Jill A Rosenfeld

List of Publications by Year
in descending order

322	19,864	16791 66	20625 120
papers	citations	h-index	g-index

342	342	342	27985
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. Brain, 2022, 145, 208-223.	3.7	15
2	Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. Brain, 2022, 145, 909-924.	3.7	17
3	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.	1.5	27
4	Retrospective analysis of a clinical exome sequencing cohort reveals the mutational spectrum and identifies candidate diseaseâ€“associated loci for BAFopathies. Genetics in Medicine, 2022, 24, 364-373.	1.1	12
5	Gain-of-function mutations in RPA1 cause a syndrome with short telomeres and somatic genetic rescue. Blood, 2022, 139, 1039-1051.	0.6	29
6	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.	1.4	5
7	Stankiewicz-Isidor syndrome: expanding the clinical and molecular phenotype. Genetics in Medicine, 2022, 24, 179-191.	1.1	9
8	The clinical and molecular spectrum of QRICH1 associated neurodevelopmental disorder. Human Mutation, 2022, 43, 266-282.	1.1	7
9	What Has the Undiagnosed Diseases Network Taught Us About the Clinical Applications of Genomic Testing?. Annual Review of Medicine, 2022, 73, 575-585.	5.0	11
10	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.	2.6	6
11	Delineation of a novel neurodevelopmental syndrome associated with PAX5 haploinsufficiency. Human Mutation, 2022, 43, 461-470.	1.1	5
12	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, .	3.3	9
13	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	1.1	44
14	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.	0.7	2
15	Loss of Neuron Navigator 2 Impairs Brain and Cerebellar Development. Cerebellum, 2022, , 1.	1.4	5
16	Drosophila functional screening of de novo variants in autism uncovers damaging variants and facilitates discovery of rare neurodevelopmental diseases. Cell Reports, 2022, 38, 110517.	2.9	24
17	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.	0.6	4
18	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.	0.5	4

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19	Novel CIC variants identified in individuals with neurodevelopmental phenotypes. Human Mutation, 2022, 43, 889-899.	1.1	1
20	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.	0.7	4
21	The microRNA processorDROSHAis a candidate gene for a severe progressive neurological disorder. Human Molecular Genetics, 2022, 31, 2934-2950.	1.4	6
22	Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis. Brain, 2022, 145, 2687-2703.	3.7	11
23	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.	1.0	7
24	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.	1.1	1
25	Heterozygous variants in CTR9, which encodes a major component of the PAF1 complex, are associated with a neurodevelopmental disorder. Genetics in Medicine, 2022, , .	1.1	1
26	Discovering a new part of the phenotypic spectrum of Coffin-Siris syndrome in a fetal cohort. Genetics in Medicine, 2022, 24, 1753-1760.	1.1	6
27	Front Cover, Volume 43, Issue 7. Human Mutation, 2022, 43, .	1.1	0
28	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.	1.1	6
29	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.	1.4	2
30	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	1.1	20
31	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.	1.6	7
32	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.	2.6	15
33	Germline mutation in POLR2A: a heterogeneous, multi-systemic developmental disorder characterized by transcriptional dysregulation. Human Genetics and Genomics Advances, 2021, 2, 100014.	1.0	10
34	Wilms tumor in patients with osteopathia striata with cranial sclerosis. European Journal of Human Genetics, 2021, 29, 396-401.	1.4	10
35	Response to Mounts and Besser. Genetics in Medicine, 2021, 23, 240-242.	1.1	1
36	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.	1.5	10

