Rikke S Mller

List of Publications by Citations

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	Analysis of shared heritability in common disorders of the brain. *Science*, **2018**, 360,	33.3	666
195	Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. *Nature Genetics*, **2013**, 45, 825-30	36.3	500
194	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. *Nature Genetics*, **2009**, 41, 160-2	36.3	454
193	Recurrent microdeletions at 15q11.2 and 16p13.11 predispose to idiopathic generalized epilepsies. *Brain*, **2010**, 133, 23-32	11.2	347
192	Mutations in GRIN2A cause idiopathic focal epilepsy with rolandic spikes. *Nature Genetics*, **2013**, 45, 1067-72	36.3	301
191	De novo mutations in synaptic transmission genes including DNM1 cause epileptic encephalopathies. *American Journal of Human Genetics*, **2014**, 95, 360-70	11	299
190	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. *Brain*, **2017**, 140, 1316-1336	11.2	285
189	The phenotypic spectrum of SCN8A encephalopathy. *Neurology*, **2015**, 84, 480-9	6.5	199
188	Genetic determinants of common epilepsies: a meta-analysis of genome-wide association studies. *Lancet Neurology, The*, **2014**, 13, 893-903	24.1	194
187	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
186	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
185	GABRA1 and STXBP1: novel genetic causes of Dravet syndrome. *Neurology*, **2014**, 82, 1245-53	6.5	180
184	De Novo Mutations in SLC1A2 and CACNA1A Are Important Causes of Epileptic Encephalopathies. *American Journal of Human Genetics*, **2016**, 99, 287-98	11	180
183	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. *Nature Genetics*, **2015**, 47, 39-46	36.3	177
182	De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy. *Nature Genetics*, **2015**, 47, 393-399	36.3	162
181	STXBP1 encephalopathy: A neurodevelopmental disorder including epilepsy. *Neurology*, **2016**, 86, 954-62	6.5	159
180	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, 967-75	11	152

179	Extending the KCNQ2 encephalopathy spectrum: clinical and neuroimaging findings in 17 patients. *Neurology*, **2013**, 81, 1697-703	6.5	151
178	De novo mutations in HCN1 cause early infantile epileptic encephalopathy. *Nature Genetics*, **2014**, 46, 640-5	36.3	145
177	De novo variants in neurodevelopmental disorders with epilepsy. *Nature Genetics*, **2018**, 50, 1048-1053	36.3	139
176	Mutations in STX1B, encoding a presynaptic protein, cause fever-associated epilepsy syndromes. *Nature Genetics*, **2014**, 46, 1327-32	36.3	138
175	Progress in Understanding and Treating SCN2A-Mediated Disorders. *Trends in Neurosciences*, **2018**, 41, 442-456	13.3	128
174	Mutations in SYNGAP1 cause intellectual disability, autism, and a specific form of epilepsy by inducing haploinsufficiency. *Human Mutation*, **2013**, 34, 385-94	4.7	126
173	Benign infantile seizures and paroxysmal dyskinesia caused by an SCN8A mutation. *Annals of Neurology*, **2016**, 79, 428-36	9.4	124
172	Truncation of the Down syndrome candidate gene DYRK1A in two unrelated patients with microcephaly. *American Journal of Human Genetics*, **2008**, 82, 1165-70	11	118
171	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. *American Journal of Human Genetics*, **2015**, 96, 808-15	11	114
170	Genome-wide association analysis of genetic generalized epilepsies implicates susceptibility loci at 1q43, 2p16.1, 2q22.3 and 17q21.32. *Human Molecular Genetics*, **2012**, 21, 5359-72	5.6	114
169	encephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. *Journal of Medical Genetics*, **2017**, 54, 460-470	5.8	109
168	Delineating the GRIN1 phenotypic spectrum: A distinct genetic NMDA receptor encephalopathy. *Neurology*, **2016**, 86, 2171-8	6.5	108
167	The incidence of SCN1A-related Dravet syndrome in Denmark is 1:22,000: a population-based study from 2004 to 2009. *Epilepsia*, **2015**, 56, e36-9	6.4	83
166	Mutations in KCNT1 cause a spectrum of focal epilepsies. *Epilepsia*, **2015**, 56, e114-20	6.4	83
165	CHD2 variants are a risk factor for photosensitivity in epilepsy. *Brain*, **2015**, 138, 1198-207	11.2	81
164	The phenotype of developmental and epileptic encephalopathy. *Neurology*, **2018**, 91, e1112-e1124	6.5	80
163	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. *Human Molecular Genetics*, **2015**, 24, 5250-9	5.6	78
162	Phenotypic spectrum of GABRA1: From generalized epilepsies to severe epileptic encephalopathies. *Neurology*, **2016**, 87, 1140-51	6.5	78

