Jennifer E Posey

List of Publications by Year in Descending Order

128 papers	2,998 citations	28 h-index	51 g-index
150 ext. papers	4,293 ext. citations	7.1 avg, IF	5.12 L-index

#	Paper	IF	Citations
128	Centers for Mendelian Genomics: A decade of facilitating gene discovery.. *Genetics in Medicine*, **2022**,	8.1	5
127	Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability.. *Human Genetics and Genomics Advances*, **2022**, 3, 100074	0.8	3
126	Advances in Next-Generation Sequencing Technologies and Functional Investigation of Candidate Variants in Neurological and Behavioral Disorders **2022**, 390-404		
125	Expanding the mutation and phenotype spectrum of MYH3-associated skeletal disorders.. *Npj Genomic Medicine*, **2022**, 7, 11	6.2	1
124	Novel RETREG1 (FAM134B) founder allele is linked to HSAN2B and renal disease in a Turkish family.. *American Journal of Medical Genetics, Part A*, **2022**,	2.5	0
123	Variant-level matching for diagnosis and discovery: challenges and opportunities.. *Human Mutation*, **2022**,	4.7	1
122	Novel Biallelic Variants in KIF21A Cause a Novel Phenotype of Fetal Akinesia with Neurodevelopmental Defects **2021**, 52,		
121	Expanding the phenotypic and allelic spectrum of SMG8: Clinical observations reveal overlap with SMG9-associated disease trait. *American Journal of Medical Genetics, Part A*, **2021**, 188, 648	2.5	1
120	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*, **2021**,	2.5	3
119	Phenotypic and protein localization heterogeneity associated with AHDC1 pathogenic protein-truncating alleles in Xia-Gibbs syndrome. *Human Mutation*, **2021**, 42, 577-591	4.7	3
118	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. *Genetics in Medicine*, **2021**, 23, 1028-1040	8.1	7
117	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	6.3	6
116	Heterozygous variants in SPTBN1 cause intellectual disability and autism. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2037-2045	2.5	2
115	A novel homozygous SLC13A5 whole-gene deletion generated by Alu/Alu-mediated rearrangement in an Iraqi family with epileptic encephalopathy. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1972-1980	2.5	5
114	Biallelic Pathogenic Variants in Associated With Congenital Myopathy. *Neurology: Genetics*, **2021**, 7, e589	3.8	4
113	Two novel bi-allelic KDELR2 missense variants cause osteogenesis imperfecta with neurodevelopmental features. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2241-2249	2.5	2
112	Clinical, neuroimaging, and molecular spectrum of TECPR2-associated hereditary sensory and autonomic neuropathy with intellectual disability. *Human Mutation*, **2021**, 42, 762-776	4.7	5

111	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	8.1	6
110	Exome sequencing reveals genetic architecture in patients with isolated or syndromic short stature. *Journal of Genetics and Genomics*, **2021**, 48, 396-402	4	5
109	Detection of a mosaic CDKL5 deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. *Molecular Genetics & Genomic Medicine*, **2021**, 9, e1665	2.3	1
108	A Case Report of Calcium-Sensing Receptor Gene Variant and Primary Hyperparathyroidism. *Journal of the Endocrine Society*, **2021**, 5, A173-A174	0.4	78
107	Risk of sudden cardiac death in EXOSC5-related disease. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2532-2540	2.5	1
106	Haploinsufficiency of ARFGEF1 is associated with developmental delay, intellectual disability, and epilepsy with variable expressivity. *Genetics in Medicine*, **2021**, 23, 1901-1911	8.1	1
105	IFIH1 loss-of-function variants contribute to very early-onset inflammatory bowel disease. *Human Genetics*, **2021**, 140, 1299-1312	6.3	3
104	Exome variant discrepancies due to reference-genome differences. *American Journal of Human Genetics*, **2021**, 108, 1239-1250	11	5
103	Diagnostic yield and clinical impact of exome sequencing in early-onset scoliosis (EOS). *Journal of Medical Genetics*, **2021**, 58, 41-47	5.8	20
102	Dominant mitochondrial membrane protein-associated neurodegeneration (MPAN) variants cluster within a specific C19orf12 isoform. *Parkinsonism and Related Disorders*, **2021**, 82, 84-86	3.6	4
101	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 185, 119-133	2.5	6
100	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. *Genetics in Medicine*, **2021**, 23, 259-271	8.1	6
99	Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. *Brain*, **2021**,	11.2	2
98	MED27 Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. *Annals of Neurology*, **2021**, 89, 828-833	9.4	2
97	Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. *Genetics in Medicine*, **2021**, 23, 1075-1085	8.1	2
96	Neurodevelopmental disorder in an Egyptian family with a biallelic ALKBH8 variant. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1288-1293	2.5	3
95	Perturbations of genes essential for Müllerian duct and Wolffian duct development in Mayer-Rokitansky-Küster-Hauser syndrome. *American Journal of Human Genetics*, **2021**, 108, 337-345	11	15
94	Clinical characterization of individuals with the distal 1q21.1 microdeletion. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1388-1398	2.5	1

