Heather C Mefford

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

169 papers	14,208 citations	52 h-index	118 g-index
209 ext. papers	17,214 ext. citations	9.5 avg, IF	5.81 L-index

#	Paper	IF	Citations
169	A recurrent, pathogenic variant in disrupts actin filament formation and causes microcephaly and speech delay.. *Human Genetics and Genomics Advances*, **2022**, 3, 100072	0.8	0
168	Genome sequencing reveals novel noncoding variants in PLA2G6 and LMNB1 causing progressive neurologic disease.. *Molecular Genetics & Genomic Medicine*, **2022**, e1892	2.3	2
167	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	4.9	1
166	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome.. *American Journal of Human Genetics*, **2022**, 109, 601-617	11	0
165	Missense variants in CTNNB1 can be associated with vitreoretinopathy-Seven new cases of CTNNB1-associated neurodevelopmental disorder including a previously unreported retinal phenotype. *Molecular Genetics & Genomic Medicine*, **2021**, 9, e1542	2.3	0
164	De novo FZR1 loss-of-function variants cause developmental and epileptic encephalopathies. *Brain*, **2021**,	11.2	2
163	Phenotypic Spectrum of Seizure Disorders in MBD5-Associated Neurodevelopmental Disorder. *Neurology: Genetics*, **2021**, 7, e579	3.8	2
162	Severe speech impairment is a distinguishing feature of FOXP1-related disorder. *Developmental Medicine and Child Neurology*, **2021**, 63, 1417-1426	3.3	3
161	Rare variants in KDR, encoding VEGF Receptor 2, are associated with tetralogy of Fallot. *Genetics in Medicine*, **2021**, 23, 1952-1960	8.1	1
160	Genetic convergence of developmental and epileptic encephalopathies and intellectual disability. *Developmental Medicine and Child Neurology*, **2021**, 63, 1441-1447	3.3	1
159	Pathogenic MAST3 Variants in the STK Domain Are Associated with Epilepsy. *Annals of Neurology*, **2021**, 90, 274-284	9.4	1
158	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. *Genetics in Medicine*, **2021**, 23, 363-373	8.1	4
157	FBXO28 causes developmental and epileptic encephalopathy with profound intellectual disability. *Epilepsia*, **2021**, 62, e13-e21	6.4	1
156	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. *Genetics in Medicine*, **2021**, 23, 653-660	8.1	5
155	Developmental and epileptic encephalopathy: Personal utility of a genetic diagnosis for families. *Epilepsia Open*, **2021**, 6, 149-159	4	2
154	Variants in GNAI1 cause a syndrome associated with variable features including developmental delay, seizures, and hypotonia. *Genetics in Medicine*, **2021**, 23, 881-887	8.1	1
153	The severe epilepsy syndromes of infancy: A population-based study. *Epilepsia*, **2021**, 62, 358-370	6.4	7

