Rikke S MÃ¸ller

List of Publications by Year
in descending order

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203 papers	15,119 citations	20817 60 h-index	24258 110 g-index
221 all docs	221 docs citations	221 times ranked	17438 citing authors

#	Article	IF	Citations
1	Assessing the landscape of STXBP1-related disorders in 534 individuals. Brain, 2022, 145, 1668-1683.	7.6	46
2	Adult phenotype of KCNQ2 encephalopathy. Journal of Medical Genetics, 2022, 59, 528-535.	3.2	14
3	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. Brain, 2022, 145, 2991-3009.	7.6	69
4	Gain-of-function variants in GABRD reveal a novel pathway for neurodevelopmental disorders and epilepsy. Brain, 2022, 145, 1299-1309.	7.6	34
5	De novo FZR1 loss-of-function variants cause developmental and epileptic encephalopathies. Brain, 2022, 145, 1684-1697.	7.6	5
6	L-Serine Treatment is Associated with Improvements in Behavior, EEG, and Seizure Frequency in Individuals with GRIN-Related Disorders Due to Null Variants. Neurotherapeutics, 2022, 19, 334-341.	4.4	21
7	Pyridoxine or pyridoxalâ€5â€phosphate treatment for seizures in glycosylphosphatidylinositol deficiency: A cohort study. Developmental Medicine and Child Neurology, 2022, 64, 789-798.	2.1	6
8	Association of ultraâ€rare coding variants with genetic generalized epilepsy: A caseâ€“control whole exome sequencing study. Epilepsia, 2022, 63, 723-735.	5.1	8
9	Development and Validation of a Prediction Model for Early Diagnosis of SCN1A -Related Epilepsies. Neurology, 2022, 98, .	1.1	24
10	OUP accepted manuscript. Brain, 2022, , .	7.6	0
11	Epilepsy Genetics and Precision Medicine in Adults: A New Landscape for Developmental and Epileptic Encephalopathies. Frontiers in Neurology, 2022, 13, 777115.	2.4	21
12	PIGNencephalopathy: Characterizing the epileptology. Epilepsia, 2022, 63, 974-991.	5.1	4
13	Structural mapping of GABRB3 variants reveals genotypeâ€“phenotype correlations. Genetics in Medicine, 2022, 24, 681-693.	2.4	10
14	Gain-of-function and loss-of-function GABRB3 variants lead to distinct clinical phenotypes in patients with developmental and epileptic encephalopathies. Nature Communications, 2022, 13, 1822.	12.8	32
15	Genetic paroxysmal neurological disorders featuring episodic ataxia and epilepsy. European Journal of Medical Genetics, 2022, 65, 104450.	1.3	10
16	SLC7A3: In Silico Prediction of a Potential New Cause of Childhood Epilepsy. Neuropediatrics, 2022, 53, 046-051.	0.6	0
17	Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis. Brain, 2022, 145, 2687-2703.	7.6	11
18	Biallelic variants in <scp>ZNF142</scp> lead to a syndromic neurodevelopmental disorder. Clinical Genetics, 2022, 102, 98-109.	2.0	6

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19	Natural History Study of STXBP1-Developmental and Epileptic Encephalopathy Into Adulthood. Neurology, 2022, 99, .	1.1	23
20	Impact of Genetic Testing on Therapeutic Decision-Making in Childhood-Onset Epilepsiesâ€"a Study in a Tertiary Epilepsy Center. Neurotherapeutics, 2022, 19, 1353-1367.	4.4	14
21	The de novo GABRA4 p.Thr300Ile variant found in a patient with earlyâ€onset intractable epilepsy and neurodevelopmental abnormalities displays gainâ€ofâ€function traits. Epilepsia, 2022, 63, 2439-2441.	5.1	6
22	KCNQ2 R144 variants cause neurodevelopmental disability with language impairment and autistic features without neonatal seizures through a gain-of-function mechanism. EBioMedicine, 2022, 81, 104130.	6.1	19
23	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	2.4	28