93	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	8.1	1
92	Exome sequencing in children with clinically suspected maturity-onset diabetes of the young. *Pediatric Diabetes*, **2021**, 22, 960-968	3.6	2
91	PhenoDB, GeneMatcher and VariantMatcher, tools for analysis and sharing of sequence data. *Orphanet Journal of Rare Diseases*, **2021**, 16, 365	4.2	5
90	Deep clinicopathological phenotyping identifies a previously unrecognized pathogenic EMD splice variant. *Annals of Clinical and Translational Neurology*, **2021**, 8, 2052-2058	5.3	
89	High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population. *American Journal of Human Genetics*, **2021**, 108, 1981-2005	11	4
88	Response to Biesecker et al. *American Journal of Human Genetics*, **2021**, 108, 1807-1808	11	0
87	missense mutations in Xia-Gibbs syndrome.. *Human Genetics and Genomics Advances*, **2021**, 2,	0.8	1
86	Multilocus inheritance and variable disease expressivity in rare disease **2021**, 185-204		
85	Clinical presentation and evolution of Xia-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	2.5	3
84	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	5.3	15
83	Wolff-Parkinson-White 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	2.5	5
82	Basic concepts of genetics and genomics **2020**, 9-19		
81	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	9.9	6
80	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. *Human Mutation*, **2020**, 41, 487-501	4.7	24
79	TBX6 missense variants expand the mutational spectrum in a non-Mendelian inheritance disease. *Human Mutation*, **2020**, 41, 182-195	4.7	14
78	Deficiencies in vesicular transport mediated by TRAPPC4 are associated with severe syndromic intellectual disability. *Brain*, **2020**, 143, 112-130	11.2	19
77	Phenotypic expansion of POGZ-related intellectual disability syndrome (White-Sutton syndrome). *American Journal of Medical Genetics, Part A*, **2020**, 182, 38-52	2.5	19
76	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	4.7	14