152	Biallelic PI4KA variants cause a novel neurodevelopmental syndrome with hypomyelinating leukodystrophy. *Brain*, **2021**, 144, 2659-2669	11.2	2
151	Targeted long-read sequencing identifies missing disease-causing variation. *American Journal of Human Genetics*, **2021**, 108, 1436-1449	11	16
150	Diagnostic Considerations in the Epilepsies-Testing Strategies, Test Type Advantages, and Limitations. *Neurotherapeutics*, **2021**, 18, 1468-1477	6.4	0
149	The phenotypic spectrum of X-linked, infantile onset ALG13-related developmental and epileptic encephalopathy. *Epilepsia*, **2021**, 62, 325-334	6.4	4
148	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. *American Journal of Human Genetics*, **2020**, 107, 727-742	11	2
147	Phenotype to Genotype and Back Again. *Epilepsy Currents*, **2020**, 20, 88-89	1.3	0
146	Inherited RORB pathogenic variants: Overlap of photosensitive genetic generalized and occipital lobe epilepsy. *Epilepsia*, **2020**, 61, e23-e29	6.4	5
145	Poison exons in neurodevelopment and disease. *Current Opinion in Genetics and Development*, **2020**, 65, 98-102	4.9	8
144	Pathogenic Variants in CEP85L Cause Sporadic and Familial Posterior Predominant Lissencephaly. *Neuron*, **2020**, 106, 237-245.e8	13.9	10
143	Epilepsy Benchmarks Area III: Improved Treatment Options for Controlling Seizures and Epilepsy-Related Conditions Without Side Effects. *Epilepsy Currents*, **2020**, 20, 23S-30S	1.3	5
142	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	11	5
141	Recent advances in epilepsy genomics and genetic testing. *F1000Research*, **2020**, 9,	3.6	25
140	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	4.7	14
139	Three novel patients with epileptic encephalopathy due to biallelic mutations in the PLCB1 gene. *Clinical Genetics*, **2020**, 97, 477-482	4	3
138	Levetiracetam efficacy in PCDH19 Girls Clustering Epilepsy. *European Journal of Paediatric Neurology*, **2020**, 24, 142-147	3.8	9
137	BRAT1 encephalopathy: a recessive cause of epilepsy of infancy with migrating focal seizures. *Developmental Medicine and Child Neurology*, **2020**, 62, 1096-1099	3.3	7
136	The Impact of Rapid Exome Sequencing on Medical Management of Critically Ill Children. *Journal of Pediatrics*, **2020**, 226, 202-212.e1	3.6	9
135	Antiepileptic Drugs as Teratogens: The Mechanism Remains a Mystery. *Epilepsy Currents*, **2020**, 20, 365-366	1.3	0

134	Developmental and epilepsy spectrum of KCNB1 encephalopathy with long-term outcome. *Epilepsia*, **2020**, 61, 2461-2473	6.4	4
133	Clinical Phenotypes of Carriers of Mutations in CHD8 or Its Conserved Target Genes. *Biological Psychiatry*, **2020**, 87, 123-131	7.9	14
132	The epileptology of GNB5 encephalopathy. *Epilepsia*, **2019**, 60, e121-e127	6.4	9
131	The Road to Diagnosis: Shortening the Diagnostic Odyssey in Epilepsy. *Epilepsy Currents*, **2019**, 19, 307-309	2.3	5
130	Autism and developmental disability caused by KCNQ3 gain-of-function variants. *Annals of Neurology*, **2019**, 86, 181-192	9.4	30
129	Double somatic mosaicism in a child with Dravet syndrome. *Neurology: Genetics*, **2019**, 5, e333	3.8	5
128	De novo and biallelic DEAF1 variants cause a phenotypic spectrum. *Genetics in Medicine*, **2019**, 21, 2059-2069	8.1	11
127	Diagnostic implications of genetic copy number variation in epilepsy plus. *Epilepsia*, **2019**, 60, 689-706	6.4	37
126	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. *American Journal of Human Genetics*, **2019**, 104, 948-956	11	17
125	Genetic heterogeneity in infantile spasms. *Epilepsy Research*, **2019**, 156, 106181	3	20
124	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. *Nature Communications*, **2019**, 10, 3094	17.4	76
123	Somatic mutation: The hidden genetics of brain malformations and focal epilepsies. *Epilepsy Research*, **2019**, 155, 106161	3	24
122	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. *Nature Communications*, **2019**, 10, 4679	17.4	21
121	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. *Annals of Neurology*, **2019**, 86, 821-831	9.4	55
120	SCN1A mutations in focal epilepsy with auditory features: widening the spectrum of GEFS plus **2019**, 21, 185-191		2
119	Reanalysis and optimisation of bioinformatic pipelines is critical for mutation detection. *Human Mutation*, **2019**, 40, 374-379	4.7	6
118	encephalopathy: A distinctive generalized developmental and epileptic encephalopathy. *Neurology*, **2019**, 92, e96-e107	6.5	55
117	ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. *American Journal of Human Genetics*, **2019**, 104, 319-330	11	19