24	Basal Ganglia Dysmorphism in Patients With Aicardi Syndrome. Neurology, 2021, 96, e1319-e1333.	1.1	6
25	Deciphering the premature mortality in PIGA-CDG â€“ An untold story. Epilepsy Research, 2021, 170, 106530.	1.6	15
26	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	2.4	20
27	Characterization of the <scp>GABRB2</scp>â€Associated Neurodevelopmental Disorders. Annals of Neurology, 2021, 89, 573-586.	5.3	14
28	Refining Genotypes and Phenotypes in KCNA2-Related Neurological Disorders. International Journal of Molecular Sciences, 2021, 22, 2824.	4.1	20
29	Real-life survey of pitfalls and successes of precision medicine in genetic epilepsies. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 1044-1052.	1.9	30
30	Electroclinical features of MEF2C haploinsufficiency-related epilepsy: A multicenter European study. Seizure: the Journal of the British Epilepsy Association, 2021, 88, 60-72.	2.0	6
31	Deep-Phenotyping the Less Severe Spectrum of PIGT Deficiency and Linking the Gene to Myoclonic Atonic Seizures. Frontiers in Genetics, 2021, 12, 663643.	2.3	6
32	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. Brain, 2021, 144, 1435-1450.	7.6	35
33	Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations. Epilepsia, 2021, 62, 1518-1527.	5.1	5
34	KCNT1-related epilepsies and epileptic encephalopathies: phenotypic and mutational spectrum. Brain, 2021, 144, 3635-3650.	7.6	34
35	PRICKLE2 revisitedâ€"further evidence implicating PRICKLE2 in neurodevelopmental disorders. European Journal of Human Genetics, 2021, 29, 1235-1244.	2.8	5
36	Expansion of the CCDC22 associated Ritscher-Schinzel/3C syndrome and review of the literature: Should the minimal diagnostic criteria be revised?. European Journal of Medical Genetics, 2021, 64, 104246.	1.3	11

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37	Two de novo GluN2B mutations affect multiple NMDAR-functions and instigate severe pediatric encephalopathy. ELife, 2021, 10, .	6.0	14
38	Epilepsy Syndromes in the First Year of Life and Usefulness of Genetic Testing for Precision Therapy. Genes, 2021, 12, 1051.	2.4	36
39	<scp>ZMYND11</scp> variants are a novel cause of centrotemporal and generalised epilepsies with neurodevelopmental disorder. Clinical Genetics, 2021, 100, 412-429.	2.0	5
40	Clinical and molecular delineation of <scp>PUS3</scp>â€associated neurodevelopmental disorders. Clinical Genetics, 2021, 100, 628-633.	2.0	23
41	Integrative approach to interpret DYRK1A variants, leading to a frequent neurodevelopmental disorder. Genetics in Medicine, 2021, 23, 2150-2159.	2.4	21
42	RHOBTB2 Mutations Expand the Phenotypic Spectrum of Alternating Hemiplegia of Childhood. Neurology, 2021, 96, e1539-e1550.	1.1	15
43	The Angelman Syndrome Online Registry â€“ A multilingual approach to support global research. European Journal of Medical Genetics, 2021, 64, 104349.	1.3	1
44	Using common genetic variants to find drugs for common epilepsies. Brain Communications, 2021, 3, fcab287.	3.3	9
45	PURA- Related Developmental and Epileptic Encephalopathy. Neurology: Genetics, 2021, 7, e613.	1.9	15
46	STXBP1 Syndrome Is Characterized by Inhibition-Dominated Dynamics of Resting-State EEG. Frontiers in Physiology, 2021, 12, 775172.	2.8	14
47	Differential excitatory vs inhibitory SCN expression at single cell level regulates brain sodium channel function in neurodevelopmental disorders. European Journal of Paediatric Neurology, 2020, 24, 129-133.	1.6	18
