Jill A Rosenfeld

List of Publications by Year
in descending order

322	19,864	14655 66	18130 120
papers	citations	h-index	g-index

342	342	342	25886
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A copy number variation morbidity map of developmental delay. Nature Genetics, 2011, 43, 838-846.	21.4	1,141
2	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. Cell, 2014, 158, 263-276.	28.9	637
3	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	21.4	583
4	Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. New England Journal of Medicine, 2017, 376, 21-31.	27.0	565
5	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. Nature Genetics, 2010, 42, 203-209.	21.4	539
6	Recurrent reciprocal 1q21.1 deletions and duplications associated with microcephaly or macrocephaly and developmental and behavioral abnormalities. Nature Genetics, 2008, 40, 1466-1471.	21.4	535
7	Phenotypic Heterogeneity of Genomic Disorders and Rare Copy-Number Variants. New England Journal of Medicine, 2012, 367, 1321-1331.	27.0	519
8	A Higher Mutational Burden in Females Supports a â€œFemale Protective Modelâ€•in Neurodevelopmental Disorders. American Journal of Human Genetics, 2014, 94, 415-425.	6.2	457
9	Recurrent reciprocal 16p11.2 rearrangements associated with global developmental delay, behavioural problems, dysmorphism, epilepsy, and abnormal head size. Journal of Medical Genetics, 2010, 47, 332-341.	3.2	447
10	Evolution of Human-Specific Neural SRGAP2 Genes by Incomplete Segmental Duplication. Cell, 2012, 149, 912-922.	28.9	341
11	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
12	Paternally inherited microdeletion at 15q11.2 confirms a significant role for the SNORD116 C/D box snoRNA cluster in Praderâ€“Willi syndrome. European Journal of Human Genetics, 2010, 18, 1196-1201.	2.8	287
13	Estimates of penetrance for recurrent pathogenic copy-number variations. Genetics in Medicine, 2013, 15, 478-481.	2.4	277
14	TBX6 Null Variants and a Common Hypomorphic Allele in Congenital Scoliosis. New England Journal of Medicine, 2015, 372, 341-350.	27.0	239
15	Detection rates of clinically significant genomic alterations by microarray analysis for specific anomalies detected by ultrasound. Prenatal Diagnosis, 2012, 32, 986-995.	2.3	222
16	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	27.0	205
17	Assessment of 2q23.1 Microdeletion Syndrome Implicates MBD5 as a Single Causal Locus of Intellectual Disability, Epilepsy, and Autism Spectrum Disorder. American Journal of Human Genetics, 2011, 89, 551-563.	6.2	195
18	Molecular diagnostic experience of whole-exome sequencing in adult patients. Genetics in Medicine, 2016, 18, 678-685.	2.4	186

#	Article	IF	Citations
19	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	8.2	184
20	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.	6.2	181
21	Recurrent 200-kb deletions of 16p11.2 that include the SH2B1 gene are associated with developmental delay and obesity. Genetics in Medicine, 2010, 12, 641-647.	2.4	178
22	Experience with microarrayâ€based comparative genomic hybridization for prenatal diagnosis in over 5000 pregnancies. Prenatal Diagnosis, 2012, 32, 976-985.	2.3	178
23	CpG Island Hypermethylation Mediated by DNMT3A Is a Consequence of AML Progression. Cell, 2017, 168, 801-816.e13.	28.9	177
24	The array CGH and its clinical applications. Drug Discovery Today, 2008, 13, 760-770.	6.4	171
25	A small recurrent deletion within 15q13.3 is associated with a range of neurodevelopmental phenotypes. Nature Genetics, 2009, 41, 1269-1271.	21.4	171
26	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	2.4	161
27	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
28	Speech delays and behavioral problems are the predominant features in individuals with developmental delays and 16p11.2 microdeletions and microduplications. Journal of Neurodevelopmental Disorders, 2010, 2, 26-38.	3.1	147
29	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.	6.2	146
30	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	6.2	142
31	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
32	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. Genetics in Medicine, 2018, 20, 1175-1185.	2.4	133
33	Heterozygous Truncating Variants in POMP Escape Nonsense-Mediated Decay and Cause a Unique Immune Dysregulatory Syndrome. American Journal of Human Genetics, 2018, 102, 1126-1142.	6.2	128
34	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. Genetics in Medicine, 2019, 21, 816-825.	2.4	127
35	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. American Journal of Human Genetics, 2017, 100, 907-925.	6.2	125
36	NAHR-mediated copy-number variants in a clinical population: Mechanistic insights into both genomic disorders and Mendelizing traits. Genome Research, 2013, 23, 1395-1409.	5.5	120

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37	Copy number variations associated with autism spectrum disorders contribute to a spectrum of neurodevelopmental disorders. Genetics in Medicine, 2010, 12, 694-702.	2.4	116
38	Structures and molecular mechanisms for common 15q13.3 microduplications involving CHRNA7: benign or pathological?. Human Mutation, 2010, 31, 840-850.	2.5	111
39	De Novo Mutations in CHD4 , an ATP-Dependent Chromatin Remodeler Gene, Cause an Intellectual Disability Syndrome with Distinctive Dysmorphisms. American Journal of Human Genetics, 2016, 99, 934-941.	6.2	111
40	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	108
41	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	12.8	105
42	A Mild PUM1 Mutation Is Associated with Adult-Onset Ataxia, whereas Haploinsufficiency Causes Developmental Delay and Seizures. Cell, 2018, 172, 924-936.e11.	28.9	103
43	Germline De Novo Mutations in GNB1 Cause Severe Neurodevelopmental Disability, Hypotonia, and Seizures. American Journal of Human Genetics, 2016, 98, 1001-1010.	6.2	102
44	Copy number variants of schizophrenia susceptibility loci are associated with a spectrum of speech and developmental delays and behavior problems. Genetics in Medicine, 2011, 13, 868-880.	2.4	98
45	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. American Journal of Human Genetics, 2016, 98, 347-357.	6.2	98
46	TBC1D24 genotypeâ€“phenotype correlation. Neurology, 2016, 87, 77-85.	1.1	97
47	Palindromic GOLGA8 core duplicons promote chromosome 15q13.3 microdeletion and evolutionary instability. Nature Genetics, 2014, 46, 1293-1302.	21.4	96
48	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137.	6.2	96
49	Investigation of NRXN1 deletions: Clinical and molecular characterization. American Journal of Medical Genetics, Part A, 2013, 161, 717-731.	1.2	94
50	The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. Genetics in Medicine, 2017, 19, 45-52.	2.4	94
51	Small Deletions of SATB2 Cause Some of the Clinical Features of the 2q33.1 Microdeletion Syndrome. PLoS ONE, 2009, 4, e6568.	2.5	94
52	An assessment of sex bias in neurodevelopmental disorders. Genome Medicine, 2015, 7, 94.	8.2	90
53	A recurrent p.Arg92Trp variant in steroidogenic factor-1 (NR5A1) can act as a molecular switch in human sex development. Human Molecular Genetics, 2016, 25, 3446-3453.	2.9	90
54	A large and complex structural polymorphism at 16p12.1 underlies microdeletion disease risk. Nature Genetics, 2010, 42, 745-750.	21.4	89

