High Rate of Recurrent De Novo Mutations in Developn Encephalopathies

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Citation Report

#	Article	IF	Citations
1	A novel missense SNAP25b mutation in two affected siblings from an Israeli family showing seizures and cerebellar ataxia. Journal of Human Genetics, 2018, 63, 673-676.	1.1	22
2	Systematic reanalysis of genomic data improves quality of variant interpretation. Clinical Genetics, 2018, 94, 174-178.	1.0	30
3	De novo mutations of the ATP6V1A gene cause developmental encephalopathy with epilepsy. Brain, 2018, 141, 1703-1718.	3.7	69
4	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. Annals of Neurology, 2018, 83, 926-934.	2.8	20
5	A novel mutation in the transmembrane 6 domain of GABBR2 leads to a Rettâ€like phenotype. Annals of Neurology, 2018, 83, 437-439.	2.8	19
6	Reply to â€œa novel mutation in the transmembrane 6 domain of GABBR2 leads to a rettâ€like phenotypeâ€. Annals of Neurology, 2018, 83, 439-439.	2.8	1
7	Genetics of Epilepsy in the Era of Precision Medicine: Implications for Testing, Treatment, and Genetic Counseling. Current Genetic Medicine Reports, 2018, 6, 73-82.	1.9	3
8	Perspectives on Glycosylation and Its Congenital Disorders. Trends in Genetics, 2018, 34, 466-476.	2.9	184
9	Chromosomal Abnormalities in Patients with Intellectual Disability: A 21-Year Retrospective Study. Human Heredity, 2018, 83, 274-282.	0.4	11
10	PHACTRing in actin: actin deregulation in genetic epilepsies. Brain, 2018, 141, 3084-3088.	3.7	0
11	Remodeling the endoplasmic reticulum proteostasis network restores proteostasis of pathogenic GABAA receptors. PLoS ONE, 2018, 13, e0207948.	1.1	26
12	matchbox: An open-source tool for patient matching via the Matchmaker Exchange. Human Mutation, 2018, 39, 1827-1834.	1.1	20
13	Prioritization of Variants Detected by Next Generation Sequencing According to the Mutation Tolerance and Mutational Architecture of the Corresponding Genes. International Journal of Molecular Sciences, 2018, 19, 1584.	1.8	16
14	Utility of trio-based exome sequencing in the elucidation of the genetic basis of isolated syndromic intellectual disability: illustrative cases. The Application of Clinical Genetics, 2018, Volume 11, 93-98.	1.4	34
15	Genotype and phenotype analysis using an epilepsyâ€associated gene panel in Chinese pediatric epilepsy patients. Clinical Genetics, 2018, 94, 512-520.	1.0	37
16	Structural determinants of Rab11 activation by the guanine nucleotide exchange factor SH3BP5. Nature Communications, 2018, 9, 3772.	5.8	29
17	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	2.6	36
18	De novo variants in GABRA2 and GABRA5 alter receptor function and contribute to early-onset epilepsy. Brain, 2018, 141, 2392-2405.	3.7	71

#	Article	IF	Citations
19	Whole genome sequencing identifies a de novo 2.1â€¯Mb balanced paracentric inversion disrupting FOXP1 and leading to severe intellectual disability. Clinica Chimica Acta, 2018, 485, 218-223.	0.5	8
20	Rare coding variants in genes encoding GABAA receptors in genetic generalised epilepsies: an exome-based case-control study. Lancet Neurology, The, 2018, 17, 699-708.	4.9	67
21	SYT1-associated neurodevelopmental disorder: a case series. Brain, 2018, 141, 2576-2591.	3.7	98
22	Biallelic UFM1 and UFC1 mutations expand the essential role of ufmylation in brain development. Brain, 2018, 141, 1934-1945.	3.7	70
23	De novo mutations of STXBP1 in Chinese children with early onset epileptic encephalopathy. Genes, Brain and Behavior, 2018, 17, e12492.	1.1	27
24	SNAP-25 Puts SNAREs at Center Stage in Metabolic Disease. Neuroscience, 2019, 420, 86-96.	1.1	9
25	Epilepsy genetics: Current knowledge, applications, and future directions. Clinical Genetics, 2019, 95, 95-111.	1.0	87
26	Novel SZT2 mutations in three patients with developmental and epileptic encephalopathies. Molecular Genetics & Genomic Medicine, 2019, 7, e926.	0.6	11
27	New avenues in molecular genetics for the diagnosis and application of therapeutics to the epilepsies. Epilepsy and Behavior, 2021, 121, 106428.	0.9	6
28	Coatopathies: Genetic Disorders of Protein Coats. Annual Review of Cell and Developmental Biology, 2019, 35, 131-168.	4.0	65
29	Early-Life Stress in D2 Heterozygous Mice Promotes Autistic-like Behaviors through the Downregulation of the BDNF-TrkB Pathway in the Dorsal Striatum. Experimental Neurobiology, 2019, 28, 337-351.	0.7	16
30	Entire FGF12 duplication by complex chromosomal rearrangements associated with West syndrome. Journal of Human Genetics, 2019, 64, 1005-1014.	1.1	9
31	Genomics: the power, potential and pitfalls of the new technologies and how they are transforming healthcare. Clinical Medicine, 2019, 19, 269-272.	0.8	6
32	De novo substitutions of TRPM3 cause intellectual disability and epilepsy. European Journal of Human Genetics, 2019, 27, 1611-1618.	1.4	45
33	Pathogenic Variants in STXBP1 and in Genes for GABAa Receptor Subunities Cause Atypical Rett/Rett-like Phenotypes. International Journal of Molecular Sciences, 2019, 20, 3621.	1.8	29
34	Scn2a Haploinsufficiency in Mice Suppresses Hippocampal Neuronal Excitability, Excitatory Synaptic Drive, and Long-Term Potentiation, and Spatial Learning and Memory. Frontiers in Molecular Neuroscience, 2019, 12, 145.	1.4	39
35	Both gainâ€ofâ€function and lossâ€ofâ€function de novo <scp>CACNA</scp>1A mutations cause severe developmental epileptic encephalopathies in the spectrum of Lennoxâ€Gastaut syndrome. Epilepsia, 2019, 60, 1881-1894.	2.6	57
36	Gene4Denovo: an integrated database and analytic platform for de novo mutations in humans. Nucleic Acids Research, 2020, 48, D913-D926.	6.5	41

