Jennifer E Posey

List of Publications by Year
in descending order

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138 papers	5,020 citations	126708 33 h-index	123241 61 g-index
150 all docs	150 docs citations	150 times ranked	8548 citing authors

#	Article	IF	Citations
1	Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. New England Journal of Medicine, 2017, 376, 21-31.	13.9	565
2	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	13.9	205
3	Molecular diagnostic experience of whole-exome sequencing in adult patients. Genetics in Medicine, 2016, 18, 678-685.	1.1	186
4	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	3.6	184
5	MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. American Journal of Human Genetics, 2017, 100, 843-853.	2.6	181
6	Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research. Genetics, 2017, 207, 9-27.	1.2	165
7	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	1.1	161
8	Recurrent De Novo and Biallelic Variation of ATAD3A , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. American Journal of Human Genetics, 2016, 99, 831-845.	2.6	146
9	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	2.6	142
10	Phenotypic expansion illuminates multilocus pathogenic variation. Genetics in Medicine, 2018, 20, 1528-1537.	1.1	104
11	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137.	2.6	96
12	Genome sequencing and implications for rare disorders. Orphanet Journal of Rare Diseases, 2019, 14, 153.	1.2	83
13	Exome sequencing in mostly consanguineous Arab families with neurologic disease provides a high potential molecular diagnosis rate. BMC Medical Genomics, 2016, 9, 42.	0.7	80
14	POGZ truncating alleles cause syndromic intellectual disability. Genome Medicine, 2016, 8, 3.	3.6	78
15	The Genomics of Arthrogryposis, a Complex Trait: Candidate Genes and Further Evidence for Oligogenic Inheritance. American Journal of Human Genetics, 2019, 105, 132-150.	2.6	74
16	Perturbations of BMP/TGF-Î² and VEGF/VEGFR signalling pathways in non-syndromic sporadic brain arteriovenous malformations (BAVM). Journal of Medical Genetics, 2018, 55, 675-684.	1.5	70
17	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	2.6	69
18	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 991-999.	2.6	68

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19	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 101, 716-724.	2.6	66
20	Phenotypic expansion in <scp>DDX</scp>3X â€“ a common cause of intellectual disability in females. Annals of Clinical and Translational Neurology, 2018, 5, 1277-1285.	1.7	66
21	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	1.1	60
22	TBX6-associated congenital scoliosis (TACS) as a clinically distinguishable subtype of congenital scoliosis: further evidence supporting the compound inheritance and TBX6 gene dosage model. Genetics in Medicine, 2019, 21, 1548-1558.	1.1	60
23	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
24	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. Human Mutation, 2020, 41, 487-501.	1.1	58
25	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. Human Genetics, 2018, 137, 553-567.	1.8	57
26	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. American Journal of Human Genetics, 2019, 105, 302-316.	2.6	56
27	Exome Sequencing of a Primary Ovarian Insufficiency Cohort Reveals Common Molecular Etiologies for a Spectrum of Disease. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 3049-3067.	1.8	53
28	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	1.1	52
29	NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. Nature Communications, 2020, 11, 4625.	5.8	47
30	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	1.1	44
31	Copy number variant and runs of homozygosity detection by microarrays enabled more precise molecular diagnoses in 11,020 clinical exome cases. Genome Medicine, 2019, 11, 30.	3.6	42
32	Perturbations of genes essential for MÃ¼llerian duct and WÃ¶lffian duct development in Mayer-Rokitansky-KÃ¼ster-Hauser syndrome. American Journal of Human Genetics, 2021, 108, 337-345.	2.6	41
33	Diagnostic yield and clinical impact of exome sequencing in early-onset scoliosis (EOS). Journal of Medical Genetics, 2021, 58, 41-47.	1.5	40
34	Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders. Genome Medicine, 2017, 9, 73.	3.6	39
35	High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population. American Journal of Human Genetics, 2021, 108, 1981-2005.	2.6	38
36	A visual and curatorial approach to clinical variant prioritization and disease gene discovery in genome-wide diagnostics. Genome Medicine, 2016, 8, 13.	3.6	37

