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	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	6.5	699
2	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. Nucleic Acids Research, 2014, 42, D966-D974.	6.5	698
3	The Human Phenotype Ontology in 2021. Nucleic Acids Research, 2021, 49, D1207-D1217.	6.5	652
4	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. Nature Genetics, 2009, 41, 160-162.	9.4	511
5	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. Brain, 2017, 140, 1316-1336.	3.7	426
6	Genome-Wide Copy Number Variation in Epilepsy: Novel Susceptibility Loci in Idiopathic Generalized and Focal Epilepsies. PLoS Genetics, 2010, 6, e1000962.	1.5	414
7	Recurrent microdeletions at 15q11.2 and 16p13.11 predispose to idiopathic generalized epilepsies. Brain, 2010, 133, 23-32.	3.7	406
8	Mutations in GRIN2A cause idiopathic focal epilepsy with rolandic spikes. Nature Genetics, 2013, 45, 1067-1072.	9.4	391
9	De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	2.6	388
10	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
11	STXBP1 encephalopathy. Neurology, 2016, 86, 954-962.	1.5	264
12	Navigating the channels and beyond: unravelling the genetics of the epilepsies. Lancet Neurology, The, 2008, 7, 231-245.	4.9	249
13	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
14	De novo variants in neurodevelopmental disorders with epilepsy. Nature Genetics, 2018, 50, 1048-1053.	9.4	230
15	GABRA1 and STXBP1 : Novel genetic causes of Dravet syndrome. Neurology, 2014, 82, 1245-1253.	1.5	229
16	De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy. Nature Genetics, 2015, 47, 393-399.	9.4	224
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	De novo mutations in HCN1 cause early infantile epileptic encephalopathy. Nature Genetics, 2014, 46, 640-645.	9.4	192

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19	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
20	Recurrent 200-kb deletions of 16p11.2 that include the SH2B1 gene are associated with developmental delay and obesity. Genetics in Medicine, 2010, 12, 641-647.	1.1	178
21	Mutations in STX1B, encoding a presynaptic protein, cause fever-associated epilepsy syndromes. Nature Genetics, 2014, 46, 1327-1332.	9.4	178
22	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
23	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. American Journal of Human Genetics, 2015, 96, 808-815.	2.6	173
24	Delineating the GRIN1 phenotypic spectrum. Neurology, 2016, 86, 2171-2178.	1.5	157
25	Connexin36 Mediates Spike Synchrony in Olfactory Bulb Glomeruli. Neuron, 2005, 46, 761-772.	3.8	152
26	GRIN2A-related disorders: genotype and functional consequence predict phenotype. Brain, 2019, 142, 80-92.	3.7	143
27	Consensus on diagnosis and management of JME: From founder's observations to current trends. Epilepsy and Behavior, 2013, 28, S87-S90.	0.9	142
28	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
29	Clinical spectrum and genotypeâ€“phenotype associations of KCNA2-related encephalopathies. Brain, 2017, 140, 2337-2354.	3.7	117
30	The phenotype of SCN8A developmental and epileptic encephalopathy. Neurology, 2018, 91, e1112-e1124.	1.5	114
31	Phenotypic spectrum of GABRA1. Neurology, 2016, 87, 1140-1151.	1.5	113
32	CHD2variants are a risk factor for photosensitivity in epilepsy. Brain, 2015, 138, 1198-1208.	3.7	112
33	Analyzing 2,589 child neurology telehealth encounters necessitated by the COVID-19 pandemic. Neurology, 2020, 95, e1257-e1266.	1.5	108
34	Defining the phenotypic spectrum of SLC6A1 mutations. Epilepsia, 2018, 59, 389-402.	2.6	99
35	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	5.8	99
36	Understanding Genotypes and Phenotypes in Epileptic Encephalopathies. Molecular Syndromology, 2016, 7, 172-181.	0.3	97

