Heather C Mefford

List of Publications by Year
in descending order

177 papers	19,110 citations	20759 60 h-index	13338 130 g-index
209 all docs	209 docs citations	209 times ranked	21243 citing authors

#	Article	IF	Citations
1	De novo mutations in epileptic encephalopathies. Nature, 2013, 501, 217-221.	13.7	1,351
2	Exome sequencing identifies MLL2 mutations as a cause of Kabuki syndrome. Nature Genetics, 2010, 42, 790-793.	9.4	1,238
3	Multiplex Targeted Sequencing Identifies Recurrently Mutated Genes in Autism Spectrum Disorders. Science, 2012, 338, 1619-1622.	6.0	1,133
4	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	13.9	663
5	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. Cell, 2014, 158, 263-276.	13.5	637
6	SFARI Gene 2.0: a community-driven knowledgebase for the autism spectrum disorders (ASDs). Molecular Autism, 2013, 4, 36.	2.6	632
7	Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. Nature Genetics, 2013, 45, 825-830.	9.4	589
8	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	9.4	583
9	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. Nature Genetics, 2010, 42, 203-209.	9.4	539
10	Population Analysis of Large Copy Number Variants and Hotspots of Human Genetic Disease. American Journal of Human Genetics, 2009, 84, 148-161.	2.6	530
11	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. Nature Genetics, 2009, 41, 160-162.	9.4	511
12	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. Nature Genetics, 2008, 40, 322-328.	9.4	509
13	Genome-Wide Copy Number Variation in Epilepsy: Novel Susceptibility Loci in Idiopathic Generalized and Focal Epilepsies. PLoS Genetics, 2010, 6, e1000962.	1.5	414
14	Recurrent microdeletions at 15q11.2 and 16p13.11 predispose to idiopathic generalized epilepsies. Brain, 2010, 133, 23-32.	3.7	406
15	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
16	GRIN2A mutations cause epilepsy-aphasia spectrum disorders. Nature Genetics, 2013, 45, 1073-1076.	9.4	326
17	Genomics, Intellectual Disability, and Autism. New England Journal of Medicine, 2012, 366, 733-743.	13.9	276
18	The phenotypic spectrum of SCN8A encephalopathy. Neurology, 2015, 84, 480-489.	1.5	246

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19	GABRA1 and STXBP1 : Novel genetic causes of Dravet syndrome. Neurology, 2014, 82, 1245-1253.	1.5	229
20	Recurrent Reciprocal Genomic Rearrangements of 17q12 Are Associated with Renal Disease, Diabetes, and Epilepsy. American Journal of Human Genetics, 2007, 81, 1057-1069.	2.6	222
21	Rare copy number variants are an important cause of epileptic encephalopathies. Annals of Neurology, 2011, 70, 974-985.	2.8	222
22	Clinical whole-genome sequencing in severe early-onset epilepsy reveals new genes and improves molecular diagnosis. Human Molecular Genetics, 2014, 23, 3200-3211.	1.4	222
23	Familial and sporadic 15q13.3 microdeletions in idiopathic generalized epilepsy: precedent for disorders with complex inheritance. Human Molecular Genetics, 2009, 18, 3626-3631.	1.4	211
24	SCN2A encephalopathy. Neurology, 2015, 85, 958-966.	1.5	211
25	Duplication hotspots, rare genomic disorders, and common disease. Current Opinion in Genetics and Development, 2009, 19, 196-204.	1.5	191
26	Ultra-rare genetic variation in common epilepsies: a case-control sequencing study. Lancet Neurology, The, 2017, 16, 135-143.	4.9	190
27	GRIN2Bencephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. Journal of Medical Genetics, 2017, 54, 460-470.	1.5	190
28	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.	1.1	178
29	Advancing epilepsy genetics in the genomic era. Genome Medicine, 2015, 7, 91.	3.6	173
30	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. American Journal of Human Genetics, 2015, 96, 808-815.	2.6	173
31	The genetic landscape of infantile spasms. Human Molecular Genetics, 2014, 23, 4846-4858.	1.4	156
32	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	5.8	150
33	The Genetics of Microdeletion and Microduplication Syndromes: An Update. Annual Review of Genomics and Human Genetics, 2014, 15, 215-244.	2.5	145
34	Copy number variants are frequent in genetic generalized epilepsy with intellectual disability. Neurology, 2013, 81, 1507-1514.	1.5	140
35	Genetic and neurodevelopmental spectrum ofSYNGAP1-associated intellectual disability and epilepsy. Journal of Medical Genetics, 2016, 53, 511-522.	1.5	135
36	Clinical phenotype of the recurrent 1q21.1 copy-number variant. Genetics in Medicine, 2016, 18, 341-349.	1.1	134