161	Germline and somatic mutations in the gene in focal cortical dysplasia and epilepsy. *Neurology: Genetics*, **2016**, 2, e118	3.8	76
160	The landscape of epilepsy-related GATOR1 variants. *Genetics in Medicine*, **2019**, 21, 398-408	8.1	75
159	Clinical spectrum and genotype-phenotype associations of KCNA2-related encephalopathies. *Brain*, **2017**, 140, 2337-2354	11.2	71
158	Burden analysis of rare microdeletions suggests a strong impact of neurodevelopmental genes in genetic generalised epilepsies. *PLoS Genetics*, **2015**, 11, e1005226	6	70
157	Mutations in GABRB3: From febrile seizures to epileptic encephalopathies. *Neurology*, **2017**, 88, 483-492	6.5	68
156	RBFOX1 and RBFOX3 mutations in rolandic epilepsy. *PLoS ONE*, **2013**, 8, e73323	3.7	68
155	GRIN2A-related disorders: genotype and functional consequence predict phenotype. *Brain*, **2019**, 142, 80-92	11.2	66
154	Gene Panel Testing in Epileptic Encephalopathies and Familial Epilepsies. *Molecular Syndromology*, **2016**, 7, 210-219	1.5	65
153	Targeted sequencing of 351 candidate genes for epileptic encephalopathy in a large cohort of patients. *Molecular Genetics & Genomic Medicine*, **2016**, 4, 568-80	2.3	60
152	Recessive mutations in SLC13A5 result in a loss of citrate transport and cause neonatal epilepsy, developmental delay and teeth hypoplasia. *Brain*, **2015**, 138, 3238-50	11.2	57
151	Neurologic phenotypes associated with / mutations: Expanding the spectrum of disease. *Neurology*, **2018**, 91, e2078-e2088	6.5	55
150	Defining the phenotypic spectrum of SLC6A1 mutations. *Epilepsia*, **2018**, 59, 389-402	6.4	54
149	Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. *Genome Medicine*, **2018**, 10, 3	14.4	53
148	Neuronal mechanisms of mutations in SCN8A causing epilepsy or intellectual disability. *Brain*, **2019**, 142, 376-390	11.2	53
147	The role of SLC2A1 mutations in myoclonic astatic epilepsy and absence epilepsy, and the estimated frequency of GLUT1 deficiency syndrome. *Epilepsia*, **2015**, 56, e203-8	6.4	52
146	Aberrant expression of miR-218 and miR-204 in human mesial temporal lobe epilepsy and hippocampal sclerosis-convergence on axonal guidance. *Epilepsia*, **2014**, 55, 2017-27	6.4	51
145	Mutations in NRXN1 in a family multiply affected with brain disorders: NRXN1 mutations and brain disorders. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2012**, 159B, 354-8	3.5	51
144	Rare exonic deletions of the RBFOX1 gene increase risk of idiopathic generalized epilepsy. *Epilepsia* , **2013**, 54, 265-71	6.4	51

