

Rikke S MÃ¸ller

List of Publications by Year in descending order

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Version: 2024-02-01

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	Assessing the landscape of <i>STXBP1</i>-related disorders in 534 individuals. <i>Brain</i> , 2022, 145, 1668-1683.	3.7	46
2	Adult phenotype of <i>KCNQ2</i> encephalopathy. <i>Journal of Medical Genetics</i> , 2022, 59, 528-535.	1.5	14
3	Genotype-phenotype correlations in <i>SCN8A</i>-related disorders reveal prognostic and therapeutic implications. <i>Brain</i> , 2022, 145, 2991-3009.	3.7	69
4	Gain-of-function variants in <i>GABRD</i> reveal a novel pathway for neurodevelopmental disorders and epilepsy. <i>Brain</i> , 2022, 145, 1299-1309.	3.7	34
5	<i>De novo FZR1</i> loss-of-function variants cause developmental and epileptic encephalopathies. <i>Brain</i> , 2022, 145, 1684-1697.	3.7	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. <i>Neurotherapeutics</i> , 2022, 19, 334-341.	2.1	21
7	Pyridoxine or pyridoxalâ€”phosphate treatment for seizures in glycosylphosphatidylinositol deficiency: A cohort study. <i>Developmental Medicine and Child Neurology</i> , 2022, 64, 789-798.	1.1	6
8	Association of ultraâ€”rare coding variants with genetic generalized epilepsy: A caseâ€”control whole exome sequencing study. <i>Epilepsia</i> , 2022, 63, 723-735.	2.6	8
9	Development and Validation of a Prediction Model for Early Diagnosis of <i>SCN1A</i>-Related Epilepsies. <i>Neurology</i> , 2022, 98, .	1.5	24
10	OUP accepted manuscript. <i>Brain</i> , 2022, , .	3.7	0
11	Epilepsy Genetics and Precision Medicine in Adults: A New Landscape for Developmental and Epileptic Encephalopathies. <i>Frontiers in Neurology</i> , 2022, 13, 777115.	1.1	21
12	<i>PIGN</i> encephalopathy: Characterizing the epileptology. <i>Epilepsia</i> , 2022, 63, 974-991.	2.6	4
13	Structural mapping of GABRB3 variants reveals genotypeâ€”phenotype correlations. <i>Genetics in Medicine</i> , 2022, 24, 681-693.	1.1	10
14	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> , 2022, 13, 1822.	5.8	32
15	Genetic paroxysmal neurological disorders featuring episodic ataxia and epilepsy. <i>European Journal of Medical Genetics</i> , 2022, 65, 104450.	0.7	10
16	SLC7A3: In Silico Prediction of a Potential New Cause of Childhood Epilepsy. <i>Neuropediatrics</i> , 2022, 53, 046-051.	0.3	0
17	Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis. <i>Brain</i> , 2022, 145, 2687-2703.	3.7	11
18	Biallelic variants in <sc><i>ZNF142</i></sc> lead to a syndromic neurodevelopmental disorder. <i>Clinical Genetics</i> , 2022, 102, 98-109.	1.0	6