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37	Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing. Journal of Clinical Investigation, 2021, 131, .	3.9	87
38	Paroxysmal Kinesigenic Dyskinesia in Twins With Chromosome 16p11.2 Duplication Syndrome. Neurology: Genetics, 2021, 7, e549.	0.9	1
39	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	2.6	30
40	RCL1 copy number variants are associated with a range of neuropsychiatric phenotypes. Molecular Psychiatry, 2021, 26, 1706-1718.	4.1	10
41	Clinical characterization of individuals with the distal 1q21.1 microdeletion. American Journal of Medical Genetics, Part A, 2021, 185, 1388-1398.	0.7	6
42	Bi-allelic variants in HOPS complex subunit VPS41 cause cerebellar ataxia and abnormal membrane trafficking. Brain, 2021, 144, 769-780.	3.7	33
43	Expanding the phenotype, genotype and biochemical knowledge of <scp>ALG3â€CDG</scp>. Journal of Inherited Metabolic Disease, 2021, 44, 987-1000.	1.7	4
44	Molecular characterisation of rare loss-of-function NPAS3 and NPAS4 variants identified in individuals with neurodevelopmental disorders. Scientific Reports, 2021, 11, 6602.	1.6	6
45	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	3.6	50
46	Heterozygous variants in SPTBN1 cause intellectual disability and autism. American Journal of Medical Genetics, Part A, 2021, 185, 2037-2045.	0.7	9
47	Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia, and insensitivity to pain. Genetics in Medicine, 2021, 23, 1465-1473.	1.1	10
48	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.	2.6	15
49	A novel de novo intronic variant in ITPR1 causes Gillespie syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2315-2324.	0.7	2
50	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.	0.7	4
51	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.	1.1	24
52	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.	2.6	10
53	Disruption of NEUROD2 causes a neurodevelopmental syndrome with autistic features via cell-autonomous defects in forebrain glutamatergic neurons. Molecular Psychiatry, 2021, 26, 6125-6148.	4.1	21
54	Heterozygous loss-of-function variants significantly expand the phenotypes associated with loss of GDF11. Genetics in Medicine, 2021, 23, 1889-1900.	1.1	13

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55	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. Genetics in Medicine, 2021, 23, 1922-1932.	1.1	16
56	<scp>PPP3CA</scp> truncating variants clustered in the regulatory domain cause earlyâ€onset refractory epilepsy. Clinical Genetics, 2021, 100, 227-233.	1.0	7
57	Saturation mutagenesis defines novel mouse models of severe spine deformity. DMM Disease Models and Mechanisms, 2021, 14, .	1.2	4
58	Functional and epigenetic phenotypes of humans and mice with DNMT3A Overgrowth Syndrome. Nature Communications, 2021, 12, 4549.	5.8	21
59	Autosomal Dominant ANO5-Related Disorder Associated With Myopathy and Gnathodiaphyseal Dysplasia. Neurology: Genetics, 2021, 7, e612.	0.9	4
60	Integrative analysis of genomic variants reveals new associations of candidate haploinsufficient genes with congenital heart disease. PLoS Genetics, 2021, 17, e1009679.	1.5	17
61	A Case Series of Familial ARID1B Variants Illustrating Variable Expression and Suggestions to Update the ACMG Criteria. Genes, 2021, 12, 1275.	1.0	5
62	COPB2 loss of function causes a coatopathy with osteoporosis and developmental delay. American Journal of Human Genetics, 2021, 108, 1710-1724.	2.6	18
63	AHDC1 missense mutations in Xia-Gibbs syndrome. Human Genetics and Genomics Advances, 2021, 2, 100049.	1.0	5
64	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.	2.6	12
65	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.	0.6	15
66	<scp>Genotypeâ€phenotype</scp> study and expansion of <scp>ARL6IP1â€related</scp> complicated hereditary spastic paraplegia. Clinical Genetics, 2021, 99, 477-480.	1.0	3
67	Identification of disease-linked hyperactivating mutations in UBE3A through large-scale functional variant analysis. Nature Communications, 2021, 12, 6809.	5.8	10
68	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.	1.7	7
69	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.	1.1	21
70	Intragenic CNTN4 copy number variants associated with a spectrum of neurobehavioral phenotypes. European Journal of Medical Genetics, 2020, 63, 103736.	0.7	11
71	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.	0.7	42
72	Sorting nexin 27 (SNX27) variants associated with seizures, developmental delay, behavioral disturbance, and subcortical brain abnormalities. Clinical Genetics, 2020, 97, 437-446.	1.0	10

