Kym Boycott

List of Publications by Year
in descending order

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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	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
2	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
3	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
4	Heterozygous De Novo <scp>KPNA3</scp> Mutations Cause Complex Hereditary Spastic Paraplegia. Annals of Neurology, 2022, 91, 730-732.	2.8	1
5	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
6	The complexity of diagnosing rare disease: An organizing framework for outcomes research and health economics based on real-world evidence. Genetics in Medicine, 2022, 24, 694-702.	1.1	4
7	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
8	PhenomeCentral: 7 years of rare disease matchmaking. Human Mutation, 2022, , .	1.1	9
9	ModelMatcher: A scientistâ€centric online platform to facilitate collaborations between stakeholders of rare and undiagnosed disease research. Human Mutation, 2022, , .	1.1	5
10	Genetic, structural and clinical analysis of spastic paraplegia 4. Parkinsonism and Related Disorders, 2022, 98, 62-69.	1.1	7
11	Biallelic Variants in the Ectonucleotidase <scp>ENTPD1</scp> Cause a Complex Neurodevelopmental Disorder with Intellectual Disability, Distinct White Matter Abnormalities, and Spastic Paraplegia. Annals of Neurology, 2022, 92, 304-321.	2.8	2
12	Genomics4RD: An integrated platform to share Canadian deep-phenotype and multiomic data for international rare disease gene discovery.. Human Mutation, 2022, , .	1.1	4
13	Seven years since the launch of the Matchmaker Exchange: The evolution of genomic matchmaking. Human Mutation, 2022, 43, 659-667.	1.1	11
14	A novel intragenic DPF2 deletion identified by genome sequencing in an adult with clinical features of <scp>Coffinâ€Siris</scp> syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 2493-2496.	0.7	3
15	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
16	Cost-effectiveness of genome-wide sequencing for unexplained developmental disabilities and multiple congenital anomalies. Genetics in Medicine, 2021, 23, 451-460.	1.1	34
17	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
18	Evidence for Nonâ€Mendelian Inheritance in Spastic Paraplegia 7. Movement Disorders, 2021, 36, 1664-1675.	2.2	11

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19	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
20	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
21	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
22	Homozygous <scp>WNT9B</scp> variants in two families with bilateral renal agenesis/hypoplasia/dysplasia. American Journal of Medical Genetics, Part A, 2021, 185, 3005-3011.	0.7	5
23	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
24	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
25	Neurophysiological Characteristics of Allgrove (Triple A) Syndrome: Case Report and Literature Review. Child Neurology Open, 2021, 8, 2329048X2110310.	0.5	1
26	Correspondence on â€œcost or price of sequencing? implications for economic evaluations in genomic medicineâ€•by Grosse and Gudgeon. Genetics in Medicine, 2021, , .	1.1	0
27	Intrafamilial variability of limb-girdle muscular dystrophy, LGMD1D type. European Journal of Medical Genetics, 2020, 63, 103655.	0.7	10
28	Implementation of Epilepsy Multigene Panel Testing in Ontario, Canada. Canadian Journal of Neurological Sciences, 2020, 47, 61-68.	0.3	6
29	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
30	A call for global action for rare diseases in Africa. Nature Genetics, 2020, 52, 21-26.	9.4	31
31	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
32	Germline AGO2 mutations impair RNA interference and human neurological development. Nature Communications, 2020, 11, 5797.	5.8	43
33	Assessing non-Mendelian inheritance in inherited axonopathies. Genetics in Medicine, 2020, 22, 2114-2119.	1.1	15
34	Channelopathies Are a Frequent Cause of Genetic Ataxias Associated with Cerebellar Atrophy. Movement Disorders Clinical Practice, 2020, 7, 940-949.	0.8	7
35	<scp>SMG9</scp>â€deficiency syndrome caused by a homozygous missense variant: Expanding the genotypic and phenotypic spectrum of this developmental disorder. American Journal of Medical Genetics, Part A, 2020, 182, 1829-1831.	0.7	5
36	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