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37	SYNGAP1 encephalopathy. Neurology, 2019, 92, e96-e107.	1.5	131
38	A method for rapid, targeted CNV genotyping identifies rare variants associated with neurocognitive disease. Genome Research, 2009, 19, 1579-1585.	2.4	118
39	Mutations in <scp>KCNT</scp>1 cause a spectrum of focal epilepsies. Epilepsia, 2015, 56, e114-20.	2.6	117
40	CHD2variants are a risk factor for photosensitivity in epilepsy. Brain, 2015, 138, 1198-1208.	3.7	112
41	Targeted long-read sequencing identifies missing disease-causing variation. American Journal of Human Genetics, 2021, 108, 1436-1449.	2.6	105
42	Parental Mosaicism in â€œDe Novoâ€•Epileptic Encephalopathies. New England Journal of Medicine, 2018, 378, 1646-1648.	13.9	104
43	Germline De Novo Mutations in GNB1 Cause Severe Neurodevelopmental Disability, Hypotonia, and Seizures. American Journal of Human Genetics, 2016, 98, 1001-1010.	2.6	102
44	SCN8A encephalopathy: Research progress and prospects. Epilepsia, 2016, 57, 1027-1035.	2.6	101
45	Defining the phenotypic spectrum of SLC6A1 mutations. Epilepsia, 2018, 59, 389-402.	2.6	99
46	Not all SCN1A epileptic encephalopathies are Dravet syndrome. Neurology, 2017, 89, 1035-1042.	1.5	97
47	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. Annals of Neurology, 2019, 86, 821-831.	2.8	96
48	TRIOloss of function is associated with mild intellectual disability and affects dendritic branching and synapse function. Human Molecular Genetics, 2016, 25, 892-902.	1.4	94
49	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. American Journal of Human Genetics, 2019, 104, 213-228.	2.6	90
50	Mutations in GABRB3. Neurology, 2017, 88, 483-492.	1.5	87
51	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. American Journal of Human Genetics, 2018, 103, 666-678.	2.6	87
52	GGC Repeat Expansion and Exon 1 Methylation of XYLT1 Is a Common Pathogenic Variant in Baratela-Scott Syndrome. American Journal of Human Genetics, 2019, 104, 35-44.	2.6	81
53	CHD2 myoclonic encephalopathy is frequently associated with self-induced seizures. Neurology, 2015, 84, 951-958.	1.5	79
54	Primer Part 1â€”The building blocks of epilepsy genetics. Epilepsia, 2016, 57, 861-868.	2.6	77