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55	MEF2C Haploinsufficiency features consistent hyperkinesis, variable epilepsy, and has a role in dorsal and ventral neuronal developmental pathways. Neurogenetics, 2013, 14, 99-111.	1.4	89
56	Diagnostic testing for uniparental disomy: a points to consider statement from the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 1133-1141.	2.4	89
57	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. American Journal of Human Genetics, 2018, 102, 27-43.	6.2	88
58	Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing. Journal of Clinical Investigation, 2021, 131, .	8.2	87
59	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	6.2	86
60	Haploinsufficiency ofSOX5at 12p12.1 is associated with developmental delays with prominent language delay, behavior problems, and mild dysmorphic features. Human Mutation, 2012, 33, 728-740.	2.5	85
61	The Exome Clinic and the role of medical genetics expertise in the interpretation of exome sequencing results. Genetics in Medicine, 2017, 19, 1040-1048.	2.4	85
62	11p14.1 microdeletions associated with ADHD, autism, developmental delay, and obesity. American Journal of Medical Genetics, Part A, 2011, 155, 1272-1280.	1.2	84
63	Proximal microdeletions and microduplications of 1q21.1 contribute to variable abnormal phenotypes. European Journal of Human Genetics, 2012, 20, 754-761.	2.8	84
64	Exome sequencing in mostly consanguineous Arab families with neurologic disease provides a high potential molecular diagnosis rate. BMC Medical Genomics, 2016, 9, 42.	1.5	80
65	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. PLoS Genetics, 2017, 13, e1006905.	3.5	80
66	POGZ truncating alleles cause syndromic intellectual disability. Genome Medicine, 2016, 8, 3.	8.2	78
67	Syndromic thrombocytopenia and predisposition to acute myelogenous leukemia caused by constitutional microdeletions on chromosome 21q. Blood, 2008, 112, 1042-1047.	1.4	74
68	Copy number gain at Xp22.31 includes complex duplication rearrangements and recurrent triplications. Human Molecular Genetics, 2011, 20, 1975-1988.	2.9	74
69	NUDT21-spanning CNVs lead to neuropsychiatric disease and altered MeCP2 abundance via alternative polyadenylation. ELife, 2015, 4, .	6.0	74
70	Novel features of 3q29 deletion syndrome: Results from the 3q29 registry. American Journal of Medical Genetics, Part A, 2016, 170, 999-1006.	1.2	73
71	Whole-exome sequencing in the molecular diagnosis of individuals with congenital anomalies of the kidney and urinary tract and identification of a new causative gene. Genetics in Medicine, 2017, 19, 412-420.	2.4	73
72	Mutations in the Chromatin Regulator Gene BRPF1 Cause Syndromic Intellectual Disability and Deficient Histone Acetylation. American Journal of Human Genetics, 2017, 100, 91-104.	6.2	72

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73	DNA Polymerase Epsilon Deficiency Causes IMAGe Syndrome with Variable Immunodeficiency. American Journal of Human Genetics, 2018, 103, 1038-1044.	6.2	71
74	Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. Neuron, 2020, 106, 589-606.e6.	8.1	71
75	TSHZ3 deletion causes an autism syndrome and defects in cortical projection neurons. Nature Genetics, 2016, 48, 1359-1369.	21.4	69
76	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
77	The Cognitive and Behavioral Phenotypes of Individuals with CHRNA7 Duplications. Journal of Autism and Developmental Disorders, 2017, 47, 549-562.	2.7	68
78	Neuroligin 2 nonsense variant associated with anxiety, autism, intellectual disability, hyperphagia, and obesity. American Journal of Medical Genetics, Part A, 2017, 173, 213-216.	1.2	68
79	De Novo Mutations of RERE Cause a Genetic Syndrome with Features that Overlap Those Associated with Proximal 1p36 Deletions. American Journal of Human Genetics, 2016, 98, 963-970.	6.2	67
80	De novo mutation screening in childhood-onset cerebellar atrophy identifies gain-of-function mutations in the CACNA1G calcium channel gene. Brain, 2018, 141, 1998-2013.	7.6	67
81	Monoallelic and Biallelic Variants in EMC1 Identified in Individuals with Global Developmental Delay, Hypotonia, Scoliosis, and Cerebellar Atrophy. American Journal of Human Genetics, 2016, 98, 562-570.	6.2	66
82	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 101, 716-724.	6.2	66
83	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.	3.7	66
84	De Novo GMNN Mutations Cause Autosomal-Dominant Primordial Dwarfism Associated with Meier-Gorlin Syndrome. American Journal of Human Genetics, 2015, 97, 904-913.	6.2	65
85	The expanding clinical phenotype of Bosch-Boonstra-Schaaf optic atrophy syndrome: 20 new cases and possible genotypeâ€“phenotype correlations. Genetics in Medicine, 2016, 18, 1143-1150.	2.4	64
86	Dominant-negative mutations in human IL6ST underlie hyper-IgE syndrome. Journal of Experimental Medicine, 2020, 217, .	8.5	64
87	Microdeletion of 6q16.1 encompassing EPHA7 in a child with mild neurological abnormalities and dysmorphic features: case report. Molecular Cytogenetics, 2009, 2, 17.	0.9	63
88	Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. American Journal of Human Genetics, 2017, 101, 503-515.	6.2	61
89	Genomic and genetic variation inÂE2FÂtranscription factor-1 in men withÂnonobstructive azoospermia. Fertility and Sterility, 2015, 103, 44-52.e1.	1.0	59
90	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.	6.2	59

