Rikke S MÃ¸ller

List of Publications by Year
in descending order

203 papers	15,119 citations	20797 60 h-index	24232 110 g-index
221 all docs	221 docs citations	221 times ranked	17438 citing authors

#	Article	IF	Citations
1	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	6.0	1,085
2	Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. Nature Genetics, 2013, 45, 825-830.	9.4	589
3	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. Nature Genetics, 2009, 41, 160-162.	9.4	511
4	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. Brain, 2017, 140, 1316-1336.	3.7	426
5	Recurrent microdeletions at 15q11.2 and 16p13.11 predispose to idiopathic generalized epilepsies. Brain, 2010, 133, 23-32.	3.7	406
6	Mutations in GRIN2A cause idiopathic focal epilepsy with rolandic spikes. Nature Genetics, 2013, 45, 1067-1072.	9.4	391
7	De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	2.6	388
8	Genetic determinants of common epilepsies: a meta-analysis of genome-wide association studies. Lancet Neurology, The, 2014, 13, 893-903.	4.9	264
9	STXBP1 encephalopathy. Neurology, 2016, 86, 954-962.	1.5	264
10	De Novo Mutations in SLC1A2 and CACNA1A Are Important Causes of Epileptic Encephalopathies. American Journal of Human Genetics, 2016, 99, 287-298.	2.6	247
11	The phenotypic spectrum of SCN8A encephalopathy. Neurology, 2015, 84, 480-489.	1.5	246
12	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. Nature Genetics, 2015, 47, 39-46.	9.4	245
13	De novo variants in neurodevelopmental disorders with epilepsy. Nature Genetics, 2018, 50, 1048-1053.	9.4	230
14	GABRA1 and STXBP1 : Novel genetic causes of Dravet syndrome. Neurology, 2014, 82, 1245-1253.	1.5	229
15	De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy. Nature Genetics, 2015, 47, 393-399.	9.4	224
16	Recurrent Reciprocal Genomic Rearrangements of 17q12 Are Associated with Renal Disease, Diabetes, and Epilepsy. American Journal of Human Genetics, 2007, 81, 1057-1069.	2.6	222
17	Familial and sporadic 15q13.3 microdeletions in idiopathic generalized epilepsy: precedent for disorders with complex inheritance. Human Molecular Genetics, 2009, 18, 3626-3631.	1.4	211
18	Progress in Understanding and Treating SCN2A-Mediated Disorders. Trends in Neurosciences, 2018, 41, 442-456.	4.2	210

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19	Extending the KCNQ2 encephalopathy spectrum. Neurology, 2013, 81, 1697-1703.	1.5	198
20	Mutations inSYNGAP1Cause Intellectual Disability, Autism, and a Specific Form of Epilepsy by Inducing Haploinsufficiency. Human Mutation, 2013, 34, 385-394.	1.1	196
21	De novo mutations in HCN1 cause early infantile epileptic encephalopathy. Nature Genetics, 2014, 46, 640-645.	9.4	192
22	GRIN2Bencephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. Journal of Medical Genetics, 2017, 54, 460-470.	1.5	190
23	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.	2.6	188
24	Mutations in STX1B, encoding a presynaptic protein, cause fever-associated epilepsy syndromes. Nature Genetics, 2014, 46, 1327-1332.	9.4	178
25	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. American Journal of Human Genetics, 2015, 96, 808-815.	2.6	173
26	Benign infantile seizures and paroxysmal dyskinesia caused by an SCN8A mutation. Annals of Neurology, 2016, 79, 428-436.	2.8	159
27	Delineating the GRIN1 phenotypic spectrum. Neurology, 2016, 86, 2171-2178.	1.5	157
28	Truncation of the Down Syndrome Candidate Gene DYRK1A in Two Unrelated Patients with Microcephaly. American Journal of Human Genetics, 2008, 82, 1165-1170.	2.6	145
29	GRIN2A-related disorders: genotype and functional consequence predict phenotype. Brain, 2019, 142, 80-92.	3.7	143
30	The landscape of epilepsy-related GATOR1 variants. Genetics in Medicine, 2019, 21, 398-408.	1.1	137
31	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, 5359-5372.	1.4	134
32	â€˜North Seaâ€™ progressive myoclonus epilepsy: phenotype of subjects with GOSR2 mutation. Brain, 2013, 136, 1146-1154.	3.7	129
33	Germline and somatic mutations in the MTOR gene in focal cortical dysplasia and epilepsy. Neurology: Genetics, 2016, 2, e118.	0.9	125
34	Mutations in <scp>KCNT</scp>1 cause a spectrum of focal epilepsies. Epilepsia, 2015, 56, e114-20.	2.6	117
35	Clinical spectrum and genotypeâ€“phenotype associations of KCNA2-related encephalopathies. Brain, 2017, 140, 2337-2354.	3.7	117
36	The phenotype of SCN8A developmental and epileptic encephalopathy. Neurology, 2018, 91, e1112-e1124.	1.5	114