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55	A populationâ€based costâ€effectiveness study of early genetic testing in severe epilepsies of infancy. Epilepsia, 2018, 59, 1177-1187.	2.6	77
56	Aberrant Inclusion of a Poison Exon Causes Dravet Syndrome and Related SCN1A-Associated Genetic Epilepsies. American Journal of Human Genetics, 2018, 103, 1022-1029.	2.6	76
57	Autism and developmental disability caused by KCNQ3 gainâ€ofâ€function variants. Annals of Neurology, 2019, 86, 181-192.	2.8	73
58	De novo mutations in GRIN1 cause extensive bilateral polymicrogyria. Brain, 2018, 141, 698-712.	3.7	72
59	Dominant KCNA2 mutation causes episodic ataxia and pharmacoresponsive epilepsy. Neurology, 2016, 87, 1975-1984.	1.5	71
60	Copy number variation analysis in singleâ€suture craniosynostosis: Multiple rare variants including RUNX2 duplication in two cousins with metopic craniosynostosis. American Journal of Medical Genetics, Part A, 2010, 152A, 2203-2210.	0.7	69
61	Seizures Are Regulated by Ubiquitin-specific Peptidase 9 X-linked (USP9X), a De-Ubiquitinase. PLoS Genetics, 2015, 11, e1005022.	1.5	66
62	Epileptic spasms are a feature of DEPDC5 mTORopathy. Neurology: Genetics, 2015, 1, e17.	0.9	63
63	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. Human Molecular Genetics, 2014, 23, 6069-6080.	1.4	61
64	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	2.6	61
65	Exonâ€disrupting deletions of <scp>NRXN1</scp> in idiopathic generalized epilepsy. Epilepsia, 2013, 54, 256-264.	2.6	59
66	Mutations Affecting the SAND Domain of DEAF1 Cause Intellectual Disability with Severe Speech Impairment and Behavioral Problems. American Journal of Human Genetics, 2014, 94, 649-661.	2.6	59
67	Copy number variant analysis from exome data in 349 patients with epileptic encephalopathy. Annals of Neurology, 2015, 78, 323-328.	2.8	59
68	Epilepsy and the new cytogenetics. Epilepsia, 2011, 52, 423-432.	2.6	56
69	Clinical phenotype of ASD-associated DYRK1A haploinsufficiency. Molecular Autism, 2017, 8, 54.	2.6	55
70	Comparative sequencing of a multicopy subtelomeric region containing olfactory receptor genes reveals multiple interactions between non-homologous chromosomes. Human Molecular Genetics, 2001, 10, 2363-2372.	1.4	51
71	A targeted resequencing gene panel for focal epilepsy. Neurology, 2016, 86, 1605-1612.	1.5	48
72	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.	2.6	48

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73	De novo mutations ofKIAA2022in females cause intellectual disability and intractable epilepsy. Journal of Medical Genetics, 2016, 53, 850-858.	1.5	47
74	Recent advances in epilepsy genomics and genetic testing. F1000Research, 2020, 9, 185.	0.8	47
75	Somatic mutation: The hidden genetics of brain malformations and focal epilepsies. Epilepsy Research, 2019, 155, 106161.	0.8	45
76	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	2.6	45
77	A homozygous B3GAT3 mutation causes a severe syndrome with multiple fractures, expanding the phenotype of linkeropathy syndromes. American Journal of Medical Genetics, Part A, 2015, 167, 2691-2696.	0.7	44
78	Severe infantile onset developmental and epileptic encephalopathy caused by mutations in autophagy gene <scp>WDR</scp>45. Epilepsia, 2018, 59, e5-e13.	2.6	44
79	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. Annals of Neurology, 2018, 84, 788-795.	2.8	44
80	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. American Journal of Human Genetics, 2017, 101, 516-524.	2.6	43
81	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	5.8	43
82	Microdeletion syndromes. Current Opinion in Genetics and Development, 2013, 23, 232-239.	1.5	42
83	Genetic contribution to common epilepsies. Current Opinion in Neurology, 2011, 24, 140-145.	1.8	41
84	TANGO2: expanding the clinical phenotype and spectrum of pathogenic variants. Genetics in Medicine, 2019, 21, 601-607.	1.1	41
85	Further clinical and molecular delineation of the 15q24 microdeletion syndrome. Journal of Medical Genetics, 2012, 49, 110-118.	1.5	40
86	Simultaneous impairment of neuronal and metabolic function of mutated gephyrin in a patient with epileptic encephalopathy. EMBO Molecular Medicine, 2015, 7, 1580-1594.	3.3	39
87	Genetic heterogeneity in infantile spasms. Epilepsy Research, 2019, 156, 106181.	0.8	38
88	Genotype to phenotypeâ€”discovery and characterization of novel genomic disorders in a â€œgenotype-firstâ€•era. Genetics in Medicine, 2009, 11, 836-842.	1.1	37
89	Loss of function of the retinoid-related nuclear receptor (RORB) gene and epilepsy. European Journal of Human Genetics, 2016, 24, 1761-1770.	1.4	36
90	The Impact of Rapid Exome Sequencing on Medical Management of Critically Ill Children. Journal of Pediatrics, 2020, 226, 202-212.e1.	0.9	35