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37	When to think outside the autozygome: Best practices for exome sequencing in â€œconsanguineousâ€• families. Clinical Genetics, 2020, 97, 835-843.	1.0	11
38	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
39	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
40	The Deep Genome Project. Genome Biology, 2020, 21, 18.	3.8	30
41	New Diagnostic Approaches for Undiagnosed Rare Genetic Diseases. Annual Review of Genomics and Human Genetics, 2020, 21, 351-372.	2.5	58
42	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
43	p21 proteinâ€activated kinase 1 is associated with severe regressive autism, and epilepsy. Clinical Genetics, 2019, 96, 449-455.	1.0	13
44	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
45	Phenotype delineation of ZNF462 related syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2075-2082.	0.7	23
46	PLPHP deficiency: clinical, genetic, biochemical, and mechanistic insights. Brain, 2019, 142, 542-559.	3.7	67
47	The value of diagnostic testing for parents of children with rare genetic diseases. Genetics in Medicine, 2019, 21, 2798-2806.	1.1	31
48	Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts. Nature Medicine, 2019, 25, 911-919.	15.2	221
49	PDXK mutations cause polyneuropathy responsive to pyridoxal 5â€²â€phosphate supplementation. Annals of Neurology, 2019, 86, 225-240.	2.8	54
50	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
51	International collaborative actions and transparency to understand, diagnose, and develop therapies for rare diseases. EMBO Molecular Medicine, 2019, 11, .	3.3	24
52	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
53	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
54	A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. Cell, 2019, 177, 32-37.	13.5	113

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55	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
56	39th Annual David W. Smith Workshop on Malformations and Morphogenesis: Abstracts of the 2018 Annual Meeting. American Journal of Medical Genetics, Part A, 2019, 179, 674-746.	0.7	4
57	Development of Criteria for Epilepsy Genetic Testing in Ontario, Canada. Canadian Journal of Neurological Sciences, 2019, 46, 7-13.	0.3	18
58	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
59	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
60	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
61	Mutations in glycyl-tRNA synthetase impair mitochondrial metabolism in neurons. Human Molecular Genetics, 2018, 27, 2187-2204.	1.4	26
62	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
63	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
64	Genome-wide sequencing technologies: A primer for paediatricians. Paediatrics and Child Health, 2018, 23, 191-197.	0.3	10
65	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
66	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
67	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
68	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
69	Evaluation of exome filtering techniques for the analysis of clinically relevant genes. Human Mutation, 2018, 39, 197-201.	1.1	13
70	Addressing challenges in the diagnosis and treatment of rare genetic diseases. Nature Reviews Drug Discovery, 2018, 17, 151-152.	21.5	34
71	Progress in Rare Diseases Research 2010â€“2016: An IRDiRC Perspective. Clinical and Translational Science, 2018, 11, 11-20.	1.5	104
72	Future of Rare Diseases Research 2017â€“2027: An IRDiRC Perspective. Clinical and Translational Science, 2018, 11, 21-27.	1.5	154

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73	Nablus syndrome: Easy to diagnose yet difficult to solve. , 2018, 178, 447-457.		2
74	Unsolved recognizable patterns of human malformation: Challenges and opportunities. , 2018, 178, 382-386.		13
75	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
76	Lysosomal dysfunction in TMEM106B hypomyelinating leukodystrophy. Neurology: Genetics, 2018, 4, e288.	0.9	11
77	The unsolved rare genetic disease atlas? An analysis of the unexplained phenotypic descriptions in OMIMÂ®. , 2018, 178, 458-463.		25
78	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
79	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
80	Novel ELOVL4 mutation associated with erythrokeratodermia and spinocerebellar ataxia (SCA) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	0.9	27
81	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
82	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	2.6	36
83	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
84	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
85	Registered access: authorizing data access. European Journal of Human Genetics, 2018, 26, 1721-1731.	1.4	33
86	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
87	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	6.5	699
88	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
89	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
90	Debunking Occam's razor: Diagnosing multiple genetic diseases in families by wholeâ€exome sequencing. Clinical Genetics, 2017, 92, 281-289.	1.0	92