143	Pitfalls in genetic testing: the story of missed SCN1A mutations. *Molecular Genetics & Genomic Medicine*, **2016**, 4, 457-64	2.3	50
142	Loss of SYNJ1 dual phosphatase activity leads to early onset refractory seizures and progressive neurological decline. *Brain*, **2016**, 139, 2420-30	11.2	49
141	QNorth SeaQprogressive myoclonus epilepsy: phenotype of subjects with GOSR2 mutation. *Brain*, **2013**, 136, 1146-54	11.2	49
140	Exon-disrupting deletions of NRXN1 in idiopathic generalized epilepsy. *Epilepsia*, **2013**, 54, 256-64	6.4	48
139	Neurodevelopmental Disorders Caused by De Novo Variants in KCNB1 Genotypes and Phenotypes. *JAMA Neurology*, **2017**, 74, 1228-1236	17.2	47
138	encephalopathy: A new disease of vesicle fission. *Neurology*, **2017**, 89, 385-394	6.5	46
137	The contribution of next generation sequencing to epilepsy genetics. *Expert Review of Molecular Diagnostics*, **2015**, 15, 1531-8	3.8	45
136	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. *Human Molecular Genetics*, **2014**, 23, 6069-80	5.6	45
135	Rare coding variants in genes encoding GABA receptors in genetic generalised epilepsies: an exome-based case-control study. *Lancet Neurology, The*, **2018**, 17, 699-708	24.1	44
134	Delineating SPTAN1 associated phenotypes: from isolated epilepsy to encephalopathy with progressive brain atrophy. *Brain*, **2017**, 140, 2322-2336	11.2	44
133	Reduced ceramide synthase 2 activity causes progressive myoclonic epilepsy. *Annals of Clinical and Translational Neurology*, **2014**, 1, 88-98	5.3	42
132	Reduction of seizure frequency after epilepsy surgery in a patient with STXBP1 encephalopathy and clinical description of six novel mutation carriers. *Epilepsia*, **2013**, 54, e74-80	6.4	42
131	Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU. *Human Genetics*, **2017**, 136, 463-479	6.3	41
130	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
129	Myoclonus epilepsy and ataxia due to KCNC1 mutation: Analysis of 20 cases and K channel properties. *Annals of Neurology*, **2017**, 81, 677-689	9.4	39
128	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	11	39
127	The spectrum of intermediate SCN8A-related epilepsy. *Epilepsia*, **2019**, 60, 830-844	6.4	38
126	Early mortality in SCN8A-related epilepsies. *Epilepsy Research*, **2018**, 143, 79-81	3	38

125	Diagnostic implications of genetic copy number variation in epilepsy plus. *Epilepsia*, **2019**, 60, 689-706	6.4	37
124	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, 2218-27	5.6	37
123	Phenotype and genotype of 87 patients with Mowat-Wilson syndrome and recommendations for care. *Genetics in Medicine*, **2018**, 20, 965-975	8.1	37
122	Carbamazepine- and oxcarbazepine-induced hyponatremia in people with epilepsy. *Epilepsia*, **2017**, 58, 1227-1233	6.4	36
121	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. *American Journal of Human Genetics*, **2017**, 100, 676-688	11	36
120	Biological concepts in human sodium channel epilepsies and their relevance in clinical practice. *Epilepsia*, **2020**, 61, 387-399	6.4	35
119	Utility of genetic testing for therapeutic decision-making in adults with epilepsy. *Epilepsia*, **2020**, 61, 1234-1239	6.4	32
118	De novo mutations of KIAA2022 in females cause intellectual disability and intractable epilepsy. *Journal of Medical Genetics*, **2016**, 53, 850-858	5.8	32
117	Incorporating epilepsy genetics into clinical practice: a 360°evaluation. *Npj Genomic Medicine*, **2018**, 3, 13	6.2	32
116	Phenotypic and genetic spectrum of SCN8A-related disorders, treatment options, and outcomes. *Epilepsia*, **2019**, 60 Suppl 3, S77-S85	6.4	32
115	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. *Genetics in Medicine*, **2019**, 21, 837-849	8.1	32
114	Deletion of 7q34-q36.2 in two siblings with mental retardation, language delay, primary amenorrhea, and dysmorphic features. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 3115-9	2.5	31
113	Treatment Responsiveness in KCNT1-Related Epilepsy. *Neurotherapeutics*, **2019**, 16, 848-857	6.4	30
112	From next-generation sequencing to targeted treatment of non-acquired epilepsies. *Expert Review of Molecular Diagnostics*, **2019**, 19, 217-228	3.8	29
111	Precision Medicine: SCN8A Encephalopathy Treated with Sodium Channel Blockers. *Neurotherapeutics*, **2016**, 13, 190-1	6.4	29
110	Neuroimaging findings in Mowat-Wilson syndrome: a study of 54 patients. *Genetics in Medicine*, **2017**, 19, 691-700	8.1	28
109	Genome-wide linkage meta-analysis identifies susceptibility loci at 2q34 and 13q31.3 for genetic generalized epilepsies. *Epilepsia*, **2012**, 53, 308-18	6.4	28
108	Novel congenital disorder of O-linked glycosylation caused by GALNT2 loss of function. *Brain*, **2020**, 143, 1114-1126	11.2	28