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91	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. American Journal of Human Genetics, 2019, 104, 164-178.	6.2	59
92	Bi-allelic Mutations in PKD1L1 Are Associated with Laterality Defects in Humans. American Journal of Human Genetics, 2016, 99, 886-893.	6.2	57
93	De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. American Journal of Human Genetics, 2018, 103, 154-162.	6.2	56
94	Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. American Journal of Human Genetics, 2020, 106, 234-245.	6.2	56
95	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. American Journal of Human Genetics, 2017, 100, 676-688.	6.2	54
96	The duplication 17p13.3 phenotype: Analysis of 21 families delineates developmental, behavioral and brain abnormalities, and rare variant phenotypes. American Journal of Medical Genetics, Part A, 2013, 161, 1833-1852.	1.2	53
97	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	2.4	52
98	Diagnostic utility of microarray testing in pregnancy loss. Ultrasound in Obstetrics and Gynecology, 2015, 46, 478-486.	1.7	50
99	Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83.	8.2	50
100	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	8.2	50
101	A Founder Mutation in VPS11 Causes an Autosomal Recessive Leukoencephalopathy Linked to Autophagic Defects. PLoS Genetics, 2016, 12, e1005848.	3.5	50
102	Mutations in GPAA1 , Encoding a GPI Transamidase Complex Protein, Cause Developmental Delay, Epilepsy, Cerebellar Atrophy, and Osteopenia. American Journal of Human Genetics, 2017, 101, 856-865.	6.2	49
103	Mutations inCOQ4, an essential component of coenzyme Q biosynthesis, cause lethal neonatal mitochondrial encephalomyopathy. Journal of Medical Genetics, 2015, 52, 627-635.	3.2	48
104	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 103, 305-316.	6.2	48
105	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. Genetics in Medicine, 2019, 21, 2723-2733.	2.4	48
106	Mutations in RABL3 alter KRAS prenylation and are associated with hereditary pancreatic cancer. Nature Genetics, 2019, 51, 1308-1314.	21.4	47
107	Subtelomeric deletion of chromosome 10p15.3: Clinical findings and molecular cytogenetic characterization. American Journal of Medical Genetics, Part A, 2012, 158A, 2152-2161.	1.2	45
108	Recurrent deletions and reciprocal duplications of 10q11.21q11.23 including CHAT and SLC18A3 are likely mediated by complex low-copy repeats. Human Mutation, 2012, 33, 165-179.	2.5	45

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109	The complex behavioral phenotype of 15q13.3 microdeletion syndrome. Genetics in Medicine, 2016, 18, 1111-1118.	2.4	45
110	De novo and inherited mutations in the X-linked gene CLCN4 are associated with syndromic intellectual disability and behavior and seizure disorders in males and females. Molecular Psychiatry, 2018, 23, 222-230.	7.9	45
111	De novo substitutions of TRPM3 cause intellectual disability and epilepsy. European Journal of Human Genetics, 2019, 27, 1611-1618.	2.8	45
112	Mutations in PIGS, Encoding a GPI Transamidase, Cause a Neurological Syndrome Ranging from Fetal Akinesia to Epileptic Encephalopathy. American Journal of Human Genetics, 2018, 103, 602-611.	6.2	44
113	Review of the phenotypic spectrum associated with haploinsufficiency of MYRF. American Journal of Medical Genetics, Part A, 2019, 179, 1376-1382.	1.2	44
114	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	2.4	44
115	TM4SF20 Ancestral Deletion and Susceptibility to a Pediatric Disorder of Early Language Delay and Cerebral White Matter Hyperintensities. American Journal of Human Genetics, 2013, 93, 197-210.	6.2	43
116	MIPEP recessive variants cause a syndrome of left ventricular non-compaction, hypotonia, and infantile death. Genome Medicine, 2016, 8, 106.	8.2	43
117	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	12.8	43
118	Distinctive phenotype in 9 patients with deletion of chromosome 1q24â€q25. American Journal of Medical Genetics, Part A, 2011, 155, 1336-1351.	1.2	42
119	Reciprocal deletion and duplication at 2q23.1 indicates a role for MBD5 in autism spectrum disorder. European Journal of Human Genetics, 2014, 22, 57-63.	2.8	42
120	The spectrum of DNMT3A variants in Tattonâ€“Brownâ€“Rahman syndrome overlaps with that in hematologic malignancies. American Journal of Medical Genetics, Part A, 2017, 173, 3022-3028.	1.2	42
121	KCTD7 deficiency defines a distinct neurodegenerative disorder with a conserved autophagyâ€lysosome defect. Annals of Neurology, 2018, 84, 766-780.	5.3	42
122	Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing. Journal of Genetic Counseling, 2019, 28, 1107-1118.	1.6	42
123	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
124	WACloss-of-function mutations cause a recognisable syndrome characterised by dysmorphic features, developmental delay and hypotonia and recapitulate 10p11.23 microdeletion syndrome. Journal of Medical Genetics, 2015, 52, 754-761.	3.2	41
125	A Novel Mutation in Isoform 3 of the Plasma Membrane Ca2+ Pump Impairs Cellular Ca2+ Homeostasis in a Patient with Cerebellar Ataxia and Laminin Subunit 1Î± Mutations. Journal of Biological Chemistry, 2015, 290, 16132-16141.	3.4	41
126	Pathogenic variants in USP7 cause a neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies. Genetics in Medicine, 2019, 21, 1797-1807.	2.4	41