#	Article	IF	Citations
37	De novo Mutations From Whole Exome Sequencing in Neurodevelopmental and Psychiatric Disorders: From Discovery to Application. Frontiers in Genetics, 2019, 10, 258.	1.1	49
38	Diagnostik genetisch bedingter Epilepsien. Medizinische Genetik, 2019, 31, 303-312.	0.1	1
39	Recurrent NUS1 canonical splice donor site mutation in two unrelated individuals with epilepsy, myoclonus, ataxia and scoliosis - a case report. BMC Neurology, 2019, 19, 253.	0.8	20
40	A novel homozygous truncating variant of NECAP1 in early infantile epileptic encephalopathy: the second case report of EIEE21. Journal of Human Genetics, 2019, 64, 347-350.	1.1	4
41	Epidemiology and etiology of infantile developmental and epileptic encephalopathies in Tasmania. Epilepsia Open, 2019, 4, 504-510.	1.3	11
42	Clinical Application of Targeted Next-Generation Sequencing Panels and Whole Exome Sequencing in Childhood Epilepsy. Neuroscience, 2019, 418, 291-310.	1.1	69
43	Expanding the clinical spectrum associated with PACS2 mutations. Clinical Genetics, 2019, 95, 525-531.	1.0	18
44	From next-generation sequencing to targeted treatment of non-acquired epilepsies. Expert Review of Molecular Diagnostics, 2019, 19, 217-228.	1.5	38
45	Structural insights to heterodimeric cis-prenyltransferases through yeast dehydrodolichyl diphosphate synthase subunit Nus1. Biochemical and Biophysical Research Communications, 2019, 515, 621-626.	1.0	19
46	Increased diagnostic and new genes identification outcome using research reanalysis of singleton exome sequencing. European Journal of Human Genetics, 2019, 27, 1519-1531.	1.4	43
47	Rearrangement of the transmembrane domain interfaces associated with the activation of a GPCR hetero-oligomer. Nature Communications, 2019, 10, 2765.	5.8	40
48	Biallelic mutations in PIGP cause developmental and epileptic encephalopathy. Annals of Clinical and Translational Neurology, 2019, 6, 968-973.	1.7	7
49	GenPipes: an open-source framework for distributed and scalable genomic analyses. GigaScience, 2019, 8, .	3.3	121
50	Drugâ€resistant epilepsy classified by a phenotyping algorithm associates with NTRK2. Acta Neurologica Scandinavica, 2019, 140, 169-176.	1.0	6
51	Multi-gene testing in neurological disorders showed an improved diagnostic yield: data from over 1000 Indian patients. Journal of Neurology, 2019, 266, 1919-1926.	1.8	26
52	Altered inhibitory synapses in de novo GABRA5 and GABRA1 mutations associated with early onset epileptic encephalopathies. Brain, 2019, 142, 1938-1954.	3.7	32
53	A Recurrent Missense Variant in AP2M1 Impairs Clathrin-Mediated Endocytosis and Causes Developmental and Epileptic Encephalopathy. American Journal of Human Genetics, 2019, 104, 1060-1072.	2.6	78
54	Novel GABRA2 variants in epileptic encephalopathy and intellectual disability with seizures. Brain, 2019, 142, e15-e15.	3.7	12

#	Article	IF	Citations
55	Novel HIVEP2 Variants in Patients with Intellectual Disability. Molecular Syndromology, 2019, 10, 195-201.	0.3	9
56	Predicting Parkinson's Disease Genes Based on Node2vec and Autoencoder. Frontiers in Genetics, 2019, 10, 226.	1.1	91
57	VariantÂrecurrence in neurodevelopmental disorders: the use of publicly available genomic data identifies clinically relevant pathogenic missense variants. Genetics in Medicine, 2019, 21, 2504-2511.	1.1	21
58	Clinical study of 19 patients with <scp>SCN</scp>8Aâ€related epilepsy: Two modes of onset regarding <scp>EEG</scp> and seizures. Epilepsia, 2019, 60, 845-856.	2.6	28
59	A structural look at GABAA receptor mutations linked to epilepsy syndromes. Brain Research, 2019, 1714, 234-247.	1.1	68
60	Expression of Slc35f1 in the murine brain. Cell and Tissue Research, 2019, 377, 167-176.	1.5	3
61	Unique bioinformatic approach and comprehensive reanalysis improve diagnostic yield of clinical exomes. European Journal of Human Genetics, 2019, 27, 1398-1405.	1.4	60
62	Germline de novo variants in CSNK2B in Chinese patients with epilepsy. Scientific Reports, 2019, 9, 17909.	1.6	26
63	Structural Characterization of Full-Length Human Dehydrodolichyl Diphosphate Synthase Using an Integrative Computational and Experimental Approach. Biomolecules, 2019, 9, 660.	1.8	10
64	Rethinking what constitutes a diagnosis in the genomics era: a critical illness perspective. Current Opinion in Pediatrics, 2019, 31, 317-321.	1.0	3
65	Cryo-EM of multiple cage architectures reveals a universal mode of clathrin self-assembly. Nature Structural and Molecular Biology, 2019, 26, 890-898.	3.6	56
66	De novo truncating variants in the intronless IRF2BPL are responsible for developmental epileptic encephalopathy. Genetics in Medicine, 2019, 21, 1008-1014.	1.1	34
67	Persistent Lin28 Expression Impairs Neurite Outgrowth and Cognitive Function in the Developing Mouse Neocortex. Molecular Neurobiology, 2019, 56, 3780-3795.	1.9	9
68	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. Genetics in Medicine, 2019, 21, 837-849.	1.1	47
69	Diagnostic exome sequencing in 100 consecutive patients with both epilepsy and intellectual disability. Epilepsia, 2019, 60, 155-164.	2.6	65
70	The role of recessive inheritance in early-onset epileptic encephalopathies: a combined whole-exome sequencing and copy number study. European Journal of Human Genetics, 2019, 27, 408-421.	1.4	52
71	De Novo Variants in MAPK8IP3 Cause Intellectual Disability with Variable Brain Anomalies. American Journal of Human Genetics, 2019, 104, 203-212.	2.6	44
72	The role of de novo mutations in adult-onset neurodegenerative disorders. Acta Neuropathologica, 2019, 137, 183-207.	3.9	39

