Ingo Helbig

List of Publications by Year in descending order

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206 papers	14,638 citations	25014 57 h-index	25770 108 g-index
236 all docs	236 docs citations	236 times ranked	17603 citing authors

#	Article	IF	Citations
1	Assessing the landscape of STXBP1-related disorders in 534 individuals. Brain, 2022, 145, 1668-1683.	3.7	46
2	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. Brain, 2022, 145, 2991-3009.	3.7	69
3	Atypical development of Brocaâ€™s area in a large family with inherited stuttering. Brain, 2022, 145, 1177-1188.	3.7	6
4	De novo FZR1 loss-of-function variants cause developmental and epileptic encephalopathies. Brain, 2022, 145, 1684-1697.	3.7	5
5	Association of ultraâ€rare coding variants with genetic generalized epilepsy: A caseâ€“control whole exome sequencing study. Epilepsia, 2022, 63, 723-735.	2.6	8
6	Molecular Diagnostic Outcomes from 700 Cases. Journal of Molecular Diagnostics, 2022, 24, 274-286.	1.2	7
7	High-throughput evaluation of epilepsy-associated KCNQ2 variants reveals functional and pharmacological heterogeneity. JCI Insight, 2022, 7, .	2.3	27
8	Multicenter Study of the Impact of COVID-19 Shelter-In-Place on Tertiary Hospital-based Care for Pediatric Neurologic Disease. Neurohospitalist, The, 2022, 12, 194187442110630.	0.3	0
9	Caregiver assessment of quality of life in individuals with genetic developmental and epileptic encephalopathies. Developmental Medicine and Child Neurology, 2022, 64, 957-964.	1.1	11
10	Spectrum of Phenotypic, Genetic, and Functional Characteristics in Patients With Epilepsy With KCNC2 Pathogenic Variants. Neurology, 2022, 98, .	1.5	11
11	Computational analysis of neurodevelopmental phenotypes: Harmonization empowers clinical discovery. Human Mutation, 2022, 43, 1642-1658.	1.1	10
12	SvAnna: efficient and accurate pathogenicity prediction of coding and regulatory structural variants in long-read genome sequencing. Genome Medicine, 2022, 14, 44.	3.6	7
13	Visits of concern in child neurology telemedicine. Developmental Medicine and Child Neurology, 2022, 64, 1351-1358.	1.1	5
14	Whole exome sequencing and coâ€expression analysis identify an SCN1A variant that modifies pathogenicity in a family with genetic epilepsy and febrile seizures plus. Epilepsia, 2022, 63, 1970-1980.	2.6	2
15	Epilepsy Course and Developmental Trajectories in STXBP1 -DEE. Neurology: Genetics, 2022, 8, .	0.9	24
16	Base editing the synapse: Modeling a complex neurological disorder in non-human primates. Molecular Therapy, 2022, 30, 2114-2116.	3.7	0
17	Natural History Study of STXBP1-Developmental and Epileptic Encephalopathy Into Adulthood. Neurology, 2022, 99, .	1.5	23
18	Common risk variants for epilepsy are enriched in families previously targeted for rare monogenic variant discovery. EBioMedicine, 2022, 81, 104079.	2.7	10

