

# Rikke S Mller

## List of Publications by Year in Descending Order

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**Version:** 2024-04-27

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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.

196  
papers

10,052  
citations

51  
h-index

96  
g-index

221  
ext. papers

12,931  
ext. citations

7.7  
avg, IF

5.62  
L-index

#	Paper	IF	Citations
196	Pyridoxine or pyridoxal-5-phosphate treatment for seizures in glycosylphosphatidylinositol deficiency: A cohort study.. <i>Developmental Medicine and Child Neurology</i> , <b>2022</b> ,	3.3	1
195	Development and Validation of a Prediction Model for Early Diagnosis of -Related Epilepsies.. <i>Neurology</i> , <b>2022</b> ,	6.5	2
194	L-Serine Treatment is Associated with Improvements in Behavior, EEG, and Seizure Frequency in Individuals with GRIN-Related Disorders Due to Null Variants.. <i>Neurotherapeutics</i> , <b>2022</b> , 1	6.4	3
193	Epilepsy Genetics and Precision Medicine in Adults: A New Landscape for Developmental and Epileptic Encephalopathies.. <i>Frontiers in Neurology</i> , <b>2022</b> , 13, 777115	4.1	1
192	Gain-of-function and loss-of-function GABRB3 variants lead to distinct clinical phenotypes in patients with developmental and epileptic encephalopathies.. <i>Nature Communications</i> , <b>2022</b> , 13, 1822	17.4	2
191	Genetic paroxysmal neurological disorders featuring episodic ataxia and epilepsy.. <i>European Journal of Medical Genetics</i> , <b>2022</b> , 65, 104450	2.6	0
190	Using common genetic variants to find drugs for common epilepsies.. <i>Brain Communications</i> , <b>2021</b> , 3, fcb287	4.5	0
189	Related Developmental and Epileptic Encephalopathy: Phenotypic and Genotypic Spectrum. <i>Neurology: Genetics</i> , <b>2021</b> , 7, e613	3.8	1
188	Gain-of-function variants in GABRD reveal a novel pathway for neurodevelopmental disorders and epilepsy. <i>Brain</i> , <b>2021</b> ,	11.2	6
187	The Angelman Syndrome Online Registry - A multilingual approach to support global research. <i>European Journal of Medical Genetics</i> , <b>2021</b> , 64, 104349	2.6	
186	De novo FZR1 loss-of-function variants cause developmental and epileptic encephalopathies. <i>Brain</i> , <b>2021</b> ,	11.2	2
185	Refining Genotypes and Phenotypes in -Related Neurological Disorders. <i>International Journal of Molecular Sciences</i> , <b>2021</b> , 22,	6.3	6
184	Adult phenotype of encephalopathy. <i>Journal of Medical Genetics</i> , <b>2021</b> ,	5.8	6
183	Real-life survey of pitfalls and successes of precision medicine in genetic epilepsies. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2021</b> , 92, 1044-1052	5.5	8
182	Electroclinical features of MEF2C haploinsufficiency-related epilepsy: A multicenter European study. <i>Seizure: the Journal of the British Epilepsy Association</i> , <b>2021</b> , 88, 60-72	3.2	3
181	Deep-Phenotyping the Less Severe Spectrum of Deficiency and Linking the Gene to Myoclonic Atonic Seizures. <i>Frontiers in Genetics</i> , <b>2021</b> , 12, 663643	4.5	2
180	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. <i>Brain</i> , <b>2021</b> , 144, 1435-1450	11.2	4

