Kym Boycott

List of Publications by Year
in descending order

188	10,472	41627	49824
papers	citations	51 h-index	91 g-index

194	194	194	18797
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	6.5	699
2	Rare-disease genetics in the era of next-generation sequencing: discovery to translation. Nature Reviews Genetics, 2013, 14, 681-691.	7.7	608
3	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. Human Mutation, 2015, 36, 915-921.	1.1	390
4	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
5	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	2.6	305
6	Biallelic Mutations in BRCA1 Cause a New Fanconi Anemia Subtype. Cancer Discovery, 2015, 5, 135-142.	7.7	251
7	SLC39A8 Deficiency: A Disorder of Manganese Transport and Glycosylation. American Journal of Human Genetics, 2015, 97, 894-903.	2.6	242
8	Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts. Nature Medicine, 2019, 25, 911-919.	15.2	221
9	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
10	An siRNA-based functional genomics screen for theÂidentification of regulators of ciliogenesis and ciliopathyÂgenes. Nature Cell Biology, 2015, 17, 1074-1087.	4.6	215
11	PhenoTips: Patient Phenotyping Software for Clinical and Research Use. Human Mutation, 2013, 34, 1057-1065.	1.1	207
12	Homozygous Deletion of the Very Low Density Lipoprotein Receptor Gene Causes Autosomal Recessive Cerebellar Hypoplasia with Cerebral Gyral Simplification. American Journal of Human Genetics, 2005, 77, 477-483.	2.6	192
13	The clinical application of genome-wide sequencing for monogenic diseases in Canada: Position Statement of the Canadian College of Medical Geneticists. Journal of Medical Genetics, 2015, 52, 431-437.	1.5	187
14	Autosomal-Recessive Intellectual Disability with Cerebellar Atrophy Syndrome Caused by Mutation of the Manganese and Zinc Transporter Gene SLC39A8. American Journal of Human Genetics, 2015, 97, 886-893.	2.6	171
15	Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research. Genetics, 2017, 207, 9-27.	1.2	165
16	Mutations in SRCAP, Encoding SNF2-Related CREBBP Activator Protein, Cause Floating-Harbor Syndrome. American Journal of Human Genetics, 2012, 90, 308-313.	2.6	157
17	Future of Rare Diseases Research 2017â€“2027: An IRDiRC Perspective. Clinical and Translational Science, 2018, 11, 21-27.	1.5	154
18	Genomic DNA Methylation Signatures Enable Concurrent Diagnosis and Clinical Genetic Variant Classification in Neurodevelopmental Syndromes. American Journal of Human Genetics, 2018, 102, 156-174.	2.6	135

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19	Missense mutations in ITPR1 cause autosomal dominant congenital nonprogressive spinocerebellar ataxia. Orphanet Journal of Rare Diseases, 2012, 7, 67.	1.2	124
20	Mutations in C5ORF42 Cause Joubert Syndrome in the French Canadian Population. American Journal of Human Genetics, 2012, 90, 693-700.	2.6	118
21	De novo CCND2 mutations leading to stabilization of cyclin D2 cause megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome. Nature Genetics, 2014, 46, 510-515.	9.4	118
22	De Novo Mutations in the Motor Domain of KIF1A Cause Cognitive Impairment, Spastic Paraparesis, Axonal Neuropathy, and Cerebellar Atrophy. Human Mutation, 2015, 36, 69-78.	1.1	114
23	DNM1L-related mitochondrial fission defect presenting as refractory epilepsy. European Journal of Human Genetics, 2016, 24, 1084-1088.	1.4	113
24	A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. Cell, 2019, 177, 32-37.	13.5	113
25	Neu-Laxova Syndrome Is a Heterogeneous Metabolic Disorder Caused by Defects in Enzymes of the L-Serine Biosynthesis Pathway. American Journal of Human Genetics, 2014, 95, 285-293.	2.6	110
26	PhenomeCentral: A Portal for Phenotypic and Genotypic Matchmaking of Patients with Rare Genetic Diseases. Human Mutation, 2015, 36, 931-940.	1.1	107
27	Progress in Rare Diseases Research 2010â€“2016: An IRDiRC Perspective. Clinical and Translational Science, 2018, 11, 11-20.	1.5	104
28	A Mild PUM1 Mutation Is Associated with Adult-Onset Ataxia, whereas Haploinsufficiency Causes Developmental Delay and Seizures. Cell, 2018, 172, 924-936.e11.	13.5	103
29	A summary of 20 CACNA1F mutations identified in 36 families with incomplete X-linked congenital stationary night blindness, and characterization of splice variants. Human Genetics, 2001, 108, 91-97.	1.8	99
30	Mutations in LAMA1 Cause Cerebellar Dysplasia and Cysts with and without Retinal Dystrophy. American Journal of Human Genetics, 2014, 95, 227-234.	2.6	92
31	Debunking Occam's razor: Diagnosing multiple genetic diseases in families by wholeâ€exome sequencing. Clinical Genetics, 2017, 92, 281-289.	1.0	92
32	Mosaic Activating Mutations in FGFR1 Cause Encephalocraniocutaneous Lipomatosis. American Journal of Human Genetics, 2016, 98, 579-587.	2.6	88
33	Next-generation sequencing for diagnosis of rare diseases in the neonatal intensive care unit. Cmaj, 2016, 188, E254-E260.	0.9	86
34	BAFopathiesâ€™ DNA methylation epi-signatures demonstrate diagnostic utility and functional continuum of Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes. Nature Communications, 2018, 9, 4885.	5.8	83
35	Attitudes of parents toward the return of targeted and incidental genomic research findings in children. Genetics in Medicine, 2014, 16, 633-640.	1.1	82
36	Pyridoxine-Dependent Epilepsy in Zebrafish Caused by Aldh7a1 Deficiency. Genetics, 2017, 207, 1501-1518.	1.2	81

