David A Dyment

List of Publications by Year
in descending order

135	7,320	71004	71088
papers	citations	h-index	g-index

136	136	136	12171
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A diseaseâ€causing variant in <scp>HNRNPH2</scp> inherited from an unaffected mother with skewed Xâ€inactivation. American Journal of Medical Genetics, Part A, 2022, 188, 668-671.	0.7	4
2	Heterozygous De Novo <scp>KPNA3</scp> Mutations Cause Complex Hereditary Spastic Paraplegia. Annals of Neurology, 2022, 91, 730-732.	2.8	1
3	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.0	42
4	Phenotypic spectrum of the recurrent TRPM3 p.(<scp>Val837Met</scp>) substitution in seven individuals with global developmental delay and hypotonia. American Journal of Medical Genetics, Part A, 2022, 188, 1667-1675.	0.7	8
5	The Benefit of Multigene Panel Testing for the Diagnosis and Management of the Genetic Epilepsies. Genes, 2022, 13, 872.	1.0	2
6	A 79â€kb paternally inherited 7q32.2 microdeletion involving <scp> MEST </scp> in a patient with a <scp>Silverâ€Russell</scp> syndromeâ€like phenotype. American Journal of Medical Genetics, Part A, 2022, , .	0.7	0
7	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 119-133.	0.7	17
8	Whole genome sequencing reveals biallelic <scp>PLA2G6</scp> mutations in siblings with cerebellar atrophy and cap myopathy. Clinical Genetics, 2021, 99, 746-748.	1.0	3
9	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	2.6	30
10	A splice site and copy number variant responsible for TTC25-related primary ciliary dyskinesia. European Journal of Medical Genetics, 2021, 64, 104193.	0.7	4
11	Heterozygous loss-of-function variants significantly expand the phenotypes associated with loss of GDF11. Genetics in Medicine, 2021, 23, 1889-1900.	1.1	13
12	Biallelic variants in PCDHGC4 cause a novel neurodevelopmental syndrome with progressive microcephaly, seizures, and joint anomalies. Genetics in Medicine, 2021, 23, 2138-2149.	1.1	11
13	Whole genome sequencing identifies pathogenic <scp>RNU4ATAC</scp> variants in a child with recurrent encephalitis, microcephaly, and normal stature. American Journal of Medical Genetics, Part A, 2021, 185, 3502-3506.	0.7	3
14	Enhanced cGAS-STINGâ€“dependent interferon signaling associated with mutations in ATAD3A. Journal of Experimental Medicine, 2021, 218, .	4.2	43
15	Expanding the Phenotypic Spectrum of GPI Anchoring Deficiency Due to Biallelic Variants in GPAA1. Neurology: Genetics, 2021, 7, e631.	0.9	2
16	Implementation of Epilepsy Multigene Panel Testing in Ontario, Canada. Canadian Journal of Neurological Sciences, 2020, 47, 61-68.	0.3	6
17	Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918.	4.9	139
18	Germline AGO2 mutations impair RNA interference and human neurological development. Nature Communications, 2020, 11, 5797.	5.8	43

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19	Channelopathies Are a Frequent Cause of Genetic Ataxias Associated with Cerebellar Atrophy. Movement Disorders Clinical Practice, 2020, 7, 940-949.	0.8	7
20	Genetic mechanisms of neurodevelopmental disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2020, 173, 307-326.	1.0	3
21	Electrophysiological Alterations of Pyramidal Cells and Interneurons of the CA1 Region of the Hippocampus in a Novel Mouse Model of Dravet Syndrome. Genetics, 2020, 215, 1055-1066.	1.2	10
22	A splice variant in ATAD3A expands the clinical and genetic spectrum of Harel-Yoon syndrome. Neurology: Genetics, 2020, 6, e452.	0.9	14