#	Article	IF	Citations
73	Diagnostic yield of genetic tests in epilepsy. Neurology, 2019, 92, .	1.5	102
74	Neurotransmitter trafficking defect in a patient with clathrin (CLTC) variation presenting with intellectual disability and early-onset parkinsonism. Parkinsonism and Related Disorders, 2019, 61, 207-210.	1.1	17
75	IQSEC2mutation update and review of the female-specific phenotype spectrum including intellectual disability and epilepsy. Human Mutation, 2019, 40, 5-24.	1.1	36
76	Childhood disintegrative disorder and autism spectrum disorder: aÂsystematic review. Developmental Medicine and Child Neurology, 2019, 61, 523-534.	1.1	18
77	A new microdeletion syndrome involving TBC1D24, ATP6V0C, and PDPK1 causes epilepsy, microcephaly, and developmental delay. Genetics in Medicine, 2019, 21, 1058-1064.	1.1	22
78	The SNAP-25 Protein Family. Neuroscience, 2019, 420, 50-71.	1.1	57
79	A De Novo Variant Identified in the PPP2R1A Gene in an Infant Induces Neurodevelopmental Abnormalities. Neuroscience Bulletin, 2020, 36, 179-182.	1.5	10
80	Phenotypeâ€toâ€genotype approach reveals headâ€circumferenceâ€associated genes in an autism spectrum disorder cohort. Clinical Genetics, 2020, 97, 338-346.	1.0	29
81	Three novel patients with epileptic encephalopathy due to biallelic mutations in the <scp>PLCB1</scp> gene. Clinical Genetics, 2020, 97, 477-482.	1.0	9
82	Epilepsy and developmental disorders: Next generation sequencing in the clinic. European Journal of Paediatric Neurology, 2020, 24, 15-23.	0.7	98
83	Molecular genetic management of epilepsy. , 2020, , 289-308.		0
84	Clinical and neuroimaging phenotypes of genetic parkinsonism from infancy to adolescence. Brain, 2020, 143, 751-770.	3.7	22
85	De novo CLTC variants are associated with a variable phenotype from mild to severe intellectual disability, microcephaly, hypoplasia of the corpus callosum, and epilepsy. Genetics in Medicine, 2020, 22, 797-802.	1.1	15
86	Phenotypic spectrum of patients with GABRB2 variants: from mild febrile seizures to severe epileptic encephalopathy. Developmental Medicine and Child Neurology, 2020, 62, 1213-1220.	1.1	11
87	Genomic variability. , 2020, , 63-75.		0
88	Genetic Testing in Epilepsy. Seminars in Neurology, 2020, 40, 730-738.	0.5	7
89	Diagnostic yield and treatment impact of wholeâ€genome sequencing in paediatric neurological disorders. Developmental Medicine and Child Neurology, 2021, 63, 934-938.	1.1	14
90	Extended Study of NUS1 Gene Variants in Parkinson's Disease. Frontiers in Neurology, 2020, 11, 583182.	1.1	4

#	Article	IF	Citations
91	Structural elucidation of the cis -prenyltransferase NgBR/DHDDS complex reveals insights in regulation of protein glycosylation. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 20794-20802.	3.3	30
92	GNAO1organizes the cytoskeletal remodeling and firing of developing neurons. FASEB Journal, 2020, 34, 16601-16621.	0.2	14
93	Structural basis of heterotetrameric assembly and disease mutations in the human cis-prenyltransferase complex. Nature Communications, 2020, 11, 5273.	5.8	23
94	Complex Neurological Phenotype Associated with a De Novo DHDDS Mutation in a Boy with Intellectual Disability, Refractory Epilepsy, and Movement Disorder. Journal of Pediatric Genetics, 2021, 10, 236-238.	0.3	10
95	Epilepsy, an orphan disorder within the neurodevelopmental family. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 1245-1247.	0.9	19
96	Pre- and Post-Zygotic TP53 De Novo Mutations in SHH-Medulloblastoma. Cancers, 2020, 12, 2503.	1.7	1
97	The substrate specificity of the human TRAPPII complexâ€™s Rab-guanine nucleotide exchange factor activity. Communications Biology, 2020, 3, 735.	2.0	16
98	CACNA1A Gene Variants in Eight Chinese Patients With a Wide Range of Phenotypes. Frontiers in Pediatrics, 2020, 8, 577544.	0.9	9
99	å…¨åŸºå›é»”å®šåºåºå…’ç«¥ç¥žç¶“ç–¾ç—…çš„è¨ºæ–·ç‹€æ³ã€‚ Developmental Medicine and Child Neurology, 2020, 63,	1.1	
100	NUS1 mutation in a family with epilepsy, cerebellar ataxia, and tremor. Epilepsy Research, 2020, 164, 106371.	0.8	18
101	mTADA is a framework for identifying risk genes from de novo mutations in multiple traits. Nature Communications, 2020, 11, 2929.	5.8	10
102	SNAREopathies: Diversity in Mechanisms and Symptoms. Neuron, 2020, 107, 22-37.	3.8	77
103	A case of GABRA5-related developmental and epileptic encephalopathy with response to a combination of antiepileptic drugs and a GABAering agent. Brain and Development, 2020, 42, 546-550.	0.6	1
104	Structural basis of the activation of a metabotropic GABA receptor. Nature, 2020, 584, 298-303.	13.7	92
105	Coexpression enrichment analysis at the single-cell level reveals convergent defects in neural progenitor cells and their cell-type transitions in neurodevelopmental disorders. Genome Research, 2020, 30, 835-848.	2.4	25
106	A catalogue of new incidence estimates of monogenic neurodevelopmental disorders caused by de novo variants. Brain, 2020, 143, 1099-1105.	3.7	64
107	A 2020 View on the Genetics of Developmental and Epileptic Encephalopathies. Epilepsy Currents, 2020, 20, 90-96.	0.4	39
108	The Human Gene Mutation Database (HGMDÂ®): optimizing its use in a clinical diagnostic or research setting. Human Genetics, 2020, 139, 1197-1207.	1.8	353

