

Jennifer E Posey

List of Publications by Year in descending order

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138
papers

5,020
citations

126907

33
h-index

123424

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. <i>New England Journal of Medicine</i> , 2017, 376, 21-31.	27.0	565
2	Reanalysis of Clinical Exome Sequencing Data. <i>New England Journal of Medicine</i> , 2019, 380, 2478-2480.	27.0	205
3	Molecular diagnostic experience of whole-exome sequencing in adult patients. <i>Genetics in Medicine</i> , 2016, 18, 678-685.	2.4	186
4	Lessons learned from additional research analyses of unsolved clinical exome cases. <i>Genome Medicine</i> , 2017, 9, 26.	8.2	184
5	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.	6.2	181
6	Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research. <i>Genetics</i> , 2017, 207, 9-27.	2.9	165
7	Insights into genetics, human biology and disease gleaned from family based genomic studies. <i>Genetics in Medicine</i> , 2019, 21, 798-812.	2.4	161
8	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.	6.2	146
9	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. <i>American Journal of Human Genetics</i> , 2017, 100, 185-192.	6.2	142
10	Phenotypic expansion illuminates multilocus pathogenic variation. <i>Genetics in Medicine</i> , 2018, 20, 1528-1537.	2.4	104
11	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. <i>American Journal of Human Genetics</i> , 2017, 100, 128-137.	6.2	96
12	Genome sequencing and implications for rare disorders. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 153.	2.7	83
13	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.	1.5	80
14	POGZ truncating alleles cause syndromic intellectual disability. <i>Genome Medicine</i> , 2016, 8, 3.	8.2	78
15	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.	6.2	74
16	Perturbations of BMP/TGF- β and VEGF/VEGFR signalling pathways in non-syndromic sporadic brain arteriovenous malformations (BAVM). <i>Journal of Medical Genetics</i> , 2018, 55, 675-684.	3.2	70
17	IRF2BPL Is Associated with Neurological Phenotypes. <i>American Journal of Human Genetics</i> , 2018, 103, 245-260.	6.2	69
18	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.	6.2	68

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19	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.	6.2	66
20	Phenotypic expansion in <i>DDX3X</i> – a common cause of intellectual disability in females. <i>Annals of Clinical and Translational Neurology</i> , 2018, 5, 1277-1285.	3.7	66
21	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. <i>Genetics in Medicine</i> , 2019, 21, 161-172.	2.4	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. <i>Genetics in Medicine</i> , 2019, 21, 1548-1558.	2.4	60
23	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.	6.2	59
24	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. <i>Human Mutation</i> , 2020, 41, 487-501.	2.5	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. <i>Human Genetics</i> , 2018, 137, 553-567.	3.8	57
26	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. <i>American Journal of Human Genetics</i> , 2019, 105, 302-316.	6.2	56
27	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.	3.6	53
28	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. <i>Genetics in Medicine</i> , 2019, 21, 663-675.	2.4	52
29	NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. <i>Nature Communications</i> , 2020, 11, 4625.	12.8	47
30	Centers for Mendelian Genomics: A decade of facilitating gene discovery. <i>Genetics in Medicine</i> , 2022, 24, 784-797.	2.4	44
31	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.	8.2	42
32	Perturbations of genes essential for Müllerian duct and Wolffian duct development in Mayer-Rokitansky-Küster-Hauser syndrome. <i>American Journal of Human Genetics</i> , 2021, 108, 337-345.	6.2	41
33	Diagnostic yield and clinical impact of exome sequencing in early-onset scoliosis (EOS). <i>Journal of Medical Genetics</i> , 2021, 58, 41-47.	3.2	40
34	Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders. <i>Genome Medicine</i> , 2017, 9, 73.	8.2	39
35	High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population. <i>American Journal of Human Genetics</i> , 2021, 108, 1981-2005.	6.2	38
36	A visual and curatorial approach to clinical variant prioritization and disease gene discovery in genome-wide diagnostics. <i>Genome Medicine</i> , 2016, 8, 13.	8.2	37

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37	Exome variant discrepancies due to reference-genome differences. American Journal of Human Genetics, 2021, 108, 1239-1250.	6.2	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.	6.2	35
39	Phenotypic expansion of <i>POGZ</i> -related intellectual disability syndrome (White-Sutton) Tj ETQq1 1 0.784314 rgBT /Overlock	1.2	35
40	The phenotypic spectrum of Xia-Gibbs syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1315-1326.	1.2	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.	2.4	34
42	Deficiencies in vesicular transport mediated by TRAPPC4 are associated with severe syndromic intellectual disability. Brain, 2020, 143, 112-130.	7.6	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.	6.2	30
44	A Genocentric Approach to Discovery of Mendelian Disorders. American Journal of Human Genetics, 2019, 105, 974-986.	6.2	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.	6.2	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.	2.4	30
47	Hutterite-type cataract maps to chromosome 6p21.32-p21.31, cosegregates with a homozygous mutation in <i>LEMD2</i> , and is associated with sudden cardiac death. Molecular Genetics & Genomic Medicine, 2016, 4, 77-94.	1.2	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.	6.2	27
49	<i>TBX6</i> missense variants expand the mutational spectrum in a non-Mendelian inheritance disease. Human Mutation, 2020, 41, 182-195.	2.5	27
50	Recurrent arginine substitutions in the <i>ACTG2</i> gene are the primary driver of disease burden and severity in visceral myopathy. Human Mutation, 2020, 41, 641-654.	2.5	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.	2.8	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.	3.8	24
53	Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. American Journal of Human Genetics, 2019, 105, 1005-1015.	6.2	24
54	PhenoDB, GeneMatcher and VariantMatcher, tools for analysis and sharing of sequence data. Orphanet Journal of Rare Diseases, 2021, 16, 365.	2.7	24