23	The Canadian Rare Diseases Models and Mechanisms (RDMM) Network: Connecting Understudied Genes to Model Organisms. American Journal of Human Genetics, 2020, 106, 143-152.	2.6	30
24	Overlapping phenotypes between SHORT and Noonan syndromes in patients with PTPN11 pathogenic variants. Clinical Genetics, 2020, 98, 10-18.	1.0	9
25	Epilepsy genetics: Current knowledge, applications, and future directions. Clinical Genetics, 2019, 95, 95-111.	1.0	87
26	p21 proteinâ€activated kinase 1 is associated with severe regressive autism, and epilepsy. Clinical Genetics, 2019, 96, 449-455.	1.0	13
27	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	5.8	150
28	De novo substitutions of TRPM3 cause intellectual disability and epilepsy. European Journal of Human Genetics, 2019, 27, 1611-1618.	1.4	45
29	B.06 Whole exome sequencing in genetic ataxias associated with cerebellar atrophy: the Canadian experience. Canadian Journal of Neurological Sciences, 2019, 46, S11.	0.3	0
30	PLPHP deficiency: clinical, genetic, biochemical, and mechanistic insights. Brain, 2019, 142, 542-559.	3.7	67
31	Pathogenic variants in USP7 cause a neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies. Genetics in Medicine, 2019, 21, 1797-1807.	1.1	41
32	Development of Criteria for Epilepsy Genetic Testing in Ontario, Canada. Canadian Journal of Neurological Sciences, 2019, 46, 7-13.	0.3	18
33	Atypical Hepatic Mesenchymal Hamartoma: Histologic Appearance, Immunophenotype, and Molecular Findings. Pediatric and Developmental Pathology, 2019, 22, 365-369.	0.5	3
34	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	0.7	108
35	Reply: The recurrent mutation in TMEM106B also causes hypomyelinating leukodystrophy in China and is a CpG hotspot. Brain, 2018, 141, e37-e37.	3.7	0
36	Wholeâ€exome sequencing is a valuable diagnostic tool for inherited peripheral neuropathies: Outcomes from a cohort of 50 families. Clinical Genetics, 2018, 93, 301-309.	1.0	48

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37	A de novo mutation in RPL10 causes a rare X-linked ribosomopathy characterized by syndromic intellectual disability and epilepsy: A new case and review of the literature. European Journal of Medical Genetics, 2018, 61, 89-93.	0.7	22
38	Genomic study of severe fetal anomalies and discovery of GREB1L mutations in renal agenesis. Genetics in Medicine, 2018, 20, 745-753.	1.1	60
39	Evaluation of exome filtering techniques for the analysis of clinically relevant genes. Human Mutation, 2018, 39, 197-201.	1.1	13
40	Unsolved recognizable patterns of human malformation: Challenges and opportunities. , 2018, 178, 382-386.		13
41	Clinical and genetic heterogeneity in Dubowitz syndrome: Implications for diagnosis, management and further research. , 2018, 178, 387-397.		15
42	Lysosomal dysfunction in TMEM106B hypomyelinating leukodystrophy. Neurology: Genetics, 2018, 4, e288.	0.9	11
43	The unsolved rare genetic disease atlas? An analysis of the unexplained phenotypic descriptions in OMIMÂ®. , 2018, 178, 458-463.		25
44	The ClinGen Epilepsy Gene Curation Expert Panelâ€"Bridging the divide between clinical domain knowledge and formal gene curation criteria. Human Mutation, 2018, 39, 1476-1484.	1.1	33
45	Atypical Rett Syndrome and Intractable Epilepsy With Novel GRIN2B Mutation. Child Neurology Open, 2018, 5, 2329048X1878794.	0.5	5