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91	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
92	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
93	Expansion of the clinical phenotype of the distal 10q26.3 deletion syndrome to include ataxia and hyperemia of the hands and feet. American Journal of Medical Genetics, Part A, 2017, 173, 1611-1619.	0.7	4
94	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	2.6	305
95	â€œMatchingâ€•consent to purpose: The example of the Matchmaker Exchange. Human Mutation, 2017, 38, 1281-1285.	1.1	13
96	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
97	Yunis-VarÃ³n syndrome caused by biallelic VAC14 mutations. European Journal of Human Genetics, 2017, 25, 1049-1054.	1.4	21
98	MCM3AP in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability. Brain, 2017, 140, 2093-2103.	3.7	31
99	Compound heterozygous mutations in the gene PIGP are associated with early infantile epileptic encephalopathy. Human Molecular Genetics, 2017, 26, 1706-1715.	1.4	39
100	When One Diagnosis Is Not Enough. New England Journal of Medicine, 2017, 376, 83-85.	13.9	27
101	Matchmaker Exchange. Current Protocols in Human Genetics, 2017, 95, 9.31.1-9.31.15.	3.5	47
102	Fragile X testing as a second-tier test. Genetics in Medicine, 2017, 19, 1380-1380.	1.1	7
103	Pyridoxine-Dependent Epilepsy in Zebrafish Caused by Aldh7a1 Deficiency. Genetics, 2017, 207, 1501-1518.	1.2	81
104	Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research. Genetics, 2017, 207, 9-27.	1.2	165
105	H3.1 K36M mutation in a congenitalâ€onset soft tissue neoplasm. Pediatric Blood and Cancer, 2017, 64, e26633.	0.8	7
106	The International Rare Diseases Research Consortium: Policies and Guidelines to maximize impact. European Journal of Human Genetics, 2017, 25, 1293-1302.	1.4	62
107	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
108	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

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109	Identification of epigenetic signature associated with alpha thalassemia/mental retardation X-linked syndrome. Epigenetics and Chromatin, 2017, 10, 10.	1.8	60
110	Loss of the arginine methyltranserase PRMT7 causes syndromic intellectual disability with microcephaly and brachydactyly. Clinical Genetics, 2017, 91, 708-716.	1.0	23
111	Expansion of the <scp>GLE1</scp>â€associated arthrogryposis multiplex congenita clinical spectrum. Clinical Genetics, 2017, 91, 426-430.	1.0	15
112	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
113	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
114	Novel 25 kb Deletion of MERTK Causes Retinitis Pigmentosa With Severe Progression. , 2017, 58, 1736.		17
115	Consent Codes: Upholding Standard Data Use Conditions. PLoS Genetics, 2016, 12, e1005772.	1.5	65
116	Late diagnosis of cerebral folate deficiency: Fewer seizures with folinic acid in adult siblings. Neurology: Genetics, 2016, 2, e38.	0.9	11
117	Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. Human Mutation, 2016, 37, 148-154.	1.1	45
118	The defining DNA methylation signature of Floating-Harbor Syndrome. Scientific Reports, 2016, 6, 38803.	1.6	55
119	Mutations in GALC cause late-onset Krabbe disease with predominant cerebellar ataxia. Neurogenetics, 2016, 17, 137-141.	0.7	16
120	Identification of a methylation profile for DNMT1-associated autosomal dominant cerebellar ataxia, deafness, and narcolepsy. Clinical Epigenetics, 2016, 8, 91.	1.8	66
121	Development of a diagnostic <scp>DNA</scp> chip to screen for 30 autosomal recessive disorders in the Hutterite population. Molecular Genetics & Genomic Medicine, 2016, 4, 312-321.	0.6	5
122	Concordance between wholeâ€exome sequencing and clinical Sanger sequencing: implications for patient care. Molecular Genetics & Genomic Medicine, 2016, 4, 504-512.	0.6	30
123	Next-generation sequencing for diagnosis of rare diseases in the neonatal intensive care unit. Cmaj, 2016, 188, E254-E260.	0.9	86
124	Severe connective tissue laxity including aortic dilatation in Sotos syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 531-535.	0.7	9
125	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
126	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