116	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	11	58
115	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	11	47
114	TANGO2: expanding the clinical phenotype and spectrum of pathogenic variants. *Genetics in Medicine*, **2019**, 21, 601-607	8.1	18
113	Parental Mosaicism in "De Novo" Epileptic Encephalopathies. *New England Journal of Medicine*, **2018**, 378, 1646-1648	59.2	67
112	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. *Annals of Neurology*, **2018**, 83, 926-934	9.4	11
111	De novo mutations in GRIN1 cause extensive bilateral polymicrogyria. *Brain*, **2018**, 141, 698-712	11.2	46
110	Defining the phenotypic spectrum of SLC6A1 mutations. *Epilepsia*, **2018**, 59, 389-402	6.4	54
109	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	11	21
108	Expanding role of GABA receptors in generalised epilepsies. *Lancet Neurology, The*, **2018**, 17, 657-658	24.1	2
107	Genetic literacy series: Primer part 2-Paradigm shifts in epilepsy genetics. *Epilepsia*, **2018**, 59, 1138-1147	6.4	9
106	A population-based cost-effectiveness study of early genetic testing in severe epilepsies of infancy. *Epilepsia*, **2018**, 59, 1177-1187	6.4	46
105	Return of individual results in epilepsy genomic research: A view from the field. *Epilepsia*, **2018**, 59, 1635-1642	6.4	7
104	Severe neurocognitive and growth disorders due to variation in THOC2, an essential component of nuclear mRNA export machinery. *Human Mutation*, **2018**, 39, 1126-1138	4.7	8
103	Severe infantile onset developmental and epileptic encephalopathy caused by mutations in autophagy gene WDR45. *Epilepsia*, **2018**, 59, e5-e13	6.4	34
102	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. *European Journal of Human Genetics*, **2018**, 26, 54-63	5.3	23
101	Aberrant Inclusion of a Poison Exon Causes Dravet Syndrome and Related SCN1A-Associated Genetic Epilepsies. *American Journal of Human Genetics*, **2018**, 103, 1022-1029	11	40
100	Expanding clinical phenotype in CACNA1C related disorders: From neonatal onset severe epileptic encephalopathy to late-onset epilepsy. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2733-2739	2.5	16
99	A de novo in-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	2.5	4

98	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. *Annals of Neurology*, **2018**, 84, 788-795	9.4	18
97	The ClinGen Epilepsy Gene Curation Expert Panel-Bridging the divide between clinical domain knowledge and formal gene curation criteria. *Human Mutation*, **2018**, 39, 1476-1484	4.7	21
96	Dravet syndrome in South African infants: Tools for an early diagnosis. *Seizure: the Journal of the British Epilepsy Association*, **2018**, 62, 99-105	3.2	5
95	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	11	44
94	Ultra-rare genetic variation in common epilepsies: a case-control sequencing study. *Lancet Neurology, The*, **2017**, 16, 135-143	24.1	133
93	Loss of function in is associated with tetralogy of Fallot and septal defects. *Journal of Medical Genetics*, **2017**, 54, 825-829	5.8	13
92	encephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. *Journal of Medical Genetics*, **2017**, 54, 460-470	5.8	109
91	Mutations in GABRB3: From febrile seizures to epileptic encephalopathies. *Neurology*, **2017**, 88, 483-492	6.5	68
90	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. *American Journal of Human Genetics*, **2017**, 101, 664-685	11	214
89	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. *American Journal of Human Genetics*, **2017**, 101, 516-524	11	29
88	Clinical phenotype of ASD-associated haploinsufficiency. *Molecular Autism*, **2017**, 8, 54	6.5	29
87	Not all epileptic encephalopathies are Dravet syndrome: Early profound Thr226Met phenotype. *Neurology*, **2017**, 89, 1035-1042	6.5	62
86	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	2.5	4
85	Clinical phenotype of the recurrent 1q21.1 copy-number variant. *Genetics in Medicine*, **2016**, 18, 341-9	8.1	84
84	Finding the Missing Pieces: The Microdeletion Burden in GGE. *Epilepsy Currents*, **2016**, 16, 16-7	1.3	
83	Genetic investigations of the epileptic encephalopathies: Recent advances. *Progress in Brain Research*, **2016**, 226, 35-60	2.9	16
82	De novo mutations of KIAA2022 in females cause intellectual disability and intractable epilepsy. *Journal of Medical Genetics*, **2016**, 53, 850-858	5.8	32
81	Loss of function of the retinoid-related nuclear receptor (RORB) gene and epilepsy. *European Journal of Human Genetics*, **2016**, 24, 1761-1770	5.3	23