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127	Further clinical and molecular delineation of the 15q24 microdeletion syndrome. Journal of Medical Genetics, 2012, 49, 110-118.	3.2	40
128	Recurrent HERV-H-Mediated 3q13.2-q13.31 Deletions Cause a Syndrome of Hypotonia and Motor, Language, and Cognitive Delays. Human Mutation, 2013, 34, 1415-1423.	2.5	40
129	NR2F1 haploinsufficiency is associated with optic atrophy, dysmorphism and global developmental delay. American Journal of Medical Genetics, Part A, 2013, 161, 377-381.	1.2	40
130	Loss-of-Function Mutations in FRRS1L Lead to an Epileptic-Dyskinetic Encephalopathy. American Journal of Human Genetics, 2016, 98, 1249-1255.	6.2	40
131	De Novo Truncating Mutations in the Kinetochore-Microtubules Attachment GeneCHAMP1Cause Syndromic Intellectual Disability. Human Mutation, 2016, 37, 354-358.	2.5	40
132	Germline mutations in ABL1 cause an autosomal dominant syndrome characterized by congenital heart defects and skeletal malformations. Nature Genetics, 2017, 49, 613-617.	21.4	40
133	Further Evidence of Contrasting Phenotypes Caused by Reciprocal Deletions and Duplications: Duplication of NSD1 Causes Growth Retardation and Microcephaly. Molecular Syndromology, 2012, 3, 247-254.	0.8	39
134	Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders. Genome Medicine, 2017, 9, 73.	8.2	39
135	Targeted knockout of a chemokine-like gene increases anxiety and fear responses. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E1041-E1050.	7.1	39
136	Whole exome sequencing in 342 congenital cardiac left sided lesion cases reveals extensive genetic heterogeneity and complex inheritance patterns. Genome Medicine, 2017, 9, 95.	8.2	37
137	Mutations in PIGB Cause an Inherited GPI Biosynthesis Defect with an Axonal Neuropathy and Metabolic Abnormality in Severe Cases. American Journal of Human Genetics, 2019, 105, 384-394.	6.2	37
138	Haploinsufficiency of the E3 ubiquitin-protein ligase gene TRIP12 causes intellectual disability with or without autism spectrum disorders, speech delay, and dysmorphic features. Human Genetics, 2017, 136, 377-386.	3.8	36
139	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
140	Interpreting Incidentally Identified Variants in Genes Associated With Catecholaminergic Polymorphic Ventricular Tachycardia in a Large Cohort of Clinical Whole-Exome Genetic Test Referrals. Circulation: Arrhythmia and Electrophysiology, 2017, 10, .	4.8	35
141	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	10.3	35
142	Phenotypic expansion of POGZâ€related intellectual disability syndrome (Whiteâ€Sutton)	1.2	35
143	Lowâ€level mosaicism of trisomy 14: Phenotypic and molecular characterization. American Journal of Medical Genetics, Part A, 2008, 146A, 1395-1405.	1.2	34
144	Gain-of-Function Mutations inRARBCause Intellectual Disability with Progressive Motor Impairment. Human Mutation, 2016, 37, 786-793.	2.5	34

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145	Genotype-phenotype correlations in individuals with pathogenicREREvariants. Human Mutation, 2018, 39, 666-675.	2.5	34
146	Loss of Oxidation Resistance 1, OXR1, Is Associated with an Autosomal-Recessive Neurological Disease with Cerebellar Atrophy and Lysosomal Dysfunction. American Journal of Human Genetics, 2019, 105, 1237-1253.	6.2	34
147	Deficiencies in vesicular transport mediated by TRAPPC4 are associated with severe syndromic intellectual disability. Brain, 2020, 143, 112-130.	7.6	33
148	Bi-allelic variants in HOPS complex subunit VPS41 cause cerebellar ataxia and abnormal membrane trafficking. Brain, 2021, 144, 769-780.	7.6	33
149	Referral patterns for microarray testing in prenatal diagnosis. Prenatal Diagnosis, 2012, 32, 344-350.	2.3	32
150	A recurrent de novo CTBP1 mutation is associated with developmental delay, hypotonia, ataxia, and tooth enamel defects. Neurogenetics, 2016, 17, 173-178.	1.4	32
151	FHF1 (FGF12) epileptic encephalopathy. Neurology: Genetics, 2016, 2, e115.	1.9	32
152	Biallelic mutations in the ferredoxin reductase gene cause novel mitochondriopathy with optic atrophy. Human Molecular Genetics, 2017, 26, 4937-4950.	2.9	32
153	BICRA, a SWI/SNF Complex Member, Is Associated with BAF-Disorder Related Phenotypes in Humans and Model Organisms. American Journal of Human Genetics, 2020, 107, 1096-1112.	6.2	32
154	De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. American Journal of Human Genetics, 2020, 107, 311-324.	6.2	32
155	Two novel RAD21 mutations in patients with mild Cornelia de Lange syndrome-like presentation and report of the first familial case. Gene, 2014, 537, 279-284.	2.2	31
156	A Genocentric Approach to Discovery of Mendelian Disorders. American Journal of Human Genetics, 2019, 105, 974-986.	6.2	30
157	Disruption of PHF21A causes syndromic intellectual disability with craniofacial anomalies, epilepsy, hypotonia, and neurobehavioral problems including autism. Molecular Autism, 2019, 10, 35.	4.9	30
158	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
159	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	6.2	30
160	A homozygous deletion of 8q24.3 including the NIBP gene associated with severe developmental delay, dysgenesis of the corpus callosum, and dysmorphic facial features. American Journal of Medical Genetics, Part A, 2010, 152A, 1268-1272.	1.2	29
161	PIAS4 is associated with macro/microcephaly in the novel interstitial 19p13.3 microdeletion/microduplication syndrome. European Journal of Human Genetics, 2015, 23, 1615-1626.	2.8	29
162	Gain-of-function mutations in RPA1 cause a syndrome with short telomeres and somatic genetic rescue. Blood, 2022, 139, 1039-1051.	1.4	29

#	Article	IF	Citations
163	IncreasedSTAG2dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. Human Molecular Genetics, 2015, 24, 7171-7181.	2.9	28
164	Variants in MED12L, encoding a subunit of the mediator kinase module, are responsible for intellectual disability associated with transcriptional defect. Genetics in Medicine, 2019, 21, 2713-2722.	2.4	28
165	Heterozygous variants inACTL6A, encoding a component of the BAF complex, are associated with intellectual disability. Human Mutation, 2017, 38, 1365-1371.	2.5	27
166	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
167	Clinical exome sequencing data reveal high diagnostic yields for congenital diaphragmatic hernia plus (CDH+) and new phenotypic expansions involving CDH. Journal of Medical Genetics, 2022, 59, 270-278.	3.2	27
168	Contribution of genomic copy-number variations in prenatal oral clefts: a multicenter cohort study. Genetics in Medicine, 2016, 18, 1052-1055.	2.4	25
169	Phenotypic and molecular characterization of 19q12q13.1 deletions: A report of five patients. American Journal of Medical Genetics, Part A, 2014, 164, 62-69.	1.2	24
170	Chromosomal Imbalances in Patients with Congenital Cardiac Defects: A Meta-analysis Reveals Novel Potential Critical Regions Involved in Heart Development. Congenital Heart Disease, 2015, 10, 193-208.	0.2	24
171	Congenital heart defects and left ventricular non-compaction in males with loss-of-function variants inNONO. Journal of Medical Genetics, 2017, 54, 47-53.	3.2	24
172	Chromosomal Microarrays: Understanding Genetics of Neurodevelopmental Disorders and Congenital Anomalies. Journal of Pediatric Genetics, 2017, 06, 042-050.	0.7	24
173	Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. American Journal of Human Genetics, 2019, 105, 1005-1015.	6.2	24
174	Predominant and novel de novo variants in 29 individuals with <scp>ALG13</scp> deficiency: Clinical description, biomarker status, biochemical analysis, and treatment suggestions. Journal of Inherited Metabolic Disease, 2020, 43, 1333-1348.	3.6	24
175	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. Human Genetics, 2020, 139, 575-592.	3.8	24
176	Loss-of-function and missense variants in NSD2 cause decreased methylation activity and are associated with a distinct developmental phenotype. Genetics in Medicine, 2021, 23, 1474-1483.	2.4	24
177	Drosophila functional screening of de novo variants in autism uncovers damaging variants and facilitates discovery of rare neurodevelopmental diseases. Cell Reports, 2022, 38, 110517.	6.4	24
178	Mixed gonadal dysgenesis in a child with isodicentric y chromosome: Does the relative proportion of the 45,X line really matter?. American Journal of Medical Genetics, Part A, 2010, 152A, 1832-1837.	1.2	23
179	A Recurrent De Novo Nonsense Variant in ZSWIM6 Results in Severe Intellectual Disability without Frontonasal or Limb Malformations. American Journal of Human Genetics, 2017, 101, 995-1005.	6.2	23
180	Gnathodiaphyseal dysplasia: Severe atypical presentation with novel heterozygous mutation of the anoctamin gene (ANO5). Bone, 2018, 107, 161-171.	2.9	23