179	Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations. <i>Epilepsia</i> , <b>2021</b> , 62, 1518-1527	6.4	1
178	KCNT1-related epilepsies and epileptic encephalopathies: phenotypic and mutational spectrum. <i>Brain</i> , <b>2021</b> ,	11.2	6
177	PRICKLE2 revisited-further evidence implicating PRICKLE2 in neurodevelopmental disorders. <i>European Journal of Human Genetics</i> , <b>2021</b> , 29, 1235-1244	5.3	2
176	Expansion of the CCDC22 associated Ritscher-Schinzel/3C syndrome and review of the literature: Should the minimal diagnostic criteria be revised?. <i>European Journal of Medical Genetics</i> , <b>2021</b> , 64, 104246 <sup>2,6</sup>	2.6	2
175	Two GluN2B mutations affect multiple NMDAR-functions and instigate severe pediatric encephalopathy. <i>ELife</i> , <b>2021</b> , 10,	8.9	4
174	Epilepsy Syndromes in the First Year of Life and Usefulness of Genetic Testing for Precision Therapy. <i>Genes</i> , <b>2021</b> , 12,	4.2	12
173	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 363-373	8.1	4
172	Basal Ganglia Dysmorphism in Patients With Aicardi Syndrome. <i>Neurology</i> , <b>2021</b> , 96, e1319-e1333	6.5	3
171	Deciphering the premature mortality in PIGA-CDG - An untold story. <i>Epilepsy Research</i> , <b>2021</b> , 170, 106530	3	5
170	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 653-660	8.1	5
169	Characterization of the GABRB2-Associated Neurodevelopmental Disorders. <i>Annals of Neurology</i> , <b>2021</b> , 89, 573-586	9.4	2
168	Assessing the landscape of STXBP1-related disorders in 534 individuals.. <i>Brain</i> , <b>2021</b> ,	11.2	4
167	ZMYND11 variants are a novel cause of centrotemporal and generalised epilepsies with neurodevelopmental disorder. <i>Clinical Genetics</i> , <b>2021</b> , 100, 412-429	4	0
166	Clinical and molecular delineation of PUS3-associated neurodevelopmental disorders. <i>Clinical Genetics</i> , <b>2021</b> , 100, 628-633	4	1
165	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. <i>Brain</i> , <b>2021</b> ,	11.2	8
164	Integrative approach to interpret DYRK1A variants, leading to a frequent neurodevelopmental disorder. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 2150-2159	8.1	4
163	Mutations Expand the Phenotypic Spectrum of Alternating Hemiplegia of Childhood. <i>Neurology</i> , <b>2021</b> , 96, e1539-e1550	6.5	3
162	Structural mapping of GABRB3 variants reveals genotype-phenotype correlations.. <i>Genetics in Medicine</i> , <b>2021</b> ,	8.1	3