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19	Natural History Study of STXBP1-Developmental and Epileptic Encephalopathy Into Adulthood. <i>Neurology</i> , 2022, 99, .	1.5	23
20	Impact of Genetic Testing on Therapeutic Decision-Making in Childhood-Onset Epilepsies—a Study in a Tertiary Epilepsy Center. <i>Neurotherapeutics</i> , 2022, 19, 1353-1367.	2.1	14
21	The de novo <i>GABRA4</i> p.Thr300Ile variant found in a patient with early-onset intractable epilepsy and neurodevelopmental abnormalities displays gain-of-function traits. <i>Epilepsia</i> , 2022, 63, 2439-2441.	2.6	6
22	KCNQ2 R144 variants cause neurodevelopmental disability with language impairment and autistic features without neonatal seizures through a gain-of-function mechanism. <i>EBioMedicine</i> , 2022, 81, 104130.	2.7	19
23	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. <i>Genetics in Medicine</i> , 2021, 23, 363-373.	1.1	28
24	Basal Ganglia Dymorphism in Patients With Aicardi Syndrome. <i>Neurology</i> , 2021, 96, e1319-e1333.	1.5	6
25	Deciphering the premature mortality in PIGA-CDG — An untold story. <i>Epilepsy Research</i> , 2021, 170, 106530.	0.8	15
26	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. <i>Genetics in Medicine</i> , 2021, 23, 653-660.	1.1	20
27	Characterization of the <i>GABRB2</i> —Associated Neurodevelopmental Disorders. <i>Annals of Neurology</i> , 2021, 89, 573-586.	2.8	14
28	Refining Genotypes and Phenotypes in KCNA2-Related Neurological Disorders. <i>International Journal of Molecular Sciences</i> , 2021, 22, 2824.	1.8	20
29	Real-life survey of pitfalls and successes of precision medicine in genetic epilepsies. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2021, 92, 1044-1052.	0.9	30
30	Electroclinical features of MEF2C haploinsufficiency-related epilepsy: A multicenter European study. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2021, 88, 60-72.	0.9	6
31	Deep-Phenotyping the Less Severe Spectrum of PIGT Deficiency and Linking the Gene to Myoclonic Atonic Seizures. <i>Frontiers in Genetics</i> , 2021, 12, 663643.	1.1	6
32	<i>ATP1A2</i> and <i>ATP1A3</i> associated early profound epileptic encephalopathy and polymicrogyria. <i>Brain</i> , 2021, 144, 1435-1450.	3.7	35
33	Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations. <i>Epilepsia</i> , 2021, 62, 1518-1527.	2.6	5
34	<i>KCNT1</i> -related epilepsies and epileptic encephalopathies: phenotypic and mutational spectrum. <i>Brain</i> , 2021, 144, 3635-3650.	3.7	34
35	PRICKLE2 revisited—further evidence implicating PRICKLE2 in neurodevelopmental disorders. <i>European Journal of Human Genetics</i> , 2021, 29, 1235-1244.	1.4	5
36	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> , 2021, 64, 104246.	0.7	11

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37	Two de novo GluN2B mutations affect multiple NMDAR-functions and instigate severe pediatric encephalopathy. <i>ELife</i> , 2021, 10, .	2.8	14
38	Epilepsy Syndromes in the First Year of Life and Usefulness of Genetic Testing for Precision Therapy. <i>Genes</i> , 2021, 12, 1051.	1.0	36
39	<scp><i>ZMYND11</i></scp> variants are a novel cause of centrotemporal and generalised epilepsies with neurodevelopmental disorder. <i>Clinical Genetics</i> , 2021, 100, 412-429.	1.0	5
40	Clinical and molecular delineation of <scp><i>PUS3</i></scp>â€associated neurodevelopmental disorders. <i>Clinical Genetics</i> , 2021, 100, 628-633.	1.0	23
41	Integrative approach to interpret DYRK1A variants, leading to a frequent neurodevelopmental disorder. <i>Genetics in Medicine</i> , 2021, 23, 2150-2159.	1.1	21
42	<i>RHOBTB2</i> Mutations Expand the Phenotypic Spectrum of Alternating Hemiplegia of Childhood. <i>Neurology</i> , 2021, 96, e1539-e1550.	1.5	15
43	The Angelman Syndrome Online Registry â€“ A multilingual approach to support global research. <i>European Journal of Medical Genetics</i> , 2021, 64, 104349.	0.7	1
44	Using common genetic variants to find drugs for common epilepsies. <i>Brain Communications</i> , 2021, 3, fcab287.	1.5	9
45	<i>PURA</i> Related Developmental and Epileptic Encephalopathy. <i>Neurology: Genetics</i> , 2021, 7, e613.	0.9	15
46	STXBP1 Syndrome Is Characterized by Inhibition-Dominated Dynamics of Resting-State EEG. <i>Frontiers in Physiology</i> , 2021, 12, 775172.	1.3	14
47	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> , 2020, 24, 129-133.	0.7	18
48	Recent advances in treatment of epilepsy-related sodium channelopathies. <i>European Journal of Paediatric Neurology</i> , 2020, 24, 123-128.	0.7	40
49	Neonatal developmental and epileptic encephalopathy due to autosomal recessive variants in <i>SLC13A5</i> gene. <i>Epilepsia</i> , 2020, 61, 2474-2485.	2.6	31
50	The impact of severe pediatric epilepsy on experienced stress and psychopathology in parents. <i>Epilepsy and Behavior</i> , 2020, 113, 107538.	0.9	23
51	Current knowledge of SLC6A1-related neurodevelopmental disorders. <i>Brain Communications</i> , 2020, 2, fcaa170.	1.5	44
52	Expanding the clinical and EEG spectrum of CNKSR2-related encephalopathy with status epilepticus during slow sleep (ESES). <i>Clinical Neurophysiology</i> , 2020, 131, 1030-1039.	0.7	11
53	The impact of low-risk genetic variants in self-limited epilepsy with centrotemporal spikes aka Rolandic epilepsy. <i>EBioMedicine</i> , 2020, 58, 102896.	2.7	2
54	Genetic testing in adult epilepsy patients: A call to action for clinicians. <i>Epilepsia</i> , 2020, 61, 2055-2056.	2.6	2