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37	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
38	Burden Analysis of Rare Microdeletions Suggests a Strong Impact of Neurodevelopmental Genes in Genetic Generalised Epilepsies. PLoS Genetics, 2015, 11, e1005226.	1.5	91
39	Deletions in 16p13 including GRIN2A in patients with intellectual disability, various dysmorphic features, and seizure disorders of the rolandic region. Epilepsia, 2010, 51, 1870-1873.	2.6	87
40	Mutations in GABRB3. Neurology, 2017, 88, 483-492.	1.5	87
41	DNM1 encephalopathy. Neurology, 2017, 89, 385-394.	1.5	87
42	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
43	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
44	De novo GABRG2mutations associated with epileptic encephalopathies. Brain, 2017, 140, 49-67.	3.7	80
45	Neurodevelopmental Disorders Caused by De Novo Variants in KCNB1 Genotypes and Phenotypes. JAMA Neurology, 2017, 74, 1228.	4.5	79
46	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
47	Primer Part 1â€”The building blocks of epilepsy genetics. Epilepsia, 2016, 57, 861-868.	2.6	77
48	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
49	Genetics of the epilepsies. Current Opinion in Neurology, 2013, 26, 179-185.	1.8	75
50	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
51	Loss of SYNJ1 dual phosphatase activity leads to early onset refractory seizures and progressive neurological decline. Brain, 2016, 139, 2420-2430.	3.7	70
52	The spectrum of intermediate <scp>SCN</scp>8Aâ€related epilepsy. Epilepsia, 2019, 60, 830-844.	2.6	70
53	Mutations in SCN3A cause early infantile epileptic encephalopathy. Annals of Neurology, 2018, 83, 703-717.	2.8	69
54	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

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55	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. Brain, 2022, 145, 2991-3009.	3.7	69
56	The contribution of next generation sequencing to epilepsy genetics. Expert Review of Molecular Diagnostics, 2015, 15, 1531-1538.	1.5	68
57	Pitfalls in genetic testing: the story of missed SCN1A mutations. Molecular Genetics & Genomic Medicine, 2016, 4, 457-464.	0.6	67
58	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
59	Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. Genome Medicine, 2018, 10, 3.	3.6	67
60	Idiopathic focal epilepsies: the â€œlost tribeâ€• Epileptic Disorders, 2016, 18, 252-288.	0.7	65
61	Biological concepts in human sodium channel epilepsies and their relevance in clinical practice. Epilepsia, 2020, 61, 387-399.	2.6	65
62	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. Human Molecular Genetics, 2014, 23, 6069-6080.	1.4	61
63	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	2.6	61
64	Treatment Responsiveness in KCNT1-Related Epilepsy. Neurotherapeutics, 2019, 16, 848-857.	2.1	60
65	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
66	Exonâ€disrupting deletions of <scp>NRXN1</scp> in idiopathic generalized epilepsy. Epilepsia, 2013, 54, 256-264.	2.6	59
67	Precision medicine in genetic epilepsies: break of dawn?. Expert Review of Neurotherapeutics, 2017, 17, 381-392.	1.4	57
68	The Pediatric Cell Atlas: Defining the Growth Phase of Human Development at Single-Cell Resolution. Developmental Cell, 2019, 49, 10-29.	3.1	57
69	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
70	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	2.6	56
71	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
72	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