161	Syndrome Is Characterized by Inhibition-Dominated Dynamics of Resting-State EEG.. <i>Frontiers in Physiology</i> , <b>2021</b> , 12, 775172	4.6	1
160	The Phenotypic Spectrum of PRRT2-Associated Paroxysmal Neurologic Disorders in Childhood. <i>Biomedicines</i> , <b>2020</b> , 8,	4.8	12
159	Utility of genetic testing for therapeutic decision-making in adults with epilepsy. <i>Epilepsia</i> , <b>2020</b> , 61, 1234-1239	6.4	32
158	Pharmacoresponse in genetic generalized epilepsy: a genome-wide association study. <i>Pharmacogenomics</i> , <b>2020</b> , 21, 325-335	2.6	8
157	Phenotypic and genetic spectrum of epilepsy with myoclonic atonic seizures. <i>Epilepsia</i> , <b>2020</b> , 61, 995-1007	6.4	18
156	Lessons learned from 40 novel PIGA patients and a review of the literature. <i>Epilepsia</i> , <b>2020</b> , 61, 1142-1155	6.4	15
155	Mowat-Wilson syndrome: growth charts. <i>Orphanet Journal of Rare Diseases</i> , <b>2020</b> , 15, 151	4.2	3
154	Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. <i>Genome Medicine</i> , <b>2020</b> , 12, 28	14.4	13
153	Testing association of rare genetic variants with resistance to three common antiseizure medications. <i>Epilepsia</i> , <b>2020</b> , 61, 657-666	6.4	9
152	A catalogue of new incidence estimates of monogenic neurodevelopmental disorders caused by de novo variants. <i>Brain</i> , <b>2020</b> , 143, 1099-1105	11.2	18
151	Biological concepts in human sodium channel epilepsies and their relevance in clinical practice. <i>Epilepsia</i> , <b>2020</b> , 61, 387-399	6.4	35
150	Genotype-phenotype correlations in patients with de novo pathogenic variants. <i>Neurology: Genetics</i> , <b>2020</b> , 6, e528	3.8	9
149	Reader response: encephalopathy: A distinctive generalized developmental and epileptic encephalopathy. <i>Neurology</i> , <b>2020</b> , 94, 368-369	6.5	1
148	Gain-of-function variants identified in vigabatrin-hypersensitive epileptic encephalopathies. <i>Brain Communications</i> , <b>2020</b> , 2, fcaa162	4.5	6
147	Differential excitatory vs inhibitory SCN expression at single cell level regulates brain sodium channel function in neurodevelopmental disorders. <i>European Journal of Paediatric Neurology</i> , <b>2020</b> , 24, 129-133	3.8	6
146	Recent advances in treatment of epilepsy-related sodium channelopathies. <i>European Journal of Paediatric Neurology</i> , <b>2020</b> , 24, 123-128	3.8	18
145	Neonatal developmental and epileptic encephalopathy due to autosomal recessive variants in SLC13A5 gene. <i>Epilepsia</i> , <b>2020</b> , 61, 2474-2485	6.4	8
144	The impact of severe pediatric epilepsy on experienced stress and psychopathology in parents. <i>Epilepsy and Behavior</i> , <b>2020</b> , 113, 107538	3.2	12

143	Current knowledge of SLC6A1-related neurodevelopmental disorders. <i>Brain Communications</i> , <b>2020</b> , 2, fcaa170	4.5	11
142	Expanding the clinical and EEG spectrum of CNKSR2-related encephalopathy with status epilepticus during slow sleep (ESES). <i>Clinical Neurophysiology</i> , <b>2020</b> , 131, 1030-1039	4.3	4
141	The impact of low-risk genetic variants in self-limited epilepsy with centrotemporal spikes aka Rolandic epilepsy. <i>EBioMedicine</i> , <b>2020</b> , 58, 102896	8.8	0
140	Genetic testing in adult epilepsy patients: A call to action for clinicians. <i>Epilepsia</i> , <b>2020</b> , 61, 2055-2056	6.4	2
139	Predicting functional effects of missense variants in voltage-gated sodium and calcium channels. <i>Science Translational Medicine</i> , <b>2020</b> , 12,	17.5	27
138	Novel congenital disorder of O-linked glycosylation caused by GALNT2 loss of function. <i>Brain</i> , <b>2020</b> , 143, 1114-1126	11.2	28
137	From next-generation sequencing to targeted treatment of non-acquired epilepsies. <i>Expert Review of Molecular Diagnostics</i> , <b>2019</b> , 19, 217-228	3.8	29
136	Chewing induced reflex seizures ("eating epilepsy") and eye closure sensitivity as a common feature in pediatric patients with SYNGAP1 mutations: Review of literature and report of 8 cases. <i>Seizure: the Journal of the British Epilepsy Association</i> , <b>2019</b> , 65, 131-137	3.2	23
135	A Recurrent Missense Variant in AP2M1 Impairs Clathrin-Mediated Endocytosis and Causes Developmental and Epileptic Encephalopathy. <i>American Journal of Human Genetics</i> , <b>2019</b> , 104, 1060-1072 <sup>11</sup>	11	39
134	Parental mosaicism in epilepsies due to alleged de novo variants. <i>Epilepsia</i> , <b>2019</b> , 60, e63-e66	6.4	16
133	Treatment Responsiveness in KCNT1-Related Epilepsy. <i>Neurotherapeutics</i> , <b>2019</b> , 16, 848-857	6.4	30
132	First report of the neuropathological findings in a patient with leukodystrophy and compound heterozygous variants in the PIGT gene. <i>Neuropathology and Applied Neurobiology</i> , <b>2019</b> , 45, 732-735	5.2	6
131	A novel in-frame mutation in leads to Juvenile neuronal ceroid lipofuscinosis in a large Pakistani family. <i>International Journal of Neuroscience</i> , <b>2019</b> , 129, 890-895	2	
130	Diagnostic implications of genetic copy number variation in epilepsy plus. <i>Epilepsia</i> , <b>2019</b> , 60, 689-706	6.4	37
129	The spectrum of intermediate SCN8A-related epilepsy. <i>Epilepsia</i> , <b>2019</b> , 60, 830-844	6.4	38
128	PIGT-CDG, a disorder of the glycosylphosphatidylinositol anchor: description of 13 novel patients and expansion of the clinical characteristics. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 2216-2223	8.1	18
127	No evidence for a BRD2 promoter hypermethylation in blood leukocytes of Europeans with juvenile myoclonic epilepsy. <i>Epilepsia</i> , <b>2019</b> , 60, e31-e36	6.4	4
126	Clinical spectrum of -related epileptic disorders. <i>Neurology</i> , <b>2019</b> , 92, e1238-e1249	6.5	25