107	Predicting functional effects of missense variants in voltage-gated sodium and calcium channels. *Science Translational Medicine*, **2020**, 12,	17.5	27
106	Epilepsy in patients with GRIN2A alterations: Genetics, neurodevelopment, epileptic phenotype and response to anticonvulsive drugs. *European Journal of Paediatric Neurology*, **2017**, 21, 530-541	3.8	25
105	Clinical spectrum of -related epileptic disorders. *Neurology*, **2019**, 92, e1238-e1249	6.5	25
104	NIPA1 mutation in complex hereditary spastic paraplegia with epilepsy. *European Journal of Neurology*, **2011**, 18, 1197-9	6	25
103	Structural genomic variation in childhood epilepsies with complex phenotypes. *European Journal of Human Genetics*, **2014**, 22, 896-901	5.3	24
102	Spectrum of GABAA receptor variants in epilepsy. *Current Opinion in Neurology*, **2019**, 32, 183-190	7.1	24
101	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. *Seizure: the Journal of the British Epilepsy Association*, **2019**, 65, 131-137	3.2	23
100	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
99	Estimating the effect size of the 15Q11.2 BP1-BP2 deletion and its contribution to neurodevelopmental symptoms: recommendations for practice. *Journal of Medical Genetics*, **2019**, 56, 701-710	5.8	22
98	Genetic heterogeneity in infantile spasms. *Epilepsy Research*, **2019**, 156, 106181	3	20
97	A balanced translocation disrupts SYNGAP1 in a patient with intellectual disability, speech impairment, and epilepsy with myoclonic absences (EMA). *Epilepsia*, **2011**, 52, e190-3	6.4	20
96	High frequency of rare copy number variants affecting functionally related genes in patients with structural brain malformations. *Human Mutation*, **2011**, 32, 1427-35	4.7	20
95	Fluorescently labelled bovine acyl-CoA-binding protein acting as an acyl-CoA sensor: interaction with CoA and acyl-CoA esters and its use in measuring free acyl-CoA esters and non-esterified fatty acids. *Biochemical Journal*, **2002**, 365, 165-72	3.8	20
94	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	8.1	18
93	Phenotypic and genetic spectrum of epilepsy with myoclonic atonic seizures. *Epilepsia*, **2020**, 61, 995-1007	6.4	18
92	A catalogue of new incidence estimates of monogenic neurodevelopmental disorders caused by de novo variants. *Brain*, **2020**, 143, 1099-1105	11.2	18
91	Clinical Phenotype of De Novo Mutation: Case Report and Review of Literature. *Child Neurology Open*, **2015**, 2, 2329048X15583717	1.3	18
90	Recent advances in treatment of epilepsy-related sodium channelopathies. *European Journal of Paediatric Neurology*, **2020**, 24, 123-128	3.8	18

89	Mild malformations of cortical development in sleep-related hypermotor epilepsy due to mutations. *Annals of Clinical and Translational Neurology*, **2019**, 6, 386-391	5.3	17
88	Parental mosaicism in epilepsies due to alleged de novo variants. *Epilepsia*, **2019**, 60, e63-e66	6.4	16
87	Lessons learned from 40 novel PIGA patients and a review of the literature. *Epilepsia*, **2020**, 61, 1142-1156	5.4	15
86	Characterization of a t(5;8)(q31;q21) translocation in a patient with mental retardation and congenital heart disease: implications for involvement of RUNX1T1 in human brain and heart development. *European Journal of Human Genetics*, **2009**, 17, 1010-8	5.3	15
85	MECP2 Duplication Syndrome: Evidence of Enhanced Oxidative Stress. A Comparison with Rett Syndrome. *PLoS ONE*, **2016**, 11, e0150101	3.7	15
84	SLC35A2-related congenital disorder of glycosylation: Defining the phenotype. *European Journal of Paediatric Neurology*, **2018**, 22, 1095-1102	3.8	15
83	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, 625-38	6.3	14
82	Clinician's guide to genes associated with Rett-like phenotypes-Investigation of a Danish cohort and review of the literature. *Clinical Genetics*, **2019**, 95, 221-230	4	14
81	Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. *Genome Medicine*, **2020**, 12, 28	14.4	13
80	Mowat-Wilson syndrome: an underdiagnosed syndrome?. *Clinical Genetics*, **2008**, 73, 579-84	4	13
79	The Phenotypic Spectrum of PRRT2-Associated Paroxysmal Neurologic Disorders in Childhood. *Biomedicines*, **2020**, 8,	4.8	12
78	Exome Sequencing Fails to Identify the Genetic Cause of Aicardi Syndrome. *Molecular Syndromology*, **2016**, 7, 234-238	1.5	12
77	The role of SLC2A1 in early onset and childhood absence epilepsies. *Epilepsy Research*, **2013**, 105, 229-33	3	12
76	Genetic studies in congenital anterior midline cervical cleft. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2021-6	2.5	12
75	The impact of severe pediatric epilepsy on experienced stress and psychopathology in parents. *Epilepsy and Behavior*, **2020**, 113, 107538	3.2	12
74	Epilepsy Syndromes in the First Year of Life and Usefulness of Genetic Testing for Precision Therapy. *Genes*, **2021**, 12,	4.2	12
73	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. *Annals of Neurology*, **2018**, 83, 926-934	9.4	11
72	Letter to the editor: confirming neonatal seizure and late onset ataxia in SCN2A Ala263Val. *Journal of Neurology*, **2016**, 263, 1459-60	5.5	11