#	Article	IF	Citations
109	Genomic analyses implicate noncoding de novo variants in congenital heart disease. Nature Genetics, 2020, 52, 769-777.	9.4	97
110	Fifteen-year follow-up of a patient with a DHDDS variant with non-progressive early onset myoclonic tremor and rare generalized epilepsy. Brain and Development, 2020, 42, 696-699.	0.6	14
111	Mitochondrial Dysfunction in Autism Spectrum Disorder: Unique Abnormalities and Targeted Treatments. Seminars in Pediatric Neurology, 2020, 35, 100829.	1.0	77
112	Retinal Degeneration Caused by Rod-Specific Dhdds Ablation Occurs without Concomitant Inhibition of Protein N-Glycosylation. IScience, 2020, 23, 101198.	1.9	14
113	From Genetic Testing to Precision Medicine in Epilepsy. Neurotherapeutics, 2020, 17, 609-615.	2.1	62
114	Ion Channel Functions in Early Brain Development. Trends in Neurosciences, 2020, 43, 103-114.	4.2	67
115	Genetics of Parkinson's disease: An introspection of its journey towards precision medicine. Neurobiology of Disease, 2020, 137, 104782.	2.1	241
116	Tattonâ€Brownâ€Rahman syndrome: Six individuals with novel features. American Journal of Medical Genetics, Part A, 2020, 182, 673-680.	0.7	11
117	Structural analysis of pathogenic missense mutations in GABRA2 and identification of a novel de novo variant in the desensitization gate. Molecular Genetics & Genomic Medicine, 2020, 8, e1106.	0.6	9
118	A comparison of genomic diagnostics in adults and children with epilepsy and comorbid intellectual disability. European Journal of Human Genetics, 2020, 28, 1066-1077.	1.4	30
119	Increased diagnostic yield in complex dystonia through exome sequencing. Parkinsonism and Related Disorders, 2020, 74, 50-56.	1.1	34
120	Bi-allelic GAD1 variants cause a neonatal onset syndromic developmental and epileptic encephalopathy. Brain, 2020, 143, 1447-1461.	3.7	18
121	Presynaptic dysfunction in neurodevelopmental disorders: Insights from the synaptic vesicle life cycle. Journal of Neurochemistry, 2021, 157, 179-207.	2.1	51
122	Role of Aberrant Spontaneous Neurotransmission in SNAP25-Associated Encephalopathies. Neuron, 2021, 109, 59-72.e5.	3.8	31
123	HeterotopÃas periventriculares: ampliaciÃ³n del espectro clÃnico de las variantes patogÃ©nicas del gen de la clatrina 1 (CLTC). NeurologÃa, 2021, 36, 327-329.	0.3	2
124	The neurodevelopmental spectrum of synaptic vesicle cycling disorders. Journal of Neurochemistry, 2021, 157, 208-228.	2.1	37
125	Genetic Testing in Children with Epilepsy: Report of a Single-Center Experience. Canadian Journal of Neurological Sciences, 2021, 48, 233-244.	0.3	4
126	Clinical phenotypes of infantile onset CACNA1A-related disorder. European Journal of Paediatric Neurology, 2021, 30, 144-154.	0.7	13