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37	Wholeâ€exome sequencing broadens the phenotypic spectrum of rare pediatric epilepsy: a retrospective study. Clinical Genetics, 2015, 88, 34-40.	1.0	79
38	Mutations in CSPP1, Encoding a Core Centrosomal Protein, Cause a Range of Ciliopathy Phenotypes in Humans. American Journal of Human Genetics, 2014, 94, 73-79.	2.6	77
39	Returning incidental findings from genetic research to children: views of parents of children affected by rare diseases. Journal of Medical Ethics, 2014, 40, 691-696.	1.0	75
40	Receptor tyrosine kinase mutations in developmental syndromes and cancer: two sides of the same coin. Human Molecular Genetics, 2015, 24, R60-R66.	1.4	70
41	PLPHP deficiency: clinical, genetic, biochemical, and mechanistic insights. Brain, 2019, 142, 542-559.	3.7	67
42	Identification of a methylation profile for DNMT1-associated autosomal dominant cerebellar ataxia, deafness, and narcolepsy. Clinical Epigenetics, 2016, 8, 91.	1.8	66
43	Consent Codes: Upholding Standard Data Use Conditions. PLoS Genetics, 2016, 12, e1005772.	1.5	65
44	Targeted exome analysis identifies the genetic basis of disease in over 50% of patients with a wide range of ataxia-related phenotypes. Genetics in Medicine, 2019, 21, 195-206.	1.1	65
45	Mutations in the enzyme glutathione peroxidase 4 cause Sedaghatian-type spondylometaphyseal dysplasia. Journal of Medical Genetics, 2014, 51, 470-474.	1.5	64
46	The International Rare Diseases Research Consortium: Policies and Guidelines to maximize impact. European Journal of Human Genetics, 2017, 25, 1293-1302.	1.4	62
47	Homozygous mutations inMFN2cause multiple symmetric lipomatosis associated with neuropathy. Human Molecular Genetics, 2015, 24, 5109-5114.	1.4	61
48	Mutations in VLDLR as a Cause for Autosomal Recessive Cerebellar Ataxia With Mental Retardation (Dysequilibrium Syndrome). Journal of Child Neurology, 2009, 24, 1310-1315.	0.7	60
49	Identification of epigenetic signature associated with alpha thalassemia/mental retardation X-linked syndrome. Epigenetics and Chromatin, 2017, 10, 10.	1.8	60
50	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994.	2.6	59
51	New Diagnostic Approaches for Undiagnosed Rare Genetic Diseases. Annual Review of Genomics and Human Genetics, 2020, 21, 351-372.	2.5	58
52	Joubert Syndrome in French Canadians and Identification of Mutations in CEP104. American Journal of Human Genetics, 2015, 97, 744-753.	2.6	56
53	The defining DNA methylation signature of Floating-Harbor Syndrome. Scientific Reports, 2016, 6, 38803.	1.6	55
54	The role of the clinician in the multiâ€omics era: are you ready?. Journal of Inherited Metabolic Disease, 2018, 41, 571-582.	1.7	55