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37	Phenotypic spectrum of GABRA1. Neurology, 2016, 87, 1140-1151.	1.5	113
38	CHD2variants are a risk factor for photosensitivity in epilepsy. Brain, 2015, 138, 1198-1208.	3.7	112
39	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.	2.6	103
40	Gene Panel Testing in Epileptic Encephalopathies and Familial Epilepsies. Molecular Syndromology, 2016, 7, 210-219.	0.3	103
41	Defining the phenotypic spectrum of SLC6A1 mutations. Epilepsia, 2018, 59, 389-402.	2.6	99
42	Neurologic phenotypes associated with COL4A1 / 2 mutations. Neurology, 2018, 91, e2078-e2088.	1.5	97
43	Recessive mutations inSLC13A5result in a loss of citrate transport and cause neonatal epilepsy, developmental delay and teeth hypoplasia. Brain, 2015, 138, 3238-3250.	3.7	96
44	RBFOX1 and RBFOX3 Mutations in Rolandic Epilepsy. PLoS ONE, 2013, 8, e73323.	1.1	94
45	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. Human Molecular Genetics, 2015, 24, 5250-5259.	1.4	93
46	Neuronal mechanisms of mutations in SCN8A causing epilepsy or intellectual disability. Brain, 2019, 142, 376-390.	3.7	92
47	Burden Analysis of Rare Microdeletions Suggests a Strong Impact of Neurodevelopmental Genes in Genetic Generalised Epilepsies. PLoS Genetics, 2015, 11, e1005226.	1.5	91
48	Mutations in GABRB3. Neurology, 2017, 88, 483-492.	1.5	87
49	DNM1 encephalopathy. Neurology, 2017, 89, 385-394.	1.5	87
50	Predicting functional effects of missense variants in voltage-gated sodium and calcium channels. Science Translational Medicine, 2020, 12, .	5.8	84
51	Targeted sequencing of 351 candidate genes for epileptic encephalopathy in a large cohort of patients. Molecular Genetics & Genomic Medicine, 2016, 4, 568-580.	0.6	83
52	Delineating SPTAN1 associated phenotypes: from isolated epilepsy to encephalopathy with progressive brain atrophy. Brain, 2017, 140, 2322-2336.	3.7	82
53	Neurodevelopmental Disorders Caused by De Novo Variants in KCNB1 Genotypes and Phenotypes. JAMA Neurology, 2017, 74, 1228.	4.5	79
54	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.	2.6	78