48	Recent advances in treatment of epilepsy-related sodium channelopathies. European Journal of Paediatric Neurology, 2020, 24, 123-128.	1.6	40
49	Neonatal developmental and epileptic encephalopathy due to autosomal recessive variants in SLC13A5 gene. Epilepsia, 2020, 61, 2474-2485.	5.1	31
50	The impact of severe pediatric epilepsy on experienced stress and psychopathology in parents. Epilepsy and Behavior, 2020, 113, 107538.	1.7	23
51	Current knowledge of SLC6A1-related neurodevelopmental disorders. Brain Communications, 2020, 2, fcaa170.	3.3	44
52	Expanding the clinical and EEG spectrum of CNKSR2-related encephalopathy with status epilepticus during slow sleep (ESES). Clinical Neurophysiology, 2020, 131, 1030-1039.	1.5	11
53	The impact of low-risk genetic variants in self-limited epilepsy with centrotemporal spikes aka Rolandic epilepsy. EBioMedicine, 2020, 58, 102896.	6.1	2
54	Genetic testing in adult epilepsy patients: A call to action for clinicians. Epilepsia, 2020, 61, 2055-2056.	5.1	2

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55	Predicting functional effects of missense variants in voltage-gated sodium and calcium channels. Science Translational Medicine, 2020, 12, .	12.4	84
56	The Phenotypic Spectrum of PRRT2-Associated Paroxysmal Neurologic Disorders in Childhood. Biomedicines, 2020, 8, 456.	3.2	23
57	Utility of genetic testing for therapeutic decisionâ€making in adults with epilepsy. Epilepsia, 2020, 61, 1234-1239.	5.1	60
58	Pharmacoresponse in genetic generalized epilepsy: a genome-wide association study. Pharmacogenomics, 2020, 21, 325-335.	1.3	21
59	Phenotypic and genetic spectrum of epilepsy with myoclonic atonic seizures. Epilepsia, 2020, 61, 995-1007.	5.1	30
60	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	5.1	32
61	Mowat-Wilson syndrome: growth charts. Orphanet Journal of Rare Diseases, 2020, 15, 151.	2.7	12
62	Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. Genome Medicine, 2020, 12, 28.	8.2	42
63	Testing association of rare genetic variants with resistance to three common antiseizure medications. Epilepsia, 2020, 61, 657-666.	5.1	22
64	A catalogue of new incidence estimates of monogenic neurodevelopmental disorders caused by de novo variants. Brain, 2020, 143, 1099-1105.	7.6	64
65	Biological concepts in human sodium channel epilepsies and their relevance in clinical practice. Epilepsia, 2020, 61, 387-399.	5.1	65
66	Novel congenital disorder of O-linked glycosylation caused by GALNT2 loss of function. Brain, 2020, 143, 1114-1126.	7.6	46
67	Gain-of-function GABRB3 variants identified in vigabatrin-hypersensitive epileptic encephalopathies. Brain Communications, 2020, 2, fcaa162.	3.3	21
68	Genotype-phenotype correlations in patients with de novo KCNQ2 pathogenic variants. Neurology: Genetics, 2020, 6, e528.	1.9	24
69	Reader response: SYNGAP1 encephalopathy: A distinctive generalized developmental and epileptic encephalopathy. Neurology, 2020, 94, 368.2-369.	1.1	2
70	The landscape of epilepsy-related GATOR1 variants. Genetics in Medicine, 2019, 21, 398-408.	2.4	137
71	Genetic heterogeneity in infantile spasms. Epilepsy Research, 2019, 156, 106181.	1.6	38
72	Idiopathic encephalopathy related to status epilepticus during slow sleep (ESES) as a â€œpureâ€•model of epileptic encephalopathy. An electroclinical, genetic, and follow-up study. Epilepsy and Behavior, 2019, 97, 244-252.	1.7	16

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73	Biallelic inherited SCN8A variants, a rare cause of SCN8A â€related developmental and epileptic encephalopathy. Epilepsia, 2019, 60, 2277-2285.	5.1	18