80	Multiplex families with epilepsy: Success of clinical and molecular genetic characterization. *Neurology*, **2016**, 86, 713-22	6.5	22
79	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, 1145-53	5.3	23
78	TRIO loss of function is associated with mild intellectual disability and affects dendritic branching and synapse function. *Human Molecular Genetics*, **2016**, 25, 892-902	5.6	56
77	Copy number variants in the population: unselected does not mean unaffected. *Epilepsy Currents*, **2016**, 16, 91-3	1.3	2
76	2014 Epilepsy Benchmarks Area I: Understanding the Causes of the Epilepsies and Epilepsy-Related Neurologic, Psychiatric, and Somatic Conditions. *Epilepsy Currents*, **2016**, 16, 182-6	1.3	5
75	SCN8A encephalopathy: Research progress and prospects. *Epilepsia*, **2016**, 57, 1027-35	6.4	82
74	Genetic and neurodevelopmental spectrum of SYNGAP1-associated intellectual disability and epilepsy. *Journal of Medical Genetics*, **2016**, 53, 511-22	5.8	76
73	A targeted resequencing gene panel for focal epilepsy. *Neurology*, **2016**, 86, 1605-12	6.5	37
72	Germline De Novo Mutations in GNB1 Cause Severe Neurodevelopmental Disability, Hypotonia, and Seizures. *American Journal of Human Genetics*, **2016**, 98, 1001-1010	11	70
71	Dominant KCNA2 mutation causes episodic ataxia and pharmacoresponsive epilepsy. *Neurology*, **2016**, 87, 1975-1984	6.5	50
70	Primer Part 1-The building blocks of epilepsy genetics. *Epilepsia*, **2016**, 57, 861-8	6.4	54
69	CHD2 variants are a risk factor for photosensitivity in epilepsy. *Brain*, **2015**, 138, 1198-207	11.2	81
68	Seizures are regulated by ubiquitin-specific peptidase 9 X-linked (USP9X), a de-ubiquitinase. *PLoS Genetics*, **2015**, 11, e1005022	6	49
67	CHD2 myoclonic encephalopathy is frequently associated with self-induced seizures. *Neurology*, **2015**, 84, 951-8	6.5	57
66	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. *American Journal of Human Genetics*, **2015**, 96, 808-15	11	114
65	Evaluation of multiple putative risk alleles within the 15q13.3 region for genetic generalized epilepsy. *Epilepsy Research*, **2015**, 117, 70-3	3	3
64	Investigating the genetic basis of fever-associated syndromic epilepsies using copy number variation analysis. *Epilepsia*, **2015**, 56, e26-32	6.4	12
63	Mutations in KCNT1 cause a spectrum of focal epilepsies. *Epilepsia*, **2015**, 56, e114-20	6.4	83