125	Mild malformations of cortical development in sleep-related hypermotor epilepsy due to mutations. <i>Annals of Clinical and Translational Neurology</i> , <b>2019</b> , 6, 386-391	5.3	17
124	The landscape of epilepsy-related GATOR1 variants. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 398-408	8.1	75
123	Genetic heterogeneity in infantile spasms. <i>Epilepsy Research</i> , <b>2019</b> , 156, 106181	3	20
122	Idiopathic encephalopathy related to status epilepticus during slow sleep (ESES) as a "pure" model of epileptic encephalopathy. An electroclinical, genetic, and follow-up study. <i>Epilepsy and Behavior</i> , <b>2019</b> , 97, 244-252	3.2	9
121	Biallelic inherited SCN8A variants, a rare cause of SCN8A-related developmental and epileptic encephalopathy. <i>Epilepsia</i> , <b>2019</b> , 60, 2277-2285	6.4	9
120	The first step towards personalized risk prediction for common epilepsies. <i>Brain</i> , <b>2019</b> , 142, 3316-3318	11.2	0
119	Update on the genetics of the epilepsy-aphasia spectrum and role of GRIN2A mutations <b>2019</b> , 21, 41-47		8
118	Defining and expanding the phenotype of -associated developmental epileptic encephalopathy. <i>Neurology: Genetics</i> , <b>2019</b> , 5, e373	3.8	3
117	Estimating the effect size of the 15Q11.2 BP1-BP2 deletion and its contribution to neurodevelopmental symptoms: recommendations for practice. <i>Journal of Medical Genetics</i> , <b>2019</b> , 56, 701-710	5.8	22
116	Spectrum of GABAA receptor variants in epilepsy. <i>Current Opinion in Neurology</i> , <b>2019</b> , 32, 183-190	7.1	24
115	Phenotypic and genetic spectrum of SCN8A-related disorders, treatment options, and outcomes. <i>Epilepsia</i> , <b>2019</b> , 60 Suppl 3, S77-S85	6.4	32
114	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 837-849	8.1	32
113	GRIN2A-related disorders: genotype and functional consequence predict phenotype. <i>Brain</i> , <b>2019</b> , 142, 80-92	11.2	66
112	Neuronal mechanisms of mutations in SCN8A causing epilepsy or intellectual disability. <i>Brain</i> , <b>2019</b> , 142, 376-390	11.2	53
111	ClinicianQ guide to genes associated with Rett-like phenotypes-Investigation of a Danish cohort and review of the literature. <i>Clinical Genetics</i> , <b>2019</b> , 95, 221-230	4	14
110	Early mortality in SCN8A-related epilepsies. <i>Epilepsy Research</i> , <b>2018</b> , 143, 79-81	3	38
109	Progress in Understanding and Treating SCN2A-Mediated Disorders. <i>Trends in Neurosciences</i> , <b>2018</b> , 41, 442-456	13.3	128
108	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. <i>Annals of Neurology</i> , <b>2018</b> , 83, 926-934	9.4	11