74	The first step towards personalized risk prediction for common epilepsies. Brain, 2019, 142, 3316-3318.	7.6	2
75	From next-generation sequencing to targeted treatment of non-acquired epilepsies. Expert Review of Molecular Diagnostics, 2019, 19, 217-228.	3.1	38
76	Chewing induced reflex seizures (â€œeating epilepsyâ€) and eye closure sensitivity as a common feature in pediatric patients with SYNGAP1 mutations: Review of literature and report of 8 cases. Seizure: the Journal of the British Epilepsy Association, 2019, 65, 131-137.	2.0	30
77	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.	6.2	78
78	Parental mosaicism in epilepsies due to alleged de novo variants. Epilepsia, 2019, 60, e63-e66.	5.1	29
79	Treatment Responsiveness in KCNT1-Related Epilepsy. Neurotherapeutics, 2019, 16, 848-857.	4.4	60
80	First report of the neuropathological findings in a patient with leukodystrophy and compound heterozygous variants in the PIGT gene. Neuropathology and Applied Neurobiology, 2019, 45, 732-735.	3.2	6
81	A novel in-frame mutation in CLN3 leads to Juvenile neuronal ceroid lipofuscinosis in a large Pakistani family. International Journal of Neuroscience, 2019, 129, 890-895.	1.6	0
82	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	5.1	61
83	The spectrum of intermediate <scp>SCN</scp>8Aâ€related epilepsy. Epilepsia, 2019, 60, 830-844.	5.1	70
84	PIGT-CDG, a disorder of the glycosylphosphatidylinositol anchor: description of 13 novel patients and expansion of the clinical characteristics. Genetics in Medicine, 2019, 21, 2216-2223.	2.4	21
85	No evidence for a BRD 2 promoter hypermethylation inÂblood leukocytes of Europeans with juvenile myoclonicÂepilepsy. Epilepsia, 2019, 60, e31-e36.	5.1	4
86	Clinical spectrum of STX1B -related epileptic disorders. Neurology, 2019, 92, e1238-e1249.	1.1	43
87	Mild malformations of cortical development in sleepâ€related hypermotor epilepsy due to KCNT1 mutations. Annals of Clinical and Translational Neurology, 2019, 6, 386-391.	3.7	25
88	Defining and expanding the phenotype of QARS-associated developmental epileptic encephalopathy. Neurology: Genetics, 2019, 5, e373.	1.9	5
89	Estimating the effect size of the 15Q11.2 BP1â€“BP2 deletion and its contribution to neurodevelopmental symptoms: recommendations for practice. Journal of Medical Genetics, 2019, 56, 701-710.	3.2	43
90	Spectrum of GABAA receptor variants in epilepsy. Current Opinion in Neurology, 2019, 32, 183-190.	3.6	59

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91	Phenotypic and genetic spectrum of <scp>SCN</scp>8Aâ€related disorders, treatment options, and outcomes. Epilepsia, 2019, 60, S77-S85.	5.1	58
92	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. Genetics in Medicine, 2019, 21, 837-849.	2.4	47
93	GRIN2A-related disorders: genotype and functional consequence predict phenotype. Brain, 2019, 142, 80-92.	7.6	143
94	Neuronal mechanisms of mutations in SCN8A causing epilepsy or intellectual disability. Brain, 2019, 142, 376-390.	7.6	92
95	Clinicianâ€™s guide to genes associated with Rettâ€like phenotypesâ€”Investigation of a Danish cohort and review of the literature. Clinical Genetics, 2019, 95, 221-230.	2.0	26
96	Update on the genetics of the epilepsyâ€aphasia spectrum and role of GRIN2A mutations. Epileptic Disorders, 2019, 21, 41-47.	1.3	20
97	Early mortality in SCN8A -related epilepsies. Epilepsy Research, 2018, 143, 79-81.	1.6	48