46	A ZPR1 mutation is associated with a novel syndrome of growth restriction, distinct craniofacial features, alopecia, and hypoplastic kidneys. Clinical Genetics, 2018, 94, 303-312.	1.0	5
47	Periodic breathing in patients with NALCN mutations. Journal of Human Genetics, 2018, 63, 1093-1096.	1.1	13
48	Diagnostic clarity of exome sequencing following negative comprehensive panel testing in the neonatal intensive care unit. American Journal of Medical Genetics, Part A, 2018, 176, 1688-1691.	0.7	28
49	P.131 De novo PIK3CB mutation associated with macrocephaly and diffuse polymicrogyria. Canadian Journal of Neurological Sciences, 2018, 45, S51-S51.	0.3	0
50	Autosomal dominant cutis laxa with progeroid features due to a novel, de novo mutation in ALDH18A1. Journal of Human Genetics, 2017, 62, 661-663.	1.1	12
51	Debunking Occam's razor: Diagnosing multiple genetic diseases in families by wholeâ€exome sequencing. Clinical Genetics, 2017, 92, 281-289.	1.0	92
52	Whole-transcriptome sequencing in blood provides a diagnosis of spinal muscular atrophy with progressive myoclonic epilepsy. Human Mutation, 2017, 38, 611-614.	1.1	25
53	Matchmaking facilitates the diagnosis of an autosomal-recessive mitochondrial disease caused by biallelic mutation of the tRNA isopentenyltransferase (TRIT1) gene. Human Mutation, 2017, 38, 511-516.	1.1	39
54	Benchmarking outcomes in the Neonatal Intensive Care Unit: Cytogenetic and molecular diagnostic rates in a retrospective cohort. American Journal of Medical Genetics, Part A, 2017, 173, 1839-1847.	0.7	25

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55	Yunis-VarÃ³n syndrome caused by biallelic VAC14 mutations. European Journal of Human Genetics, 2017, 25, 1049-1054.	1.4	21
56	Compound heterozygous mutations in the gene PIGP are associated with early infantile epileptic encephalopathy. Human Molecular Genetics, 2017, 26, 1706-1715.	1.4	39
57	Two females with mutations in USP9X highlight the variable expressivity of the intellectual disability syndrome. European Journal of Medical Genetics, 2017, 60, 359-364.	0.7	12
58	Pyridoxine-Dependent Epilepsy in Zebrafish Caused by Aldh7a1 Deficiency. Genetics, 2017, 207, 1501-1518.	1.2	81
59	Pointed rhythmic theta waves: a unique EEG pattern in KCNQ2-related neonatal epileptic encephalopathy. Epileptic Disorders, 2017, 19, 351-356.	0.7	5
60	Loss of the arginine methyltranserase PRMT7 causes syndromic intellectual disability with microcephaly and brachydactyly. Clinical Genetics, 2017, 91, 708-716.	1.0	23
61	SHORT syndrome due to a novel de novo mutation in PRKCE (Protein Kinase CÉ›) impairing TORC2-dependent AKT activation. Human Molecular Genetics, 2017, 26, 3713-3721.	1.4	22
62	A recurrent de novo mutation in TMEM106B causes hypomyelinating leukodystrophy. Brain, 2017, 140, 3105-3111.	3.7	64
63	Late diagnosis of cerebral folate deficiency: Fewer seizures with folinic acid in adult siblings. Neurology: Genetics, 2016, 2, e38.	0.9	11
64	Clinical reappraisal of <scp>SHORT</scp> syndrome with <scp>PIK3R1</scp> mutations: toward recommendation for molecular testing and management. Clinical Genetics, 2016, 89, 501-506.	1.0	66
65	Concordance between wholeâ€exome sequencing and clinical Sanger sequencing: implications for patient care. Molecular Genetics & Genomic Medicine, 2016, 4, 504-512.	0.6	30
66	Next-generation sequencing for diagnosis of rare diseases in the neonatal intensive care unit. Cmaj, 2016, 188, E254-E260.	0.9	86