107	Defining the phenotypic spectrum of SLC6A1 mutations. <i>Epilepsia</i> , <b>2018</b> , 59, 389-402	6.4	54
106	Phenotype and genotype of 87 patients with Mowat-Wilson syndrome and recommendations for care. <i>Genetics in Medicine</i> , <b>2018</b> , 20, 965-975	8.1	37
105	Analysis of shared heritability in common disorders of the brain. <i>Science</i> , <b>2018</b> , 360,	33.3	666
104	Rare coding variants in genes encoding GABA receptors in genetic generalised epilepsies: an exome-based case-control study. <i>Lancet Neurology</i> , <b>2018</b> , 17, 699-708	24.1	44
103	Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. <i>Genome Medicine</i> , <b>2018</b> , 10, 3	14.4	53
102	Incorporating epilepsy genetics into clinical practice: a 360° evaluation. <i>Npj Genomic Medicine</i> , <b>2018</b> , 3, 13	6.2	32
101	Neurologic phenotypes associated with / mutations: Expanding the spectrum of disease. <i>Neurology</i> , <b>2018</b> , 91, e2078-e2088	6.5	55
100	Aberrant Inclusion of a Poison Exon Causes Dravet Syndrome and Related SCN1A-Associated Genetic Epilepsies. <i>American Journal of Human Genetics</i> , <b>2018</b> , 103, 1022-1029	11	40
99	SLC35A2-related congenital disorder of glycosylation: Defining the phenotype. <i>European Journal of Paediatric Neurology</i> , <b>2018</b> , 22, 1095-1102	3.8	15
98	The phenotype of developmental and epileptic encephalopathy. <i>Neurology</i> , <b>2018</b> , 91, e1112-e1124	6.5	80
97	De novo variants in neurodevelopmental disorders with epilepsy. <i>Nature Genetics</i> , <b>2018</b> , 50, 1048-1053	36.3	139
96	Epilepsy in patients with GRIN2A alterations: Genetics, neurodevelopment, epileptic phenotype and response to anticonvulsive drugs. <i>European Journal of Paediatric Neurology</i> , <b>2017</b> , 21, 530-541	3.8	25
95	Application of rare variant transmission disequilibrium tests to epileptic encephalopathy trio sequence data. <i>European Journal of Human Genetics</i> , <b>2017</b> , 25, 894-899	5.3	6
94	Carbamazepine- and oxcarbazepine-induced hyponatremia in people with epilepsy. <i>Epilepsia</i> , <b>2017</b> , 58, 1227-1233	6.4	36
93	Myoclonus epilepsy and ataxia due to KCNC1 mutation: Analysis of 20 cases and K channel properties. <i>Annals of Neurology</i> , <b>2017</b> , 81, 677-689	9.4	39
92	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. <i>American Journal of Human Genetics</i> , <b>2017</b> , 100, 676-688	11	36
91	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. <i>Brain</i> , <b>2017</b> , 140, 1316-1336	11.2	285
90	encephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. <i>Journal of Medical Genetics</i> , <b>2017</b> , 54, 460-470	5.8	109