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55	PDXK mutations cause polyneuropathy responsive to pyridoxal 5â€²â€phosphate supplementation. Annals of Neurology, 2019, 86, 225-240.	2.8	54
56	Homozygous nonsense mutation in SYNJ1 associated with intractable epilepsy and tau pathology. Neurobiology of Aging, 2015, 36, 1222.e1-1222.e5.	1.5	50
57	Biallelic Mutations in LRRC56, Encoding a Protein Associated with Intraflagellar Transport, Cause Mucociliary Clearance and Laterality Defects. American Journal of Human Genetics, 2018, 103, 727-739.	2.6	49
58	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
59	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	2.6	48
60	Congenital Visual Impairment and Progressive Microcephaly Due to Lysylâ€“Transfer Ribonucleic Acid (RNA) Synthetase (KARS) Mutations. Journal of Child Neurology, 2015, 30, 1037-1043.	0.7	47
61	Matchmaker Exchange. Current Protocols in Human Genetics, 2017, 95, 9.31.1-9.31.15.	3.5	47
62	SPG7 mutations explain a significant proportion of French Canadian spastic ataxia cases. European Journal of Human Genetics, 2016, 24, 1016-1021.	1.4	46
63	A de novo non-sense mutation in ZBTB18 in a patient with features of the 1q43q44 microdeletion syndrome. European Journal of Human Genetics, 2014, 22, 844-846.	1.4	45
64	Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. Human Mutation, 2016, 37, 148-154.	1.1	45
65	Clinical genetics and the Hutterite population: A review of Mendelian disorders. American Journal of Medical Genetics, Part A, 2008, 146A, 1088-1098.	0.7	44
66	Germline AGO2 mutations impair RNA interference and human neurological development. Nature Communications, 2020, 11, 5797.	5.8	43
67	Evidence for clinical, genetic and biochemical variability in spinal muscular atrophy with progressive myoclonic epilepsy. Clinical Genetics, 2014, 86, 558-563.	1.0	42
68	Spinocerebellar ataxia type 29 due to mutations in ITPR1: a case series and review of this emerging congenital ataxia. Orphanet Journal of Rare Diseases, 2017, 12, 121.	1.2	42
69	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
70	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
71	Compound heterozygous mutations in the gene PIGP are associated with early infantile epileptic encephalopathy. Human Molecular Genetics, 2017, 26, 1706-1715.	1.4	39
72	Homozygous mutation in the eukaryotic translation initiation factor 2alpha phosphatase gene,PPP1R15B, is associated with severe microcephaly, short stature and intellectual disability. Human Molecular Genetics, 2015, 24, 6293-6300.	1.4	36

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73	A novel multisystem disease associated with recessive mutations in the tyrosylâ€tRNA synthetase (YARS) gene. American Journal of Medical Genetics, Part A, 2017, 173, 126-134.	0.7	36
74	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	2.6	36
75	Addressing challenges in the diagnosis and treatment of rare genetic diseases. Nature Reviews Drug Discovery, 2018, 17, 151-152.	21.5	34
76	Cost-effectiveness of genome-wide sequencing for unexplained developmental disabilities and multiple congenital anomalies. Genetics in Medicine, 2021, 23, 451-460.	1.1	34
77	Registered access: authorizing data access. European Journal of Human Genetics, 2018, 26, 1721-1731.	1.4	33
78	Matching Two Independent Cohorts ValidatesDPH1as a Gene Responsible for Autosomal Recessive Intellectual Disability with Short Stature, Craniofacial, and Ectodermal Anomalies. Human Mutation, 2015, 36, 1015-1019.	1.1	32
79	Localization of a gene for incomplete X-linked congenital stationary night blindness to the interval between DXS6849 and DXS8023 in Xp11.23. Human Genetics, 1998, 103, 124-130.	1.8	31
80	MCM3AP in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability. Brain, 2017, 140, 2093-2103.	3.7	31
81	The value of diagnostic testing for parents of children with rare genetic diseases. Genetics in Medicine, 2019, 21, 2798-2806.	1.1	31
82	A call for global action for rare diseases in Africa. Nature Genetics, 2020, 52, 21-26.	9.4	31
83	Identification of Genes for Childhood Heritable Diseases. Annual Review of Medicine, 2014, 65, 19-31.	5.0	30
84	Concordance between wholeâ€exome sequencing and clinical Sanger sequencing: implications for patient care. Molecular Genetics & Genomic Medicine, 2016, 4, 504-512.	0.6	30
85	â€˜IRDiRC Recognized Resourcesâ€™: a new mechanism to support scientists to conduct efficient, high-quality research for rare diseases. European Journal of Human Genetics, 2017, 25, 162-165.	1.4	30
86	A family segregating lethal neonatal coenzyme Q₁₀ deficiency caused by mutations in COQ9. Journal of Inherited Metabolic Disease, 2018, 41, 719-729.	1.7	30
87	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
88	The Deep Genome Project. Genome Biology, 2020, 21, 18.	3.8	30
89	Whole exome sequencing identifies the TNNI3K gene as a cause of familial conduction system disease and congenital junctional ectopic tachycardia. International Journal of Cardiology, 2015, 185, 114-116.	0.8	29
90	Motherâ€toâ€daughter transmission of Kennyâ€“Caffey syndrome associated with the recurrent, dominant <scp>FAM111A</scp> mutation p.<scp>Arg569His</scp>. Clinical Genetics, 2014, 86, 394-395.	1.0	28