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55	Aberrant Inclusion of a Poison Exon Causes Dravet Syndrome and Related SCN1A-Associated Genetic Epilepsies. American Journal of Human Genetics, 2018, 103, 1022-1029.	2.6	76
56	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.	2.6	71
57	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.	2.6	71
58	Loss of SYNJ1 dual phosphatase activity leads to early onset refractory seizures and progressive neurological decline. Brain, 2016, 139, 2420-2430.	3.7	70
59	The spectrum of intermediate <scp>SCN</scp>8Aâ€related epilepsy. Epilepsia, 2019, 60, 830-844.	2.6	70
60	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.	2.8	69
61	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. Brain, 2022, 145, 2991-3009.	3.7	69
62	The contribution of next generation sequencing to epilepsy genetics. Expert Review of Molecular Diagnostics, 2015, 15, 1531-1538.	1.5	68
63	Pitfalls in genetic testing: the story of missed SCN1A mutations. Molecular Genetics & Genomic Medicine, 2016, 4, 457-464.	0.6	67
64	Phenotype and genotype of 87 patients with Mowatâ€"Wilson syndrome and recommendations for care. Genetics in Medicine, 2018, 20, 965-975.	1.1	67
65	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.	4.9	67
66	Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. Genome Medicine, 2018, 10, 3.	3.6	67
67	Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU. Human Genetics, 2017, 136, 463-479.	1.8	66
68	Biological concepts in human sodium channel epilepsies and their relevance in clinical practice. Epilepsia, 2020, 61, 387-399.	2.6	65
69	A catalogue of new incidence estimates of monogenic neurodevelopmental disorders caused by de novo variants. Brain, 2020, 143, 1099-1105.	3.7	64
70	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, 354-358.	1.1	63
71	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. Human Molecular Genetics, 2014, 23, 6069-6080.	1.4	61
72	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	2.6	61

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73	Treatment Responsiveness in KCNT1-Related Epilepsy. Neurotherapeutics, 2019, 16, 848-857.	2.1	60
74	Utility of genetic testing for therapeutic decisionâ€making in adults with epilepsy. Epilepsia, 2020, 61, 1234-1239.	2.6	60
75	Rare exonic deletions of the <scp>RBFOX1</scp> gene increase risk of idiopathic generalized epilepsy. Epilepsia, 2013, 54, 265-271.	2.6	59
76	Reduction of seizure frequency after epilepsy surgery in a patient with <scp>STXBP1</scp> encephalopathy and clinical description of six novel mutation carriers. Epilepsia, 2013, 54, e74-80.	2.6	59
77	Exonâ€disrupting deletions of <scp>NRXN1</scp> in idiopathic generalized epilepsy. Epilepsia, 2013, 54, 256-264.	2.6	59
78	Spectrum of GABAA receptor variants in epilepsy. Current Opinion in Neurology, 2019, 32, 183-190.	1.8	59
79	Phenotypic and genetic spectrum of <scp>SCN</scp>8Aâ€related disorders, treatment options, and outcomes. Epilepsia, 2019, 60, S77-S85.	2.6	58
80	Carbamazepineâ€and oxcarbazepineâ€induced hyponatremia in people with epilepsy. Epilepsia, 2017, 58, 1227-1233.	2.6	54
81	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. American Journal of Human Genetics, 2017, 100, 676-688.	2.6	54
82	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.	1.4	53
83	Reduced ceramide synthase 2 activity causes progressive myoclonic epilepsy. Annals of Clinical and Translational Neurology, 2014, 1, 88-98.	1.7	50
84	Early mortality in SCN8A -related epilepsies. Epilepsy Research, 2018, 143, 79-81.	0.8	48
85	De novo mutations ofKIAA2022in females cause intellectual disability and intractable epilepsy. Journal of Medical Genetics, 2016, 53, 850-858.	1.5	47
86	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. Genetics in Medicine, 2019, 21, 837-849.	1.1	47
87	Incorporating epilepsy genetics into clinical practice: a 360Â°evaluation. Npj Genomic Medicine, 2018, 3, 13.	1.7	46
88	Novel congenital disorder of O-linked glycosylation caused by GALNT2 loss of function. Brain, 2020, 143, 1114-1126.	3.7	46
89	Assessing the landscape of STXBP1-related disorders in 534 individuals. Brain, 2022, 145, 1668-1683.	3.7	46
90	Neuroimaging findings in Mowatâ€“Wilson syndrome: a study of 54 patients. Genetics in Medicine, 2017, 19, 691-700.	1.1	45