#	Article	IF	Citations
127	Association of HECW2 variants with developmental and epileptic encephalopathy and knockdown of zebrafish hecw2a. American Journal of Medical Genetics, Part A, 2021, 185, 377-383.	0.7	5
128	Mutations in G Proteinâ€“Coupled Receptors: Mechanisms, Pathophysiology and Potential Therapeutic Approaches. Pharmacological Reviews, 2021, 73, 89-119.	7.1	60
129	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	1.1	20
130	A patient with a 6q22.1 deletion and a phenotype of non-progressive early-onset generalized epilepsy with tremor. Epilepsy and Behavior Reports, 2021, 15, 100405.	0.5	2
131	Characterization of the <scp>GABRB2</scp>â€Associated Neurodevelopmental Disorders. Annals of Neurology, 2021, 89, 573-586.	2.8	14
132	De novo variants in CELF2 that disrupt the nuclear localization signal cause developmental and epileptic encephalopathy. Human Mutation, 2021, 42, 66-76.	1.1	16
133	Psychosis in <scp>NUS1</scp> de novo mutation: New phenotypical presentation. Clinical Genetics, 2021, 99, 475-476.	1.0	7
134	SNAREs and developmental disorders. Journal of Cellular Physiology, 2021, 236, 2482-2504.	2.0	13
135	Distinct genetic patterns of shared and unique genes across four neurodevelopmental disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2021, 186, 3-15.	1.1	6
136	Disorders of synaptic vesicle fusion machinery. Journal of Neurochemistry, 2021, 157, 130-164.	2.1	44
137	Developmental and epileptic encephalopathy: Personal utility of a genetic diagnosis for families. Epilepsia Open, 2021, 6, 149-159.	1.3	11
138	De novo mutations in folate-related genes associated with common developmental disorders. Computational and Structural Biotechnology Journal, 2021, 19, 1414-1422.	1.9	6
139	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. Brain, 2022, 145, 208-223.	3.7	15
140	Reliability of genomic variants across different next-generation sequencing platforms and bioinformatic processing pipelines. BMC Genomics, 2021, 22, 62.	1.2	5
141	Missense variants in the N-terminal domain of the A isoform of FHF2/FGF13 cause an X-linked developmental and epileptic encephalopathy. American Journal of Human Genetics, 2021, 108, 176-185.	2.6	20
142	Diagnostic Yield of Whole Genome Sequencing After Nondiagnostic Exome Sequencing or Gene Panel in Developmental and Epileptic Encephalopathies. Neurology, 2021, 96, e1770-e1782.	1.5	53
144	Developmental and epileptic encephalopathies: recognition and approaches to care. Epileptic Disorders, 2021, 23, 40-52.	0.7	48
146	From Genotype to Phenotype: Expanding the Clinical Spectrum of CACNA1A Variants in the Era of Next Generation Sequencing. Frontiers in Neurology, 2021, 12, 639994.	1.1	49

#	Article	IF	Citations
147	Refining Genotypes and Phenotypes in KCNA2-Related Neurological Disorders. International Journal of Molecular Sciences, 2021, 22, 2824.	1.8	20
148	Pathogenic variants in <scp>KCNQ2</scp> cause intellectual deficiency without epilepsy: Broadening the phenotypic spectrum of a potassium channelopathy. American Journal of Medical Genetics, Part A, 2021, 185, 1803-1815.	0.7	13
149	Lysosomal cholesterol accumulation contributes to the movement phenotypes associated with NUS1 haploinsufficiency. Genetics in Medicine, 2021, 23, 1305-1314.	1.1	17
150	Genetic Neonatal-Onset Epilepsies and Developmental/Epileptic Encephalopathies with Movement Disorders: A Systematic Review. International Journal of Molecular Sciences, 2021, 22, 4202.	1.8	10
151	ATP6V0A1 encoding the a1-subunit of the V0 domain of vacuolar H+-ATPases is essential for brain development in humans and mice. Nature Communications, 2021, 12, 2107.	5.8	30
152	Progressive myoclonus epilepsiesâ€"Residual unsolved cases have marked genetic heterogeneity including dolichol-dependent protein glycosylation pathway genes. American Journal of Human Genetics, 2021, 108, 722-738.	2.6	41
153	Combining exome/genome sequencing with data repository analysis reveals novel geneâ€“disease associations for a wide range of genetic disorders. Genetics in Medicine, 2021, 23, 1551-1568.	1.1	30
154	Detection of Disease-Causing SNVs/Indels and CNVs in Single Test Based on Whole Exome Sequencing: A Retrospective Case Study in Epileptic Encephalopathies. Frontiers in Pediatrics, 2021, 9, 635703.	0.9	9
155	Molecular mechanisms of metabotropic GABA _B receptor function. Science Advances, 2021, 7, .	4.7	46
156	Family-Based Genome-Wide Association Study of Autism Spectrum Disorder in Middle Eastern Families. Genes, 2021, 12, 761.	1.0	7
157	Periventricular heterotopia: broadening of the clinical spectrum of the clathrin 1 gene (CLTC) pathogenic variants. NeurologÃa (English Edition), 2021, 36, 327-329.	0.2	0
158	Cross-Disorder Analysis of De Novo Mutations in Neuropsychiatric Disorders. Journal of Autism and Developmental Disorders, 2022, 52, 1299-1313.	1.7	3
159	Confirming the contribution and genetic spectrum of de novo mutation in infantile spasms: Evidence from a Chinese cohort. Molecular Genetics & Genomic Medicine, 2021, 9, e1689.	0.6	6
160	A mutation in SLC37A4 causes a dominantly inherited congenital disorder of glycosylation characterized by liver dysfunction. American Journal of Human Genetics, 2021, 108, 1040-1052.	2.6	7
161	Phenotypic analysis of catastrophic childhood epilepsy genes. Communications Biology, 2021, 4, 680.	2.0	34
162	A novel variant of dehydrodolichol diphosphate synthase (DHDDS) mutation with adult-onset progressive myoclonus ataxia. Parkinsonism and Related Disorders, 2021, 87, 135-136.	1.1	7
163	A Chinese patient with developmental and epileptic encephalopathies (DEE) carrying a TRPM3 gene mutation: a paediatric case report. BMC Pediatrics, 2021, 21, 256.	0.7	10
164	Reversing frontal disinhibition rescues behavioural deficits in models of CACNA1A-associated neurodevelopment disorders. Molecular Psychiatry, 2021, 26, 7225-7246.	4.1	16