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37	Exome variant discrepancies due to reference-genome differences. American Journal of Human Genetics, 2021, 108, 1239-1250.	2.6	36
38	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. American Journal of Human Genetics, 2017, 100, 343-351.	2.6	35
39	Phenotypic expansion of POGZâ€related intellectual disability syndrome (Whiteâ€Sutton) Tj ETQq1 1 0.784314 rgBT /Overloc	0.7	35
40	The phenotypic spectrum of Xiaâ€Gibbs syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1315-1326.	0.7	34
41	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. Genetics in Medicine, 2021, 23, 1028-1040.	1.1	34
42	Deficiencies in vesicular transport mediated by TRAPPC4 are associated with severe syndromic intellectual disability. Brain, 2020, 143, 112-130.	3.7	33
43	Homozygous Missense Variants in NTNG2, Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 1048-1056.	2.6	30
44	A Genocentric Approach to Discovery of Mendelian Disorders. American Journal of Human Genetics, 2019, 105, 974-986.	2.6	30
45	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	2.6	30
46	Low-level parental somatic mosaic SNVs in exomes from a large cohort of trios with diverse suspected Mendelian conditions. Genetics in Medicine, 2020, 22, 1768-1776.	1.1	30
47	Hutteriteâ€type cataract maps to chromosome 6p21.32â€p21.31, cosegregates with a homozygous mutation in <scp>LEMD</scp>2, and is associated with sudden cardiac death. Molecular Genetics & Genomic Medicine, 2016, 4, 77-94.	0.6	28
48	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	2.6	27
49	TBX6 missense variants expand the mutational spectrum in a nonâ€Mendelian inheritance disease. Human Mutation, 2020, 41, 182-195.	1.1	27
50	Recurrent arginine substitutions in the ACTG2 gene are the primary driver of disease burden and severity in visceral myopathy. Human Mutation, 2020, 41, 641-654.	1.1	27
51	Functional biology of the Steel syndrome founder allele and evidence for clan genomics derivation of COL27A1 pathogenic alleles worldwide. European Journal of Human Genetics, 2020, 28, 1243-1264.	1.4	27
52	Identification of likely pathogenic and known variants in TSPEAR, LAMB3, BCOR, and WNT10A in four Turkish families with tooth agenesis. Human Genetics, 2018, 137, 689-703.	1.8	24
53	Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. American Journal of Human Genetics, 2019, 105, 1005-1015.	2.6	24
54	PhenoDB, GeneMatcher and VariantMatcher, tools for analysis and sharing of sequence data. Orphanet Journal of Rare Diseases, 2021, 16, 365.	1.2	24

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55	Target DNA Structure Plays a Critical Role in RAG Transposition. PLoS Biology, 2006, 4, e350.	2.6	23
56	22q11.2q13 duplication including SOX10 causes sexâ€reversal and peripheral demyelinating neuropathy, central dysmyelinating leukodystrophy, Waardenburg syndrome, and Hirschsprung disease. American Journal of Medical Genetics, Part A, 2017, 173, 1066-1070.	0.7	23
57	De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smithâ€“Magenis syndrome. Genome Medicine, 2019, 11, 12.	3.6	23
58	Exome sequencing reveals predominantly de novo variants in disorders with intellectual disability (ID) in the founder population of Finland. Human Genetics, 2021, 140, 1011-1029.	1.8	23
59	Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. Genetics in Medicine, 2021, 23, 1715-1725.	1.1	22
60	Exome sequencing reveals genetic architecture in patients with isolated or syndromic short stature. Journal of Genetics and Genomics, 2021, 48, 396-402.	1.7	21
61	Lysinuric protein intolerance presenting with multiple fractures. Molecular Genetics and Metabolism Reports, 2014, 1, 176-183.	0.4	20
62	Biallelic CACNA2D2 variants in epileptic encephalopathy and cerebellar atrophy. Annals of Clinical and Translational Neurology, 2019, 6, 1395-1406.	1.7	20
63	Genetic and molecular mechanism for distinct clinical phenotypes conveyed by allelic truncating mutations implicated in FBN1. Molecular Genetics & Genomic Medicine, 2020, 8, e1023.	0.6	19
64	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. Genetics in Medicine, 2020, 22, 1863-1873.	1.1	19
65	Bi-allelic CCDC47 Variants Cause a Disorder Characterized by Woolly Hair, Liver Dysfunction, Dysmorphic Features, and Global Developmental Delay. American Journal of Human Genetics, 2018, 103, 794-807.	2.6	18
66	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. Frontiers in Pediatrics, 2019, 7, 303.	0.9	18
67	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. Genetics in Medicine, 2021, 23, 259-271.	1.1	18
68	Clinical, neuroimaging, and molecular spectrum of TECPR2 â€associated hereditary sensory and autonomic neuropathy with intellectual disability. Human Mutation, 2021, 42, 762-776.	1.1	18
69	Missense variants in TAF1 and developmental phenotypes: Challenges of determining pathogenicity. Human Mutation, 2020, 41, 449-464.	1.1	17
70	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
71	Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. Brain, 2022, 145, 909-924.	3.7	17
72	IFIH1 loss-of-function variants contribute to very early-onset inflammatory bowel disease. Human Genetics, 2021, 140, 1299-1312.	1.8	17