62	Five patients with a chromosome 1q21.1 triplication show macrocephaly, increased weight and facial similarities. *European Journal of Medical Genetics*, **2015**, 58, 503-8	2.6	8
61	SCN2A encephalopathy: A major cause of epilepsy of infancy with migrating focal seizures. *Neurology*, **2015**, 85, 958-66	6.5	163
60	Intragenic deletions of ALDH7A1 in pyridoxine-dependent epilepsy caused by Alu-Alu recombination. *Neurology*, **2015**, 85, 756-62	6.5	23
59	Copy number variant analysis from exome data in 349 patients with epileptic encephalopathy. *Annals of Neurology*, **2015**, 78, 323-8	9.4	44
58	Simultaneous impairment of neuronal and metabolic function of mutated gephyrin in a patient with epileptic encephalopathy. *EMBO Molecular Medicine*, **2015**, 7, 1580-94	12	26
57	Recurrent duplications of 17q12 associated with variable phenotypes. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 3038-45	2.5	17
56	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**, 167A, 2691-6	2.5	39
55	Epileptic spasms are a feature of DEPDC5 mTORopathy. *Neurology: Genetics*, **2015**, 1, e17	3.8	44
54	Clinical Genetic Testing in Epilepsy. *Epilepsy Currents*, **2015**, 15, 197-201	1.3	17
53	Copy Number Matters in Epilepsy. *Epilepsy Currents*, **2015**, 15, 180-2	1.3	9
52	Next-Generation Sequencing in Intellectual Disability. *Journal of Pediatric Genetics*, **2015**, 4, 128-35	0.7	14
51	Advancing epilepsy genetics in the genomic era. *Genome Medicine*, **2015**, 7, 91	14.4	121
50	The phenotypic spectrum of SCN8A encephalopathy. *Neurology*, **2015**, 84, 480-9	6.5	199
49	Clinical whole-genome sequencing in severe early-onset epilepsy reveals new genes and improves molecular diagnosis. *Human Molecular Genetics*, **2014**, 23, 3200-11	5.6	179
48	Refining analyses of copy number variation identifies specific genes associated with developmental delay. *Nature Genetics*, **2014**, 46, 1063-71	36.3	429
47	Epilepsy: Beyond the single nucleotide variant in epilepsy genetics. *Nature Reviews Neurology*, **2014**, 10, 490-1	15	10
46	CNVs in Epilepsy. *Current Genetic Medicine Reports*, **2014**, 2, 162-167	2.2	26
45	GABRA1 and STXBP1: novel genetic causes of Dravet syndrome. *Neurology*, **2014**, 82, 1245-53	6.5	180

44	The genetic landscape of infantile spasms. *Human Molecular Genetics*, **2014**, 23, 4846-58	5.6	118
43	The genetics of microdeletion and microduplication syndromes: an update. *Annual Review of Genomics and Human Genetics*, **2014**, 15, 215-244	9.7	103
42	Disruptive CHD8 mutations define a subtype of autism early in development. *Cell*, **2014**, 158, 263-276	56.2	467
41	Mutations affecting the SAND domain of DEAF1 cause intellectual disability with severe speech impairment and behavioral problems. *American Journal of Human Genetics*, **2014**, 94, 649-61	11	51
40	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. *Human Molecular Genetics*, **2014**, 23, 6069-80	5.6	45
39	Iterative phenotyping of 15q11.2, 15q13.3 and 16p13.11 microdeletion carriers in pediatric epilepsies. *Epilepsy Research*, **2014**, 108, 109-16	3	17
38	De novo mutations in epileptic encephalopathies. *Nature*, **2013**, 501, 217-21	50.4	1081
37	GRIN2A mutations cause epilepsy-aphasia spectrum disorders. *Nature Genetics*, **2013**, 45, 1073-6	36.3	249
36	Microdeletion syndromes. *Current Opinion in Genetics and Development*, **2013**, 23, 232-9	4.9	37
35	The unexpected role of copy number variations in juvenile myoclonic epilepsy. *Epilepsy and Behavior*, **2013**, 28 Suppl 1, S66-8	3.2	16
34	SFARI Gene 2.0: a community-driven knowledgebase for the autism spectrum disorders (ASDs). *Molecular Autism*, **2013**, 4, 36	6.5	352
33	Copy number variants are frequent in genetic generalized epilepsy with intellectual disability. *Neurology*, **2013**, 81, 1507-14	6.5	115
32	Clarifying the role of the 22q11.2 microdeletion in juvenile myoclonic epilepsy. *Epilepsy and Behavior*, **2013**, 29, 589-90	3.2	3
31	15q13.3 microdeletions in a prospectively recruited cohort of patients with idiopathic generalized epilepsy in Bulgaria. *Epilepsy Research*, **2013**, 104, 241-5	3	7
30	Deletions of 16p11.2 and 19p13.2 in a family with intellectual disability and generalized epilepsy. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1722-5	2.5	16
29	Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. *Nature Genetics*, **2013**, 45, 825-30	36.3	500
28	Exon-disrupting deletions of NRXN1 in idiopathic generalized epilepsy. *Epilepsia*, **2013**, 54, 256-64	6.4	48
27	Multiplex targeted sequencing identifies recurrently mutated genes in autism spectrum disorders. *Science*, **2012**, 338, 1619-22	33.3	892