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127	DNM1L-related mitochondrial fission defect presenting as refractory epilepsy. European Journal of Human Genetics, 2016, 24, 1084-1088.	1.4	113
128	SPG7 mutations explain a significant proportion of French Canadian spastic ataxia cases. European Journal of Human Genetics, 2016, 24, 1016-1021.	1.4	46
129	Autosomal recessive cerebellar ataxia caused by a homozygous mutation inPMPCA. Brain, 2016, 139, e19-e19.	3.7	27
130	Mosaic Activating Mutations in FGFR1 Cause Encephalocraniocutaneous Lipomatosis. American Journal of Human Genetics, 2016, 98, 579-587.	2.6	88
131	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
132	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
133	PhenomeCentral: A Portal for Phenotypic and Genotypic Matchmaking of Patients with Rare Genetic Diseases. Human Mutation, 2015, 36, 931-940.	1.1	107
134	Very lateâ€onset Sandhoff disease presenting as Kennedy Disease. Muscle and Nerve, 2015, 52, 1135-1136.	1.0	4
135	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
136	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
137	SLC39A8 Deficiency: A Disorder of Manganese Transport and Glycosylation. American Journal of Human Genetics, 2015, 97, 894-903.	2.6	242
138	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
139	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
140	LIMS2 mutations are associated with a novel muscular dystrophy, severe cardiomyopathy and triangular tongues. Clinical Genetics, 2015, 88, 558-564.	1.0	23
141	Biallelic Mutations in BRCA1 Cause a New Fanconi Anemia Subtype. Cancer Discovery, 2015, 5, 135-142.	7.7	251
142	Homozygous mutations inMFN2cause multiple symmetric lipomatosis associated with neuropathy. Human Molecular Genetics, 2015, 24, 5109-5114.	1.4	61
143	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
144	An siRNA-based functional genomics screen for theÂidentification of regulators of ciliogenesis and ciliopathyÂgenes. Nature Cell Biology, 2015, 17, 1074-1087.	4.6	215

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145	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
146	Atypical fibrodysplasia ossificans progressiva diagnosed by wholeâ€exome sequencing. American Journal of Medical Genetics, Part A, 2015, 167, 1337-1341.	0.7	11
147	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
148	Identification of a Recognizable Progressive Skeletal Dysplasia Caused by RSPRY1 Mutations. American Journal of Human Genetics, 2015, 97, 608-615.	2.6	14
149	Joubert Syndrome in French Canadians and Identification of Mutations in CEP104. American Journal of Human Genetics, 2015, 97, 744-753.	2.6	56
150	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
151	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. Human Mutation, 2015, 36, 915-921.	1.1	390
152	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
153	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
154	Wholeâ€exome sequencing broadens the phenotypic spectrum of rare pediatric epilepsy: a retrospective study. Clinical Genetics, 2015, 88, 34-40.	1.0	79
155	Homozygous nonsense mutation in SYNJ1 associated with intractable epilepsy and tau pathology. Neurobiology of Aging, 2015, 36, 1222.e1-1222.e5.	1.5	50
156	Evidence for clinical, genetic and biochemical variability in spinal muscular atrophy with progressive myoclonic epilepsy. Clinical Genetics, 2014, 86, 558-563.	1.0	42
157	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
158	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
159	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
160	Wholeâ€exome sequencing expands the phenotype of Hunter syndrome. Clinical Genetics, 2014, 86, 172-176.	1.0	7
161	Understanding Rare Disease Pathogenesis: A Grand Challenge for Model Organisms. Genetics, 2014, 198, 443-445.	1.2	8
162	Complex genomic rearrangements in the dystrophin gene due to replicationâ€based mechanisms. Molecular Genetics & Genomic Medicine, 2014, 2, 539-547.	0.6	16

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163	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
164	The utility of exome sequencing for genetic diagnosis in a familial microcephaly epilepsy syndrome. BMC Neurology, 2014, 14, 22.	0.8	18
165	Identification of Genes for Childhood Heritable Diseases. Annual Review of Medicine, 2014, 65, 19-31.	5.0	30
166	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
167	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
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