75	Genetic and molecular mechanism for distinct clinical phenotypes conveyed by allelic truncating mutations implicated in FBN1. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1023	2.3	11
74	Biallelic in-frame deletion in TRAPPC4 in a family with developmental delay and cerebellar atrophy. *Brain*, **2020**, 143, e83	11.2	4
73	Congenital diaphragmatic hernia as a prominent feature of a SPECC1L-related syndrome. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2919-2925	2.5	2
72	TSC1 Variant Associated With Mild or Absent Clinical Features of Tuberous Sclerosis Complex in a Three-Generation Family. *Pediatric Neurology*, **2020**, 110, 89-91	2.9	2
71	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	8.1	11
70	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. *Genetics in Medicine*, **2020**, 22, 1863-1873	8.1	9
69	Missed diagnoses: Clinically relevant lessons learned through medical mysteries solved by the Undiagnosed Diseases Network. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1397	2.3	5
68	Phenotypic expansion in KIF1A-related dominant disorders: A description of novel variants and review of published cases. *Human Mutation*, **2020**, 41, 2094-2104	4.7	1
67	Clinical genomics and contextualizing genome variation in the diagnostic laboratory. *Expert Review of Molecular Diagnostics*, **2020**, 20, 995-1002	3.8	7
66	NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. *Nature Communications*, **2020**, 11, 4625	17.4	21
65	Integrated sequencing and array comparative genomic hybridization in familial Parkinson disease. *Neurology: Genetics*, **2020**, 6, e498	3.8	5
64	Exome sequencing reveals a novel variant in causing intracranial aneurysm in a Chinese family. *Journal of NeuroInterventional Surgery*, **2020**, 12, 221-226	7.8	4
63	Biallelic GRM7 variants cause epilepsy, microcephaly, and cerebral atrophy. *Annals of Clinical and Translational Neurology*, **2020**, 7, 610-627	5.3	5
62	Introduction to Human Genetics **2019**, 1-17		
61	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	5.6	30
60	Reanalysis of Clinical Exome Sequencing Data. *New England Journal of Medicine*, **2019**, 380, 2478-2480	59.2	109
59	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	14.4	27
58	De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smith-Magenis syndrome. *Genome Medicine*, **2019**, 11, 12	14.4	7

57	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. *Genetics in Medicine*, **2019**, 21, 161-172	8.1	36
56	Biallelic and De Novo Variants in DONSON Reveal a Clinical Spectrum of Cell Cycle-opathies with Microcephaly, Dwarfism and Skeletal Abnormalities. *American Journal of Medical Genetics, Part A*, **2019**, 179, 2056-2066	2.5	8
55	Novel Heterozygous Mutation in 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	3.4	9
54	Biallelic CACNA2D2 variants in epileptic encephalopathy and cerebellar atrophy. *Annals of Clinical and Translational Neurology*, **2019**, 6, 1395-1406	5.3	10
53	Genome sequencing and implications for rare disorders. *Orphanet Journal of Rare Diseases*, **2019**, 14, 153	4.2	40
52	The Genomics of Arthrogryposis, a Complex Trait: Candidate Genes and Further Evidence for Oligogenic Inheritance. *American Journal of Human Genetics*, **2019**, 105, 132-150	11	50
51	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. *American Journal of Human Genetics*, **2019**, 105, 302-316	11	19
50	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	11	13
49	A Genocentric Approach to Discovery of Mendelian Disorders. *American Journal of Human Genetics*, **2019**, 105, 974-986	11	18
48	Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. *American Journal of Human Genetics*, **2019**, 105, 1005-1015	11	20
47	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	11	17
46	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. *American Journal of Human Genetics*, **2019**, 104, 422-438	11	10
45	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. *Genetics in Medicine*, **2019**, 21, 663-675	8.1	31
44	Insights into genetics, human biology and disease gleaned from family based genomic studies. *Genetics in Medicine*, **2019**, 21, 798-812	8.1	100
43	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	8.1	36
42	Missense variants in TAF1 and developmental phenotypes: challenges of determining pathogenicity. *Human Mutation*, **2019**, 41, 449	4.7	7
41	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	2.5	9
40	The role of FREM2 and FRAS1 in the development of congenital diaphragmatic hernia. *Human Molecular Genetics*, **2018**, 27, 2064-2075	5.6	9