#	Article	IF	Citations
181	Variable cardiovascular phenotypes associated with SMAD2 pathogenic variants. Human Mutation, 2018, 39, 1875-1884.	2.5	23
182	2-Pyrrolidinone and Succinimide as Clinical Screening Biomarkers for GABA-Transaminase Deficiency: Anti-seizure Medications Impact Accurate Diagnosis. Frontiers in Neuroscience, 2019, 13, 394.	2.8	23
183	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.	8.2	23
184	Pathogenic Variants in GPC4 Cause Keipert Syndrome. American Journal of Human Genetics, 2019, 104, 914-924.	6.2	23
185	Variants in TCF20 in neurodevelopmental disability: description of 27 new patients and review of literature. Genetics in Medicine, 2019, 21, 2036-2042.	2.4	23
186	A Recurrent Gain-of-Function Mutation in CLCN6, Encoding the ClC-6 Clâ¯/H+-Exchanger, Causes Early-Onset Neurodegeneration. American Journal of Human Genetics, 2020, 107, 1062-1077.	6.2	23
187	De Novo Variants in CDK19 Are Associated with a Syndrome Involving Intellectual Disability and Epileptic Encephalopathy. American Journal of Human Genetics, 2020, 106, 717-725.	6.2	23
188	Dysfunction of the Cerebral Glucose Transporter SLC45A1 in Individuals with Intellectual Disability and Epilepsy. American Journal of Human Genetics, 2017, 100, 824-830.	6.2	22
189	6q25.1 (TAB2) microdeletion syndrome: Congenital heart defects and cardiomyopathy. American Journal of Medical Genetics, Part A, 2017, 173, 1848-1857.	1.2	22
190	Evidence for secondary-variant genetic burden and non-random distribution across biological modules in a recessive ciliopathy. Nature Genetics, 2020, 52, 1145-1150.	21.4	22
191	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	2.4	22
192	Estimating the relative frequency of leukodystrophies and recommendations for carrier screening in the era of nextâ€generation sequencing. American Journal of Medical Genetics, Part A, 2020, 182, 1906-1912.	1.2	22
193	Bi-allelic Variants in the GPI Transamidase Subunit PIGK Cause a Neurodevelopmental Syndrome with Hypotonia, Cerebellar Atrophy, and Epilepsy. American Journal of Human Genetics, 2020, 106, 484-495.	6.2	22
194	Multigenerational autosomal dominant inheritance of 5p chromosomal deletions. American Journal of Medical Genetics, Part A, 2016, 170, 583-593.	1.2	21
195	Widening of the genetic and clinical spectrum of Lambâ€“Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. Genetics in Medicine, 2020, 22, 524-537.	2.4	21
196	Disruption of NEUROD2 causes a neurodevelopmental syndrome with autistic features via cell-autonomous defects in forebrain glutamatergic neurons. Molecular Psychiatry, 2021, 26, 6125-6148.	7.9	21
197	Functional and epigenetic phenotypes of humans and mice with DNMT3A Overgrowth Syndrome. Nature Communications, 2021, 12, 4549.	12.8	21
198	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	2.4	20

#	Article	IF	Citations
199	Recognition of Smith-Lemli-Opitz syndrome (RSH) in the fetus: Utility of ultrasonography and biochemical analysis in pregnancies with low maternal serum estriol. American Journal of Medical Genetics, Part A, 2005, 138A, 56-60.	1.2	19
200	Splicing mutation in the fibrillin-1 gene associated with neonatal Marfan syndrome and severe pulmonary emphysema with tracheobronchomalacia. Pediatric Pulmonology, 2005, 39, 374-378.	2.0	19
201	The Xp contiguous deletion syndrome and autism. American Journal of Medical Genetics, Part A, 2009, 149A, 1138-1148.	1.2	19
202	Mutations in the <scp>PH</scp> Domain of <scp>DNM</scp>1 are associated with a nonepileptic phenotype characterized by developmental delay and neurobehavioral abnormalities. Molecular Genetics & Genomic Medicine, 2018, 6, 294-300.	1.2	19
203	DYRK1A-related intellectual disability: a syndrome associated with congenital anomalies of the kidney and urinary tract. Genetics in Medicine, 2019, 21, 2755-2764.	2.4	19
204	Model system identification of novel congenital heart disease gene candidates: focus on RPL13. Human Molecular Genetics, 2019, 28, 3954-3969.	2.9	19
205	De Novo Variants Disrupting the HX Repeat Motif of ATN1 Cause a Recognizable Non-Progressive Neurocognitive Syndrome. American Journal of Human Genetics, 2019, 104, 542-552.	6.2	19
206	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. Genetics in Medicine, 2020, 22, 1863-1873.	2.4	19
207	Deletions flanked by breakpoints 3 and 4 on 15q13 may contribute to abnormal phenotypes. European Journal of Human Genetics, 2011, 19, 547-554.	2.8	18
208	Pathogenic Variants in Fucokinase Cause a Congenital Disorder of Glycosylation. American Journal of Human Genetics, 2018, 103, 1030-1037.	6.2	18
209	Novel parent-of-origin-specific differentially methylated loci on chromosome 16. Clinical Epigenetics, 2019, 11, 60.	4.1	18
210	Cell-based analysis of CAD variants identifies individuals likely to benefit from uridine therapy. Genetics in Medicine, 2020, 22, 1598-1605.	2.4	18
211	COPB2 loss of function causes a coatopathy with osteoporosis and developmental delay. American Journal of Human Genetics, 2021, 108, 1710-1724.	6.2	18
212	19q13.32 microdeletion syndrome: Three new cases. European Journal of Medical Genetics, 2014, 57, 654-658.	1.3	17
213	Further delineation of the phenotypic spectrum associated with hemizygous lossâ€ofâ€function variants in NONO. American Journal of Medical Genetics, Part A, 2020, 182, 652-658.	1.2	17
214	Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. Brain, 2022, 145, 909-924.	7.6	17
215	Integrative analysis of genomic variants reveals new associations of candidate haploinsufficient genes with congenital heart disease. PLoS Genetics, 2021, 17, e1009679.	3.5	17
216	Defining the impact of maternal cell contamination on the interpretation of prenatal microarray analysis. Genetics in Medicine, 2012, 14, 914-921.	2.4	16