89	Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU. <i>Human Genetics</i> , <b>2017</b> , 136, 463-479	6.3	41
88	Mutations in GABRB3: From febrile seizures to epileptic encephalopathies. <i>Neurology</i> , <b>2017</b> , 88, 483-492	6.5	68
87	Delineating SPTAN1 associated phenotypes: from isolated epilepsy to encephalopathy with progressive brain atrophy. <i>Brain</i> , <b>2017</b> , 140, 2322-2336	11.2	44
86	Clinical spectrum and genotype-phenotype associations of KCNA2-related encephalopathies. <i>Brain</i> , <b>2017</b> , 140, 2337-2354	11.2	71
85	Neurodevelopmental Disorders Caused by De Novo Variants in KCNB1 Genotypes and Phenotypes. <i>JAMA Neurology</i> , <b>2017</b> , 74, 1228-1236	17.2	47
84	Alternating hemiplegia of childhood and a pathogenic variant of ATP1A3: a case report and pathophysiological considerations. <i>Epileptic Disorders</i> , <b>2017</b> , 19, 226-230	1.9	3
83	Filadelfia, Danish Epilepsy Center, Dianalund, Denmark. <i>Epilepsy and Behavior</i> , <b>2017</b> , 76S, S4-S8	3.2	1
82	encephalopathy: A new disease of vesicle fission. <i>Neurology</i> , <b>2017</b> , 89, 385-394	6.5	46
81	Neuroimaging findings in Mowat-Wilson syndrome: a study of 54 patients. <i>Genetics in Medicine</i> , <b>2017</b> , 19, 691-700	8.1	28
80	Phenotypic spectrum of GABRA1: From generalized epilepsies to severe epileptic encephalopathies. <i>Neurology</i> , <b>2016</b> , 87, 1140-51	6.5	78
79	Targeted sequencing of 351 candidate genes for epileptic encephalopathy in a large cohort of patients. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2016</b> , 4, 568-80	2.3	60
78	Exome Sequencing Fails to Identify the Genetic Cause of Aicardi Syndrome. <i>Molecular Syndromology</i> , <b>2016</b> , 7, 234-238	1.5	12
77	Gene Panel Testing in Epileptic Encephalopathies and Familial Epilepsies. <i>Molecular Syndromology</i> , <b>2016</b> , 7, 210-219	1.5	65
76	Loss of SYNJ1 dual phosphatase activity leads to early onset refractory seizures and progressive neurological decline. <i>Brain</i> , <b>2016</b> , 139, 2420-30	11.2	49
75	Germline and somatic mutations in the gene in focal cortical dysplasia and epilepsy. <i>Neurology: Genetics</i> , <b>2016</b> , 2, e118	3.8	76
74	De novo mutations of KIAA2022 in females cause intellectual disability and intractable epilepsy. <i>Journal of Medical Genetics</i> , <b>2016</b> , 53, 850-858	5.8	32
73	Reply. <i>Annals of Neurology</i> , <b>2016</b> , 80, 168-9	9.4	
72	Letter to the editor: confirming neonatal seizure and late onset ataxia in SCN2A Ala263Val. <i>Journal of Neurology</i> , <b>2016</b> , 263, 1459-60	5.5	11