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55	Target DNA Structure Plays a Critical Role in RAG Transposition. <i>PLoS Biology</i> , 2006, 4, e350.	5.6	23
56	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.	1.2	23
57	De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smith-Magenis syndrome. <i>Genome Medicine</i> , 2019, 11, 12.	8.2	23
58	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.	3.8	23
59	Biallelic and monoallelic variants in <i>PLXNA1</i> are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. <i>Genetics in Medicine</i> , 2021, 23, 1715-1725.	2.4	22
60	Exome sequencing reveals genetic architecture in patients with isolated or syndromic short stature. <i>Journal of Genetics and Genomics</i> , 2021, 48, 396-402.	3.9	21
61	Lysinuric protein intolerance presenting with multiple fractures. <i>Molecular Genetics and Metabolism Reports</i> , 2014, 1, 176-183.	1.1	20
62	Biallelic <i>CACNA2D2</i> variants in epileptic encephalopathy and cerebellar atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1395-1406.	3.7	20
63	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.	1.2	19
64	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.	2.4	19
65	Bi-allelic <i>CCDC47</i> 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.	6.2	18
66	Novel Heterozygous Mutation in <i>NFKB2</i> 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.	1.9	18
67	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. <i>Genetics in Medicine</i> , 2021, 23, 259-271.	2.4	18
68	Clinical, neuroimaging, and molecular spectrum of <i>TECPR2</i> -associated hereditary sensory and autonomic neuropathy with intellectual disability. <i>Human Mutation</i> , 2021, 42, 762-776.	2.5	18
69	Missense variants in <i>TAF1</i> and developmental phenotypes: Challenges of determining pathogenicity. <i>Human Mutation</i> , 2020, 41, 449-464.	2.5	17
70	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.	1.2	17
71	Biallelic variants in <i>SLC38A3</i> encoding a glutamine transporter cause epileptic encephalopathy. <i>Brain</i> , 2022, 145, 909-924.	7.6	17
72	<i>IFIH1</i> loss-of-function variants contribute to very early-onset inflammatory bowel disease. <i>Human Genetics</i> , 2021, 140, 1299-1312.	3.8	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. <i>Kidney International</i> , 2020, 98, 1020-1030.	5.2	17
74	Mechanisms for the Generation of Two Quadruplications Associated with Split-Hand Malformation. <i>Human Mutation</i> , 2016, 37, 160-164.	2.5	16
75	The role of <i>FREM2</i> and <i>FRAS1</i> in the development of congenital diaphragmatic hernia. <i>Human Molecular Genetics</i> , 2018, 27, 2064-2075.	2.9	16
76	Missed diagnoses: Clinically relevant lessons learned through medical mysteries solved by the Undiagnosed Diseases Network. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1397.	1.2	16
77	Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. <i>Genetics in Medicine</i> , 2021, 23, 1075-1085.	2.4	16
78	A novel homozygous <i>SLC13A5</i> whole-gene deletion generated by <i>Alu/Alu</i> -mediated rearrangement in an Iraqi family with epileptic encephalopathy. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1972-1980.	1.2	16
79	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.	1.2	15
80	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.	0.7	15
81	Biallelic and <i>De Novo</i> Variants in <i>DONSON</i> Reveal a Clinical Spectrum of Cell Cycleopathies with Microcephaly, Dwarfism and Skeletal Abnormalities. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2056-2066.	1.2	15
82	Biallelic <i>GRM7</i> variants cause epilepsy, microcephaly, and cerebral atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 610-627.	3.7	15
83	Paradigm switching in the germinal center. <i>Nature Immunology</i> , 2004, 5, 476-477.	14.5	14
84	Clinical genomics and contextualizing genome variation in the diagnostic laboratory. <i>Expert Review of Molecular Diagnostics</i> , 2020, 20, 995-1002.	3.1	14
85	<i>Wolff-Parkinson-White</i> 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.	1.2	14
86	<i>MED27</i> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. <i>Annals of Neurology</i> , 2021, 89, 828-833.	5.3	14
87	Phenotypic and protein localization heterogeneity associated with <i>AHDC1</i> pathogenic protein-truncating alleles in Xia-Gibbs syndrome. <i>Human Mutation</i> , 2021, 42, 577-591.	2.5	14
88	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.	1.2	14
89	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.	1.7	14
90	Identification of a pathogenic <i>PMP2</i> 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.	1.1	13