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91	Intragenic deletions of ALDH7A1 in pyridoxine-dependent epilepsy caused by Alu - Alu recombination. Neurology, 2015, 85, 756-762.	1.5	34
92	De novo loss-of-function mutations in WAC cause a recognizable intellectual disability syndrome and learning deficits in Drosophila. European Journal of Human Genetics, 2016, 24, 1145-1153.	1.4	34
93	The ClinGen Epilepsy Gene Curation Expert Panelâ€"Bridging the divide between clinical domain knowledge and formal gene curation criteria. Human Mutation, 2018, 39, 1476-1484.	1.1	33
94	Expanding the genetic and phenotypic relevance of KCNB1 variants in developmental and epileptic encephalopathies: 27 new patients and overview of the literature. Human Mutation, 2020, 41, 69-80.	1.1	33
95	Genetically complex epilepsies, copy number variants and syndrome constellations. Genome Medicine, 2010, 2, 71.	3.6	32
96	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. European Journal of Human Genetics, 2018, 26, 54-63.	1.4	32
97	The severe epilepsy syndromes of infancy: A populationâ€based study. Epilepsia, 2021, 62, 358-370.	2.6	31
98	Next-Generation Sequencing in Intellectual Disability. Journal of Pediatric Genetics, 2015, 04, 128-135.	0.3	30
99	Expanding clinical phenotype in CACNA1C related disorders: From neonatal onset severe epileptic encephalopathy to lateâ€onset epilepsy. American Journal of Medical Genetics, Part A, 2018, 176, 2733-2739.	0.7	30
100	ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 319-330.	2.6	30
101	Absence seizures with intellectual disability as a phenotype of the 15q13.3 microdeletion syndrome. Epilepsia, 2011, 52, e194-8.	2.6	29
102	CNVs in Epilepsy. Current Genetic Medicine Reports, 2014, 2, 162-167.	1.9	28
103	Severe neurocognitive and growth disorders due to variation in THOC2 , an essential component of nuclear mRNA export machinery. Human Mutation, 2018, 39, 1126-1138.	1.1	28
104	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	1.1	28
105	Loss of function inROBO1is associated with tetralogy of Fallot and septal defects. Journal of Medical Genetics, 2017, 54, 825-829.	1.5	27
106	Poison exons in neurodevelopment and disease. Current Opinion in Genetics and Development, 2020, 65, 98-102.	1.5	26
107	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. American Journal of Human Genetics, 2020, 107, 727-742.	2.6	25
108	Severe speech impairment is a distinguishing feature of FOXP1â€related disorder. Developmental Medicine and Child Neurology, 2021, 63, 1417-1426.	1.1	24

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109	Multiplex families with epilepsy. Neurology, 2016, 86, 713-722.	1.5	23
110	Recurrent duplications of 17q12 associated with variable phenotypes. American Journal of Medical Genetics, Part A, 2015, 167, 3038-3045.	0.7	22
111	Clinical Genetic Testing in Epilepsy. Epilepsy Currents, 2015, 15, 197-201.	0.4	22
112	Clinical Phenotypes of Carriers of Mutations in CHD8 or Its Conserved Target Genes. Biological Psychiatry, 2020, 87, 123-131.	0.7	22
113	Iterative phenotyping of 15q11.2, 15q13.3 and 16p13.11 microdeletion carriers in pediatric epilepsies. Epilepsy Research, 2014, 108, 109-116.	0.8	21
114	Pathogenic Variants in CEP85L Cause Sporadic and Familial Posterior Predominant Lissencephaly. Neuron, 2020, 106, 237-245.e8.	3.8	21
115	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. Annals of Neurology, 2018, 83, 926-934.	2.8	20
116	De novo and biallelic DEAF1 variants cause a phenotypic spectrum. Genetics in Medicine, 2019, 21, 2059-2069.	1.1	20
117	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	1.1	20
118	Genetic investigations of the epileptic encephalopathies. Progress in Brain Research, 2016, 226, 35-60.	0.9	19
119	Biallelic PI4KA variants cause a novel neurodevelopmental syndrome with hypomyelinating leukodystrophy. Brain, 2021, 144, 2659-2669.	3.7	19
120	Epilepsy due to 20q13.33 subtelomere deletion masquerading as pyridoxineâ€dependent epilepsy. American Journal of Medical Genetics, Part A, 2012, 158A, 3190-3195.	0.7	18
121	Deletions of 16p11.2 and 19p13.2 in a family with intellectual disability and generalized epilepsy. American Journal of Medical Genetics, Part A, 2013, 161, 1722-1725.	0.7	18
122	<scp>BRAT</scp>1 encephalopathy: a recessive cause of epilepsy of infancy with migrating focal seizures. Developmental Medicine and Child Neurology, 2020, 62, 1096-1099.	1.1	18
123	Bi-allelic Loss-of-Function Variants in NUP188 Cause a Recognizable Syndrome Characterized by Neurologic, Ocular, and Cardiac Abnormalities. American Journal of Human Genetics, 2020, 106, 623-631.	2.6	18
124	Genetic literacy series: Primer part 2â€”Paradigm shifts in epilepsy genetics. Epilepsia, 2018, 59, 1138-1147.	2.6	17
125	Developmental and epilepsy spectrum of KCNB1 encephalopathy with longâ€term outcome. Epilepsia, 2020, 61, 2461-2473.	2.6	17
126	The unexpected role of copy number variations in juvenile myoclonic epilepsy. Epilepsy and Behavior, 2013, 28, S66-S68.	0.9	16