98	Progress in Understanding and Treating SCN2A-Mediated Disorders. Trends in Neurosciences, 2018, 41, 442-456.	8.6	210
99	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. Annals of Neurology, 2018, 83, 926-934.	5.3	20
100	Defining the phenotypic spectrum of SLC6A1 mutations. Epilepsia, 2018, 59, 389-402.	5.1	99
101	Phenotype and genotype of 87 patients with Mowatâ€“Wilson syndrome and recommendations for care. Genetics in Medicine, 2018, 20, 965-975.	2.4	67
102	Neurologic phenotypes associated with COL4A1 / 2 mutations. Neurology, 2018, 91, e2078-e2088.	1.1	97
103	Aberrant Inclusion of a Poison Exon Causes Dravet Syndrome and Related SCN1A-Associated Genetic Epilepsies. American Journal of Human Genetics, 2018, 103, 1022-1029.	6.2	76
104	SLC35A2-related congenital disorder of glycosylation: Defining the phenotype. European Journal of Paediatric Neurology, 2018, 22, 1095-1102.	1.6	27
105	The phenotype of SCN8A developmental and epileptic encephalopathy. Neurology, 2018, 91, e1112-e1124.	1.1	114
106	De novo variants in neurodevelopmental disorders with epilepsy. Nature Genetics, 2018, 50, 1048-1053.	21.4	230
107	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	12.6	1,085
108	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.	10.2	67

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109	Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. Genome Medicine, 2018, 10, 3.	8.2	67
110	Incorporating epilepsy genetics into clinical practice: a 360Â°evaluation. Npj Genomic Medicine, 2018, 3, 13.	3.8	46
111	Epilepsy in patients with GRIN2A alterations: Genetics, neurodevelopment, epileptic phenotype and response to anticonvulsive drugs. European Journal of Paediatric Neurology, 2017, 21, 530-541.	1.6	37
112	Application of rare variant transmission disequilibrium tests to epileptic encephalopathy trio sequence data. European Journal of Human Genetics, 2017, 25, 894-899.	2.8	7
113	Carbamazepineâ€•and oxcarbazepineâ€•induced hyponatremia in people with epilepsy. Epilepsia, 2017, 58, 1227-1233.	5.1	54
114	Myoclonus epilepsy and ataxia due to <scp>KCNC</scp>1 mutation: Analysis of 20 cases and <scp>K</scp>⁺ channel properties. Annals of Neurology, 2017, 81, 677-689.	5.3	69
115	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. American Journal of Human Genetics, 2017, 100, 676-688.	6.2	54
116	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. Brain, 2017, 140, 1316-1336.	7.6	426
117	GRIN2Bencephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. Journal of Medical Genetics, 2017, 54, 460-470.	3.2	190
118	Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU. Human Genetics, 2017, 136, 463-479.	3.8	66
119	Mutations in GABRB3. Neurology, 2017, 88, 483-492.	1.1	87
120	Delineating SPTAN1 associated phenotypes: from isolated epilepsy to encephalopathy with progressive brain atrophy. Brain, 2017, 140, 2322-2336.	7.6	82
121	Clinical spectrum and genotypeâ€“phenotype associations of KCNA2-related encephalopathies. Brain, 2017, 140, 2337-2354.	7.6	117
122	Neurodevelopmental Disorders Caused by De Novo Variants in KCNB1 Genotypes and Phenotypes. JAMA Neurology, 2017, 74, 1228.	9.0	79
123	Alternating hemiplegia of childhood and a pathogenic variant of ATP1A3: a case report and pathophysiological considerations. Epileptic Disorders, 2017, 19, 226-230.	1.3	5
124	Filadelfia, Danish Epilepsy Center, Dianalund, Denmark. Epilepsy and Behavior, 2017, 76, S4-S8.	1.7	1
125	DNM1 encephalopathy. Neurology, 2017, 89, 385-394.	1.1	87