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55	Predicting functional effects of missense variants in voltage-gated sodium and calcium channels. <i>Science Translational Medicine</i> , 2020, 12, .	5.8	84
56	The Phenotypic Spectrum of PRRT2-Associated Paroxysmal Neurologic Disorders in Childhood. <i>Biomedicines</i> , 2020, 8, 456.	1.4	23
57	Utility of genetic testing for therapeutic decision-making in adults with epilepsy. <i>Epilepsia</i> , 2020, 61, 1234-1239.	2.6	60
58	Pharmacoresponse in genetic generalized epilepsy: a genome-wide association study. <i>Pharmacogenomics</i> , 2020, 21, 325-335.	0.6	21
59	Phenotypic and genetic spectrum of epilepsy with myoclonic atonic seizures. <i>Epilepsia</i> , 2020, 61, 995-1007.	2.6	30
60	Lessons learned from 40 novel <i>PIGA</i> patients and a review of the literature. <i>Epilepsia</i> , 2020, 61, 1142-1155.	2.6	32
61	Mowat-Wilson syndrome: growth charts. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 151.	1.2	12
62	Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. <i>Genome Medicine</i> , 2020, 12, 28.	3.6	42
63	Testing association of rare genetic variants with resistance to three common antiseizure medications. <i>Epilepsia</i> , 2020, 61, 657-666.	2.6	22
64	A catalogue of new incidence estimates of monogenic neurodevelopmental disorders caused by de novo variants. <i>Brain</i> , 2020, 143, 1099-1105.	3.7	64
65	Biological concepts in human sodium channel epilepsies and their relevance in clinical practice. <i>Epilepsia</i> , 2020, 61, 387-399.	2.6	65
66	Novel congenital disorder of <i>O</i> -linked glycosylation caused by <i>GALNT2</i> loss of function. <i>Brain</i> , 2020, 143, 1114-1126.	3.7	46
67	Gain-of-function <i>GABRB3</i> variants identified in vigabatrin-hypersensitive epileptic encephalopathies. <i>Brain Communications</i> , 2020, 2, fcaa162.	1.5	21
68	Genotype-phenotype correlations in patients with de novo <i>KCNQ2</i> pathogenic variants. <i>Neurology: Genetics</i> , 2020, 6, e528.	0.9	24
69	Reader response: <i>SYNGAP1</i> encephalopathy: A distinctive generalized developmental and epileptic encephalopathy. <i>Neurology</i> , 2020, 94, 368.2-369.	1.5	2
70	The landscape of epilepsy-related <i>GATOR1</i> variants. <i>Genetics in Medicine</i> , 2019, 21, 398-408.	1.1	137
71	Genetic heterogeneity in infantile spasms. <i>Epilepsy Research</i> , 2019, 156, 106181.	0.8	38
72	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> , 2019, 97, 244-252.	0.9	16