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91	Neurodevelopmental disorder in an Egyptian family with a biallelic <i>ALKBH8</i> variant. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1288-1293.	1.2	13
92	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.	2.4	12
93	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.	1.2	11
94	Integrated sequencing and array comparative genomic hybridization in familial Parkinson disease. <i>Neurology: Genetics</i> , 2020, 6, e498.	1.9	11
95	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.	1.2	11
96	Variant-level matching for diagnosis and discovery: Challenges and opportunities. <i>Human Mutation</i> , 2022, , .	2.5	11
97	Dominant mitochondrial membrane protein-associated neurodegeneration (MPAN) variants cluster within a specific <i>C19orf12</i> isoform. <i>Parkinsonism and Related Disorders</i> , 2021, 82, 84-86.	2.2	10
98	Syngnathia and obstructive apnea in a case of popliteal pterygium syndrome. <i>European Journal of Pediatrics</i> , 2014, 173, 1741-1744.	2.7	9
99	Heterozygous variants in <i>SPTBN1</i> cause intellectual disability and autism. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2037-2045.	1.2	9
100	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.	2.4	9
101	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.	2.4	9
102	Understanding how the V(D)J recombinase catalyzes transesterification: distinctions between DNA cleavage and transposition. <i>Nucleic Acids Research</i> , 2008, 36, 2864-2873.	14.5	8
103	Biallelic in-frame deletion in <i>TRAPPC4</i> in a family with developmental delay and cerebellar atrophy. <i>Brain</i> , 2020, 143, e83-e83.	7.6	8
104	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.	1.2	8
105	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.	2.5	8
106	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.	5.2	8
107	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.	1.2	7
108	Exome sequencing reveals a novel variant in <i>NFX1</i> causing intracranial aneurysm in a Chinese family. <i>Journal of NeuroInterventional Surgery</i> , 2020, 12, 221-226.	3.3	7

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109	Two novel biallelic <i>KDEL2</i> missense variants cause osteogenesis imperfecta with neurodevelopmental features. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2241-2249.	1.2	7
110	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.	1.2	7
111	The clinical and molecular spectrum of <i>QRICH1</i> associated neurodevelopmental disorder. <i>Human Mutation</i> , 2022, 43, 266-282.	2.5	7
112	Expanding the mutation and phenotype spectrum of MYH3-associated skeletal disorders. <i>Npj Genomic Medicine</i> , 2022, 7, 11.	3.8	7
113	<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.	2.0	7
114	Adult presentation of X-linked Conradi-Hänermann-Happle syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1309-1314.	1.2	6
115	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.	1.2	6
116	Dominant Transmission Observed in Adolescents and Families With Orthostatic Intolerance. <i>Pediatric Neurology</i> , 2017, 66, 53-58.e5.	2.1	6
117	Clinical characterization of individuals with the distal 1q21.1 microdeletion. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1388-1398.	1.2	6
118	Biallelic Pathogenic Variants in TNNT3 Associated With Congenital Myopathy. <i>Neurology: Genetics</i> , 2021, 7, e589.	1.9	6
119	Risk of sudden cardiac death in <i>EXOSC5</i> -related disease. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2532-2540.	1.2	6
120	Exome sequencing in children with clinically suspected maturity-onset diabetes of the young. <i>Pediatric Diabetes</i> , 2021, 22, 960-968.	2.9	6
121	AHDC1 missense mutations in Xia-Gibbs syndrome. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100049.	1.7	5
122	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.	1.2	4
123	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.	2.1	3
124	Response to Biesecker et al.. <i>American Journal of Human Genetics</i> , 2021, 108, 1807-1808.	6.2	3
125	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.	1.2	3
126	De novo 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.	1.2	3

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127	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.	5.3	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. <i>Journal of Genetic Counseling</i> , 2022, 31, 326-337.	1.6	1
130	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.	3.7	1
131	Front Cover, Volume 41, Issue 1. <i>Human Mutation</i> , 2020, 41, i.	2.5	0
132	A Case Report of Calcium-Sensing Receptor Gene Variant and Primary Hyperparathyroidism. <i>Journal of the Endocrine Society</i> , 2021, 5, A173-A174.	0.2	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 <i>KIF21A</i> Cause a Novel Phenotype of Fetal Akinesia with Neurodevelopmental Defects. , 2021, 52, .		0
137	Elucidating the clinical spectrum and molecular basis of <i>HYAL2</i> deficiency. <i>Genetics in Medicine</i> , 2022, 24, 631-644.	2.4	0
138	MO047: Biallelic pathogenic variants in <i>ROBO1</i> associate with syndromic CAKUT. <i>Nephrology Dialysis Transplantation</i> , 2022, 37, .	0.7	0