndrome with a 9185T>C mutation in the MTATP6 gene. *Neuropediatrics*, **2007**, 38, 313-6	1.6	35
33	The spectrum of SCN1A-related infantile epileptic encephalopathies. *Brain*, **2007**, 130, 843-52	11.2	423
32	Focal dermal hypoplasia with subependymal heterotopia and hypoplastic corpus callosum. *Clinical Dysmorphology*, **2007**, 16, 59-61	0.9	2
31	Heterozygous mutations in TREX1 cause familial chilblain lupus and dominant Aicardi-Goutieres syndrome. *American Journal of Human Genetics*, **2007**, 80, 811-5	11	286
30	Clinical and molecular phenotype of Aicardi-Goutieres syndrome. *American Journal of Human Genetics*, **2007**, 81, 713-25	11	310
29	Reduced penetrance alleles for Huntington's disease: a multi-centre direct observational study. *Journal of Medical Genetics*, **2007**, 44, e68	5.8	52
28	A newly recognized, likely autosomal recessive syndrome comprising agammaglobulinemia, microcephaly, craniosynostosis, severe dermatitis, and other features. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1131-5	2.5	1
27	Sedaghatian spondylometaphyseal dysplasia with pachygyria and absence of the corpus callosum. *American Journal of Medical Genetics, Part A*, **2006**, 140A, 1854-8	2.5	5
26	Elevated interferon-alpha in fetal blood in the prenatal diagnosis of Aicardi-Goutières syndrome. *Fetal Diagnosis and Therapy*, **2006**, 21, 153-5	2.4	9
25	Severe childhood SMA and axonal CMT due to anticodon binding domain mutations in the GARS gene. *Neurology*, **2006**, 67, 1710-2	6.5	62
24	Clinical phenotype associated with homozygosity for a HOXD13 7-residue polyalanine tract expansion. *European Journal of Medical Genetics*, **2006**, 49, 396-401	2.6	17
23	Mutations in genes encoding ribonuclease H2 subunits cause Aicardi-Goutières syndrome and mimic congenital viral brain infection. *Nature Genetics*, **2006**, 38, 910-6	36.3	505
22	Mutations in the gene encoding the 3'-5' DNA exonuclease TREX1 cause Aicardi-Goutières syndrome at the AGS1 locus. *Nature Genetics*, **2006**, 38, 917-20	36.3	633
21	A second locus for Aicardi-Goutieres syndrome at chromosome 13q14-21. *Journal of Medical Genetics*, **2006**, 43, 444-50	5.8	27
20	Gross rearrangements of the MECP2 gene are found in both classical and atypical Rett syndrome patients. *Journal of Medical Genetics*, **2006**, 43, 451-6	5.8	58
19	Genetic syndromes mimic congenital infections. *Journal of Pediatrics*, **2005**, 146, 701-5	3.6	46
18	Clinical and mutational spectrum of Mowat-Wilson syndrome. *European Journal of Medical Genetics*, **2005**, 48, 97-111	2.6	103
17	Mental retardation, keratoconus, febrile seizures and sinoatrial block: a previously undescribed autosomal recessive disorder. *Clinical Genetics*, **2005**, 67, 448-9	4	8

16	Brown-Vialetto-Van Laere syndrome; variability in age at onset and disease progression highlighting the phenotypic overlap with Fazio-Londe disease. *Brain and Development*, **2005**, 27, 443-6	2.2	38
15	Natural history of cardiac involvement in geleophysic dysplasia **2005**, 132A, 320-3		14
14	Congenital palmar polyonychia with postaxial limb defects may be the same as the ulnar-mammary syndrome. *American Journal of Medical Genetics, Part A*, **2005**, 137, 233; author reply 234	2.5	
13	Chromosome 1q42 deletion and agenesis of the corpus callosum. *American Journal of Medical Genetics, Part A*, **2005**, 138, 68-9	2.5	17
12	CoatsNplus: a progressive familial syndrome of bilateral CoatsNdisease, characteristic cerebral calcification, leukoencephalopathy, slow pre- and post-natal linear growth and defects of bone marrow and integument. *Neuropediatrics*, **2004**, 35, 10-9	1.6	66
11	Congenital glaucoma and brain stem atrophy as features of Aicardi-Goutiﾭes syndrome **2004**, 129A, 303-7		24
10	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	11	279
9	The genetics of Aicardi-Goutiﾭes syndrome. *European Journal of Paediatric Neurology*, **2002**, 6 Suppl A, A33-5; discussion A37-9, A77-86	3.8	5
8	ASPM is a major determinant of cerebral cortical size. *Nature Genetics*, **2002**, 32, 316-20	36.3	472
7	Identification of microcephalin, a protein implicated in determining the size of the human brain. *American Journal of Human Genetics*, **2002**, 71, 136-42	11	455
6	Aicardi-Goutiﾭes syndrome displays genetic heterogeneity with one locus (AGS1) on chromosome 3p21. *American Journal of Human Genetics*, **2000**, 67, 213-21	11	70
5	A fifth locus for primary autosomal recessive microcephaly maps to chromosome 1q31. *American Journal of Human Genetics*, **2000**, 67, 1578-80	11	91
4	The molecular basis of GROD-storing neuronal ceroid lipofuscinoses in Scotland. *Molecular Genetics and Metabolism*, **1999**, 66, 245-7	3.7	9
3	Mutations in the palmitoyl-protein thioesterase gene (PPT; CLN1) causing juvenile neuronal ceroid lipofuscinosis with granular osmiophilic deposits. *Human Molecular Genetics*, **1998**, 7, 291-7	5.6	102
2	Spondylocostal dysostosis associated with a 46, XX,+15,dic(6;15)(q25;q11.2) translocation. *Clinical Dysmorphology*, **1997**, 6, 347-50	0.9	13
1	Maternal serum alpha-fetoprotein levels in congenital nephrosis **1997**, 17, 1089-1089		4