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19	Precision medicine for genetic epilepsy on the horizon: Recent advances, present challenges, and suggestions for continued progress. Epilepsia, 2022, 63, 2461-2475.	2.6	50
20	Predicting the functional effects of voltage-gated potassium channel missense variants with multi-task learning. EBioMedicine, 2022, 81, 104115.	2.7	8
21	Ketamine for Management of Neonatal and Pediatric Refractory Status Epilepticus. Neurology, 2022, 99, .	1.5	18
22	Child Neurology Telemedicine: Understanding the Data We Have and Finding the Patients We Do Not See. Pediatric Neurology, 2021, 116, 84.	1.0	8
23	The Human Phenotype Ontology in 2021. Nucleic Acids Research, 2021, 49, D1207-D1217.	6.5	652
24	Computational analysis of 10,860 phenotypic annotations in individuals with SCN2A-related disorders. Genetics in Medicine, 2021, 23, 1263-1272.	1.1	38
25	Phenotypic homogeneity in childhood epilepsies evolves in gene-specific patterns across 3251 patient-years of clinical data. European Journal of Human Genetics, 2021, 29, 1690-1700.	1.4	13
26	Agammaglobulinemia with normal B-cell numbers in a patient lacking Bob1. Journal of Allergy and Clinical Immunology, 2021, 147, 1977-1980.	1.5	12
27	Modeling seizures in the Human Phenotype Ontology according to contemporary ILAE concepts makes big phenotypic data tractable. Epilepsia, 2021, 62, 1293-1305.	2.6	15
28	Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations. Epilepsia, 2021, 62, 1518-1527.	2.6	5
29	Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. American Journal of Human Genetics, 2021, 108, 965-982.	2.6	35
30	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. American Journal of Human Genetics, 2021, 108, 1138-1150.	2.6	17
31	Assessing seizure burden in pediatric epilepsy using an electronic medical recordâ€“based tool through a common data element approach. Epilepsia, 2021, 62, 1617-1628.	2.6	19
32	Clinical Phenotypic Spectrum of 4095 Individuals with Down Syndrome from Text Mining of Electronic Health Records. Genes, 2021, 12, 1159.	1.0	6
33	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	9.4	44
34	Long-Term Risk of Epilepsy After Pediatric Stroke and Potential Genetic Vulnerabilities. Stroke, 2021, 52, 3541-3542.	1.0	1
35	The dose makes the poisonâ€”Novel insights into Dravet syndrome and SCN1A regulation through nonproductive splicing. PLoS Genetics, 2021, 17, e1009214.	1.5	4
36	Design and implementation of electronic health record common data elements for pediatric epilepsy: Foundations for a learning health care system. Epilepsia, 2021, 62, 198-216.	2.6	30

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37	Variants in ATP6V0A1 cause progressive myoclonus epilepsy and developmental and epileptic encephalopathy. Brain Communications, 2021, 3, fcab245.	1.5	10
38	Using common genetic variants to find drugs for common epilepsies. Brain Communications, 2021, 3, fcab287.	1.5	9
39	PURA- Related Developmental and Epileptic Encephalopathy. Neurology: Genetics, 2021, 7, e613.	0.9	15
40	Bi-allelic variants in OGDHL cause a neurodevelopmental spectrum disease featuring epilepsy, hearing loss, visual impairment, and ataxia. American Journal of Human Genetics, 2021, 108, 2368-2384.	2.6	12
41	The Genomics Research and Innovation Network: creating an interoperable, federated, genomics learning system. Genetics in Medicine, 2020, 22, 371-380.	1.1	30
42	The Monarch Initiative in 2019: an integrative data and analytic platform connecting phenotypes to genotypes across species. Nucleic Acids Research, 2020, 48, D704-D715.	6.5	178
43	RARS1â€related hypomyelinating leukodystrophy: Expanding the spectrum. Annals of Clinical and Translational Neurology, 2020, 7, 83-93.	1.7	18
44	Wholeâ€exome and HLA sequencing in Febrile infectionâ€related epilepsy syndrome. Annals of Clinical and Translational Neurology, 2020, 7, 1429-1435.	1.7	15
45	Current knowledge of SLC6A1-related neurodevelopmental disorders. Brain Communications, 2020, 2, fcaa170.	1.5	44
46	Semantic Similarity Analysis Reveals Robust Gene-Disease Relationships in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2020, 107, 683-697.	2.6	23
47	A longitudinal footprint of genetic epilepsies using automated electronic medical record interpretation. Genetics in Medicine, 2020, 22, 2060-2070.	1.1	22
48	Phenotypic and genetic spectrum of epilepsy with myoclonic atonic seizures. Epilepsia, 2020, 61, 995-1007.	2.6	30
49	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	2.6	32
50	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	3.7	47
51	<scp>SCN3A</scp>â€Related Neurodevelopmental Disorder: A Spectrum of Epilepsy and Brain Malformation. Annals of Neurology, 2020, 88, 348-362.	2.8	42
52	Analyzing 2,589 child neurology telehealth encounters necessitated by the COVID-19 pandemic. Neurology, 2020, 95, e1257-e1266.	1.5	108
53	Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. Genome Medicine, 2020, 12, 28.	3.6	42
54	Phenotypic and Imaging Spectrum Associated With WDR45. Pediatric Neurology, 2020, 109, 56-62.	1.0	16