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73	The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. Epilepsia, 2019, 60, 797-806.	2.6	52
74	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
75	Febrile infectionâ€related epilepsy syndrome (FIRES) is not caused by SCN1A, POLG, PCDH19 mutations or rare copy number variations. Developmental Medicine and Child Neurology, 2012, 54, 1144-1148.	1.1	49
76	A recurrent mutation in KCNA2 as a novel cause of hereditary spastic paraplegia and ataxia. Annals of Neurology, 2016, 80, .	2.8	49
77	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
78	Early mortality in SCN8A -related epilepsies. Epilepsy Research, 2018, 143, 79-81.	0.8	48
79	De novo mutations ofKIAA2022in females cause intellectual disability and intractable epilepsy. Journal of Medical Genetics, 2016, 53, 850-858.	1.5	47
80	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. Genetics in Medicine, 2019, 21, 837-849.	1.1	47
81	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	3.7	47
82	Status Epilepticus and Refractory Status Epilepticus Management. Seminars in Pediatric Neurology, 2014, 21, 263-274.	1.0	46
83	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
84	Assessing the landscape of STXBP1-related disorders in 534 individuals. Brain, 2022, 145, 1668-1683.	3.7	46
85	Targeted gene sequencing in 6994 individuals with neurodevelopmental disorder with epilepsy. Genetics in Medicine, 2019, 21, 2496-2503.	1.1	45
86	Use of a Dynamic Genetic Testing Approach for Childhood-Onset Epilepsy. JAMA Network Open, 2019, 2, e192129.	2.8	45
87	Investigation of GRIN2A in common epilepsy phenotypes. Epilepsy Research, 2015, 115, 95-99.	0.8	44
88	Genetic Causes of Generalized Epilepsies. Seminars in Neurology, 2015, 35, 288-292.	0.5	44
89	Current knowledge of SLC6A1-related neurodevelopmental disorders. Brain Communications, 2020, 2, fcaa170.	1.5	44
90	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	9.4	44

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91	A retrospective study of the relation between vaccination and occurrence of seizures in Dravet syndrome. Epilepsia, 2011, 52, 175-178.	2.6	43
92	Clinical spectrum of STX1B -related epileptic disorders. Neurology, 2019, 92, e1238-e1249.	1.5	43
93	<scp>SCN3A</scp>â€Related Neurodevelopmental Disorder: A Spectrum of Epilepsy and Brain Malformation. Annals of Neurology, 2020, 88, 348-362.	2.8	42
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	TANGO2: expanding the clinical phenotype and spectrum of pathogenic variants. Genetics in Medicine, 2019, 21, 601-607.	1.1	41
96	Biallelic VARS variants cause developmental encephalopathy with microcephaly that is recapitulated in vars knockout zebrafish. Nature Communications, 2019, 10, 708.	5.8	40
97	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
98	Genetic heterogeneity in infantile spasms. Epilepsy Research, 2019, 156, 106181.	0.8	38
99	Computational analysis of 10,860 phenotypic annotations in individuals with SCN2A-related disorders. Genetics in Medicine, 2021, 23, 1263-1272.	1.1	38
100	Genetics of febrile seizure subtypes and syndromes: A twin study. Epilepsy Research, 2013, 105, 103-109.	0.8	36
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	Personalized medicine in genetic epilepsies â€“ possibilities, challenges, and new frontiers. Neuropharmacology, 2020, 172, 107970.	2.0	35
103	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
104	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	5.8	35
105	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
106	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	2.6	32
107	Bi-allelic ADARB1 Variants Associated with Microcephaly, Intellectual Disability, and Seizures. American Journal of Human Genetics, 2020, 106, 467-483.	2.6	31
108	Genetic risk perception and reproductive decision making among people with epilepsy. Epilepsia, 2010, 51, 1874-1877.	2.6	30

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109	The Genomics Research and Innovation Network: creating an interoperable, federated, genomics learning system. Genetics in Medicine, 2020, 22, 371-380.	1.1	30
110	Phenotypic and genetic spectrum of epilepsy with myoclonic atonic seizures. Epilepsia, 2020, 61, 995-1007.	2.6	30
111	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
112	In vivo evidence for the involvement of the carboxy terminal domain in assembling connexin 36 at the electrical synapse. Molecular and Cellular Neurosciences, 2010, 45, 47-58.	1.0	29
113	Absence seizures with intellectual disability as a phenotype of the 15q13.3 microdeletion syndrome. Epilepsia, 2011, 52, e194-8.	2.6	29
114	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
115	Structural genomic variation in childhood epilepsies with complex phenotypes. European Journal of Human Genetics, 2014, 22, 896-901.	1.4	28
116	PRRT2 mutations: exploring the phenotypical boundaries. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 462-465.	0.9	27
117	High-throughput evaluation of epilepsy-associated KCNQ2 variants reveals functional and pharmacological heterogeneity. JCI Insight, 2022, 7, .	2.3	27
118	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
119	Association Study of TRPC4 as a Candidate Gene for Generalized Epilepsy with Photosensitivity. NeuroMolecular Medicine, 2010, 12, 292-299.	1.8	25
120	Gene expression analysis in absence epilepsy using a monozygotic twin design. Epilepsia, 2008, 49, 1546-1554.	2.6	24
121	Epilepsy Course and Developmental Trajectories in STXBP1 -DEE. Neurology: Genetics, 2022, 8, .	0.9	24
122	Galanin pathogenic mutations in temporal lobe epilepsy. Human Molecular Genetics, 2015, 24, 3082-3091.	1.4	23
123	Epileptic Encephalopathiesâ€”Clinical Syndromes and Pathophysiological Concepts. Current Neurology and Neuroscience Reports, 2017, 17, 10.	2.0	23
124	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
125	Natural History Study of STXBP1-Developmental and Epileptic Encephalopathy Into Adulthood. Neurology, 2022, 99, .	1.5	23
126	Heterogeneous contribution of microdeletions in the development of common generalised and focal epilepsies. Journal of Medical Genetics, 2017, 54, 598-606.	1.5	22