#	Article	IF	Citations
217	CEDNIK. Child Neurology Open, 2017, 4, 2329048X1773321.	1.1	16
218	A pathogenic CtBP1 missense mutation causes altered cofactor binding and transcriptional activity. Neurogenetics, 2019, 20, 129-143.	1.4	16
219	Missed diagnoses: Clinically relevant lessons learned through medical mysteries solved by the Undiagnosed Diseases Network. Molecular Genetics & Genomic Medicine, 2020, 8, e1397.	1.2	16
220	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. Genetics in Medicine, 2021, 23, 1922-1932.	2.4	16
221	Clinical features associated with copy number variations of the 14q32 imprinted gene cluster. American Journal of Medical Genetics, Part A, 2015, 167, 345-353.	1.2	15
222	Five patients with a chromosome 1q21.1 triplication show macrocephaly, increased weight and facial similarities. European Journal of Medical Genetics, 2015, 58, 503-508.	1.3	15
223	Inactivation of AMMECR1 is associated with growth, bone, and heart alterations. Human Mutation, 2018, 39, 281-291.	2.5	15
224	Variants in DOCK3 cause developmental delay and hypotonia. European Journal of Human Genetics, 2019, 27, 1225-1234.	2.8	15
225	Truncating variants in UBAP1 associated with childhoodâ€onset nonsyndromic hereditary spastic paraplegia. Human Mutation, 2020, 41, 632-640.	2.5	15
226	Biallelic GRM7 variants cause epilepsy, microcephaly, and cerebral atrophy. Annals of Clinical and Translational Neurology, 2020, 7, 610-627.	3.7	15
227	UBR7 functions with UBR5 in the Notch signaling pathway and is involved in a neurodevelopmental syndrome with epilepsy, ptosis, and hypothyroidism. American Journal of Human Genetics, 2021, 108, 134-147.	6.2	15
228	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. Brain, 2022, 145, 208-223.	7.6	15
229	Haploinsufficiency of the Sin3/HDAC corepressor complex member SIN3B causes a syndromic intellectual disability/autism spectrum disorder. American Journal of Human Genetics, 2021, 108, 929-941.	6.2	15
230	Missense variants in CTNNB1 can be associated with vitreoretinopathyâ€"Seven new cases of CTNNB1â€associated neurodevelopmental disorder including a previously unreported retinal phenotype. Molecular Genetics & Genomic Medicine, 2021, 9, e1542.	1.2	15
231	Hyperhomocysteinemia and cobalamin disorders. Molecular Genetics and Metabolism, 2007, 90, 113-121.	1.1	14
232	Parental somatic mosaicism for CNV deletions â€“ A need for more sensitive and precise detection methods in clinical diagnostics settings. Genomics, 2020, 112, 2937-2941.	2.9	14
233	Lymphedema of the Lower Extremity: Is It Genetic or Nongenetic?. Clinical Pediatrics, 2007, 46, 835-841.	0.8	13
234	Amino acidâ€"level signal-to-noise analysis of incidentally identified variants in genes associated with long QT syndrome during pediatric whole exome sequencing reflects background genetic noise. Heart Rhythm, 2018, 15, 1042-1050.	0.7	13

#	Article	IF	Citations
235	A mutation in Siteâ€1 Protease is associated with a complex phenotype that includes episodic hyperCKemia and focal myoedema. Molecular Genetics & Genomic Medicine, 2019, 7, e00733.	1.2	13
236	Incidentally identified genetic variants in arrhythmogenic right ventricular cardiomyopathyâ€associated genes among children undergoing exome sequencing reflect healthy population variation. Molecular Genetics & Genomic Medicine, 2019, 7, e593.	1.2	13
237	Variants in SCAF4 Cause a Neurodevelopmental Disorder and Are Associated with Impaired mRNA Processing. American Journal of Human Genetics, 2020, 107, 544-554.	6.2	13
238	Heterozygous loss-of-function variants significantly expand the phenotypes associated with loss of GDF11. Genetics in Medicine, 2021, 23, 1889-1900.	2.4	13
239	Chromosome 19p13.3 deletion in a child with Peutz-Jeghers syndrome, congenital heart defect, high myopia, learning difficulties and dysmorphic features: clinical and molecular characterization of a new contiguous gene syndrome. Genetics and Molecular Biology, 2011, 34, 557-561.	1.3	12
240	De novo variants in CACNA1E found in patients with intellectual disability, developmental regression and social cognition deficit but no seizures. Molecular Autism, 2021, 12, 69.	4.9	12
241	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.	2.4	12
242	Genotypeâ€“phenotype characterization in 13 individuals with chromosome Xp11.22 duplications. American Journal of Medical Genetics, Part A, 2016, 170, 967-977.	1.2	11
243	DeSanto-Shinawi Syndrome: First Case in South America. Molecular Syndromology, 2018, 9, 154-158.	0.8	11
244	Recurrent mosaic MTOR c.5930C > T (p.Thr1977Ile) variant causing megalencephaly, asymmetric polymicrogyria, and cutaneous pigmentary mosaicism: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2019, 179, 475-479.	1.2	11
245	Lamin B receptor-related disorder is associated with a spectrum of skeletal dysplasia phenotypes. Bone, 2019, 120, 354-363.	2.9	11
246	Intragenic CNTN4 copy number variants associated with a spectrum of neurobehavioral phenotypes. European Journal of Medical Genetics, 2020, 63, 103736.	1.3	11
247	Overcoming presynaptic effects of VAMP2 mutations with 4â€aminopyridine treatment. Human Mutation, 2020, 41, 1999-2011.	2.5	11
248	BAZ2B haploinsufficiency as a cause of developmental delay, intellectual disability, and autism spectrum disorder. Human Mutation, 2020, 41, 921-925.	2.5	11
249	What Has the Undiagnosed Diseases Network Taught Us About the Clinical Applications of Genomic Testing?. Annual Review of Medicine, 2022, 73, 575-585.	12.2	11
250	Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis. Brain, 2022, 145, 2687-2703.	7.6	11
251	De novo deletions and duplications of 17q25.3 cause susceptibility to cardiovascular malformations. Orphanet Journal of Rare Diseases, 2015, 10, 75.	2.7	10
252	Duplication of HEY2 in cardiac and neurologic development. American Journal of Medical Genetics, Part A, 2015, 167, 2145-2149.	1.2	10