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55	Whole-Exome Sequencing in NF1-Related West Syndrome Leads to the Identification of KCNC2 as a Novel Candidate Gene for Epilepsy. Neuropediatrics, 2020, 51, 368-372.	0.3	15
56	Biological concepts in human sodium channel epilepsies and their relevance in clinical practice. Epilepsia, 2020, 61, 387-399.	2.6	65
57	Personalized medicine in genetic epilepsies â€“ possibilities, challenges, and new frontiers. Neuropharmacology, 2020, 172, 107970.	2.0	35
58	Bi-allelic ADARB1 Variants Associated with Microcephaly, Intellectual Disability, and Seizures. American Journal of Human Genetics, 2020, 106, 467-483.	2.6	31
59	Bi-allelic GAD1 variants cause a neonatal onset syndromic developmental and epileptic encephalopathy. Brain, 2020, 143, 1447-1461.	3.7	18
60	Early-onset genetic epilepsies reaching adult clinics. Brain, 2020, 143, e19-e19.	3.7	10
61	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	5.8	35
62	A Framework for Analysis, Ontological Evaluation, and Visualization in Preparation to Predictive Analytics in Pediatric Brain Tumor Research. , 2020, , .		1
63	Genetic heterogeneity in infantile spasms. Epilepsy Research, 2019, 156, 106181.	0.8	38
64	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. American Journal of Human Genetics, 2019, 105, 267-282.	2.6	237
65	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	5.8	99
66	Inclusion of hemimegalencephaly into the phenotypic spectrum of <scp>NPRL</scp>3 pathogenic variants in familial focal epilepsy with variable foci. Epilepsia, 2019, 60, e67-e73.	2.6	16
67	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
68	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	2.6	56
69	Treatment Responsiveness in KCNT1-Related Epilepsy. Neurotherapeutics, 2019, 16, 848-857.	2.1	60
70	Targeted gene sequencing in 6994 individuals with neurodevelopmental disorder with epilepsy. Genetics in Medicine, 2019, 21, 2496-2503.	1.1	45
71	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	2.6	61
72	Use of a Dynamic Genetic Testing Approach for Childhood-Onset Epilepsy. JAMA Network Open, 2019, 2, e192129.	2.8	45

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73	The spectrum of intermediate <scp>SCN</scp>8Aâ€related epilepsy. Epilepsia, 2019, 60, 830-844.	2.6	70
74	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
75	The Pediatric Cell Atlas: Defining the Growth Phase of Human Development at Single-Cell Resolution. Developmental Cell, 2019, 49, 10-29.	3.1	57
76	The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. Epilepsia, 2019, 60, 797-806.	2.6	52
77	Clinical spectrum of STX1B -related epileptic disorders. Neurology, 2019, 92, e1238-e1249.	1.5	43
78	Biallelic VARS variants cause developmental encephalopathy with microcephaly that is recapitulated in vars knockout zebrafish. Nature Communications, 2019, 10, 708.	5.8	40
79	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. Genetics in Medicine, 2019, 21, 837-849.	1.1	47
80	GRIN2A-related disorders: genotype and functional consequence predict phenotype. Brain, 2019, 142, 80-92.	3.7	143
81	TANGO2: expanding the clinical phenotype and spectrum of pathogenic variants. Genetics in Medicine, 2019, 21, 601-607.	1.1	41
82	Mutations in SCN3A cause early infantile epileptic encephalopathy. Annals of Neurology, 2018, 83, 703-717.	2.8	69
83	Early mortality in SCN8A -related epilepsies. Epilepsy Research, 2018, 143, 79-81.	0.8	48
84	Commentary: The genetic architecture of the epilepsies, as told by 8500 gene panels. Epilepsia, 2018, 59, 1072-1073.	2.6	0
85	Teaching NeuroImages: Atrophy in epileptic encephalopathy. Neurology, 2018, 90, e442-e443.	1.5	1
86	Defining the phenotypic spectrum of SLC6A1 mutations. Epilepsia, 2018, 59, 389-402.	2.6	99
87	Genetic test utilization and diagnostic yield in adult patients with neurological disorders. Neurogenetics, 2018, 19, 105-110.	0.7	20
88	Mutations in PMPCB Encoding the Catalytic Subunit of the Mitochondrial Presequence Protease Cause Neurodegeneration in Early Childhood. American Journal of Human Genetics, 2018, 102, 557-573.	2.6	69
89	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
90	The ClinGen Epilepsy Gene Curation Expert Panelâ€"Bridging the divide between clinical domain knowledge and formal gene curation criteria. Human Mutation, 2018, 39, 1476-1484.	1.1	33