67	DNM1L-related mitochondrial fission defect presenting as refractory epilepsy. European Journal of Human Genetics, 2016, 24, 1084-1088.	1.4	113
68	A novel mutation in two Hmong families broadens the range of STRA6â€related malformations to include contractures and camptodactyly. American Journal of Medical Genetics, Part A, 2016, 170, 11-18.	0.7	15
69	Case report of novel DYRK1A mutations in 2 individuals with syndromic intellectual disability and a review of the literature. BMC Medical Genetics, 2016, 17, 15.	2.1	42
70	Identification of a pathogenicFTOmutation by next-generation sequencing in a newborn with growth retardation and developmental delay. Journal of Medical Genetics, 2016, 53, 200-207.	1.5	50
71	Utility of wholeâ€exome sequencing for those near the end of the diagnostic odyssey: time to address gaps in care. Clinical Genetics, 2016, 89, 275-284.	1.0	323
72	Congenital Nemaline Myopathy: The Value of Magnetic Resonance Imaging of Muscle. Canadian Journal of Neurological Sciences, 2015, 42, 338-340.	0.3	7

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73	Resolution of refractory hypotension and anuria in a premature newborn with lossâ€ofâ€function of ACE. American Journal of Medical Genetics, Part A, 2015, 167, 1654-1658.	0.7	10
74	LIMS2 mutations are associated with a novel muscular dystrophy, severe cardiomyopathy and triangular tongues. Clinical Genetics, 2015, 88, 558-564.	1.0	23
75	Biallelic Mutations in BRCA1 Cause a New Fanconi Anemia Subtype. Cancer Discovery, 2015, 5, 135-142.	7.7	251
76	Homozygous mutations inMFN2cause multiple symmetric lipomatosis associated with neuropathy. Human Molecular Genetics, 2015, 24, 5109-5114.	1.4	61
77	Brain malformations in a patient with deletion 2p16.1: A refinement ofÂthe phenotype to BCL11A. European Journal of Medical Genetics, 2015, 58, 351-354.	0.7	24
78	Detailed Biochemical and Bioenergetic Characterization of FBXL4-Related Encephalomyopathic Mitochondrial DNA Depletion. JIMD Reports, 2015, 27, 1-9.	0.7	19
79	Axons to Exons: the Molecular Diagnosis of Rare Neurological Diseases by Next-Generation Sequencing. Current Neurology and Neuroscience Reports, 2015, 15, 64.	2.0	29
80	Autosomal recessive axonal polyneuropathy in a sibling pair due to a novel homozygous mutation in IGHMBP2. Neuromuscular Disorders, 2015, 25, 794-799.	0.3	16
81	Wholeâ€exome sequencing broadens the phenotypic spectrum of rare pediatric epilepsy: a retrospective study. Clinical Genetics, 2015, 88, 34-40.	1.0	79
82	Homozygous nonsense mutation in SYNJ1 associated with intractable epilepsy and tau pathology. Neurobiology of Aging, 2015, 36, 1222.e1-1222.e5.	1.5	50
83	BCL11A deletions result in fetal hemoglobin persistence and neurodevelopmental alterations. Journal of Clinical Investigation, 2015, 125, 2363-2368.	3.9	122
84	Evidence for clinical, genetic and biochemical variability in spinal muscular atrophy with progressive myoclonic epilepsy. Clinical Genetics, 2014, 86, 558-563.	1.0	42
85	Wholeâ€exome sequencing in an individual with severe global developmental delay and intractable epilepsy identifies a novel, de novo GRIN2A mutation. Epilepsia, 2014, 55, e75-9.	2.6	36
86	Exome Sequencing as a Diagnostic Tool for Pediatricâ€Onset Ataxia. Human Mutation, 2014, 35, 45-49.	1.1	91
87	Identification of Genes for Childhood Heritable Diseases. Annual Review of Medicine, 2014, 65, 19-31.	5.0	30