126	Neuroimaging findings in Mowatâ€“Wilson syndrome: a study of 54 patients. Genetics in Medicine, 2017, 19, 691-700.	2.4	45

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127	Benign infantile seizures and paroxysmal dyskinesia caused by an SCN8A mutation. Annals of Neurology, 2016, 79, 428-436.	5.3	159
128	Delineating the GRIN1 phenotypic spectrum. Neurology, 2016, 86, 2171-2178.	1.1	157
129	De Novo Mutations in SLC1A2 and CACNA1A Are Important Causes of Epileptic Encephalopathies. American Journal of Human Genetics, 2016, 99, 287-298.	6.2	247
130	Pitfalls in genetic testing: the story of missed SCN1A mutations. Molecular Genetics & Genomic Medicine, 2016, 4, 457-464.	1.2	67
131	Phenotypic spectrum of GABRA1. Neurology, 2016, 87, 1140-1151.	1.1	113
132	Targeted sequencing of 351 candidate genes for epileptic encephalopathy in a large cohort of patients. Molecular Genetics & Genomic Medicine, 2016, 4, 568-580.	1.2	83
133	Exome Sequencing Fails to Identify the Genetic Cause of Aicardi Syndrome. Molecular Syndromology, 2016, 7, 234-238.	0.8	16
134	Gene Panel Testing in Epileptic Encephalopathies and Familial Epilepsies. Molecular Syndromology, 2016, 7, 210-219.	0.8	103
135	Loss of SYNJ1 dual phosphatase activity leads to early onset refractory seizures and progressive neurological decline. Brain, 2016, 139, 2420-2430.	7.6	70
136	Germline and somatic mutations in the MTOR gene in focal cortical dysplasia and epilepsy. Neurology: Genetics, 2016, 2, e118.	1.9	125
137	De novo mutations ofKIAA2022in females cause intellectual disability and intractable epilepsy. Journal of Medical Genetics, 2016, 53, 850-858.	3.2	47
138	Reply. Annals of Neurology, 2016, 80, 168-169.	5.3	0
139	Letter to the editor: confirming neonatal seizure and late onset ataxia in SCN2A Ala263Val. Journal of Neurology, 2016, 263, 1459-1460.	3.6	14
140	Loss of function of the retinoid-related nuclear receptor (RORB) gene and epilepsy. European Journal of Human Genetics, 2016, 24, 1761-1770.	2.8	36
141	Precision Medicine: SCN8A Encephalopathy Treated with Sodium Channel Blockers. Neurotherapeutics, 2016, 13, 190-191.	4.4	38
142	STXBP1 encephalopathy. Neurology, 2016, 86, 954-962.	1.1	264
143	MECP2 Duplication Syndrome: Evidence of Enhanced Oxidative Stress. A Comparison with Rett Syndrome. PLoS ONE, 2016, 11, e0150101.	2.5	22
144	Evaluation of Presumably Disease Causing SCN1A Variants in a Cohort of Common Epilepsy Syndromes. PLoS ONE, 2016, 11, e0150426.	2.5	22

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145	Clinical Phenotype of De Novo GNAO1 Mutation. Child Neurology Open, 2015, 2, 2329048X1558371.	1.1	25
146	The role of <scp>SLC</scp>2A1 mutations in myoclonic astatic epilepsy and absence epilepsy, and the estimated frequency of <scp>GLUT</scp>1 deficiency syndrome. Epilepsia, 2015, 56, e203-8.	5.1	71
147	Dysregulation of FOXG1 by ring chromosome 14. Molecular Cytogenetics, 2015, 8, 24.	0.9	8
148	The contribution of next generation sequencing to epilepsy genetics. Expert Review of Molecular Diagnostics, 2015, 15, 1531-1538.	3.1	68
149	The phenotypic spectrum of SCN8A encephalopathy. Neurology, 2015, 84, 480-489.	1.1	246
150	CHD2variants are a risk factor for photosensitivity in epilepsy. Brain, 2015, 138, 1198-1208.	7.6	112
151	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. Human Molecular Genetics, 2015, 24, 5250-5259.	2.9	93
152	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. American Journal of Human Genetics, 2015, 96, 808-815.	6.2	173