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73	Human and mouse studies establish TBX6 in Mendelian CAKUT and as a potential driver of kidney defects associated with the 16p11.2 microdeletion syndrome. Kidney International, 2020, 98, 1020-1030.	2.6	17
74	Mechanisms for the Generation of Two Quadruplications Associated with Split-Hand Malformation. Human Mutation, 2016, 37, 160-164.	1.1	16
75	The role of FREM2 and FRAS1 in the development of congenital diaphragmatic hernia. Human Molecular Genetics, 2018, 27, 2064-2075.	1.4	16
76	Missed diagnoses: Clinically relevant lessons learned through medical mysteries solved by the Undiagnosed Diseases Network. Molecular Genetics & Genomic Medicine, 2020, 8, e1397.	0.6	16
77	Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. Genetics in Medicine, 2021, 23, 1075-1085.	1.1	16
78	A novel homozygous <scp>SLC13A5</scp> wholeâ€gene deletion generated by <scp>Alu/Alu</scp>â€mediated rearrangement in an Iraqi family with epileptic encephalopathy. American Journal of Medical Genetics, Part A, 2021, 185, 1972-1980.	0.7	16
79	Atypical presentation of moyamoya disease in an infant with a de novo RNF213 variant. American Journal of Medical Genetics, Part A, 2015, 167, 2742-2747.	0.7	15
80	Prioritization of Candidate Genes for Congenital Diaphragmatic Hernia in a Critical Region on Chromosome 4p16 using a Machine-Learning Algorithm. Journal of Pediatric Genetics, 2018, 07, 164-173.	0.3	15
81	Biallelic and De Novo Variants in DONSON Reveal a Clinical Spectrum of Cell Cycleâ€opathies with Microcephaly, Dwarfism and Skeletal Abnormalities. American Journal of Medical Genetics, Part A, 2019, 179, 2056-2066.	0.7	15
82	Biallelic GRM7 variants cause epilepsy, microcephaly, and cerebral atrophy. Annals of Clinical and Translational Neurology, 2020, 7, 610-627.	1.7	15
83	Paradigm switching in the germinal center. Nature Immunology, 2004, 5, 476-477.	7.0	14
84	Clinical genomics and contextualizing genome variation in the diagnostic laboratory. Expert Review of Molecular Diagnostics, 2020, 20, 995-1002.	1.5	14
85	<scp>Wolffâ€“Parkinsonâ€“White</scp> syndrome: De novo variants and evidence for mutational burden in genes associated with atrial fibrillation. American Journal of Medical Genetics, Part A, 2020, 182, 1387-1399.	0.7	14
86	<scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833.	2.8	14
87	Phenotypic and protein localization heterogeneity associated with AHDC1 pathogenic proteinâ€truncating alleles in Xiaâ€“Gibbs syndrome. Human Mutation, 2021, 42, 577-591.	1.1	14
88	Quantitative dissection of multilocus pathogenic variation in an Egyptian infant with severe neurodevelopmental disorder resulting from multiple molecular diagnoses. American Journal of Medical Genetics, Part A, 2022, 188, 735-750.	0.7	14
89	Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability. Human Genetics and Genomics Advances, 2022, 3, 100074.	1.0	14
90	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. Molecular Genetics and Metabolism, 2018, 125, 302-304.	0.5	13