71	Loss of function of the retinoid-related nuclear receptor (RORB) gene and epilepsy. <i>European Journal of Human Genetics</i> , <b>2016</b> , 24, 1761-1770	5.3	23
70	Precision Medicine: SCN8A Encephalopathy Treated with Sodium Channel Blockers. <i>Neurotherapeutics</i> , <b>2016</b> , 13, 190-1	6.4	29
69	STXBP1 encephalopathy: A neurodevelopmental disorder including epilepsy. <i>Neurology</i> , <b>2016</b> , 86, 954-63.5	6.5	159
68	MECP2 Duplication Syndrome: Evidence of Enhanced Oxidative Stress. A Comparison with Rett Syndrome. <i>PLoS ONE</i> , <b>2016</b> , 11, e0150101	3.7	15
67	Evaluation of Presumably Disease Causing SCN1A Variants in a Cohort of Common Epilepsy Syndromes. <i>PLoS ONE</i> , <b>2016</b> , 11, e0150426	3.7	10
66	Benign infantile seizures and paroxysmal dyskinesia caused by an SCN8A mutation. <i>Annals of Neurology</i> , <b>2016</b> , 79, 428-36	9.4	124
65	Delineating the GRIN1 phenotypic spectrum: A distinct genetic NMDA receptor encephalopathy. <i>Neurology</i> , <b>2016</b> , 86, 2171-8	6.5	108
64	De Novo Mutations in SLC1A2 and CACNA1A Are Important Causes of Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , <b>2016</b> , 99, 287-98	11	180
63	Pitfalls in genetic testing: the story of missed SCN1A mutations. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2016</b> , 4, 457-64	2.3	50
62	CHD2 variants are a risk factor for photosensitivity in epilepsy. <i>Brain</i> , <b>2015</b> , 138, 1198-207	11.2	81
61	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 5250-9	5.6	78
60	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. <i>American Journal of Human Genetics</i> , <b>2015</b> , 96, 808-15	11	114
59	Burden analysis of rare microdeletions suggests a strong impact of neurodevelopmental genes in genetic generalised epilepsies. <i>PLoS Genetics</i> , <b>2015</b> , 11, e1005226	6	70
58	De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy. <i>Nature Genetics</i> , <b>2015</b> , 47, 393-399	36.3	162
57	Recessive loss-of-function mutations in AP4S1 cause mild fever-sensitive seizures, developmental delay and spastic paraplegia through loss of AP-4 complex assembly. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 2218-27	5.6	37
56	Recessive mutations in SLC13A5 result in a loss of citrate transport and cause neonatal epilepsy, developmental delay and teeth hypoplasia. <i>Brain</i> , <b>2015</b> , 138, 3238-50	11.2	57
55	The incidence of SCN1A-related Dravet syndrome in Denmark is 1:22,000: a population-based study from 2004 to 2009. <i>Epilepsia</i> , <b>2015</b> , 56, e36-9	6.4	83
54	Mutations in KCNT1 cause a spectrum of focal epilepsies. <i>Epilepsia</i> , <b>2015</b> , 56, e114-20	6.4	83

53	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. <i>Nature Genetics</i> , <b>2015</b> , 47, 39-46	36.3	177
52	Clinical Phenotype of De Novo Mutation: Case Report and Review of Literature. <i>Child Neurology Open</i> , <b>2015</b> , 2, 2329048X15583717	1.3	18
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46	Aberrant expression of miR-218 and miR-204 in human mesial temporal lobe epilepsy and hippocampal sclerosis-convergence on axonal guidance. <i>Epilepsia</i> , <b>2014</b> , 55, 2017-27	6.4	51
45	Mutations in STX1B, encoding a presynaptic protein, cause fever-associated epilepsy syndromes. <i>Nature Genetics</i> , <b>2014</b> , 46, 1327-32	36.3	138
44	Atypical vitamin B6 deficiency: a rare cause of unexplained neonatal and infantile epilepsies. <i>Journal of Child Neurology</i> , <b>2014</b> , 29, 704-7	2.5	11
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33	Mutations in SYNGAP1 cause intellectual disability, autism, and a specific form of epilepsy by inducing haploinsufficiency. <i>Human Mutation</i> , <b>2013</b> , 34, 385-94	4.7	126
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29	Rare exonic deletions of the RBFOX1 gene increase risk of idiopathic generalized epilepsy. <i>Epilepsia</i> , <b>2013</b> , 54, 265-71	6.4	51
28	Reduction of seizure frequency after epilepsy surgery in a patient with STXBP1 encephalopathy and clinical description of six novel mutation carriers. <i>Epilepsia</i> , <b>2013</b> , 54, e74-80	6.4	42
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15	Deletion of 7q34-q36.2 in two siblings with mental retardation, language delay, primary amenorrhea, and dysmorphic features. <i>American Journal of Medical Genetics, Part A</i> , <b>2010</b> , 152A, 3115-9	2.5	31
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3	De novo Variants in Neurodevelopmental Disorders with Epilepsy		3
2	Predicting Functional Effects of Missense Variants in Voltage-Gated Sodium and Calcium Channels		1
1	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications		2