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127	Investigating the genetic basis of feverâ€associated syndromic epilepsies using copy number variation analysis. Epilepsia, 2015, 56, e26-32.	2.6	16
128	Levetiracetam efficacy in PCDH19 Girls Clustering Epilepsy. European Journal of Paediatric Neurology, 2020, 24, 142-147.	0.7	16
129	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617.	2.6	16
130	Five patients with a chromosome 1q21.1 triplication show macrocephaly, increased weight and facial similarities. European Journal of Medical Genetics, 2015, 58, 503-508.	0.7	15
131	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
132	Inherited RORB pathogenic variants: Overlap of photosensitive genetic generalized and occipital lobe epilepsy. Epilepsia, 2020, 61, e23-e29.	2.6	14
133	The epileptology of GNB5 encephalopathy. Epilepsia, 2019, 60, e121-e127.	2.6	13
134	Variants in GNAI1 cause a syndrome associated with variable features including developmental delay, seizures, and hypotonia. Genetics in Medicine, 2021, 23, 881-887.	1.1	13
135	Copy Number Matters in Epilepsy. Epilepsy Currents, 2015, 15, 180-182.	0.4	12
136	Dravet syndrome in South African infants: Tools for an early diagnosis. Seizure: the Journal of the British Epilepsy Association, 2018, 62, 99-105.	0.9	12
137	Beyond the single nucleotide variant in epilepsy genetics. Nature Reviews Neurology, 2014, 10, 490-491.	4.9	11
138	Description of a new oncogenic mechanism for atypical teratoid rhabdoid tumors in patients with ring chromosome 22. American Journal of Medical Genetics, Part A, 2017, 173, 245-249.	0.7	11
139	Developmental and epileptic encephalopathy: Personal utility of a genetic diagnosis for families. Epilepsia Open, 2021, 6, 149-159.	1.3	11
140	The Road to Diagnosis: Shortening the Diagnostic Odyssey in Epilepsy. Epilepsy Currents, 2019, 19, 307-309.	0.4	10
141	The phenotypic spectrum of Xâ€linked, infantile onset ALG13â€related developmental and epileptic encephalopathy. Epilepsia, 2021, 62, 325-334.	2.6	10
142	2014 Epilepsy Benchmarks Area I: Understanding the Causes of the Epilepsies and Epilepsy-Related Neurologic, Psychiatric, and Somatic Conditions. Epilepsy Currents, 2016, 16, 182-186.	0.4	9
143	Return of individual results in epilepsy genomic research: A view from the field. Epilepsia, 2018, 59, 1635-1642.	2.6	9
144	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