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91	Neurodevelopmental disorder in an Egyptian family with a biallelic <scp>ALKBH8</scp> variant. American Journal of Medical Genetics, Part A, 2021, 185, 1288-1293.	0.7	13
92	Retrospective analysis of a clinical exome sequencing cohort reveals the mutational spectrum and identifies candidate diseaseâ€“associated loci for BAFopathies. Genetics in Medicine, 2022, 24, 364-373.	1.1	12
93	A biallelic ANTXR1 variant expands the anthrax toxin receptor associated phenotype to tooth agenesis. American Journal of Medical Genetics, Part A, 2018, 176, 1015-1022.	0.7	11
94	Integrated sequencing and array comparative genomic hybridization in familial Parkinson disease. Neurology: Genetics, 2020, 6, e498.	0.9	11
95	Detection of a mosaic CDKL5 deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. Molecular Genetics & Genomic Medicine, 2021, 9, e1665.	0.6	11
96	Variantâ€level matching for diagnosis and discovery: Challenges and opportunities. Human Mutation, 2022, , .	1.1	11
97	Dominant mitochondrial membrane protein-associated neurodegeneration (MPAN) variants cluster within a specific C19orf12 isoform. Parkinsonism and Related Disorders, 2021, 82, 84-86.	1.1	10
98	Syngnathia and obstructive apnea in a case of popliteal pterygium syndrome. European Journal of Pediatrics, 2014, 173, 1741-1744.	1.3	9
99	Heterozygous variants in SPTBN1 cause intellectual disability and autism. American Journal of Medical Genetics, Part A, 2021, 185, 2037-2045.	0.7	9
100	Haploinsufficiency of ARFGEF1 is associated with developmental delay, intellectual disability, and epilepsy with variable expressivity. Genetics in Medicine, 2021, 23, 1901-1911.	1.1	9
101	Biallelic loss-of-function variants in the splicing regulator NSRP1 cause a severe neurodevelopmental disorder with spastic cerebral palsy and epilepsy. Genetics in Medicine, 2021, 23, 2455-2460.	1.1	9
102	Understanding how the V(D)J recombinase catalyzes transesterification: distinctions between DNA cleavage and transposition. Nucleic Acids Research, 2008, 36, 2864-2873.	6.5	8
103	Biallelic in-frame deletion in TRAPPC4 in a family with developmental delay and cerebellar atrophy. Brain, 2020, 143, e83-e83.	3.7	8
104	Congenital diaphragmatic hernia as a prominent feature of a SPECC1L â€related syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2919-2925.	0.7	8
105	Phenotypic expansion in KIF1A â€related dominant disorders: A description of novel variants and review of published cases. Human Mutation, 2020, 41, 2094-2104.	1.1	8
106	Biallelic pathogenic variants in roundabout guidance receptor 1 associate with syndromic congenital anomalies of the kidney and urinary tract. Kidney International, 2022, 101, 1039-1053.	2.6	8
107	A comprehensive clinical and genetic study in 127 patients with ID in Kinshasa, DR Congo. American Journal of Medical Genetics, Part A, 2018, 176, 1897-1909.	0.7	7
108	Exome sequencing reveals a novel variant in NFX1 causing intracranial aneurysm in a Chinese family. Journal of NeuroInterventional Surgery, 2020, 12, 221-226.	2.0	7