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91	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. American Journal of Human Genetics, 2018, 103, 666-678.	2.6	87
92	The phenotype of SCN8A developmental and epileptic encephalopathy. Neurology, 2018, 91, e1112-e1124.	1.5	114
93	Variants in the ATP1A3 Gene Mutations within Severe Apnea Starting in Early Infancy: An Observational Study of Two Cases with a Possible Relation to Epileptic Activity. Neuropediatrics, 2018, 49, 342-346.	0.3	6
94	PCDH19 -related epilepsy in a male with Klinefelter syndrome: Additional evidence supporting PCDH19 cellular interference disease mechanism. Epilepsy Research, 2018, 145, 89-92.	0.8	20
95	De novo variants in neurodevelopmental disorders with epilepsy. Nature Genetics, 2018, 50, 1048-1053.	9.4	230
96	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
97	Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. Genome Medicine, 2018, 10, 3.	3.6	67
98	Genetic literacy series: Primer part 2â€"Paradigm shifts in epilepsy genetics. Epilepsia, 2018, 59, 1138-1147.	2.6	17
99	Reply to â€œRecurrent SCN3A p.Ile875Thr variant in patients with polymicrogyriaâ€• Annals of Neurology, 2018, 84, 161-161.	2.8	1
100	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	6.5	699
101	Epileptic Encephalopathiesâ€"Clinical Syndromes and Pathophysiological Concepts. Current Neurology and Neuroscience Reports, 2017, 17, 10.	2.0	23
102	Gene expression analysis in untreated absence epilepsy demonstrates an inconsistent pattern. Epilepsy Research, 2017, 132, 84-90.	0.8	7
103	The phenotypic spectrum of ARHGEF9 includes intellectual disability, focal epilepsy and febrile seizures. Journal of Neurology, 2017, 264, 1421-1425.	1.8	20
104	Application of rare variant transmission disequilibrium tests to epileptic encephalopathy trio sequence data. European Journal of Human Genetics, 2017, 25, 894-899.	1.4	7
105	Advancing the phenome alongside the genome in epilepsy studies. Neurology, 2017, 89, 14-15.	1.5	4
106	Recessive mutations in SLC35A3 cause early onset epileptic encephalopathy with skeletal defects. American Journal of Medical Genetics, Part A, 2017, 173, 1119-1123.	0.7	16
107	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
108	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. Brain, 2017, 140, 1316-1336.	3.7	426

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109	Understanding Genetic Test Results in Childhood Epilepsies. Current Pediatrics Reports, 2017, 5, 24-29.	1.7	0
110	Mutations in GABRB3. Neurology, 2017, 88, 483-492.	1.5	87
111	Heterogeneous contribution of microdeletions in the development of common generalised and focal epilepsies. Journal of Medical Genetics, 2017, 54, 598-606.	1.5	22
112	Epileptic encephalopathy, movement disorder, and the yin and yang of GNAO1 function. Neurology, 2017, 89, 754-755.	1.5	7
113	Reply. Annals of Neurology, 2017, 81, 328-329.	2.8	0
114	Clinical spectrum and genotypeâ€“phenotype associations of KCNA2-related encephalopathies. Brain, 2017, 140, 2337-2354.	3.7	117
115	Neurodevelopmental Disorders Caused by De Novo Variants in KCNB1 Genotypes and Phenotypes. JAMA Neurology, 2017, 74, 1228.	4.5	79
116	Epileptic Encephalopathies as Neurodegenerative Disorders. Advances in Neurobiology, 2017, 15, 295-315.	1.3	9
117	DNM1 encephalopathy. Neurology, 2017, 89, 385-394.	1.5	87
118	De novo GABRG2mutations associated with epileptic encephalopathies. Brain, 2017, 140, 49-67.	3.7	80
119	Precision medicine in genetic epilepsies: break of dawn?. Expert Review of Neurotherapeutics, 2017, 17, 381-392.	1.4	57
120	Attitudes toward Epilepsy Genetics Testing among Adult and Pediatric Epileptologistsâ€”Results of a Q-PULSE Survey. Epilepsy Currents, 2016, 16, 46-47.	0.4	6
121	Rare Noncoding Mutations Extend the Mutational Spectrum in the PGAP3 Subtype of Hyperphosphatasia with Mental Retardation Syndrome. Human Mutation, 2016, 37, 737-744.	1.1	46
122	Identity by descent fine mapping of familial adult myoclonus epilepsy (FAME) to 2p11.2â€“2q11.2. Human Genetics, 2016, 135, 1117-1125.	1.8	29
123	Seizures as presenting and prominent symptom in choreaâ€acanthocytosis with c.2343del <scp>VPS</scp>13A gene mutation. Epilepsia, 2016, 57, 549-556.	2.6	16
124	Delineating the GRIN1 phenotypic spectrum. Neurology, 2016, 86, 2171-2178.	1.5	157
125	Idiopathic focal epilepsies: the â€œlost tribeâ€. Epileptic Disorders, 2016, 18, 252-288.	0.7	65
126	Pitfalls in genetic testing: the story of missed SCN1A mutations. Molecular Genetics & Genomic Medicine, 2016, 4, 457-464.	0.6	67