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73	Biallelic inherited SCN8A variants, a rare cause of SCN8A -related developmental and epileptic encephalopathy. <i>Epilepsia</i> , 2019, 60, 2277-2285.	2.6	18
74	The first step towards personalized risk prediction for common epilepsies. <i>Brain</i> , 2019, 142, 3316-3318.	3.7	2
75	From next-generation sequencing to targeted treatment of non-acquired epilepsies. <i>Expert Review of Molecular Diagnostics</i> , 2019, 19, 217-228.	1.5	38
76	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> , 2019, 65, 131-137.	0.9	30
77	A Recurrent Missense Variant in AP2M1 Impairs Clathrin-Mediated Endocytosis and Causes Developmental and Epileptic Encephalopathy. <i>American Journal of Human Genetics</i> , 2019, 104, 1060-1072.	2.6	78
78	Parental mosaicism in epilepsies due to alleged de novo variants. <i>Epilepsia</i> , 2019, 60, e63-e66.	2.6	29
79	Treatment Responsiveness in KCNT1-Related Epilepsy. <i>Neurotherapeutics</i> , 2019, 16, 848-857.	2.1	60
80	First report of the neuropathological findings in a patient with leukodystrophy and compound heterozygous variants in the <i>PIGT</i> gene. <i>Neuropathology and Applied Neurobiology</i> , 2019, 45, 732-735.	1.8	6
81	A novel in-frame mutation in CLN3 leads to Juvenile neuronal ceroid lipofuscinosis in a large Pakistani family. <i>International Journal of Neuroscience</i> , 2019, 129, 890-895.	0.8	0
82	Diagnostic implications of genetic copy number variation in epilepsy plus. <i>Epilepsia</i> , 2019, 60, 689-706.	2.6	61
83	The spectrum of intermediate <i>SCN8A</i> -related epilepsy. <i>Epilepsia</i> , 2019, 60, 830-844.	2.6	70
84	PIGT-CDG, a disorder of the glycosylphosphatidylinositol anchor: description of 13 novel patients and expansion of the clinical characteristics. <i>Genetics in Medicine</i> , 2019, 21, 2216-2223.	1.1	21
85	No evidence for a BRD 2 promoter hypermethylation in blood leukocytes of Europeans with juvenile myoclonic epilepsy. <i>Epilepsia</i> , 2019, 60, e31-e36.	2.6	4
86	Clinical spectrum of <i>STX1B</i> -related epileptic disorders. <i>Neurology</i> , 2019, 92, e1238-e1249.	1.5	43
87	Mild malformations of cortical development in sleep-related hypermotor epilepsy due to <i>KCNT1</i> mutations. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 386-391.	1.7	25
88	Defining and expanding the phenotype of QARS-associated developmental epileptic encephalopathy. <i>Neurology: Genetics</i> , 2019, 5, e373.	0.9	5
89	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> , 2019, 56, 701-710.	1.5	43
90	Spectrum of GABAA receptor variants in epilepsy. <i>Current Opinion in Neurology</i> , 2019, 32, 183-190.	1.8	59

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91	Phenotypic and genetic spectrum of <i>SCN8A</i> -related disorders, treatment options, and outcomes. <i>Epilepsia</i> , 2019, 60, S77-S85.	2.6	58
92	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. <i>Genetics in Medicine</i> , 2019, 21, 837-849.	1.1	47
93	<i>GRIN2A</i> -related disorders: genotype and functional consequence predict phenotype. <i>Brain</i> , 2019, 142, 80-92.	3.7	143
94	Neuronal mechanisms of mutations in <i>SCN8A</i> causing epilepsy or intellectual disability. <i>Brain</i> , 2019, 142, 376-390.	3.7	92
95	Clinician's guide to genes associated with Rett-like phenotypes—Investigation of a Danish cohort and review of the literature. <i>Clinical Genetics</i> , 2019, 95, 221-230.	1.0	26
96	Update on the genetics of the epilepsy-aphasia spectrum and role of <i>GRIN2A</i> mutations. <i>Epileptic Disorders</i> , 2019, 21, 41-47.	0.7	20
97	Early mortality in <i>SCN8A</i> -related epilepsies. <i>Epilepsy Research</i> , 2018, 143, 79-81.	0.8	48
98	Progress in Understanding and Treating <i>SCN2A</i> -Mediated Disorders. <i>Trends in Neurosciences</i> , 2018, 41, 442-456.	4.2	210
99	The epilepsy phenotypic spectrum associated with a recurrent <i>CLUX2</i> variant. <i>Annals of Neurology</i> , 2018, 83, 926-934.	2.8	20
100	Defining the phenotypic spectrum of <i>SLC6A1</i> mutations. <i>Epilepsia</i> , 2018, 59, 389-402.	2.6	99
101	Phenotype and genotype of 87 patients with Mowat-Wilson syndrome and recommendations for care. <i>Genetics in Medicine</i> , 2018, 20, 965-975.	1.1	67
102	Neurologic phenotypes associated with <i>COL4A1</i> / <i>COL4A2</i> mutations. <i>Neurology</i> , 2018, 91, e2078-e2088.	1.5	97
103	Aberrant Inclusion of a Poison Exon Causes Dravet Syndrome and Related <i>SCN1A</i> -Associated Genetic Epilepsies. <i>American Journal of Human Genetics</i> , 2018, 103, 1022-1029.	2.6	76
104	<i>SLC35A2</i> -related congenital disorder of glycosylation: Defining the phenotype. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 1095-1102.	0.7	27
105	The phenotype of <i>SCN8A</i> developmental and epileptic encephalopathy. <i>Neurology</i> , 2018, 91, e1112-e1124.	1.5	114
106	De novo variants in neurodevelopmental disorders with epilepsy. <i>Nature Genetics</i> , 2018, 50, 1048-1053.	9.4	230
107	Analysis of shared heritability in common disorders of the brain. <i>Science</i> , 2018, 360, .	6.0	1,085
108	Rare coding variants in genes encoding GABAA receptors in genetic generalised epilepsies: an exome-based case-control study. <i>Lancet Neurology</i> , The, 2018, 17, 699-708.	4.9	67