88	Multiple sclerosis in the Iranian immigrant population of BC, Canada: prevalence and risk factors. Multiple Sclerosis Journal, 2014, 20, 1182-1188.	1.4	24
89	Loss-of-function HDAC8 mutations cause a phenotypic spectrum of Cornelia de Lange syndrome-like features, ocular hypertelorism, large fontanelle and X-linked inheritance. Human Molecular Genetics, 2014, 23, 2888-2900.	1.4	120
90	Biallelic mutations at PPARG cause a congenital, generalized lipodystrophy similar to the Berardinelliâ€“Seip syndrome. European Journal of Medical Genetics, 2014, 57, 524-526.	0.7	30

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91	FORGE Canada Consortium: Outcomes of a 2-Year National Rare-Disease Gene-Discovery Project. American Journal of Human Genetics, 2014, 94, 809-817.	2.6	219
92	Recent Advances in the Genetic Etiology of Brain Malformations. Current Neurology and Neuroscience Reports, 2013, 13, 364.	2.0	20
93	Identification of Novel Mutations ConfirmsPde4das a Major Gene Causing Acrodysostosis. Human Mutation, 2013, 34, 97-102.	1.1	49
94	Mutations in PIK3R1 Cause SHORT Syndrome. American Journal of Human Genetics, 2013, 93, 158-166.	2.6	156
95	Exome sequencing identifies a novel multiple sclerosis susceptibility variant in the TYK2 gene. Neurology, 2012, 79, 406-411.	1.5	56
96	Early life child exposure and the risk of multiple sclerosis: A population based study. Journal of the Neurological Sciences, 2011, 307, 162-163.	0.3	3
97	Rare variants in the CYP27B1 gene are associated with multiple sclerosis. Annals of Neurology, 2011, 70, 881-886.	2.8	204
98	HLAâ€DRB1*15, low infant sibling exposure, and multiple sclerosis geneâ€“environment interaction. Annals of Neurology, 2010, 67, 694-695.	2.8	5
99	Motherâ€“child blood group incompatibility and the risk of multiple sclerosis. Journal of Neurology, 2010, 257, 286-287.	1.8	1
100	Prevalence of MS in Iranian Immigrants to British Columbia, Canada. Journal of Neurology, 2010, 257, 667-668.	1.8	16
101	No Effect of Parental Age on Risk of Multiple Sclerosis: A Population-Based Study. Neuroepidemiology, 2010, 34, 106-109.	1.1	10
102	A genome-wide scan of male sexual orientation. Journal of Human Genetics, 2010, 55, 131-132.	1.1	25
103	Childhood cow's milk allergy and the risk of multiple sclerosis: A population based study. Journal of the Neurological Sciences, 2010, 291, 86-88.	0.3	8
104	Epistasis amongHLA-DRB1, HLA-DQA1,andHLA-DQB1loci determines multiple sclerosis susceptibility. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 7542-7547.	3.3	148
105	Age of Onset in Concordant Twins and Other Relative Pairs With Multiple Sclerosis. American Journal of Epidemiology, 2009, 170, 289-296.	1.6	26
106	Association of Infectious Mononucleosis with Multiple Sclerosis. Neuroepidemiology, 2009, 32, 257-262.	1.1	85
107	Parent-of-origin of HLA-DRB1*1501 and age of onset of multiple sclerosis. Journal of Human Genetics, 2009, 54, 547-549.	1.1	19
108	Expression of the Multiple Sclerosis-Associated MHC Class II Allele HLA-DRB1*1501 Is Regulated by Vitamin D. PLoS Genetics, 2009, 5, e1000369.	1.5	442

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109	Analysis of 45 candidate genes for disease modifying activity in multiple sclerosis. Journal of Neurology, 2008, 255, 1215-1219.	1.8	19
110	Parental transmission of HLA-DRB1*15 in multiple sclerosis. Human Genetics, 2008, 122, 661-663.	1.8	47
111	Parental non-inherited HLA resistance alleles do not confer protection against multiple sclerosis. Journal of Neuroimmunology, 2008, 196, 170-172.	1.1	3