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145	Epilepsy Benchmarks Area III: Improved Treatment Options for Controlling Seizures and Epilepsy-Related Conditions Without Side Effects. Epilepsy Currents, 2020, 20, 23S-30S.	0.4	9
146	15q13.3 microdeletions in a prospectively recruited cohort of patients with idiopathic generalized epilepsy in Bulgaria. Epilepsy Research, 2013, 104, 241-245.	0.8	8
147	FBXO28 causes developmental and epileptic encephalopathy with profound intellectual disability. Epilepsia, 2021, 62, e13-e21.	2.6	8
148	Phenotypic Spectrum of Seizure Disorders in MBD5-Associated Neurodevelopmental Disorder. Neurology: Genetics, 2021, 7, e579.	0.9	8
149	Association of ultraâ€rare coding variants with genetic generalized epilepsy: A caseâ€“control whole exome sequencing study. Epilepsia, 2022, 63, 723-735.	2.6	8
150	Biallelic ADAM22 pathogenic variants cause progressive encephalopathy and infantile-onset refractory epilepsy. Brain, 2022, 145, 2301-2312.	3.7	8
151	A de novo inâ€frame deletion of CASK gene causes early onset infantile spasms and supratentorial cerebral malformation in a female patient. American Journal of Medical Genetics, Part A, 2018, 176, 2425-2429.	0.7	7
152	Double somatic mosaicism in a child with Dravet syndrome. Neurology: Genetics, 2019, 5, e333.	0.9	7
153	Reanalysis and optimisation of bioinformatic pipelines is critical for mutation detection. Human Mutation, 2019, 40, 374-379.	1.1	7
154	Rare variants in KDR, encoding VEGF Receptor 2, are associated with tetralogy of Fallot. Genetics in Medicine, 2021, 23, 1952-1960.	1.1	7
155	Pathogenic <scp>MAST3</scp> Variants in the <scp>STK</scp> Domain Are Associated with Epilepsy. Annals of Neurology, 2021, 90, 274-284.	2.8	7
156	Evaluation of multiple putative risk alleles within the 15q13.3 region for genetic generalized epilepsy. Epilepsy Research, 2015, 117, 70-73.	0.8	6
157	De novo FZR1 loss-of-function variants cause developmental and epileptic encephalopathies. Brain, 2022, 145, 1684-1697.	3.7	5
158	SCN1A mutations in focal epilepsy with auditory features: widening the spectrum of GEFS plus. Epileptic Disorders, 2019, 21, 185-191.	0.7	5
159	Clarifying the role of the 22q11.2 microdeletion in juvenile myoclonic epilepsy. Epilepsy and Behavior, 2013, 29, 589-590.	0.9	4
160	Expanding role of GABAA receptors in generalised epilepsies. Lancet Neurology, The, 2018, 17, 657-658.	4.9	4
161	Genetic convergence of developmental and epileptic encephalopathies and intellectual disability. Developmental Medicine and Child Neurology, 2021, 63, 1441-1447.	1.1	4
162	Diagnostic Considerations in the Epilepsiesâ€”Testing Strategies, Test Type Advantages, and Limitations. Neurotherapeutics, 2021, 18, 1468-1477.	2.1	4

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163	A recurrent, de novo pathogenic variant in ARPC4 disrupts actin filament formation and causes a neurodevelopmental disorder with microcephaly and speech delay. Human Genetics and Genomics Advances, 2021, 3, 100072.	1.0	4
164	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
165	Copy number variants in the population: Unselected does not mean unaffected. Epilepsy Currents, 2016, 16, 91-93.	0.4	3
166	Mutations of the DNA repair gene PNKP in a patient with microcephaly, seizures, and developmental delay (MCSZ) presenting with a high-grade brain tumor. Scientific Reports, 2022, 12, 5386.	1.6	3
167	Phenotype to Genotype and Back Again. Epilepsy Currents, 2020, 20, 88-89.	0.4	1
168	Shedding Light on the Genomeâ€™s Dark Matter. Science Translational Medicine, 2014, 6, .	5.8	1
169	Mosaicism in clinical genetics. Journal of Physical Education and Sports Management, 2021, 7, a006162.	0.5	1
170	Thinking about cognition and epilepsy. Epilepsy and Behavior, 2014, 41, 276.	0.9	0
171	Finding the Missing Pieces: The Microdeletion Burden in GGE. Epilepsy Currents, 2016, 16, 16-17.	0.4	0
172	Antiepileptic Drugs as Teratogens: The Mechanism Remains a Mystery. Epilepsy Currents, 2020, 20, 365-366.	0.4	0
173	SNFing for Clues to the Genetics of Autism. Science Translational Medicine, 2014, 6, .	5.8	0
174	â€œBrain-Criticalâ€•Exons in Autism. Science Translational Medicine, 2014, 6, .	5.8	0
175	Whatâ€™s Your Attitude?. Science Translational Medicine, 2014, 6, .	5.8	0
176	Journey to the Center of the Cause. Science Translational Medicine, 2015, 7, .	5.8	0
177	Patchwork people: A role for somatic mutations in brain malformations. Science Translational Medicine, 2015, 7, .	5.8	0