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109	Two novel biâ€allelic <scp>KDELR2</scp> missense variants cause osteogenesis imperfecta with neurodevelopmental features. American Journal of Medical Genetics, Part A, 2021, 185, 2241-2249.	0.7	7
110	Clinical presentation and evolution of Xiaâ€Gibbs syndrome due to p.Gly375ArgfsTer3 variant in a patient from DR Congo (Central Africa). American Journal of Medical Genetics, Part A, 2021, 185, 990-994.	0.7	7
111	The clinical and molecular spectrum of QRICH1 associated neurodevelopmental disorder. Human Mutation, 2022, 43, 266-282.	1.1	7
112	Expanding the mutation and phenotype spectrum of MYH3-associated skeletal disorders. Npj Genomic Medicine, 2022, 7, 11.	1.7	7
113	<scp>Elâ€Hattabâ€Alkuraya</scp> syndrome caused by biallelic <scp>WDR45B</scp> pathogenic variants: Further delineation of the phenotype and genotype. Clinical Genetics, 2022, 101, 530-540.	1.0	7
114	Adult presentation of Xâ€linked Conradiâ€HÃ¼nermannâ€Happle syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1309-1314.	0.7	6
115	Triploidy mosaicism (45,X/68,XX) in an infant presenting with failure to thrive. American Journal of Medical Genetics, Part A, 2016, 170, 694-698.	0.7	6
116	Dominant Transmission Observed in Adolescents and Families With Orthostatic Intolerance. Pediatric Neurology, 2017, 66, 53-58.e5.	1.0	6
117	Clinical characterization of individuals with the distal 1q21.1 microdeletion. American Journal of Medical Genetics, Part A, 2021, 185, 1388-1398.	0.7	6
118	Biallelic Pathogenic Variants in TNNT3 Associated With Congenital Myopathy. Neurology: Genetics, 2021, 7, e589.	0.9	6
119	Risk of sudden cardiac death in <scp>EXOSC5</scp>â€related disease. American Journal of Medical Genetics, Part A, 2021, 185, 2532-2540.	0.7	6
120	Exome sequencing in children with clinically suspected <scp>maturityâ€onset</scp> diabetes of the young. Pediatric Diabetes, 2021, 22, 960-968.	1.2	6
121	AHDC1 missense mutations in Xia-Gibbs syndrome. Human Genetics and Genomics Advances, 2021, 2, 100049.	1.0	5
122	Novel RETREG1 (<scp>FAM134B</scp> founder allele is linked to <scp>HSAN2B</scp> and renal disease in a Turkish family. American Journal of Medical Genetics, Part A, 2022, 188, 2153-2161.	0.7	4
123	TSC1 Variant Associated With Mild or Absent Clinical Features of Tuberous Sclerosis Complex in a Three-Generation Family. Pediatric Neurology, 2020, 110, 89-91.	1.0	3
124	Response to Biesecker etÂal.. American Journal of Human Genetics, 2021, 108, 1807-1808.	2.6	3
125	Expanding the phenotypic and allelic spectrum of <scp>SMG8</scp>: Clinical observations reveal overlap with <scp>SMG9</scp>â€associated disease trait. American Journal of Medical Genetics, Part A, 2022, 188, 648-657.	0.7	3
126	De novo heterozygous variants in <scp>SLC30A7</scp> are a candidate cause for Joubert syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 2360-2366.	0.7	3

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127	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
128	Introduction to Human Genetics. , 2019, , 1-17.		1
129	Genetic counselor roles in the undiagnosed diseases network research study: Clinical care, collaboration, and curation. Journal of Genetic Counseling, 2022, 31, 326-337.	0.9	1
130	Deep clinicopathological phenotyping identifies a previously unrecognized pathogenic EMD splice variant. Annals of Clinical and Translational Neurology, 2021, 8, 2052-2058.	1.7	1
131	Front Cover, Volume 41, Issue 1. Human Mutation, 2020, 41, i.	1.1	0
132	A Case Report of Calcium-Sensing Receptor Gene Variant and Primary Hyperparathyroidism. Journal of the Endocrine Society, 2021, 5, A173-A174.	0.1	0
133	Advances in Next-Generation Sequencing Technologies and Functional Investigation of Candidate Variants in Neurological and Behavioral Disorders. , 2022, , 390-404.		0
134	Multilocus inheritance and variable disease expressivity in rare disease. , 2021, , 185-204.		0
135	Basic concepts of genetics and genomics. , 2020, , 9-19.		0
136	Novel Biallelic Variants in KIF21A Cause a Novel Phenotype of Fetal Akinesia with Neurodevelopmental Defects. , 2021, 52, .		0
137	Elucidating the clinical spectrum and molecular basis of HYAL2 deficiency. Genetics in Medicine, 2022, 24, 631-644.	1.1	0
138	MO047: Biallelic pathogenic variants in ROBO1 associate with syndromic CAKUT. Nephrology Dialysis Transplantation, 2022, 37, .	0.4	0