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109	Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. <i>Genome Medicine</i> , 2018, 10, 3.	3.6	67
110	Incorporating epilepsy genetics into clinical practice: a 360° evaluation. <i>Npj Genomic Medicine</i> , 2018, 3, 13.	1.7	46
111	Epilepsy in patients with GRIN2A alterations: Genetics, neurodevelopment, epileptic phenotype and response to anticonvulsive drugs. <i>European Journal of Paediatric Neurology</i> , 2017, 21, 530-541.	0.7	37
112	Application of rare variant transmission disequilibrium tests to epileptic encephalopathy trio sequence data. <i>European Journal of Human Genetics</i> , 2017, 25, 894-899.	1.4	7
113	Carbamazepine and oxcarbazepine induced hyponatremia in people with epilepsy. <i>Epilepsia</i> , 2017, 58, 1227-1233.	2.6	54
114	Myoclonus epilepsy and ataxia due to <i>KCNC1</i> mutation: Analysis of 20 cases and <i>K</i> channel properties. <i>Annals of Neurology</i> , 2017, 81, 677-689.	2.8	69
115	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. <i>American Journal of Human Genetics</i> , 2017, 100, 676-688.	2.6	54
116	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. <i>Brain</i> , 2017, 140, 1316-1336.	3.7	426
117	<i>GRIN2B</i> encephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. <i>Journal of Medical Genetics</i> , 2017, 54, 460-470.	1.5	190
118	Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU. <i>Human Genetics</i> , 2017, 136, 463-479.	1.8	66
119	Mutations in <i>GABRB3</i> . <i>Neurology</i> , 2017, 88, 483-492.	1.5	87
120	Delineating SPTAN1 associated phenotypes: from isolated epilepsy to encephalopathy with progressive brain atrophy. <i>Brain</i> , 2017, 140, 2322-2336.	3.7	82
121	Clinical spectrum and genotype-phenotype associations of KCNA2-related encephalopathies. <i>Brain</i> , 2017, 140, 2337-2354.	3.7	117
122	Neurodevelopmental Disorders Caused by De Novo Variants in <i>KCNB1</i> Genotypes and Phenotypes. <i>JAMA Neurology</i> , 2017, 74, 1228.	4.5	79
123	Alternating hemiplegia of childhood and a pathogenic variant of <i>ATP1A3</i> : a case report and pathophysiological considerations. <i>Epileptic Disorders</i> , 2017, 19, 226-230.	0.7	5
124	Filadelfia, Danish Epilepsy Center, Dianalund, Denmark. <i>Epilepsy and Behavior</i> , 2017, 76, S4-S8.	0.9	1
125	<i>DNM1</i> encephalopathy. <i>Neurology</i> , 2017, 89, 385-394.	1.5	87
126	Neuroimaging findings in Mowat-Wilson syndrome: a study of 54 patients. <i>Genetics in Medicine</i> , 2017, 19, 691-700.	1.1	45