112	Mutations in the hemochromatosis gene and the clinical outcome of multiple sclerosis. Journal of Neuroimmunology, 2008, 203, 104-107.	1.1	12
113	Genetic epidemiology: the use of old and new tools for multiple sclerosis. Trends in Neurosciences, 2008, 31, 645-652.	4.2	57
114	No Effect of Birth Weight on the Risk of Multiple Sclerosis. Neuroepidemiology, 2008, 31, 181-184.	1.1	13
115	Autoimmune disease in patients with multiple sclerosis and their first-degree relatives: a nationwide cohort study in Denmark. Multiple Sclerosis Journal, 2008, 14, 1288-1289.	1.4	3
116	HLA class I alleles tag HLA-DRB1 * 1501 haplotypes for differential risk in multiple sclerosis susceptibility. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 13069-13074.	3.3	86
117	Epigenetics in multiple sclerosis susceptibility: difference in transgenerational risk localizes to the major histocompatibility complex. Human Molecular Genetics, 2008, 18, 261-266.	1.4	89
118	Multiple sclerosis susceptibility and the X chromosome. Multiple Sclerosis Journal, 2007, 13, 856-864.	1.4	26
119	Transmission of class I/II multi-locus MHC haplotypes and multiple sclerosis susceptibility: accounting for linkage disequilibrium. Human Molecular Genetics, 2007, 16, 1951-1958.	1.4	33
120	No effect of APOE and PVRL2 on the clinical outcome of multiple sclerosis. Journal of Neuroimmunology, 2007, 186, 156-160.	1.1	8
121	Microsatellites and genome scans â€" A GAMES postscript. Journal of Neuroimmunology, 2007, 190, 5-7.	1.1	6
122	Clustering of autoimmune disease in families at high risk for multiple sclerosis?. Lancet Neurology, The, 2007, 6, 206-207.	4.9	14
123	A genome-wide scan in forty large pedigrees with multiple sclerosis. Journal of Human Genetics, 2007, 52, 955-962.	1.1	30
124	Maternal - offspring HLA-DRB1 compatibility in multiple sclerosis. Tissue Antigens, 2005, 66, 44-47.	1.0	6
125	A predominant role for the HLA class II region in the association of the MHC region with multiple sclerosis. Nature Genetics, 2005, 37, 1108-1112.	9.4	295
126	Complex interactions among MHC haplotypes in multiple sclerosis: susceptibility and resistance. Human Molecular Genetics, 2005, 14, 2019-2026.	1.4	212

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127	An extended genome scan in 442 Canadian multiple sclerosis-affected sibships: a report from the Canadian Collaborative Study Group. Human Molecular Genetics, 2004, 13, 1005-1015.	1.4	52
128	Genetics of multiple sclerosis. Lancet Neurology, The, 2004, 3, 104-110.	4.9	458
129	Parent-of-origin effect in multiple sclerosis: observations in half-siblings. Lancet, The, 2004, 363, 1773-1774.	6.3	249
130	Significant linkage to migraine with aura on chromosome 11q24. Human Molecular Genetics, 2003, 12, 2511-2517.	1.4	76
131	An Array of Sunshine in Multiple Sclerosis. New England Journal of Medicine, 2002, 347, 1445-1447.	13.9	27
132	No evidence to support CTLA-4 as a susceptibility gene in MS families: the Canadian Collaborative Study. Journal of Neuroimmunology, 2002, 123, 193-198.	1.1	44
133	Evidence of Linkage with HLA-DR in DRB1*15-Negative Families with Multiple Sclerosis. American Journal of Human Genetics, 2001, 69, 900-903.	2.6	72
134	Genetic susceptibility to MS: a second stage analysis in Canadian MS families. Neurogenetics, 2001, 3, 145-151.	0.7	40
135	Evidence for genetic basis of multiple sclerosis. Lancet, The, 1996, 347, 1728-1730.	6.3	321