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91	Autosomal recessive mutations in <scp>THOC6</scp> cause intellectual disability: syndrome delineation requiring forward and reverse phenotyping. Clinical Genetics, 2017, 91, 92-99.	1.0	28
92	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
93	Autosomal recessive cerebellar ataxia caused by a homozygous mutation inPMPCA. Brain, 2016, 139, e19-e19.	3.7	27
94	When One Diagnosis Is Not Enough. New England Journal of Medicine, 2017, 376, 83-85.	13.9	27
95	Novel ELOVL4 mutation associated with erythrokeratodermia and spinocerebellar ataxia (SCA) Tj ETQq1 1 0.784314 rgBT /Ove	0.9	27
96	Mutations in glycyl-tRNA synthetase impair mitochondrial metabolism in neurons. Human Molecular Genetics, 2018, 27, 2187-2204.	1.4	26
97	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
98	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
99	The unsolved rare genetic disease atlas? An analysis of the unexplained phenotypic descriptions in OMIMÂ®. , 2018, 178, 458-463.		25
100	Meconium ileus in a Lebanese family secondary to mutations in the GUCY2C gene. European Journal of Human Genetics, 2015, 23, 990-992.	1.4	24
101	International collaborative actions and transparency to understand, diagnose, and develop therapies for rare diseases. EMBO Molecular Medicine, 2019, 11, .	3.3	24
102	LIMS2 mutations are associated with a novel muscular dystrophy, severe cardiomyopathy and triangular tongues. Clinical Genetics, 2015, 88, 558-564.	1.0	23
103	Loss of the arginine methyltranserase PRMT7 causes syndromic intellectual disability with microcephaly and brachydactyly. Clinical Genetics, 2017, 91, 708-716.	1.0	23
104	Phenotype delineation of ZNF462 related syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2075-2082.	0.7	23
105	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
106	Yunis-VarÃ³n syndrome caused by biallelic VAC14 mutations. European Journal of Human Genetics, 2017, 25, 1049-1054.	1.4	21
107	Phenotype and mutation expansion of the PTPN23 associated disorder characterized by neurodevelopmental delay and structural brain abnormalities. European Journal of Human Genetics, 2020, 28, 76-87.	1.4	21
108	Meckel syndrome in the Hutterite population is actually a Joubert-related cerebello-oculo-renal syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1715-1725.	0.7	20