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91	Current knowledge of SLC6A1-related neurodevelopmental disorders. Brain Communications, 2020, 2, fcaa170.	1.5	44
92	Clinical spectrum of STX1B -related epileptic disorders. Neurology, 2019, 92, e1238-e1249.	1.5	43
93	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.	1.5	43
94	Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. Genome Medicine, 2020, 12, 28.	3.6	42
95	Recent advances in treatment of epilepsy-related sodium channelopathies. European Journal of Paediatric Neurology, 2020, 24, 123-128.	0.7	40
96	Precision Medicine: SCN8A Encephalopathy Treated with Sodium Channel Blockers. Neurotherapeutics, 2016, 13, 190-191.	2.1	38
97	Genetic heterogeneity in infantile spasms. Epilepsy Research, 2019, 156, 106181.	0.8	38
98	From next-generation sequencing to targeted treatment of non-acquired epilepsies. Expert Review of Molecular Diagnostics, 2019, 19, 217-228.	1.5	38
99	Deletion of 7q34â€“q36.2 in two siblings with mental retardation, language delay, primary amenorrhea, and dysmorphic features. American Journal of Medical Genetics, Part A, 2010, 152A, 3115-3119.	0.7	37
100	Epilepsy in patients with GRIN2A alterations: Genetics, neurodevelopment, epileptic phenotype and response to anticonvulsive drugs. European Journal of Paediatric Neurology, 2017, 21, 530-541.	0.7	37
101	Loss of function of the retinoid-related nuclear receptor (RORB) gene and epilepsy. European Journal of Human Genetics, 2016, 24, 1761-1770.	1.4	36
102	Epilepsy Syndromes in the First Year of Life and Usefulness of Genetic Testing for Precision Therapy. Genes, 2021, 12, 1051.	1.0	36
103	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. Brain, 2021, 144, 1435-1450.	3.7	35
104	NIPA1mutation in complex hereditary spastic paraplegia with epilepsy. European Journal of Neurology, 2011, 18, 1197-1199.	1.7	34
105	KCNT1-related epilepsies and epileptic encephalopathies: phenotypic and mutational spectrum. Brain, 2021, 144, 3635-3650.	3.7	34
106	Gain-of-function variants in GABRD reveal a novel pathway for neurodevelopmental disorders and epilepsy. Brain, 2022, 145, 1299-1309.	3.7	34
107	Genomeâ€wide linkage metaâ€analysis identifies susceptibility loci at 2q34 and 13q31.3 for genetic generalized epilepsies. Epilepsia, 2012, 53, 308-318.	2.6	32
108	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	2.6	32