#	Article	IF	Citations
165	Knockdown of Dehydrodolichyl Diphosphate Synthase in the Drosophila Retina Leads to a Unique Pattern of Retinal Degeneration. Frontiers in Molecular Neuroscience, 2021, 14, 693967.	1.4	5
166	Delineating the genotypic and phenotypic spectrum of HECW2-related neurodevelopmental disorders. Journal of Medical Genetics, 2022, 59, 669-677.	1.5	5
167	Developmental encephalopathy and epilepsy associated with a heterozygous de novo mutation in the IRF2BPL gene: a case report. Russkii Zhunal Detskoi Nevrologii, 2021, 16, 69-75.	0.1	0
168	Genotypeâ€“phenotype correlation of CACNA1A variants in children with epilepsy. Developmental Medicine and Child Neurology, 2021, , .	1.1	10
169	Genomic frontiers in congenital heart disease. Nature Reviews Cardiology, 2022, 19, 26-42.	6.1	93
170	Novel CLTC variants cause new brain and kidney phenotypes. Journal of Human Genetics, 2021, , .	1.1	4
171	Phenotype of heterozygous variants of dehydrodolichol diphosphate synthase. Developmental Medicine and Child Neurology, 2022, 64, 125-134.	1.1	6
172	Physiological mediators of prenatal environmental influences in autism spectrum disorder. BioEssays, 2021, 43, e2000307.	1.2	12
173	Case Report: Clinical Features of a Chinese Boy With Epileptic Seizures and Intellectual Disabilities Who Carries a Truncated NUS1 Variant. Frontiers in Pediatrics, 2021, 9, 725231.	0.9	8
174	Chromosomal microarray and exome sequencing in unexplained early infantile epileptic encephalopathies in a highly consanguineous population. International Journal of Neuroscience, 2023, 133, 683-700.	0.8	0
175	A recurrent de novo variant supports <scp>KCNC2</scp> involvement in the pathogenesis of developmental and epileptic encephalopathy. American Journal of Medical Genetics, Part A, 2021, 185, 3384-3389.	0.7	15
177	Low-frequency and rare coding variants of NUS1 contribute to susceptibility and phenotype of Parkinson's disease. Neurobiology of Aging, 2022, 110, 106-112.	1.5	2
178	Solving the Molecular Basis of the Developmental and Epileptic Encephalopathies: Are We there Yet?. Epilepsy Currents, 2021, 21, 153575972110381.	0.4	3
179	A single center experience with publicly funded clinical exome sequencing for neurodevelopmental disorders or multiple congenital anomalies. Scientific Reports, 2021, 11, 19099.	1.6	13
180	Developmental disabilities across the world: A scientometric review from 1936 to 2020. Research in Developmental Disabilities, 2021, 117, 104031.	1.2	20
181	DHDDS related epilepsyâ€“â€“Report of familial cases and review of the literature. Seizure: the Journal of the British Epilepsy Association, 2021, 91, 189-191.	0.9	7
182	The evolving genetic landscape of congenital disorders of glycosylation. Biochimica Et Biophysica Acta - General Subjects, 2021, 1865, 129976.	1.1	24
183	NTRK2-related developmental and epileptic encephalopathy: Report of 5 new cases. Seizure: the Journal of the British Epilepsy Association, 2021, 92, 52-55.	0.9	5

#	Article	IF	Citations
184	Trio exome sequencing identified a novel de novo WASF1 missense variant leading to recurrent site substitution in a Chinese patient with developmental delay, microcephaly, and early-onset seizures: A mutational hotspot p.Trp161 and literature review. Clinica Chimica Acta, 2021, 523, 10-18.	0.5	5
185	Whole exome sequencing identified a novel homozygous ARV1 mutation in an Iranian family with developmental and epileptic encephalopathy-38. Meta Gene, 2021, 30, 100953.	0.3	3
187	Wbox2: A clathrin terminal domainâ€“derived peptide inhibitor of clathrin-mediated endocytosis. Journal of Cell Biology, 2020, 219, .	2.3	13
189	A de novo GABRB2 variant associated with myoclonic status epilepticus and rhythmic highâ€amplitude delta with superimposed (poly) spikes (RHADS). Epileptic Disorders, 2020, 22, 476-481.	0.7	3
192	Stxbp1/Munc18-1 haploinsufficiency impairs inhibition and mediates key neurological features of STXBP1 encephalopathy. ELife, 2020, 9, .	2.8	42
193	GABRB2, a key player in neuropsychiatric disorders and beyond. Gene, 2022, 809, 146021.	1.0	19
194	Active site variants in STT3A cause a dominant type I congenital disorder of glycosylation with neuromusculoskeletal findings. American Journal of Human Genetics, 2021, 108, 2130-2144.	2.6	5
196	The GABA _B receptor mediates neuroprotection by coupling to G ₁₃. Science Signaling, 2021, 14, eaaz4112.	1.6	11
208	Interest of exome sequencing trioâ€like strategy based on pooled parental DNA for diagnosis and translational research in rare diseases. Molecular Genetics & Genomic Medicine, 2021, 9, e1836.	0.6	5
209	The specific role of zinc in autism spectrum disorders. , 2020, , 115-130.		0
212	Performance of meta-predictors for the classification of MED13L missense variations, implication of raw parameters. European Journal of Medical Genetics, 2022, 65, 104398.	0.7	0
213	SNAP25 mutation disrupts metabolic homeostasis, steroid hormone production and central neurobehavior. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2022, 1868, 166304.	1.8	3
214	Adultâ€onset rapidly worsening progressive myoclonic epilepsy caused by a novel variant in DHDDS. Annals of Clinical and Translational Neurology, 2021, 8, 2319-2326.	1.7	9
215	Artificial intelligence (AI)-assisted exome reanalysis greatly aids in the identification of new positive cases and reduces analysis time in a clinical diagnostic laboratory. Genetics in Medicine, 2022, 24, 192-200.	1.1	19
217	Syndromic obesity with neurodevelopmental delay: Opportunities for targeted interventions. European Journal of Medical Genetics, 2022, 65, 104443.	0.7	4
218	Genetic testing for the epilepsies: A systematic review. Epilepsia, 2022, 63, 375-387.	2.6	53
219	Congenital disorders of glycosylation: A multi-genetic disease family with multiple subcellular locations. Medycyna Wieku Rozwojowego, 2020, 24, 14-20.	0.2	7
223	Genetic analysis of developmental and epileptic encephalopathy caused by novel biallelic SZT2 gene mutations in three Chinese Han infants: a case series and literature review. Neurological Sciences, 2022, 43, 5039-5048.	0.9	2