71	Atypical vitamin B6 deficiency: a rare cause of unexplained neonatal and infantile epilepsies. *Journal of Child Neurology*, **2014**, 29, 704-7	2.5	11
70	Current knowledge of SLC6A1-related neurodevelopmental disorders. *Brain Communications*, **2020**, 2, fcaa170	4.5	11
69	Sequence analysis of 17 NRXN1 deletions. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2014**, 165B, 52-61	3.5	10
68	Balanced translocation in a patient with severe myoclonic epilepsy of infancy disrupts the sodium channel gene SCN1A. *Epilepsia*, **2008**, 49, 1091-4	6.4	10
67	Evaluation of Presumably Disease Causing SCN1A Variants in a Cohort of Common Epilepsy Syndromes. *PLoS ONE*, **2016**, 11, e0150426	3.7	10
66	Testing association of rare genetic variants with resistance to three common antiseizure medications. *Epilepsia*, **2020**, 61, 657-666	6.4	9
65	Idiopathic encephalopathy related to status epilepticus during slow sleep (ESES) as a "pure" model of epileptic encephalopathy. An electroclinical, genetic, and follow-up study. *Epilepsy and Behavior*, **2019**, 97, 244-252	3.2	9
64	Biallelic inherited SCN8A variants, a rare cause of SCN8A-related developmental and epileptic encephalopathy. *Epilepsia*, **2019**, 60, 2277-2285	6.4	9
63	Duplication of MAOA, MAOB, and NDP in a patient with mental retardation and epilepsy. *European Journal of Human Genetics*, **2011**, 19, 1-2	5.3	9
62	Interstitial deletion of chromosome 4p associated with mild mental retardation, epilepsy and polymicrogyria of the left temporal lobe. *Clinical Genetics*, **2007**, 72, 593-8	4	9
61	Genotype-phenotype correlations in patients with de novo pathogenic variants. *Neurology: Genetics* , **2020**, 6, e528	3.8	9
60	Pharmacoresponse in genetic generalized epilepsy: a genome-wide association study. *Pharmacogenomics*, **2020**, 21, 325-335	2.6	8
59	Update on the genetics of the epilepsy-aphasia spectrum and role of GRIN2A mutations **2019**, 21, 41-47		8
58	Neonatal developmental and epileptic encephalopathy due to autosomal recessive variants in SLC13A5 gene. *Epilepsia*, **2020**, 61, 2474-2485	6.4	8
57	Real-life survey of pitfalls and successes of precision medicine in genetic epilepsies. *Journal of Neurology, Neurosurgery and Psychiatry*, **2021**, 92, 1044-1052	5.5	8
56	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. *Brain*, **2021**,	11.2	8
55	Application of rare variant transmission disequilibrium tests to epileptic encephalopathy trio sequence data. *European Journal of Human Genetics*, **2017**, 25, 894-899	5.3	6
54	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	5.2	6

