

Jennifer E Posey

List of Publications by Year in descending order

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Version: 2024-02-01

138
papers

5,020
citations

145106

33
h-index

139680

61
g-index

150
all docs

150
docs citations

150
times ranked

9142
citing authors

#	ARTICLE	IF	CITATIONS
1	Biallelic variants in <i>SLC38A3</i> encoding a glutamine transporter cause epileptic encephalopathy. <i>Brain</i> , 2022, 145, 909-924.	3.7	17
2	Genetic counselor roles in the undiagnosed diseases network research study: Clinical care, collaboration, and curation. <i>Journal of Genetic Counseling</i> , 2022, 31, 326-337.	0.9	1
3	Advances in Next-Generation Sequencing Technologies and Functional Investigation of Candidate Variants in Neurological and Behavioral Disorders. , 2022, , 390-404.		0
4	Elucidating the clinical spectrum and molecular basis of <i>HYAL2</i> deficiency. <i>Genetics in Medicine</i> , 2022, 24, 631-644.	1.1	0
5	Retrospective analysis of a clinical exome sequencing cohort reveals the mutational spectrum and identifies candidate disease-associated loci for BAFopathies. <i>Genetics in Medicine</i> , 2022, 24, 364-373.	1.1	12
6	Expanding the phenotypic and allelic spectrum of <i>SMG8</i> : Clinical observations reveal overlap with <i>SMG9</i> -associated disease trait. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 648-657.	0.7	3
7	Quantitative dissection of multilocus pathogenic variation in an Egyptian infant with severe neurodevelopmental disorder resulting from multiple molecular diagnoses. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 735-750.	0.7	14
8	The clinical and molecular spectrum of <i>QRICH1</i> associated neurodevelopmental disorder. <i>Human Mutation</i> , 2022, 43, 266-282.	1.1	7
9	Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100074.	1.0	14
10	Centers for Mendelian Genomics: A decade of facilitating gene discovery. <i>Genetics in Medicine</i> , 2022, 24, 784-797.	1.1	44
11	Biallelic pathogenic variants in roundabout guidance receptor 1 associate with syndromic congenital anomalies of the kidney and urinary tract. <i>Kidney International</i> , 2022, 101, 1039-1053.	2.6	8
12	Expanding the mutation and phenotype spectrum of MYH3-associated skeletal disorders. <i>Npj Genomic Medicine</i> , 2022, 7, 11.	1.7	7
13	<i>El-Hattab-Alkuraya</i> syndrome caused by biallelic <i>WDR45B</i> pathogenic variants: Further delineation of the phenotype and genotype. <i>Clinical Genetics</i> , 2022, 101, 530-540.	1.0	7
14	Novel <i>RETREG1</i> (<i>FAM134B</i>) founder allele is linked to <i>HSAN2B</i> and renal disease in a Turkish family. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2153-2161.	0.7	4
15	Variant-level matching for diagnosis and discovery: Challenges and opportunities. <i>Human Mutation</i> , 2022, , .	1.1	11
16	Biallelic Variants in the Ectonucleotidase <i>ENTPD1</i> Cause a Complex Neurodevelopmental Disorder with Intellectual Disability, Distinct White Matter Abnormalities, and Spastic Paraplegia. <i>Annals of Neurology</i> , 2022, 92, 304-321.	2.8	2
17	MO047: Biallelic pathogenic variants in <i>ROBO1</i> associate with syndromic CAKUT. <i>Nephrology Dialysis Transplantation</i> , 2022, 37, .	0.4	0
18	<i>De novo</i> heterozygous variants in <i>SLC30A7</i> are a candidate cause for Joubert syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2360-2366.	0.7	3