#	Article	IF	Citations
224	Synaptopathies in Developmental and Epileptic Encephalopathies: A Focus on Pre-synaptic Dysfunction. Frontiers in Neurology, 2022, 13, 826211.	1.1	12
225	Genetic analysis using targeted exome sequencing of 53 Vietnamese children with developmental and epileptic encephalopathies. American Journal of Medical Genetics, Part A, 2022, 188, 2048-2060.	0.7	2
226	GABAA Receptor Variants in Epilepsy. , 0, , 95-118.		11
227	Diagnostic yield of patients with undiagnosed intellectual disability, global developmental delay and multiples congenital anomalies using karyotype, microarray analysis, whole exome sequencing from Central Brazil. PLoS ONE, 2022, 17, e0266493.	1.1	9
228	Functional characterization and potential therapeutic avenues for variants in the <scp> NTRK2 </scp> gene causing developmental and epileptic encephalopathies. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2022, 189, 37-47.	1.1	0
229	Difficulties of Prenatal Genetic Counseling for a Subsequent Child in a Family With Multiple Genetic Variations. Frontiers in Genetics, 2021, 12, 612100.	1.1	0
230	Allosteric ligands control the activation of a class C GPCR heterodimer by acting at the transmembrane interface. ELife, 2021, 10, .	2.8	14
231	Progressive myoclonus without epilepsy due to a NUS1 frameshift insertion: Dyssynergia cerebellaris myoclonica revisited. Parkinsonism and Related Disorders, 2022, 98, 53-55.	1.1	4
232	Paroxysmal limb dystonias associated with GABBR2 pathogenic variant: A case-based literature review. Brain and Development, 2022, , .	0.6	2
239	Detection of Deregulated miRNAs in Childhood Epileptic Encephalopathies. Journal of Molecular Neuroscience, 2022, 72, 1234-1242.	1.1	10
240	UBE3A-Induced Ubiquitination Changes in the Brain Reveal the Molecular Complexity of Angelman Syndrome. SSRN Electronic Journal, 0, , .	0.4	0
241	Nomenclature of Genetic Movement Disorders: Recommendations of the International Parkinson and Movement Disorder Society Task Force â€“ An Update. Movement Disorders, 2022, 37, 905-935.	2.2	49
242	Systematic Collaborative Reanalysis of Genomic Data Improves Diagnostic Yield in Neurologic Rare Diseases. Journal of Molecular Diagnostics, 2022, 24, 529-542.	1.2	6
243	Structural basis for long-chain isoprenoid synthesis by cis -prenyltransferases. Science Advances, 2022, 8, eabn1171.	4.7	3
244	Human KCNQ5 de novo mutations underlie epilepsy and intellectual disability. Journal of Neurophysiology, 2022, 128, 40-61.	0.9	8
246	NUS1 and Epilepsy-myoclonus-ataxia Syndrome: An Under-recognized Entity?. Tremor and Other Hyperkinetic Movements, 2022, 12, .	1.1	5
248	Molecular and clinical descriptions of patients with <scp>GABA_A</scp> receptor gene variants (<scp>GABRA1</scp>, <scp>GABRB2</scp>, <scp>GABRB3</scp>, <scp>GABRG2</scp>): A cohort study, review of literature, and genotypeâ€“phenotype correlation. Epilepsia, 2022, 63, 2519-2533.	2.6	23
249	Impact of Genetic Testing on Therapeutic Decision-Making in Childhood-Onset Epilepsiesâ€”a Study in a Tertiary Epilepsy Center. Neurotherapeutics, 2022, 19, 1353-1367.	2.1	14

#	Article	IF	Citations
250	Exome sequencing for patients with developmental and epileptic encephalopathies in clinical practice. Developmental Medicine and Child Neurology, 2023, 65, 50-57.	1.1	11
251	Loss of Drosophila NUS1 results in cholesterol accumulation and Parkinsonâ€™s diseaseâ€related neurodegeneration. FASEB Journal, 2022, 36, .	0.2	3
252	The role of common genetic variation in presumed monogenic epilepsies. EBioMedicine, 2022, 81, 104098.	2.7	12
253	Assessment of burden and segregation profiles of <scp>CNVs</scp> in patients with epilepsy. Annals of Clinical and Translational Neurology, 2022, 9, 1050-1058.	1.7	2
254	Modern Biomarkers for Autism Spectrum Disorder: Future Directions. Molecular Diagnosis and Therapy, 2022, 26, 483-495.	1.6	18
255	Clinical Findings on Chromosome 1 Copy Number Variations. Neuropediatrics, 2022, 53, 265-273.	0.3	1
256	First splicing variant in HECW2 with an autosomal recessive pattern of inheritance and associated with NDHSAL. Human Mutation, 0, , .	1.1	0
257	Genetic variations in GABA metabolism and epilepsy. Seizure: the Journal of the British Epilepsy Association, 2022, 101, 22-29.	0.9	20
258	Whole genomic approach in mutation discovery of infantile spasms patients. Frontiers in Neurology, 0, 13, .	1.1	4
259	Acetazolamide treatment in late onset CDG type 1 due to biallelic pathogenic DHDDS variants. Molecular Genetics and Metabolism Reports, 2022, 32, 100901.	0.4	3
260	Developmental and epileptic encephalopathies: from genetic heterogeneity to phenotypic continuum. Physiological Reviews, 2023, 103, 433-513.	13.1	38
261	Emerging Trends in the Management of Cryptogenic Epilepsy. , 0, , .		0
262	Monogenic developmental and epileptic encephalopathies of infancy and childhood, a population cohort from Norway. Frontiers in Pediatrics, 0, 10, .	0.9	2
263	Emerging evidence of genotypeâ€“phenotype associations of developmental and epileptic encephalopathy due to KCNC2 mutation: Identification of novel R405G. Frontiers in Molecular Neuroscience, 0, 15, .	1.4	2
266	Synaptic genes and neurodevelopmental disorders: From molecular mechanisms to developmental strategies of behavioral testing. Neurobiology of Disease, 2022, 173, 105856.	2.1	12
267	Clathrin and Clathrin-Mediated Membrane Traffic. , 2022, , .		0
268	Variable Expression of GABAA Receptor Subunit Gamma 2 Mutation in a Nuclear Family Displaying Developmental and Encephalopathic Phenotype. International Journal of Molecular Sciences, 2022, 23, 9683.	1.8	5
269	Interstitial deletions in the proximal regions of 6q: 12 original cases and a literature review. Intractable and Rare Diseases Research, 2022, 11, 143-148.	0.3	1