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109	Improved Diagnosis and Care for Rare Diseases through Implementation of Precision Public Health Framework. Advances in Experimental Medicine and Biology, 2017, 1031, 55-94.	0.8	20
110	Association of Early-Onset Spasticity and Risk for Cognitive Impairment With Mutations at Amino Acid 499 in SPAST. Journal of Child Neurology, 2018, 33, 329-332.	0.7	20
111	Direct health-care costs for children diagnosed with genetic diseases are significantly higher than for children with other chronic diseases. Genetics in Medicine, 2019, 21, 1049-1057.	1.1	20
112	Recessive, Deleterious Variants in SMG8 Expand the Role of Nonsense-Mediated Decay in Developmental Disorders in Humans. American Journal of Human Genetics, 2020, 107, 1178-1185.	2.6	20
113	Adult siblings with homozygous G6PC3 mutations expand our understanding of the severe congenital neutropenia type 4 (SCN4) phenotype. BMC Medical Genetics, 2012, 13, 111.	2.1	19
114	Lateral meningocele (Lehman) syndrome: A child with a novel NOTCH3 mutation. American Journal of Medical Genetics, Part A, 2016, 170, 1070-1075.	0.7	19
115	Syndrome disintegration: Exome sequencing reveals that Fitzsimmons syndrome is a coâ€occurrence of multiple events. American Journal of Medical Genetics, Part A, 2016, 170, 1820-1825.	0.7	19
116	The utility of exome sequencing for genetic diagnosis in a familial microcephaly epilepsy syndrome. BMC Neurology, 2014, 14, 22.	0.8	18
117	Development of Criteria for Epilepsy Genetic Testing in Ontario, Canada. Canadian Journal of Neurological Sciences, 2019, 46, 7-13.	0.3	18
118	A founder mutation in BBS2 is responsible for Bardetâ€Biedl syndrome in the Hutterite population: utility of SNP arrays in genetically heterogeneous disorders. Clinical Genetics, 2010, 78, 424-431.	1.0	17
119	Novel 25 kb Deletion of MERTK Causes Retinitis Pigmentosa With Severe Progression. , 2017, 58, 1736.		17
120	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
121	Complex genomic rearrangements in the dystrophin gene due to replicationâ€based mechanisms. Molecular Genetics & Genomic Medicine, 2014, 2, 539-547.	0.6	16
122	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
123	Mutations in GALC cause late-onset Krabbe disease with predominant cerebellar ataxia. Neurogenetics, 2016, 17, 137-141.	0.7	16
124	Expansion of the <scp>GLE1</scp>â€associated arthrogryposis multiplex congenita clinical spectrum. Clinical Genetics, 2017, 91, 426-430.	1.0	15
125	A novel pathogenic variant in TNPO3 in a Hungarian family with limb-girdle muscular dystrophy 1F. European Journal of Medical Genetics, 2019, 62, 103662.	0.7	15
126	Neuâ€“Laxova syndrome presenting prenatally with increased nuchal translucency and cystic hygroma: The utility of exome sequencing in deciphering the diagnosis. American Journal of Medical Genetics, Part A, 2019, 179, 813-816.	0.7	15

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127	Assessing non-Mendelian inheritance in inherited axonopathies. Genetics in Medicine, 2020, 22, 2114-2119.	1.1	15
128	Early infantile epileptic encephalopathy due to biallelic pathogenic variants in <scp>PIGQ</scp>: Report of seven new subjects and review of the literature. Journal of Inherited Metabolic Disease, 2020, 43, 1321-1332.	1.7	15
129	Outcome of over 1500 matches through the Matchmaker Exchange for rare disease gene discovery: The 2-year experience of Care4Rare Canada. Genetics in Medicine, 2022, 24, 100-108.	1.1	15
130	Identification of a Recognizable Progressive Skeletal Dysplasia Caused by RSPRY1 Mutations. American Journal of Human Genetics, 2015, 97, 608-615.	2.6	14
131	â€œMatchingâ€•consent to purpose: The example of the Matchmaker Exchange. Human Mutation, 2017, 38, 1281-1285.	1.1	13
132	Evaluation of exome filtering techniques for the analysis of clinically relevant genes. Human Mutation, 2018, 39, 197-201.	1.1	13
133	Unsolved recognizable patterns of human malformation: Challenges and opportunities. , 2018, 178, 382-386.		13
134	Is PNPT1â€related hearing loss ever nonâ€syndromic? Whole exome sequencing of adult siblings expands the natural history of PNPT1â€related disorders. American Journal of Medical Genetics, Part A, 2018, 176, 2487-2493.	0.7	13
135	p21 proteinâ€activated kinase 1 is associated with severe regressive autism, and epilepsy. Clinical Genetics, 2019, 96, 449-455.	1.0	13
136	Searching for secondary findings: considering actionability and preserving the right not to know. European Journal of Human Genetics, 2019, 27, 1481-1484.	1.4	13
137	Novel variants in TUBA1A cause congenital fibrosis of the extraocular muscles with or without malformations of cortical brain development. European Journal of Human Genetics, 2021, 29, 816-826.	1.4	13
138	Clinical application of fetal genome-wide sequencing during pregnancy: position statement of the Canadian College of Medical Geneticists. Journal of Medical Genetics, 2022, 59, 931-937.	1.5	13
139	A novel autosomal recessive malformation syndrome associated with developmental delay and distinctive facies maps to 16ptel in the Hutterite population. American Journal of Medical Genetics, Part A, 2010, 152A, 1349-1356.	0.7	12
140	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
141	A Novel Mutation in MARS in a Patient with Charcot-Marie-Tooth Disease, Axonal, Type 2U with Congenital Onset. Journal of Neuromuscular Diseases, 2019, 6, 333-339.	1.1	12
142	Compound heterozygous variants in SHQ1 are associated with a spectrum of neurological features, including early-onset dystonia. Human Molecular Genetics, 2022, 31, 614-624.	1.4	12
143	Atypical fibrodysplasia ossificans progressiva diagnosed by wholeâ€exome sequencing. American Journal of Medical Genetics, Part A, 2015, 167, 1337-1341.	0.7	11
144	Late diagnosis of cerebral folate deficiency: Fewer seizures with folinic acid in adult siblings. Neurology: Genetics, 2016, 2, e38.	0.9	11