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73	Truncating variants in UBAP1 associated with childhoodâ€onset nonsyndromic hereditary spastic paraplegia. Human Mutation, 2020, 41, 632-640.	1.1	15
74	Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. American Journal of Human Genetics, 2020, 106, 234-245.	2.6	56
75	De novo copy number variants and parental age: Is there an association?. European Journal of Medical Genetics, 2020, 63, 103829.	0.7	6
76	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.	0.7	17
77	Deficiencies in vesicular transport mediated by TRAPPC4 are associated with severe syndromic intellectual disability. Brain, 2020, 143, 112-130.	3.7	33
78	Phenotypic expansion of POGZâ€related intellectual disability syndrome (Whiteâ€Sutton) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 5	0.7	35
79	Overcoming presynaptic effects of VAMP2 mutations with 4â€aminopyridine treatment. Human Mutation, 2020, 41, 1999-2011.	1.1	11
80	Evidence for secondary-variant genetic burden and non-random distribution across biological modules in a recessive ciliopathy. Nature Genetics, 2020, 52, 1145-1150.	9.4	22
81	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.	1.7	24
82	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.	2.6	23
83	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.	2.6	32
84	Variants in SCAF4 Cause a Neurodevelopmental Disorder and Are Associated with Impaired mRNA Processing. American Journal of Human Genetics, 2020, 107, 544-554.	2.6	13
85	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. Genetics in Medicine, 2020, 22, 1863-1873.	1.1	19
86	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.	2.6	32
87	Missed diagnoses: Clinically relevant lessons learned through medical mysteries solved by the Undiagnosed Diseases Network. Molecular Genetics & Genomic Medicine, 2020, 8, e1397.	0.6	16
88	Dominant-negative mutations in human IL6ST underlie hyper-IgE syndrome. Journal of Experimental Medicine, 2020, 217, .	4.2	64
89	Phenotypic expansion in KIF1A â€related dominant disorders: A description of novel variants and review of published cases. Human Mutation, 2020, 41, 2094-2104.	1.1	8
90	The transcription factor Maz is essential for normal eye development. DMM Disease Models and Mechanisms, 2020, 13, .	1.2	8

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91	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.	2.6	23
92	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	1.1	22
93	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.	1.3	14
94	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.	0.7	22
95	Variants in ALX4 and their association with genitourinary defects. Andrology, 2020, 8, 1243-1255.	1.9	3
96	Abnormally increased carotid intima media-thickness and elasticity in patients with Morquio A disease. Orphanet Journal of Rare Diseases, 2020, 15, 73.	1.2	5
97	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. Human Genetics, 2020, 139, 575-592.	1.8	24
98	Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. Neuron, 2020, 106, 589-606.e6.	3.8	71
99	Cell-based analysis of CAD variants identifies individuals likely to benefit from uridine therapy. Genetics in Medicine, 2020, 22, 1598-1605.	1.1	18
100	BAZ2B haploinsufficiency as a cause of developmental delay, intellectual disability, and autism spectrum disorder. Human Mutation, 2020, 41, 921-925.	1.1	11
101	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.	2.6	22
102	RSRC1 loss-of-function variants cause mild to moderate autosomal recessive intellectual disability. Brain, 2020, 143, e31-e31.	3.7	6
103	Biallelic GRM7 variants cause epilepsy, microcephaly, and cerebral atrophy. Annals of Clinical and Translational Neurology, 2020, 7, 610-627.	1.7	15
104	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.	1.1	89
105	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	5.8	105
106	A complementary study approach unravels novel players in the pathoetiology of Hirschsprung disease. PLoS Genetics, 2020, 16, e1009106.	1.5	7
107	Genetic testing in adults. , 2020, , 43-57.		0
108	Abstract 53: Missense Pathogenic Variants in ANO1 Predispose to Moyamoya Disease. Stroke, 2020, 51, .	1.0	0

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109	Mutations in RABL3 alter KRAS prenylation and are associated with hereditary pancreatic cancer. Nature Genetics, 2019, 51, 1308-1314.	9.4	47
110	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	5.8	150
111	De novo substitutions of TRPM3 cause intellectual disability and epilepsy. European Journal of Human Genetics, 2019, 27, 1611-1618.	1.4	45
112	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. Genetics in Medicine, 2019, 21, 2723-2733.	1.1	48
113	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.	2.6	37
114	DYRK1A-related intellectual disability: a syndrome associated with congenital anomalies of the kidney and urinary tract. Genetics in Medicine, 2019, 21, 2755-2764.	1.1	19
115	Model system identification of novel congenital heart disease gene candidates: focus on RPL13. Human Molecular Genetics, 2019, 28, 3954-3969.	1.4	19
116	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	5.8	43
117	Characterization of the renal phenotype in RMND1 â€related mitochondrial disease. Molecular Genetics & Genomic Medicine, 2019, 7, e973.	0.6	10
118	A Genocentric Approach to Discovery of Mendelian Disorders. American Journal of Human Genetics, 2019, 105, 974-986.	2.6	30
119	Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing. Journal of Genetic Counseling, 2019, 28, 1107-1118.	0.9	42
120	Disruption of PHF21A causes syndromic intellectual disability with craniofacial anomalies, epilepsy, hypotonia, and neurobehavioral problems including autism. Molecular Autism, 2019, 10, 35.	2.6	30
121	Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. American Journal of Human Genetics, 2019, 105, 1005-1015.	2.6	24
122	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	4.7	35
123	Pathogenic variants in USP7 cause a neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies. Genetics in Medicine, 2019, 21, 1797-1807.	1.1	41
124	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	13.9	205
125	2-Pyrrolidinone and Succinimide as Clinical Screening Biomarkers for GABA-Transaminase Deficiency: Anti-seizure Medications Impact Accurate Diagnosis. Frontiers in Neuroscience, 2019, 13, 394.	1.4	23
126	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.	1.1	28