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19	Diagnostic yield and clinical impact of exome sequencing in early-onset scoliosis (EOS). <i>Journal of Medical Genetics</i> , 2021, 58, 41-47.	1.5	40
20	Dominant mitochondrial membrane protein-associated neurodegeneration (MPAN) variants cluster within a specific C19orf12 isoform. <i>Parkinsonism and Related Disorders</i> , 2021, 82, 84-86.	1.1	10
21	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 119-133.	0.7	17
22	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. <i>Genetics in Medicine</i> , 2021, 23, 259-271.	1.1	18
23	<sc><i>MED27</i></sc> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. <i>Annals of Neurology</i> , 2021, 89, 828-833.	2.8	14
24	Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. <i>Genetics in Medicine</i> , 2021, 23, 1075-1085.	1.1	16
25	Neurodevelopmental disorder in an Egyptian family with a biallelic <sc><i>ALKBH8</i></sc> variant. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1288-1293.	0.7	13
26	Perturbations of genes essential for MÅ¼llerian duct and WÅ¼lfian duct development in Mayer-Rokitansky-KÅ¼ster-Hauser syndrome. <i>American Journal of Human Genetics</i> , 2021, 108, 337-345.	2.6	41
27	Clinical characterization of individuals with the distal 1q21.1 microdeletion. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1388-1398.	0.7	6
28	Phenotypic and protein localization heterogeneity associated with <i>AHDC1</i> pathogenic proteinâ€ truncating alleles in Xiaâ€ Gibbs syndrome. <i>Human Mutation</i> , 2021, 42, 577-591.	1.1	14
29	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. <i>Genetics in Medicine</i> , 2021, 23, 1028-1040.	1.1	34
30	Exome sequencing reveals predominantly de novo variants in disorders with intellectual disability (ID) in the founder population of Finland. <i>Human Genetics</i> , 2021, 140, 1011-1029.	1.8	23
31	Heterozygous variants in SPTBN1 cause intellectual disability and autism. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2037-2045.	0.7	9
32	A novel homozygous <sc><i>SLC13A5</i></sc> wholeâ€ gene deletion generated by <sc><i>Alu/Alu</i></sc>â€ mediated rearrangement in an Iraqi family with epileptic encephalopathy. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1972-1980.	0.7	16
33	Biallelic Pathogenic Variants in TNNT3 Associated With Congenital Myopathy. <i>Neurology: Genetics</i> , 2021, 7, e589.	0.9	6
34	Two novel biâ€ allelic <sc><i>KDEL2</i></sc> missense variants cause osteogenesis imperfecta with neurodevelopmental features. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2241-2249.	0.7	7
35	Clinical, neuroimaging, and molecular spectrum of <i>TECPR2</i> â€ associated hereditary sensory and autonomic neuropathy with intellectual disability. <i>Human Mutation</i> , 2021, 42, 762-776.	1.1	18
36	Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. <i>Genetics in Medicine</i> , 2021, 23, 1715-1725.	1.1	22

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37	Exome sequencing reveals genetic architecture in patients with isolated or syndromic short stature. <i>Journal of Genetics and Genomics</i> , 2021, 48, 396-402.	1.7	21
38	Detection of a mosaic <i>CDKL5</i> deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1665.	0.6	11
39	A Case Report of Calcium-Sensing Receptor Gene Variant and Primary Hyperparathyroidism. <i>Journal of the Endocrine Society</i> , 2021, 5, A173-A174.	0.1	0
40	Risk of sudden cardiac death in <i>EXOSC5</i> -related disease. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2532-2540.	0.7	6
41	Haploinsufficiency of <i>ARFGEF1</i> is associated with developmental delay, intellectual disability, and epilepsy with variable expressivity. <i>Genetics in Medicine</i> , 2021, 23, 1901-1911.	1.1	9
42	<i>IFIH1</i> loss-of-function variants contribute to very early-onset inflammatory bowel disease. <i>Human Genetics</i> , 2021, 140, 1299-1312.	1.8	17
43	Exome variant discrepancies due to reference-genome differences. <i>American Journal of Human Genetics</i> , 2021, 108, 1239-1250.	2.6	36
44	Biallelic loss-of-function variants in the splicing regulator <i>NSRP1</i> cause a severe neurodevelopmental disorder with spastic cerebral palsy and epilepsy. <i>Genetics in Medicine</i> , 2021, 23, 2455-2460.	1.1	9
45	Exome sequencing in children with clinically suspected maturity-onset diabetes of the young. <i>Pediatric Diabetes</i> , 2021, 22, 960-968.	1.2	6
46	PhenoDB, GeneMatcher and VariantMatcher, tools for analysis and sharing of sequence data. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 365.	1.2	24
47	Deep clinicopathological phenotyping identifies a previously unrecognized pathogenic <i>EMD</i> splice variant. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 2052-2058.	1.7	1
48	High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population. <i>American Journal of Human Genetics</i> , 2021, 108, 1981-2005.	2.6	38
49	Response to Biesecker et al. <i>American Journal of Human Genetics</i> , 2021, 108, 1807-1808.	2.6	3
50	<i>AHDC1</i> missense mutations in Xia-Gibbs syndrome. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100049.	1.0	5
51	Multilocus inheritance and variable disease expressivity in rare disease. , 2021, , 185-204.		0
52	Clinical presentation and evolution of Xia-Gibbs syndrome due to p.Gly375ArgfsTer3 variant in a patient from DR Congo (Central Africa). <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 990-994.	0.7	7
53	Novel Biallelic Variants in <i>KIF21A</i> Cause a Novel Phenotype of Fetal Akinesia with Neurodevelopmental Defects. , 2021, 52, .		0
54	Missense variants in <i>TAF1</i> and developmental phenotypes: Challenges of determining pathogenicity. <i>Human Mutation</i> , 2020, 41, 449-464.	1.1	17