#	Article	IF	Citations
270	The Promising Epigenetic Regulators for Refractory Epilepsy: An Adventurous Road Ahead. NeuroMolecular Medicine, 0, , .	1.8	0
271	Exome sequencing as first-tier genetic testing in infantile-onset pharmacoresistant epilepsy: diagnostic yield and treatment impact. European Journal of Human Genetics, 2023, 31, 179-187.	1.4	7
272	Rates of rare copy number variants in different circumstances among patients with genetic developmental and epileptic encephalopathy. Science Progress, 2022, 105, 003685042211312.	1.0	1
273	Epidemiology, aetiology, interventions and genomics in children with arthrogryposis multiplex congenita: protocol for a multisite registry. BMJ Open, 2022, 12, e060591.	0.8	3
274	Transcriptional and functional consequences of alterations to MEF2C and its topological organization in neuronal models. American Journal of Human Genetics, 2022, 109, 2049-2067.	2.6	8
275	Vertebrate Animal Models of RP59: Current Status and Future Prospects. International Journal of Molecular Sciences, 2022, 23, 13324.	1.8	0
276	Anti-seizure mechanisms of midazolam and valproate at the Î²2(L51M) variant of the GABAA receptor. Neuropharmacology, 2022, 221, 109295.	2.0	0
277	Rapid genome sequencing identifies a novel de novoSNAP25variant for neonatal congenital myasthenic syndrome. Journal of Physical Education and Sports Management, 2022, 8, a006242.	0.5	3
278	Cys-loop receptors on cannabinoids: All high?. Frontiers in Physiology, 0, 13, .	1.3	2
279	A novel variant in NEUROD2 in a patient with Rett-like phenotype points to Glu130 codon as a mutational hotspot. Brain and Development, 2023, 45, 179-184.	0.6	1
280	Detection of mosaic variants using genome sequencing in a large pediatric cohort. American Journal of Medical Genetics, Part A, 2023, 191, 699-710.	0.7	8
281	DHDDS Mutation: A Rare Cause of Refractory Epilepsy and Hyperkinetic Movement Disorder. Journal of Movement Disorders, 2023, 16, 107-109.	0.7	1
282	Relating pathogenic loss-of-function mutations in humans to their evolutionary fitness costs. ELife, 0, 12, .	2.8	12
283	<scp>Fundamental Neurochemistry Review: GABA_A</scp> receptor neurotransmission and epilepsy: Principles, disease mechanisms and pharmacotherapy. Journal of Neurochemistry, 2023, 165, 6-28.	2.1	10
284	Efficacy, safety, and tolerability of soticlestat as adjunctive therapy for the treatment of seizures in patients with Dup15q syndrome or CDKL5 deficiency disorder in an open-label signal-finding phase II study (ARCADE). Epilepsy and Behavior, 2023, 142, 109173.	0.9	2
285	Genetic and clinical variations of developmental epileptic encephalopathies. Neurological Research, 2023, 45, 226-233.	0.6	1
286	The current landscape of epilepsy genetics: where are we, and where are we going?. Current Opinion in Neurology, 2023, 36, 86-94.	1.8	6
287	The current benefit of genome sequencing compared to exome sequencing in patients with developmental or epileptic encephalopathies. Molecular Genetics & Genomic Medicine, 2023, 11, .	0.6	3

#	Article	IF	Citations
288	Single nucleotide variations encoding missense mutations in G proteinâ€coupled receptors may contribute to autism. British Journal of Pharmacology, 0, , .	2.7	1
289	Rhythmic cortical myoclonus in patients with 6Q22.1 deletion. European Journal of Paediatric Neurology, 2023, 44, 25-27.	0.7	0
290	Morphological and behavioral analysis of Slc35f1-deficient mice revealed no neurodevelopmental phenotype. Brain Structure and Function, 0, , .	1.2	1
293	Genetic disorders of neurotransmitter release machinery. Frontiers in Synaptic Neuroscience, 0, 15, .	1.3	1
294	A deep intronic variant in DNM1 in a patient with developmental and epileptic encephalopathy creates a splice acceptor site and affects only transcript variants including exon 10a. Neurogenetics, 0, , .	0.7	0
303	Case report: splicing effect of a novel heterozygous variant of the NUS1 gene in a child with epilepsy. Frontiers in Genetics, 0, 14, .	1.1	0
309	GABAergic Neurotransmission Abnormalities in Pharmacoresistant Epilepsy: Experimental and Human Studies. , 2023, , 335-369.		0
315	Case report: Identification of a recurrent pathogenic DHDDS mutation in Chinese family with epilepsy, intellectual disability and myoclonus. Frontiers in Genetics, 0, 14, .	1.1	0
324	Editorial: Mitochondrial Gene Variations Increase Autism Risk: Uncovering the Complex Polygenetic Landscape of Autism. Journal of the American Academy of Child and Adolescent Psychiatry, 2023, , .	0.3	0