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127	Primer Part 1â€"The building blocks of epilepsy genetics. Epilepsia, 2016, 57, 861-868.	2.6	77
128	Phenotypic spectrum of GABRA1. Neurology, 2016, 87, 1140-1151.	1.5	113
129	A recurrent mutation in KCNA2 as a novel cause of hereditary spastic paraplegia and ataxia. Annals of Neurology, 2016, 80, .	2.8	49
130	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
131	Understanding Genotypes and Phenotypes in Epileptic Encephalopathies. Molecular Syndromology, 2016, 7, 172-181.	0.3	97
132	Loss of SYNJ1 dual phosphatase activity leads to early onset refractory seizures and progressive neurological decline. Brain, 2016, 139, 2420-2430.	3.7	70
133	SPATA5 mutations cause a distinct autosomal recessive phenotype of intellectual disability, hypotonia and hearing loss. Orphanet Journal of Rare Diseases, 2016, 11, 130.	1.2	19
134	De novo mutations ofKIAA2022in females cause intellectual disability and intractable epilepsy. Journal of Medical Genetics, 2016, 53, 850-858.	1.5	47
135	Genome-wide rare copy number variation screening in ulcerative colitis identifies potential susceptibility loci. BMC Medical Genetics, 2016, 17, 26.	2.1	14
136	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
137	Diagnostic exome sequencing provides a molecular diagnosis for a significant proportion of patients with epilepsy. Genetics in Medicine, 2016, 18, 898-905.	1.1	299
138	Autosomal dominant epilepsy with auditory features: a new LGI1 family including a phenocopy with cortical dysplasia. Journal of Neurology, 2016, 263, 11-16.	1.8	10
139	STXBP1 encephalopathy. Neurology, 2016, 86, 954-962.	1.5	264
140	Retrospective evaluation of low long-term efficacy of antiepileptic drugs and ketogenic diet in 39 patients with CDKL5-related epilepsy. European Journal of Paediatric Neurology, 2016, 20, 147-151.	0.7	56
141	Structural genomic variants in pediatric seizure disorders. Journal of Pediatric Epilepsy, 2015, 01, 161-169.	0.1	0
142	Simultaneous impairment of neuronal and metabolic function of mutated gephyrin in a patient with epileptic encephalopathy. EMBO Molecular Medicine, 2015, 7, 1580-1594.	3.3	39
143	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
144	Commentary: Pathogenic EFHC1 mutations are tolerated in healthy individuals dependent on reported ancestry. Epilepsia, 2015, 56, 195-196.	2.6	7