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55	Exome sequencing reveals a novel variant in NFX1 causing intracranial aneurysm in a Chinese family. <i>Journal of NeuroInterventional Surgery</i> , 2020, 12, 221-226.	2.0	7
56	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. <i>Human Mutation</i> , 2020, 41, 487-501.	1.1	58
57	<i>TBX6</i> missense variants expand the mutational spectrum in a non-Mendelian inheritance disease. <i>Human Mutation</i> , 2020, 41, 182-195.	1.1	27
58	Front Cover, Volume 41, Issue 1. <i>Human Mutation</i> , 2020, 41, i.	1.1	0
59	Deficiencies in vesicular transport mediated by TRAPPC4 are associated with severe syndromic intellectual disability. <i>Brain</i> , 2020, 143, 112-130.	3.7	33
60	Phenotypic expansion of <i>POGZ</i> -related intellectual disability syndrome (White-Sutton) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 5	0.7	35
61	Recurrent arginine substitutions in the <i>ACTG2</i> gene are the primary driver of disease burden and severity in visceral myopathy. <i>Human Mutation</i> , 2020, 41, 641-654.	1.1	27
62	Genetic and molecular mechanism for distinct clinical phenotypes conveyed by allelic truncating mutations implicated in <i>FBN1</i> . <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1023.	0.6	19
63	Biallelic in-frame deletion in <i>TRAPPC4</i> in a family with developmental delay and cerebellar atrophy. <i>Brain</i> , 2020, 143, e83-e83.	3.7	8
64	Congenital diaphragmatic hernia as a prominent feature of a <i>SPECC1L</i> -related syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2919-2925.	0.7	8
65	TSC1 Variant Associated With Mild or Absent Clinical Features of Tuberous Sclerosis Complex in a Three-Generation Family. <i>Pediatric Neurology</i> , 2020, 110, 89-91.	1.0	3
66	Low-level parental somatic mosaic SNVs in exomes from a large cohort of trios with diverse suspected Mendelian conditions. <i>Genetics in Medicine</i> , 2020, 22, 1768-1776.	1.1	30
67	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in <i>LARS1</i> . <i>Genetics in Medicine</i> , 2020, 22, 1863-1873.	1.1	19
68	Missed diagnoses: Clinically relevant lessons learned through medical mysteries solved by the Undiagnosed Diseases Network. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1397.	0.6	16
69	Phenotypic expansion in <i>KIF1A</i> -related dominant disorders: A description of novel variants and review of published cases. <i>Human Mutation</i> , 2020, 41, 2094-2104.	1.1	8
70	Clinical genomics and contextualizing genome variation in the diagnostic laboratory. <i>Expert Review of Molecular Diagnostics</i> , 2020, 20, 995-1002.	1.5	14
71	NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. <i>Nature Communications</i> , 2020, 11, 4625.	5.8	47
72	Integrated sequencing and array comparative genomic hybridization in familial Parkinson disease. <i>Neurology: Genetics</i> , 2020, 6, e498.	0.9	11