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145	Lysosomal dysfunction in TMEM106B hypomyelinating leukodystrophy. Neurology: Genetics, 2018, 4, e288.	0.9	11
146	Infantile Myofibromatosis With Intracranial Extradural Involvement and PDGFRB Mutation: A Case Report and Review of the Literature. Pediatric and Developmental Pathology, 2019, 22, 258-264.	0.5	11
147	Clinical delineation of GTPBP2â€associated neuroâ€ectodermal syndrome: Report of two new families and review of the literature. Clinical Genetics, 2019, 95, 601-606.	1.0	11
148	When to think outside the autozygome: Best practices for exome sequencing in â€œconsanguineousâ€• families. Clinical Genetics, 2020, 97, 835-843.	1.0	11
149	Evidence for Nonâ€Mendelian Inheritance in Spastic Paraplegia 7. Movement Disorders, 2021, 36, 1664-1675.	2.2	11
150	Deleterious variants in CRLS1 lead to cardiolipin deficiency and cause an autosomal recessive multi-system mitochondrial disease. Human Molecular Genetics, 2022, 31, 3597-3612.	1.4	11
151	Seven years since the launch of the Matchmaker Exchange: The evolution of genomic matchmaking. Human Mutation, 2022, 43, 659-667.	1.1	11
152	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
153	Genome-wide sequencing technologies: A primer for paediatricians. Paediatrics and Child Health, 2018, 23, 191-197.	0.3	10
154	Intrafamilial variability of limb-girdle muscular dystrophy, LGMD1D type. European Journal of Medical Genetics, 2020, 63, 103655.	0.7	10
155	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
156	Nablus maskâ€like facial syndrome: Deletion of chromosome 8q22.1 is necessary but not sufficient to cause the phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 2091-2099.	0.7	9
157	Severe connective tissue laxity including aortic dilatation in Sotos syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 531-535.	0.7	9
158	ABHD16A deficiency causes a complicated form of hereditary spastic paraplegia associated with intellectual disability and cerebral anomalies. American Journal of Human Genetics, 2021, 108, 2017-2023.	2.6	9
159	PhenomeCentral: 7 years of rare disease matchmaking. Human Mutation, 2022, , .	1.1	9
160	Understanding Rare Disease Pathogenesis: A Grand Challenge for Model Organisms. Genetics, 2014, 198, 443-445.	1.2	8
161	Wholeâ€exome sequencing expands the phenotype of Hunter syndrome. Clinical Genetics, 2014, 86, 172-176.	1.0	7
162	Fragile X testing as a second-tier test. Genetics in Medicine, 2017, 19, 1380-1380.	1.1	7

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163	H3.1 K36M mutation in a congenitalâ€onset soft tissue neoplasm. Pediatric Blood and Cancer, 2017, 64, e26633.	0.8	7
164	Mosaic KRAS mutation in a patient with encephalocraniocutaneous lipomatosis and renovascular hypertension. American Journal of Medical Genetics, Part A, 2018, 176, 2523-2527.	0.7	7
165	Channelopathies Are a Frequent Cause of Genetic Ataxias Associated with Cerebellar Atrophy. Movement Disorders Clinical Practice, 2020, 7, 940-949.	0.8	7
166	Genetic, structural and clinical analysis of spastic paraplegia 4. Parkinsonism and Related Disorders, 2022, 98, 62-69.	1.1	7
167	Implementation of Epilepsy Multigene Panel Testing in Ontario, Canada. Canadian Journal of Neurological Sciences, 2020, 47, 61-68.	0.3	6
168	A DNA repair disorder caused by de novo monoallelic DDB1 variants is associated with a neurodevelopmental syndrome. American Journal of Human Genetics, 2021, 108, 749-756.	2.6	6
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