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145	The contribution of next generation sequencing to epilepsy genetics. Expert Review of Molecular Diagnostics, 2015, 15, 1531-1538.	1.5	68
146	Investigation of GRIN2A in common epilepsy phenotypes. Epilepsy Research, 2015, 115, 95-99.	0.8	44
147	Galanin pathogenic mutations in temporal lobe epilepsy. Human Molecular Genetics, 2015, 24, 3082-3091.	1.4	23
148	Genetic Causes of Generalized Epilepsies. Seminars in Neurology, 2015, 35, 288-292.	0.5	44
149	CHD2variants are a risk factor for photosensitivity in epilepsy. Brain, 2015, 138, 1198-1208.	3.7	112
150	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. American Journal of Human Genetics, 2015, 96, 808-815.	2.6	173
151	Burden Analysis of Rare Microdeletions Suggests a Strong Impact of Neurodevelopmental Genes in Genetic Generalised Epilepsies. PLoS Genetics, 2015, 11, e1005226.	1.5	91
152	De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy. Nature Genetics, 2015, 47, 393-399.	9.4	224
153	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
154	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
155	Investigating the genetic basis of feverâ€associated syndromic epilepsies using copy number variation analysis. Epilepsia, 2015, 56, e26-32.	2.6	16
156	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. Human Molecular Genetics, 2014, 23, 6069-6080.	1.4	61
157	Broad Phenotypic Heterogeneity due to a Novel SCN1A Mutation in a Family With Genetic Epilepsy With Febrile Seizures Plus. Journal of Child Neurology, 2014, 29, 221-226.	0.7	48
158	Status Epilepticus and Refractory Status Epilepticus Management. Seminars in Pediatric Neurology, 2014, 21, 263-274.	1.0	46
159	Iterative phenotyping of 15q11.2, 15q13.3 and 16p13.11 microdeletion carriers in pediatric epilepsies. Epilepsy Research, 2014, 108, 109-116.	0.8	21
160	De novo mutations in HCN1 cause early infantile epileptic encephalopathy. Nature Genetics, 2014, 46, 640-645.	9.4	192
161	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. Nucleic Acids Research, 2014, 42, D966-D974.	6.5	698
162	PRRT2 mutations: exploring the phenotypical boundaries. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 462-465.	0.9	27

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163	Mutations in STX1B, encoding a presynaptic protein, cause fever-associated epilepsy syndromes. Nature Genetics, 2014, 46, 1327-1332.	9.4	178
164	Atypical Vitamin B₆Deficiency. Journal of Child Neurology, 2014, 29, 704-707.	0.7	16
165	De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	2.6	388
166	New technologies in molecular genetics. Progress in Brain Research, 2014, 213, 253-278.	0.9	6
167	GABRA1 and STXBP1 : Novel genetic causes of Dravet syndrome. Neurology, 2014, 82, 1245-1253.	1.5	229
168	Structural genomic variation in childhood epilepsies with complex phenotypes. European Journal of Human Genetics, 2014, 22, 896-901.	1.4	28
169	The unexpected role of copy number variations in juvenile myoclonic epilepsy. Epilepsy and Behavior, 2013, 28, S66-S68.	0.9	16
170	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
171	Mutations in GRIN2A cause idiopathic focal epilepsy with rolandic spikes. Nature Genetics, 2013, 45, 1067-1072.	9.4	391
172	CDKL5 Mutations as a Cause of Severe Epilepsy in Infancy. Journal of Child Neurology, 2013, 28, 937-941.	0.7	17
173	The role of SLC2A1 in early onset and childhood absence epilepsies. Epilepsy Research, 2013, 105, 229-233.	0.8	13
174	Clarifying the role of the 22q11.2 microdeletion in juvenile myoclonic epilepsy. Epilepsy and Behavior, 2013, 29, 589-590.	0.9	4
175	Consensus on diagnosis and management of JME: From founder's observations to current trends. Epilepsy and Behavior, 2013, 28, S87-S90.	0.9	142
176	15q13.3 microdeletions in a prospectively recruited cohort of patients with idiopathic generalized epilepsy in Bulgaria. Epilepsy Research, 2013, 104, 241-245.	0.8	8
177	Genetics of febrile seizure subtypes and syndromes: A twin study. Epilepsy Research, 2013, 105, 103-109.	0.8	36
178	Familial cosegregation of rare genetic variants with disease in complex disorders. European Journal of Human Genetics, 2013, 21, 444-450.	1.4	14
179	Genetics of the epilepsies. Current Opinion in Neurology, 2013, 26, 179-185.	1.8	75
180	Variability of <scp>EEG</scp>â€f<scp>MRI</scp> findings in patients with <scp>SCN1A</scp>â€positive <scp>D</scp>ravet syndrome. Epilepsia, 2013, 54, 918-926.	2.6	26

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