153	Burden Analysis of Rare Microdeletions Suggests a Strong Impact of Neurodevelopmental Genes in Genetic Generalised Epilepsies. PLoS Genetics, 2015, 11, e1005226.	3.5	91
154	De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy. Nature Genetics, 2015, 47, 393-399.	21.4	224
155	Recessive loss-of-function mutations in AP4S1 cause mild fever-sensitive seizures, developmental delay and spastic paraplegia through loss of AP-4 complex assembly. Human Molecular Genetics, 2015, 24, 2218-2227.	2.9	53
156	Recessive mutations inSLC13A5result in a loss of citrate transport and cause neonatal epilepsy, developmental delay and teeth hypoplasia. Brain, 2015, 138, 3238-3250.	7.6	96
157	The incidence of <scp>SCN</scp>1Aâ€related Dravet syndrome in <scp>D</scp>enmark is 1:22,000: A populationâ€based study from 2004 to 2009. Epilepsia, 2015, 56, e36-9.	5.1	103
158	Mutations in <scp>KCNT</scp>1 cause a spectrum of focal epilepsies. Epilepsia, 2015, 56, e114-20.	5.1	117
159	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. Nature Genetics, 2015, 47, 39-46.	21.4	245
160	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. Human Molecular Genetics, 2014, 23, 6069-6080.	2.9	61
161	Reduced ceramide synthase 2 activity causes progressive myoclonic epilepsy. Annals of Clinical and Translational Neurology, 2014, 1, 88-98.	3.7	50
162	X-linked congenital ptosis and associated intellectual disability, short stature, microcephaly, cleft palate, digital and genital abnormalities define novel Xq25q26 duplication syndrome. Human Genetics, 2014, 133, 625-638.	3.8	17

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163	De novo mutations in HCN1 cause early infantile epileptic encephalopathy. Nature Genetics, 2014, 46, 640-645.	21.4	192
164	Aberrant expression of miRâ€218 and miRâ€204 in human mesial temporal lobe epilepsy and hippocampal sclerosisâ€"Convergence on axonal guidance. Epilepsia, 2014, 55, 2017-2027.	5.1	71
165	Mutations in STX1B, encoding a presynaptic protein, cause fever-associated epilepsy syndromes. Nature Genetics, 2014, 46, 1327-1332.	21.4	178
166	Atypical Vitamin B₆Deficiency. Journal of Child Neurology, 2014, 29, 704-707.	1.4	16
167	Sequence analysis of 17 NRXN1 deletions. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2014, 165, 52-61.	1.7	11
168	De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	6.2	388
169	GABRA1 and STXBP1 : Novel genetic causes of Dravet syndrome. Neurology, 2014, 82, 1245-1253.	1.1	229
170	Genetic determinants of common epilepsies: a meta-analysis of genome-wide association studies. Lancet Neurology, The, 2014, 13, 893-903.	10.2	264
171	Structural genomic variation in childhood epilepsies with complex phenotypes. European Journal of Human Genetics, 2014, 22, 896-901.	2.8	28
172	De Novo Loss-of-Function Mutations in CHD2 Cause a Fever-Sensitive Myoclonic Epileptic Encephalopathy Sharing Features with Dravet Syndrome. American Journal of Human Genetics, 2013, 93, 967-975.	6.2	188
173	Mutations in GRIN2A cause idiopathic focal epilepsy with rolandic spikes. Nature Genetics, 2013, 45, 1067-1072.	21.4	391
174	Mutations inSYNGAP1Cause Intellectual Disability, Autism, and a Specific Form of Epilepsy by Inducing Haploinsufficiency. Human Mutation, 2013, 34, 385-394.	2.5	196
175	The role of SLC2A1 in early onset and childhood absence epilepsies. Epilepsy Research, 2013, 105, 229-233.	1.6	13
176	Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. Nature Genetics, 2013, 45, 825-830.	21.4	589
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