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109	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.	5.8	32
110	Neonatal developmental and epileptic encephalopathy due to autosomal recessive variants in SLC13A5 gene. Epilepsia, 2020, 61, 2474-2485.	2.6	31
111	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.	0.9	30
112	Phenotypic and genetic spectrum of epilepsy with myoclonic atonic seizures. Epilepsia, 2020, 61, 995-1007.	2.6	30
113	Real-life survey of pitfalls and successes of precision medicine in genetic epilepsies. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 1044-1052.	0.9	30
114	Parental mosaicism in epilepsies due to alleged de novo variants. Epilepsia, 2019, 60, e63-e66.	2.6	29
115	Structural genomic variation in childhood epilepsies with complex phenotypes. European Journal of Human Genetics, 2014, 22, 896-901.	1.4	28
116	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	1.1	28
117	SLC35A2-related congenital disorder of glycosylation: Defining the phenotype. European Journal of Paediatric Neurology, 2018, 22, 1095-1102.	0.7	27
118	A balanced translocation disruptsSYNGAP1in a patient with intellectual disability, speech impairment, and epilepsy with myoclonic absences (EMA). Epilepsia, 2011, 52, e190-e193.	2.6	26
119	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.	1.0	26
120	Clinical Phenotype of De Novo GNAO1 Mutation. Child Neurology Open, 2015, 2, 2329048X1558371.	0.5	25
121	Mild malformations of cortical development in sleepâ€related hypermotor epilepsy due to KCNT1 mutations. Annals of Clinical and Translational Neurology, 2019, 6, 386-391.	1.7	25
122	High frequency of rare copy number variants affecting functionally related genes in patients with structural brain malformations. Human Mutation, 2011, 32, 1427-1435.	1.1	24
123	Genotype-phenotype correlations in patients with de novo KCNQ2 pathogenic variants. Neurology: Genetics, 2020, 6, e528.	0.9	24
124	Development and Validation of a Prediction Model for Early Diagnosis of SCN1A -Related Epilepsies. Neurology, 2022, 98, .	1.5	24
125	The impact of severe pediatric epilepsy on experienced stress and psychopathology in parents. Epilepsy and Behavior, 2020, 113, 107538.	0.9	23
126	The Phenotypic Spectrum of PRRT2-Associated Paroxysmal Neurologic Disorders in Childhood. Biomedicines, 2020, 8, 456.	1.4	23

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127	Clinical and molecular delineation of <scp>PUS3</scp>â€associated neurodevelopmental disorders. Clinical Genetics, 2021, 100, 628-633.	1.0	23
128	Natural History Study of STXBP1-Developmental and Epileptic Encephalopathy Into Adulthood. Neurology, 2022, 99, .	1.5	23
129	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, 165-172.	1.7	22
130	Testing association of rare genetic variants with resistance to three common antiseizure medications. Epilepsia, 2020, 61, 657-666.	2.6	22
131	MECP2 Duplication Syndrome: Evidence of Enhanced Oxidative Stress. A Comparison with Rett Syndrome. PLoS ONE, 2016, 11, e0150101.	1.1	22
132	Evaluation of Presumably Disease Causing SCN1A Variants in a Cohort of Common Epilepsy Syndromes. PLoS ONE, 2016, 11, e0150426.	1.1	22
133	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.	1.1	21
134	Pharmacoresponse in genetic generalized epilepsy: a genome-wide association study. Pharmacogenomics, 2020, 21, 325-335.	0.6	21
135	Integrative approach to interpret DYRK1A variants, leading to a frequent neurodevelopmental disorder. Genetics in Medicine, 2021, 23, 2150-2159.	1.1	21
136	Gain-of-function GABRB3 variants identified in vigabatrin-hypersensitive epileptic encephalopathies. Brain Communications, 2020, 2, fcaa162.	1.5	21
137	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.	2.1	21
138	Epilepsy Genetics and Precision Medicine in Adults: A New Landscape for Developmental and Epileptic Encephalopathies. Frontiers in Neurology, 2022, 13, 777115.	1.1	21
139	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, 1010-1018.	1.4	20
140	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. Annals of Neurology, 2018, 83, 926-934.	2.8	20
141	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	1.1	20
142	Refining Genotypes and Phenotypes in KCNA2-Related Neurological Disorders. International Journal of Molecular Sciences, 2021, 22, 2824.	1.8	20
143	Update on the genetics of the epilepsyâ€aphasia spectrum and role of GRIN2A mutations. Epileptic Disorders, 2019, 21, 41-47.	0.7	20
144	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.	2.7	19