#	Article	IF	Citations
253	Child Neurology: Brown-Vialetto-Van Laere syndrome. Neurology, 2018, 91, 938-941.	1.1	10
254	Characterization of the renal phenotype in RMND1 â€related mitochondrial disease. Molecular Genetics & Genomic Medicine, 2019, 7, e973.	1.2	10
255	Sorting nexin 27 (SNX27) variants associated with seizures, developmental delay, behavioral disturbance, and subcortical brain abnormalities. Clinical Genetics, 2020, 97, 437-446.	2.0	10
256	Germline mutation in POLR2A: a heterogeneous, multi-systemic developmental disorder characterized by transcriptional dysregulation. Human Genetics and Genomics Advances, 2021, 2, 100014.	1.7	10
257	Wilms tumor in patients with osteopathia striata with cranial sclerosis. European Journal of Human Genetics, 2021, 29, 396-401.	2.8	10
258	Homozygous missense WIPI2 variants cause a congenital disorder of autophagy with neurodevelopmental impairments of variable clinical severity and disease course. Brain Communications, 2021, 3, fcab183.	3.3	10
259	RCL1 copy number variants are associated with a range of neuropsychiatric phenotypes. Molecular Psychiatry, 2021, 26, 1706-1718.	7.9	10
260	Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia, and insensitivity to pain. Genetics in Medicine, 2021, 23, 1465-1473.	2.4	10
261	A human importin-Î²-related disorder: Syndromic thoracic aortic aneurysm caused by bi-allelic loss-of-function variants in IPO8. American Journal of Human Genetics, 2021, 108, 1115-1125.	6.2	10
262	Identification of disease-linked hyperactivating mutations in UBE3A through large-scale functional variant analysis. Nature Communications, 2021, 12, 6809.	12.8	10
263	Loss-of-function mutations in Lysyl-tRNA synthetase cause various leukoencephalopathy phenotypes. Neurology: Genetics, 2019, 5, e565.	1.9	9
264	Phenotype and response to growth hormone therapy in siblings with B4GALT7 deficiency. Bone, 2019, 124, 14-21.	2.9	9
265	Heterozygous variants in SPTBN1 cause intellectual disability and autism. American Journal of Medical Genetics, Part A, 2021, 185, 2037-2045.	1.2	9
266	Stankiewicz-Isidor syndrome: expanding the clinical and molecular phenotype. Genetics in Medicine, 2022, 24, 179-191.	2.4	9
267	A dominant negative variant of RAB5B disrupts maturation of surfactant protein B and surfactant protein C. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	7.1	9
268	Microdeletions excluding YWHAE and PAFAH1B1 cause a unique leukoencephalopathy: further delineation of the 17p13.3 microdeletion spectrum. Genetics in Medicine, 2019, 21, 1652-1656.	2.4	8
269	Phenotypic expansion in KIF1A â€related dominant disorders: A description of novel variants and review of published cases. Human Mutation, 2020, 41, 2094-2104.	2.5	8
270	The transcription factor Maz is essential for normal eye development. DMM Disease Models and Mechanisms, 2020, 13, .	2.4	8

#	Article	IF	Citations
271	Atypical Alexander disease with dystonia, retinopathy, and a brain mass mimicking astrocytoma. Neurology: Genetics, 2018, 4, e248.	1.9	7
272	Amino Acid-Level Signal-to-Noise Analysis Aids in Pathogenicity Prediction of Incidentally Identified TTN -Encoded Titin Truncating Variants. Circulation Genomic and Precision Medicine, 2021, 14, e003131.	3.6	7
273	<scp>PPP3CA</scp> truncating variants clustered in the regulatory domain cause earlyâ€onset refractory epilepsy. Clinical Genetics, 2021, 100, 227-233.	2.0	7
274	A complementary study approach unravels novel players in the pathoetiology of Hirschsprung disease. PLoS Genetics, 2020, 16, e1009106.	3.5	7
275	The clinical and molecular spectrum of QRICH1 associated neurodevelopmental disorder. Human Mutation, 2022, 43, 266-282.	2.5	7
276	De novo variants in H3-3A and H3-3B are associated with neurodevelopmental delay, dysmorphic features, and structural brain abnormalities. Npj Genomic Medicine, 2021, 6, 104.	3.8	7
277	De novo variants of CSNK2B cause a new intellectual disability-craniodigital syndrome by disrupting the canonical Wnt signaling pathway. Human Genetics and Genomics Advances, 2022, 3, 100111.	1.7	7
278	Earlyâ€onset Hepatic Fibrosis in Lysinuric Protein Intolerance. Journal of Pediatric Gastroenterology and Nutrition, 2011, 53, 695-698.	1.8	6
279	De novo copy number variants and parental age: Is there an association?. European Journal of Medical Genetics, 2020, 63, 103829.	1.3	6
280	RSRC1 loss-of-function variants cause mild to moderate autosomal recessive intellectual disability. Brain, 2020, 143, e31-e31.	7.6	6
281	Clinical characterization of individuals with the distal 1q21.1 microdeletion. American Journal of Medical Genetics, Part A, 2021, 185, 1388-1398.	1.2	6
282	Molecular characterisation of rare loss-of-function NPAS3 and NPAS4 variants identified in individuals with neurodevelopmental disorders. Scientific Reports, 2021, 11, 6602.	3.3	6
283	Rare germline heterozygous missense variants in BRCA1-associated protein 1, BAP1, cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2022, 109, 361-372.	6.2	6
284	The microRNA processorDROSHAis a candidate gene for a severe progressive neurological disorder. Human Molecular Genetics, 2022, 31, 2934-2950.	2.9	6
285	Discovering a new part of the phenotypic spectrum of Coffin-Siris syndrome in a fetal cohort. Genetics in Medicine, 2022, 24, 1753-1760.	2.4	6
286	Biallelic variants in WARS1 cause a highly variable neurodevelopmental syndrome and implicate a critical exon for normal auditory function. Human Mutation, 2022, 43, 1472-1489.	2.5	6
287	ASPP2 deficiency causes features of 1q41q42 microdeletion syndrome. Cell Death and Differentiation, 2016, 23, 1973-1984.	11.2	5
288	Abnormally increased carotid intima media-thickness and elasticity in patients with Morquio A disease. Orphanet Journal of Rare Diseases, 2020, 15, 73.	2.7	5