53	Dysregulation of FOXG1 by ring chromosome 14. *Molecular Cytogenetics*, **2015**, 8, 24	2	6
52	Gain-of-function variants in GABRD reveal a novel pathway for neurodevelopmental disorders and epilepsy. *Brain*, **2021**,	11.2	6
51	Gain-of-function variants identified in vigabatrin-hypersensitive epileptic encephalopathies. *Brain Communications*, **2020**, 2, fcaa162	4.5	6
50	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	3.8	6
49	Refining Genotypes and Phenotypes in -Related Neurological Disorders. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	6
48	Adult phenotype of encephalopathy. *Journal of Medical Genetics*, **2021**,	5.8	6
47	KCNT1-related epilepsies and epileptic encephalopathies: phenotypic and mutational spectrum. *Brain*, **2021**,	11.2	6
46	Deciphering the premature mortality in PIGA-CDG - An untold story. *Epilepsy Research*, **2021**, 170, 106539	3	5
45	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. *Genetics in Medicine*, **2021**, 23, 653-660	8.1	5
44	No evidence for a BRD2 promoter hypermethylation in blood leukocytes of Europeans with juvenile myoclonic epilepsy. *Epilepsia*, **2019**, 60, e31-e36	6.4	4
43	Expanding the clinical and EEG spectrum of CNKSR2-related encephalopathy with status epilepticus during slow sleep (ESES). *Clinical Neurophysiology*, **2020**, 131, 1030-1039	4.3	4
42	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. *Brain*, **2021**, 144, 1435-1450	11.2	4
41	Two GluN2B mutations affect multiple NMDAR-functions and instigate severe pediatric encephalopathy. *ELife*, **2021**, 10,	8.9	4
40	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. *Genetics in Medicine*, **2021**, 23, 363-373	8.1	4
39	Assessing the landscape of STXBP1-related disorders in 534 individuals.. *Brain*, **2021**,	11.2	4
38	Integrative approach to interpret DYRK1A variants, leading to a frequent neurodevelopmental disorder. *Genetics in Medicine*, **2021**, 23, 2150-2159	8.1	4
37	Mowat-Wilson syndrome: growth charts. *Orphanet Journal of Rare Diseases*, **2020**, 15, 151	4.2	3
36	Alternating hemiplegia of childhood and a pathogenic variant of ATP1A3: a case report and pathophysiological considerations. *Epileptic Disorders*, **2017**, 19, 226-230	1.9	3

35	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**, 1	6.4	3
34	De novo Variants in Neurodevelopmental Disorders with Epilepsy		3
33	Electroclinical features of MEF2C haploinsufficiency-related epilepsy: A multicenter European study. *Seizure: the Journal of the British Epilepsy Association*, **2021**, 88, 60-72	3.2	3
32	Defining and expanding the phenotype of -associated developmental epileptic encephalopathy. *Neurology: Genetics*, **2019**, 5, e373	3.8	3
31	Basal Ganglia Dysmorphism in Patients With Aicardi Syndrome. *Neurology*, **2021**, 96, e1319-e1333	6.5	3
30	Mutations Expand the Phenotypic Spectrum of Alternating Hemiplegia of Childhood. *Neurology*, **2021**, 96, e1539-e1550	6.5	3
29	Structural mapping of GABRB3 variants reveals genotype-phenotype correlations.. *Genetics in Medicine*, **2021**,	8.1	3
28	A cryptic unbalanced translocation resulting in del 13q and dup 15q. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2570-3	2.5	2
27	Development and Validation of a Prediction Model for Early Diagnosis of -Related Epilepsies.. *Neurology*, **2022**,	6.5	2
26	De novo FZR1 loss-of-function variants cause developmental and epileptic encephalopathies. *Brain*, **2021**,	11.2	2
25	Genetic testing in adult epilepsy patients: A call to action for clinicians. *Epilepsia*, **2020**, 61, 2055-2056	6.4	2
24	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications		2
23	Deep-Phenotyping the Less Severe Spectrum of Deficiency and Linking the Gene to Myoclonic Atonic Seizures. *Frontiers in Genetics*, **2021**, 12, 663643	4.5	2
22	PRICKLE2 revisited-further evidence implicating PRICKLE2 in neurodevelopmental disorders. *European Journal of Human Genetics*, **2021**, 29, 1235-1244	5.3	2
21	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	2.6	2
20	Characterization of the GABRB2-Associated Neurodevelopmental Disorders. *Annals of Neurology*, **2021**, 89, 573-586	9.4	2
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