39	The phenotypic spectrum of Xia-Gibbs syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1315-1326	2.5	25
38	Phenotypic expansion illuminates multilocus pathogenic variation. *Genetics in Medicine*, **2018**, 20, 1528-1537	8.3	71
37	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	11	26
36	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**, 7, 164-173	0.7	7
35	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	6.3	13
34	IRF2BPL Is Associated with Neurological Phenotypes. *American Journal of Human Genetics*, **2018**, 103, 245-260	11	39
33	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	6.3	33
32	Perturbations of BMP/TGF-□and VEGF/VEGFR signalling pathways in non-syndromic sporadic brain arteriovenous malformations (BAVM). *Journal of Medical Genetics*, **2018**, 55, 675-684	5.8	38
31	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	2.5	4
30	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	11	10
29	Phenotypic expansion in - a common cause of intellectual disability in females. *Annals of Clinical and Translational Neurology*, **2018**, 5, 1277-1285	5.3	37
28	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	3.7	8
27	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. *American Journal of Human Genetics*, **2017**, 100, 185-192	11	102
26	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	11	23
25	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	11	104
24	22q11.2q13 duplication including SOX10 causes sex-reversal and peripheral demyelinating neuropathy, central dysmyelinating leukodystrophy, Waardenburg syndrome, and Hirschsprung disease. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1066-1070	2.5	18
23	Lessons learned from additional research analyses of unsolved clinical exome cases. *Genome Medicine*, **2017**, 9, 26	14.4	125
22	Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. *New England Journal of Medicine*, **2017**, 376, 21-31	59.2	391

21	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. *American Journal of Human Genetics*, **2017**, 100, 128-137	11	65
20	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. *American Journal of Human Genetics*, **2017**, 101, 716-724	11	38
19	Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders. *Genome Medicine*, **2017**, 9, 73	14.4	30
18	Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research. *Genetics*, **2017**, 207, 9-27	4	99
17	Dominant Transmission Observed in Adolescents and Families With Orthostatic Intolerance. *Pediatric Neurology*, **2017**, 66, 53-58.e5	2.9	4
16	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	11	113
15	Exome sequencing in mostly consanguineous Arab families with neurologic disease provides a high potential molecular diagnosis rate. *BMC Medical Genomics*, **2016**, 9, 42	3.7	58
14	POGZ truncating alleles cause syndromic intellectual disability. *Genome Medicine*, **2016**, 8, 3	14.4	49
13	A visual and curatorial approach to clinical variant prioritization and disease gene discovery in genome-wide diagnostics. *Genome Medicine*, **2016**, 8, 13	14.4	27
12	Mechanisms for the Generation of Two Quadruplications Associated with Split-Hand Malformation. *Human Mutation*, **2016**, 37, 160-4	4.7	13
11	Molecular diagnostic experience of whole-exome sequencing in adult patients. *Genetics in Medicine*, **2016**, 18, 678-85	8.1	149
10	Triploidy mosaicism (45,X/68,XX) in an infant presenting with failure to thrive. *American Journal of Medical Genetics, Part A*, **2016**, 170, 694-8	2.5	6
9	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. *American Journal of Human Genetics*, **2016**, 99, 991-999	11	42
8	Hutterite-type cataract maps to chromosome 6p21.32-p21.31, cosegregates with a homozygous mutation in LEMD2, and is associated with sudden cardiac death. *Molecular Genetics & Genomic Medicine*, **2016**, 4, 77-94	2.3	16
7	Atypical presentation of moyamoya disease in an infant with a de novo RNF213 variant. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2742-7	2.5	13
6	Adult presentation of X-linked Conradi-H□ermann-Happle syndrome. *American Journal of Medical Genetics, Part A*, **2015**, 167, 1309-14	2.5	5
5	Lysinuric Protein Intolerance Presenting with Multiple Fractures. *Molecular Genetics and Metabolism Reports*, **2014**, 1, 176-183	1.8	15
4	Syngnathia and obstructive apnea in a case of popliteal pterygium syndrome. *European Journal of Pediatrics*, **2014**, 173, 1741-4	4.1	7

3	Understanding how the V(D)J recombinase catalyzes transesterification: distinctions between DNA cleavage and transposition. *Nucleic Acids Research*, **2008**, 36, 2864-73	20.1	7
2	Target DNA structure plays a critical role in RAG transposition. *PLoS Biology*, **2006**, 4, e350	9.7	22
1	De novomutation in ancestral generations evolves haplotypes contributing to disease		2