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127	Review of the phenotypic spectrum associated with haploinsufficiency of MYRF. American Journal of Medical Genetics, Part A, 2019, 179, 1376-1382.	0.7	44
128	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.	0.6	13
129	Loss-of-function mutations in Lysyl-tRNA synthetase cause various leukoencephalopathy phenotypes. Neurology: Genetics, 2019, 5, e565.	0.9	9
130	A pathogenic CtBP1 missense mutation causes altered cofactor binding and transcriptional activity. Neurogenetics, 2019, 20, 129-143.	0.7	16
131	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.	0.6	13
132	De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smithâ€“Magenis syndrome. Genome Medicine, 2019, 11, 12.	3.6	23
133	Phenotype and response to growth hormone therapy in siblings with B4GALT7 deficiency. Bone, 2019, 124, 14-21.	1.4	9
134	Variants in DOCK3 cause developmental delay and hypotonia. European Journal of Human Genetics, 2019, 27, 1225-1234.	1.4	15
135	Novel parent-of-origin-specific differentially methylated loci on chromosome 16. Clinical Epigenetics, 2019, 11, 60.	1.8	18
136	Pathogenic Variants in GPC4 Cause Keipert Syndrome. American Journal of Human Genetics, 2019, 104, 914-924.	2.6	23
137	Variants in TCF20 in neurodevelopmental disability: description of 27 new patients and review of literature. Genetics in Medicine, 2019, 21, 2036-2042.	1.1	23
138	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	2.6	30
139	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.	2.6	19
140	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	2.6	27
141	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.	2.6	34
142	Special Therapy and Psychosocial Needs Identified in a Multidisciplinary Cancer Predisposition Syndrome Clinic. Journal of Pediatric Hematology/Oncology, 2019, 41, 133-136.	0.3	1
143	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	1.1	52
144	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. Genetics in Medicine, 2019, 21, 816-825.	1.1	127

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145	Microdeletions excluding YWHAE and PAFAH1B1 cause a unique leukoencephalopathy: further delineation of the 17p13.3 microdeletion spectrum. Genetics in Medicine, 2019, 21, 1652-1656.	1.1	8
146	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.	2.6	59
147	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.	0.7	11
148	Lamin B receptor-related disorder is associated with a spectrum of skeletal dysplasia phenotypes. Bone, 2019, 120, 354-363.	1.4	11
149	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	1.1	161
150	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	0.7	108
151	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, .	0.1	1
152	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. Genetics in Medicine, 2018, 20, 1175-1185.	1.1	133
153	A Mild PUM1 Mutation Is Associated with Adult-Onset Ataxia, whereas Haploinsufficiency Causes Developmental Delay and Seizures. Cell, 2018, 172, 924-936.e11.	13.5	103
154	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.	0.3	13
155	Genotype-phenotype correlations in individuals with pathogenicREREvariants. Human Mutation, 2018, 39, 666-675.	1.1	34
156	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.	3.3	39
157	Inactivation of AMMECR1 is associated with growth, bone, and heart alterations. Human Mutation, 2018, 39, 281-291.	1.1	15
158	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. American Journal of Human Genetics, 2018, 102, 27-43.	2.6	88
159	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994.	2.6	59
160	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.	4.1	45
161	Gnathodiaphyseal dysplasia: Severe atypical presentation with novel heterozygous mutation of the anoctamin gene (ANO5). Bone, 2018, 107, 161-171.	1.4	23
162	Pathogenic Variants in Fucokinase Cause a Congenital Disorder of Glycosylation. American Journal of Human Genetics, 2018, 103, 1030-1037.	2.6	18

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