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73	Functional biology of the Steel syndrome founder allele and evidence for clan genomics derivation of COL27A1 pathogenic alleles worldwide. <i>European Journal of Human Genetics</i> , 2020, 28, 1243-1264.	1.4	27
74	<scp>Wolffâ€“Parkinsonâ€“White</scp> syndrome: De novo variants and evidence for mutational burden in genes associated with atrial fibrillation. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1387-1399.	0.7	14
75	Biallelic <i>GRM7</i> variants cause epilepsy, microcephaly, and cerebral atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 610-627.	1.7	15
76	Human and mouse studies establish TBX6 in Mendelian CAKUT and as a potential driver of kidney defects associated with the 16p11.2 microdeletion syndrome. <i>Kidney International</i> , 2020, 98, 1020-1030.	2.6	17
77	Basic concepts of genetics and genomics. , 2020, , 9-19.		0
78	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. <i>Genetics in Medicine</i> , 2019, 21, 161-172.	1.1	60
79	Biallelic and <i>De Novo</i> Variants in <i>DONSON</i> Reveal a Clinical Spectrum of Cell Cycleâ€“opathies with Microcephaly, Dwarfism and Skeletal Abnormalities. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2056-2066.	0.7	15
80	Novel Heterozygous Mutation in NFKB2 Is Associated With Early Onset CVID and a Functional Defect in NK Cells Complicated by Disseminated CMV Infection and Severe Nephrotic Syndrome. <i>Frontiers in Pediatrics</i> , 2019, 7, 303.	0.9	18
81	Biallelic <i>CACNA2D2</i> variants in epileptic encephalopathy and cerebellar atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1395-1406.	1.7	20
82	Genome sequencing and implications for rare disorders. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 153.	1.2	83
83	The Genomics of Arthrogyrosis, a Complex Trait: Candidate Genes and Further Evidence for Oligogenic Inheritance. <i>American Journal of Human Genetics</i> , 2019, 105, 132-150.	2.6	74
84	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. <i>American Journal of Human Genetics</i> , 2019, 105, 302-316.	2.6	56
85	Homozygous Missense Variants in NTNG2, Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2019, 105, 1048-1056.	2.6	30
86	A Genocentric Approach to Discovery of Mendelian Disorders. <i>American Journal of Human Genetics</i> , 2019, 105, 974-986.	2.6	30
87	Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. <i>American Journal of Human Genetics</i> , 2019, 105, 1005-1015.	2.6	24
88	Introduction to Human Genetics. , 2019, , 1-17.		1
89	Exome Sequencing of a Primary Ovarian Insufficiency Cohort Reveals Common Molecular Etiologies for a Spectrum of Disease. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2019, 104, 3049-3067.	1.8	53
90	Reanalysis of Clinical Exome Sequencing Data. <i>New England Journal of Medicine</i> , 2019, 380, 2478-2480.	13.9	205

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91	Copy number variant and runs of homozygosity detection by microarrays enabled more precise molecular diagnoses in 11,020 clinical exome cases. <i>Genome Medicine</i> , 2019, 11, 30.	3.6	42
92	De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smithâ€™s Magenis syndrome. <i>Genome Medicine</i> , 2019, 11, 12.	3.6	23
93	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , 2019, 104, 530-541.	2.6	30
94	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. <i>American Journal of Human Genetics</i> , 2019, 104, 422-438.	2.6	27
95	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. <i>Genetics in Medicine</i> , 2019, 21, 663-675.	1.1	52
96	Insights into genetics, human biology and disease gleaned from family based genomic studies. <i>Genetics in Medicine</i> , 2019, 21, 798-812.	1.1	161
97	TBX6-associated congenital scoliosis (TACS) as a clinically distinguishable subtype of congenital scoliosis: further evidence supporting the compound inheritance and TBX6 gene dosage model. <i>Genetics in Medicine</i> , 2019, 21, 1548-1558.	1.1	60
98	A biallelic <i>ANTXR1</i> variant expands the anthrax toxin receptor associated phenotype to tooth agenesis. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1015-1022.	0.7	11
99	The role of FREM2 and FRAS1 in the development of congenital diaphragmatic hernia. <i>Human Molecular Genetics</i> , 2018, 27, 2064-2075.	1.4	16
100	The phenotypic spectrum of Xiaâ€™s Gibbs syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1315-1326.	0.7	34
101	Phenotypic expansion illuminates multilocus pathogenic variation. <i>Genetics in Medicine</i> , 2018, 20, 1528-1537.	1.1	104
102	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. <i>American Journal of Human Genetics</i> , 2018, 102, 985-994.	2.6	59
103	Bi-allelic CCDC47 Variants Cause a Disorder Characterized by Woolly Hair, Liver Dysfunction, Dysmorphic Features, and Global Developmental Delay. <i>American Journal of Human Genetics</i> , 2018, 103, 794-807.	2.6	18
104	Phenotypic expansion in <i>DDX3X</i> â€™ a common cause of intellectual disability in females. <i>Annals of Clinical and Translational Neurology</i> , 2018, 5, 1277-1285.	1.7	66
105	Identification of a pathogenic PMP2 variant in a multi-generational family with CMT type 1: Clinical gene panels versus genome-wide approaches to molecular diagnosis. <i>Molecular Genetics and Metabolism</i> , 2018, 125, 302-304.	0.5	13
106	Prioritization of Candidate Genes for Congenital Diaphragmatic Hernia in a Critical Region on Chromosome 4p16 using a Machine-Learning Algorithm. <i>Journal of Pediatric Genetics</i> , 2018, 07, 164-173.	0.3	15
107	Identification of likely pathogenic and known variants in TSPEAR, LAMB3, BCOR, and WNT10A in four Turkish families with tooth agenesis. <i>Human Genetics</i> , 2018, 137, 689-703.	1.8	24
108	IRF2BPL Is Associated with Neurological Phenotypes. <i>American Journal of Human Genetics</i> , 2018, 103, 245-260.	2.6	69