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145	Biallelic inherited SCN8A variants, a rare cause of SCN8A â€related developmental and epileptic encephalopathy. Epilepsia, 2019, 60, 2277-2285.	2.6	18
146	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.	0.7	18
147	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.	1.8	17
148	Atypical Vitamin B₆Deficiency. Journal of Child Neurology, 2014, 29, 704-707.	0.7	16
149	Exome Sequencing Fails to Identify the Genetic Cause of Aicardi Syndrome. Molecular Syndromology, 2016, 7, 234-238.	0.3	16
150	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.	0.9	16
151	Mowatâ€“Wilson syndrome: an underdiagnosed syndrome?. Clinical Genetics, 2008, 73, 579-584.	1.0	15
152	Deciphering the premature mortality in PIGA-CDG â€“ An untold story. Epilepsy Research, 2021, 170, 106530.	0.8	15
153	RHOBTB2 Mutations Expand the Phenotypic Spectrum of Alternating Hemiplegia of Childhood. Neurology, 2021, 96, e1539-e1550.	1.5	15
154	PURA- Related Developmental and Epileptic Encephalopathy. Neurology: Genetics, 2021, 7, e613.	0.9	15
155	Letter to the editor: confirming neonatal seizure and late onset ataxia in SCN2A Ala263Val. Journal of Neurology, 2016, 263, 1459-1460.	1.8	14
156	Characterization of the <scp>GABRB2</scp>â€Associated Neurodevelopmental Disorders. Annals of Neurology, 2021, 89, 573-586.	2.8	14
157	Adult phenotype of KCNQ2 encephalopathy. Journal of Medical Genetics, 2022, 59, 528-535.	1.5	14
158	Two de novo GluN2B mutations affect multiple NMDAR-functions and instigate severe pediatric encephalopathy. ELife, 2021, 10, .	2.8	14
159	STXBP1 Syndrome Is Characterized by Inhibition-Dominated Dynamics of Resting-State EEG. Frontiers in Physiology, 2021, 12, 775172.	1.3	14
160	Impact of Genetic Testing on Therapeutic Decision-Making in Childhood-Onset Epilepsiesâ€”a Study in a Tertiary Epilepsy Center. Neurotherapeutics, 2022, 19, 1353-1367.	2.1	14
161	The role of SLC2A1 in early onset and childhood absence epilepsies. Epilepsy Research, 2013, 105, 229-233.	0.8	13
162	Genetic studies in congenital anterior midline cervical cleft. American Journal of Medical Genetics, Part A, 2012, 158A, 2021-2026.	0.7	12

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163	Mowat-Wilson syndrome: growth charts. Orphanet Journal of Rare Diseases, 2020, 15, 151.	1.2	12
164	Sequence analysis of 17 NRXN1 deletions. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2014, 165, 52-61.	1.1	11
165	Expanding the clinical and EEG spectrum of CNKSR2-related encephalopathy with status epilepticus during slow sleep (ESES). Clinical Neurophysiology, 2020, 131, 1030-1039.	0.7	11
166	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.	0.7	11
167	Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis. Brain, 2022, 145, 2687-2703.	3.7	11
168	Interstitial deletion of chromosome 4p associated with mild mental retardation, epilepsy and polymicrogyria of the left temporal lobe. Clinical Genetics, 2007, 72, 593-598.	1.0	10
169	Balanced translocation in a patient with severe myoclonic epilepsy of infancy disrupts the sodium channel gene SCN1A. Epilepsia, 2008, 49, 1091-1094.	2.6	10
170	Structural mapping of GABRB3 variants reveals genotypeâ€“phenotype correlations. Genetics in Medicine, 2022, 24, 681-693.	1.1	10
171	Genetic paroxysmal neurological disorders featuring episodic ataxia and epilepsy. European Journal of Medical Genetics, 2022, 65, 104450.	0.7	10
172	Duplication of MAOA, MAOB, and NDP in a patient with mental retardation and epilepsy. European Journal of Human Genetics, 2011, 19, 1-2.	1.4	9
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174	Dysregulation of FOXG1 by ring chromosome 14. Molecular Cytogenetics, 2015, 8, 24.	0.4	8
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