#	Article	IF	Citations
289	A Case Series of Familial ARID1B Variants Illustrating Variable Expression and Suggestions to Update the ACMG Criteria. Genes, 2021, 12, 1275.	2.4	5
290	AHDC1 missense mutations in Xia-Gibbs syndrome. Human Genetics and Genomics Advances, 2021, 2, 100049.	1.7	5
291	Mutations of the histone linker H1â€“4 in neurodevelopmental disorders and functional characterization of neurons expressing C-terminus frameshift mutant H1.4. Human Molecular Genetics, 2022, 31, 1430-1442.	2.9	5
292	Delineation of a novel neurodevelopmental syndrome associated with PAX5 haploinsufficiency. Human Mutation, 2022, 43, 461-470.	2.5	5
293	Loss of Neuron Navigator 2 Impairs Brain and Cerebellar Development. Cerebellum, 2022, , 1.	2.5	5
294	Expanding the phenotype, genotype and biochemical knowledge of <scp>ALG3â€CDG</scp>. Journal of Inherited Metabolic Disease, 2021, 44, 987-1000.	3.6	4
295	Biallelic <scp>ASCC1</scp> variants including a novel intronic variant result in expanded phenotypic spectrum of spinal muscular atrophy with congenital bone fractures 2 (<scp>SMABF2</scp>). American Journal of Medical Genetics, Part A, 2021, 185, 2190-2197.	1.2	4
296	Saturation mutagenesis defines novel mouse models of severe spine deformity. DMM Disease Models and Mechanisms, 2021, 14, .	2.4	4
297	Autosomal Dominant ANO5-Related Disorder Associated With Myopathy and Gnathodiaphyseal Dysplasia. Neurology: Genetics, 2021, 7, e612.	1.9	4
298	Genome sequencing reveals novel noncoding variants in <scp>PLA2G6</scp> and <scp>LMNB1</scp> causing progressive neurologic disease. Molecular Genetics & Genomic Medicine, 2022, 10, e1892.	1.2	4
299	Functional analysis of a novel de novo variant in PPP5C associated with microcephaly, seizures, and developmental delay. Molecular Genetics and Metabolism, 2022, 136, 65-73.	1.1	4
300	A novel, de novo intronic variant in <scp>POGZ</scp> causes <scp>Whiteâ€“Sutton</scp> syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 2198-2203.	1.2	4
301	Variants in ALX4 and their association with genitourinary defects. Andrology, 2020, 8, 1243-1255.	3.5	3
302	<scp>Genotypeâ€•phenotype</scp> study and expansion of <scp>ARL6IP1â€•related</scp> complicated hereditary spastic paraplegia. Clinical Genetics, 2021, 99, 477-480.	2.0	3
303	When â€œNof 2â€•is not enough: integrating statistical and functional data in gene discovery. Journal of Physical Education and Sports Management, 2017, 3, a001099.	1.2	2
304	Delineation of the 1q24.3 microdeletion syndrome provides further evidence for the potential role of non-coding RNAs in regulating the skeletal phenotype. Bone, 2021, 142, 115705.	2.9	2
305	A novel de novo intronic variant in ITPR1 causes Gillespie syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2315-2324.	1.2	2
306	PRUNE1 c.<scp>933G</scp>>A synonymous variant induces exon 7 skipping, disrupts the <scp>DHHA2</scp> domain, and leads to an atypical <scp>NMIHBA</scp> syndrome presentation: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2022, 188, 1868-1874.	1.2	2

#	Article	IF	Citations
307	Adult-onset dystonia with marfanoid features. Neurology: Clinical Practice, 2017, 7, e31-e34.	1.6	1
308	Special Therapy and Psychosocial Needs Identified in a Multidisciplinary Cancer Predisposition Syndrome Clinic. Journal of Pediatric Hematology/Oncology, 2019, 41, 133-136.	0.6	1
309	Response to Mounts and Besser. Genetics in Medicine, 2021, 23, 240-242.	2.4	1
310	Paroxysmal Kinesigenic Dyskinesia in Twins With Chromosome 16p11.2 Duplication Syndrome. Neurology: Genetics, 2021, 7, e549.	1.9	1
311	SAT-LB085 First Report of Burosumab (Anti-FGF23 Monoclonal Antibody) for Rickets Complicating HRAS-Associated Cutaneous Skeletal Hypophosphatemia Syndrome. Journal of the Endocrine Society, 2019, 3, .	0.2	1
312	Novel CIC variants identified in individuals with neurodevelopmental phenotypes. Human Mutation, 2022, 43, 889-899.	2.5	1
313	Signal-to-Noise Analysis Can Inform the Likelihood That Incidentally Identified Variants in Sarcomeric Genes Are Associated with Pediatric Cardiomyopathy. Journal of Personalized Medicine, 2022, 12, 733.	2.5	1
314	Heterozygous variants in CTR9, which encodes a major component of the PAF1 complex, are associated with a neurodevelopmental disorder. Genetics in Medicine, 2022, , .	2.4	1
315	Extensive primary cutaneous herpes simplex virus type 1 infection in an infant following acute rotavirus gastroenteritis. European Journal of Pediatrics, 2005, 164, 175-176.	2.7	0
316	A 5-Month-Old Boy with Delay in Growth and Development and Decreased Muscle Tone. Clinical Chemistry, 2015, 61, 50-54.	3.2	0
317	Support for the Diagnosis of CHARGE Syndromeâ€"Reply. JAMA Otolaryngology - Head and Neck Surgery, 2017, 143, 635.	2.2	0
318	Genetic testing in adults. , 2020, , 43-57.		0
319	Abstract 53: Missense Pathogenic Variants in ANO1 Predispose to Moyamoya Disease. Stroke, 2020, 51, .	2.0	0
320	Chromosomal Microarrays: Understanding Genetics of Neurodevelopmental Disorders and Congenital Anomalies. Journal of Pediatric Genetics, 0, 06, .	0.7	0
321	Front Cover, Volume 43, Issue 7. Human Mutation, 2022, 43, .	2.5	0
322	Characterization of functionally deficient SIM2 variants found in patients with neurological phenotypes. Biochemical Journal, 0, , .	3.7	0