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127	Benign infantile seizures and paroxysmal dyskinesia caused by an <i>SCN8A</i> mutation. <i>Annals of Neurology</i> , 2016, 79, 428-436.	2.8	159
128	Delineating the <i>GRIN1</i> phenotypic spectrum. <i>Neurology</i> , 2016, 86, 2171-2178.	1.5	157
129	De Novo Mutations in <i>SLC1A2</i> and <i>CACNA1A</i> Are Important Causes of Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2016, 99, 287-298.	2.6	247
130	Pitfalls in genetic testing: the story of missed <i>SCN1A</i> mutations. <i>Molecular Genetics & Genomic Medicine</i> , 2016, 4, 457-464.	0.6	67
131	Phenotypic spectrum of <i>GABRA1</i> . <i>Neurology</i> , 2016, 87, 1140-1151.	1.5	113
132	Targeted sequencing of 351 candidate genes for epileptic encephalopathy in a large cohort of patients. <i>Molecular Genetics & Genomic Medicine</i> , 2016, 4, 568-580.	0.6	83
133	Exome Sequencing Fails to Identify the Genetic Cause of Aicardi Syndrome. <i>Molecular Syndromology</i> , 2016, 7, 234-238.	0.3	16
134	Gene Panel Testing in Epileptic Encephalopathies and Familial Epilepsies. <i>Molecular Syndromology</i> , 2016, 7, 210-219.	0.3	103
135	Loss of <i>SYNJ1</i> dual phosphatase activity leads to early onset refractory seizures and progressive neurological decline. <i>Brain</i> , 2016, 139, 2420-2430.	3.7	70
136	Germline and somatic mutations in the <i>MTOR</i> gene in focal cortical dysplasia and epilepsy. <i>Neurology: Genetics</i> , 2016, 2, e118.	0.9	125
137	De novo mutations of <i>KIAA2022</i> in females cause intellectual disability and intractable epilepsy. <i>Journal of Medical Genetics</i> , 2016, 53, 850-858.	1.5	47
138	Reply. <i>Annals of Neurology</i> , 2016, 80, 168-169.	2.8	0
139	Letter to the editor: confirming neonatal seizure and late onset ataxia in <i>SCN2A</i> Ala263Val. <i>Journal of Neurology</i> , 2016, 263, 1459-1460.	1.8	14
140	Loss of function of the retinoid-related nuclear receptor (<i>RORB</i>) gene and epilepsy. <i>European Journal of Human Genetics</i> , 2016, 24, 1761-1770.	1.4	36
141	Precision Medicine: <i>SCN8A</i> Encephalopathy Treated with Sodium Channel Blockers. <i>Neurotherapeutics</i> , 2016, 13, 190-191.	2.1	38
142	<i>STXBP1</i> encephalopathy. <i>Neurology</i> , 2016, 86, 954-962.	1.5	264
143	<i>MECP2</i> Duplication Syndrome: Evidence of Enhanced Oxidative Stress. A Comparison with Rett Syndrome. <i>PLoS ONE</i> , 2016, 11, e0150101.	1.1	22
144	Evaluation of Presumably Disease Causing <i>SCN1A</i> Variants in a Cohort of Common Epilepsy Syndromes. <i>PLoS ONE</i> , 2016, 11, e0150426.	1.1	22

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145	Clinical Phenotype of De Novo <i>GNAO1</i> Mutation. <i>Child Neurology Open</i> , 2015, 2, 2329048X1558371.	0.5	25
146	The role of <i>SLC2A1</i> mutations in myoclonic astatic epilepsy and absence epilepsy, and the estimated frequency of <i>GLUT1</i> deficiency syndrome. <i>Epilepsia</i> , 2015, 56, e203-8.	2.6	71
147	Dysregulation of <i>FOXP1</i> by ring chromosome 14. <i>Molecular Cytogenetics</i> , 2015, 8, 24.	0.4	8
148	The contribution of next generation sequencing to epilepsy genetics. <i>Expert Review of Molecular Diagnostics</i> , 2015, 15, 1531-1538.	1.5	68
149	The phenotypic spectrum of <i>SCN8A</i> encephalopathy. <i>Neurology</i> , 2015, 84, 480-489.	1.5	246
150	<i>CHD2</i> variants are a risk factor for photosensitivity in epilepsy. <i>Brain</i> , 2015, 138, 1198-1208.	3.7	112
151	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. <i>Human Molecular Genetics</i> , 2015, 24, 5250-5259.	1.4	93
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154	De novo loss- or gain-of-function mutations in <i>KCNA2</i> cause epileptic encephalopathy. <i>Nature Genetics</i> , 2015, 47, 393-399.	9.4	224
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157	The incidence of <i>SCN1A</i> -related Dravet syndrome in Denmark is 1:22,000: A population-based study from 2004 to 2009. <i>Epilepsia</i> , 2015, 56, e36-9.	2.6	103
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186	Genome-wide linkage meta-analysis identifies susceptibility loci at 2q34 and 13q31.3 for genetic generalized epilepsies. <i>Epilepsia</i> , 2012, 53, 308-318.	2.6	32
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