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109	The coexistence of copy number variations (CNVs) and single nucleotide polymorphisms (SNPs) at a locus can result in distorted calculations of the significance in associating SNPs to disease. <i>Human Genetics</i> , 2018, 137, 553-567.	1.8	57
110	Perturbations of BMP/TGF- β 2 and VEGF/VEGFR signalling pathways in non-syndromic sporadic brain arteriovenous malformations (BAVM). <i>Journal of Medical Genetics</i> , 2018, 55, 675-684.	1.5	70
111	A comprehensive clinical and genetic study in 127 patients with ID in Kinshasa, DR Congo. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1897-1909.	0.7	7
112	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. <i>American Journal of Human Genetics</i> , 2017, 100, 185-192.	2.6	142
113	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. <i>American Journal of Human Genetics</i> , 2017, 100, 343-351.	2.6	35
114	MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. <i>American Journal of Human Genetics</i> , 2017, 100, 843-853.	2.6	181
115	22q11.2q13 duplication including <i>SOX10</i> causes sex reversal and peripheral demyelinating neuropathy, central dysmyelinating leukodystrophy, Waardenburg syndrome, and Hirschsprung disease. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1066-1070.	0.7	23
116	Lessons learned from additional research analyses of unsolved clinical exome cases. <i>Genome Medicine</i> , 2017, 9, 26.	3.6	184
117	Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. <i>New England Journal of Medicine</i> , 2017, 376, 21-31.	13.9	565
118	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. <i>American Journal of Human Genetics</i> , 2017, 100, 128-137.	2.6	96
119	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2017, 101, 716-724.	2.6	66
120	Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research. <i>Genetics</i> , 2017, 207, 9-27.	1.2	165
121	Dominant Transmission Observed in Adolescents and Families With Orthostatic Intolerance. <i>Pediatric Neurology</i> , 2017, 66, 53-58.e5.	1.0	6
122	Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders. <i>Genome Medicine</i> , 2017, 9, 73.	3.6	39
123	Triploidy mosaicism (45,X/68,XX) in an infant presenting with failure to thrive. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 694-698.	0.7	6
124	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. <i>American Journal of Human Genetics</i> , 2016, 99, 991-999.	2.6	68
125	Hutterite cataract maps to chromosome 6p21.32-p21.31, cosegregates with a homozygous mutation in <i>LEMD2</i> , and is associated with sudden cardiac death. <i>Molecular Genetics & Genomic Medicine</i> , 2016, 4, 77-94.	0.6	28
126	Recurrent De Novo and Biallelic Variation of ATAD3A, Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. <i>American Journal of Human Genetics</i> , 2016, 99, 831-845.	2.6	146

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127	Exome sequencing in mostly consanguineous Arab families with neurologic disease provides a high potential molecular diagnosis rate. <i>BMC Medical Genomics</i> , 2016, 9, 42.	0.7	80
128	POGZ truncating alleles cause syndromic intellectual disability. <i>Genome Medicine</i> , 2016, 8, 3.	3.6	78
129	A visual and curatorial approach to clinical variant prioritization and disease gene discovery in genome-wide diagnostics. <i>Genome Medicine</i> , 2016, 8, 13.	3.6	37
130	Mechanisms for the Generation of Two Quadruplications Associated with Split-Hand Malformation. <i>Human Mutation</i> , 2016, 37, 160-164.	1.1	16
131	Molecular diagnostic experience of whole-exome sequencing in adult patients. <i>Genetics in Medicine</i> , 2016, 18, 678-685.	1.1	186
132	Atypical presentation of moyamoya disease in an infant with a de novo <i>RNF213</i> variant. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2742-2747.	0.7	15
133	Adult presentation of X-linked Conradi-Hayashi syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1309-1314.	0.7	6
134	Syngnathia and obstructive apnea in a case of popliteal pterygium syndrome. <i>European Journal of Pediatrics</i> , 2014, 173, 1741-1744.	1.3	9
135	Lysinuric protein intolerance presenting with multiple fractures. <i>Molecular Genetics and Metabolism Reports</i> , 2014, 1, 176-183.	0.4	20
136	Understanding how the V(D)J recombinase catalyzes transesterification: distinctions between DNA cleavage and transposition. <i>Nucleic Acids Research</i> , 2008, 36, 2864-2873.	6.5	8
137	Target DNA Structure Plays a Critical Role in RAG Transposition. <i>PLoS Biology</i> , 2006, 4, e350.	2.6	23
138	Paradigm switching in the germinal center. <i>Nature Immunology</i> , 2004, 5, 476-477.	7.0	14