26	Epilepsy due to 20q13.33 subtelomere deletion masquerading as pyridoxine-dependent epilepsy. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 3190-5	2.5	13
25	Genomics, intellectual disability, and autism. *New England Journal of Medicine*, **2012**, 366, 733-43	59.2	218
24	Further clinical and molecular delineation of the 15q24 microdeletion syndrome. *Journal of Medical Genetics*, **2012**, 49, 110-8	5.8	32
23	Genetic contribution to common epilepsies. *Current Opinion in Neurology*, **2011**, 24, 140-5	7.1	35
22	Epilepsy and the new cytogenetics. *Epilepsia*, **2011**, 52, 423-32	6.4	51
21	Absence seizures with intellectual disability as a phenotype of the 15q13.3 microdeletion syndrome. *Epilepsia*, **2011**, 52, e194-8	6.4	22
20	Rare copy number variants are an important cause of epileptic encephalopathies. *Annals of Neurology*, **2011**, 70, 974-85	9.4	176
19	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. *Nature Genetics*, **2010**, 42, 203-9	36.3	461
18	Exome sequencing identifies MLL2 mutations as a cause of Kabuki syndrome. *Nature Genetics*, **2010** , 42, 790-3	36.3	1041
17	Recurrent microdeletions at 15q11.2 and 16p13.11 predispose to idiopathic generalized epilepsies. *Brain*, **2010**, 133, 23-32	11.2	347
16	Genome-wide copy number variation in epilepsy: novel susceptibility loci in idiopathic generalized and focal epilepsies. *PLoS Genetics*, **2010**, 6, e1000962	6	348
15	Recurrent 200-kb deletions of 16p11.2 that include the SH2B1 gene are associated with developmental delay and obesity. *Genetics in Medicine*, **2010**, 12, 641-7	8.1	152
14	Genetically complex epilepsies, copy number variants and syndrome constellations. *Genome Medicine*, **2010**, 2, 71	14.4	30
13	Copy number variation analysis in single-suture craniosynostosis: multiple rare variants including RUNX2 duplication in two cousins with metopic craniosynostosis. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2203-10	2.5	66
12	Familial and sporadic 15q13.3 microdeletions in idiopathic generalized epilepsy: precedent for disorders with complex inheritance. *Human Molecular Genetics*, **2009**, 18, 3626-31	5.6	190
11	Genotype to phenotype-discovery and characterization of novel genomic disorders in a "genotype-first" era. *Genetics in Medicine*, **2009**, 11, 836-42	8.1	30
10	A method for rapid, targeted CNV genotyping identifies rare variants associated with neurocognitive disease. *Genome Research*, **2009**, 19, 1579-85	9.7	106
9	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. *Nature Genetics*, **2009**, 41, 160-2	36.3	454

8	Population analysis of large copy number variants and hotspots of human genetic disease. *American Journal of Human Genetics*, **2009**, 84, 148-61	11	454
7	Duplication hotspots, rare genomic disorders, and common disease. *Current Opinion in Genetics and Development*, **2009**, 19, 196-204	4.9	167
6	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. *Nature Genetics*, **2008**, 40, 322-8	36.3	463
5	Recurrent rearrangements of chromosome 1q21.1 and variable pediatric phenotypes. *New England Journal of Medicine*, **2008**, 359, 1685-99	59.2	587
4	Recurrent reciprocal genomic rearrangements of 17q12 are associated with renal disease, diabetes, and epilepsy. *American Journal of Human Genetics*, **2007**, 81, 1057-69	11	193
3	Comparative sequencing of a multicopy subtelomeric region containing olfactory receptor genes reveals multiple interactions between non-homologous chromosomes. *Human Molecular Genetics*, **2001**, 10, 2363-72	5.6	35
2	Targeted long-read sequencing resolves complex structural variants and identifies missing disease-causing variants		6
1	Pathogenic MAST3 variants in the STK domain are associated with epilepsy		1