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127	A longitudinal footprint of genetic epilepsies using automated electronic medical record interpretation. Genetics in Medicine, 2020, 22, 2060-2070.	1.1	22
128	Iterative phenotyping of 15q11.2, 15q13.3 and 16p13.11 microdeletion carriers in pediatric epilepsies. Epilepsy Research, 2014, 108, 109-116.	0.8	21
129	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
130	The phenotypic spectrum of ARHGEF9 includes intellectual disability, focal epilepsy and febrile seizures. Journal of Neurology, 2017, 264, 1421-1425.	1.8	20
131	Genetic test utilization and diagnostic yield in adult patients with neurological disorders. Neurogenetics, 2018, 19, 105-110.	0.7	20
132	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
133	Role of GRM4 in idiopathic generalized epilepsies analysed by genetic association and sequence analysis. Epilepsy Research, 2010, 89, 319-326.	0.8	19
134	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
135	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
136	RARS1â€related hypomyelinating leukodystrophy: Expanding the spectrum. Annals of Clinical and Translational Neurology, 2020, 7, 83-93.	1.7	18
137	Bi-allelic GAD1 variants cause a neonatal onset syndromic developmental and epileptic encephalopathy. Brain, 2020, 143, 1447-1461.	3.7	18
138	Ketamine for Management of Neonatal and Pediatric Refractory Status Epilepticus. Neurology, 2022, 99, .	1.5	18
139	A retrospective populationâ€based study on seizures related to childhood vaccination. Epilepsia, 2011, 52, 1506-1512.	2.6	17
140	CDKL5 Mutations as a Cause of Severe Epilepsy in Infancy. Journal of Child Neurology, 2013, 28, 937-941.	0.7	17
141	Genetic literacy series: Primer part 2â€”Paradigm shifts in epilepsy genetics. Epilepsia, 2018, 59, 1138-1147.	2.6	17
142	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
143	The unexpected role of copy number variations in juvenile myoclonic epilepsy. Epilepsy and Behavior, 2013, 28, S66-S68.	0.9	16
144	Atypical Vitamin B₆Deficiency. Journal of Child Neurology, 2014, 29, 704-707.	0.7	16

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145	Investigating the genetic basis of feverâ€associated syndromic epilepsies using copy number variation analysis. Epilepsia, 2015, 56, e26-32.	2.6	16
146	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
147	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
148	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
149	Phenotypic and Imaging Spectrum Associated With WDR45. Pediatric Neurology, 2020, 109, 56-62.	1.0	16
150	Wholeâ€exome and HLA sequencing in Febrile infectionâ€related epilepsy syndrome. Annals of Clinical and Translational Neurology, 2020, 7, 1429-1435.	1.7	15
151	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
152	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
153	PURA- Related Developmental and Epileptic Encephalopathy. Neurology: Genetics, 2021, 7, e613.	0.9	15
154	Familial cosegregation of rare genetic variants with disease in complex disorders. European Journal of Human Genetics, 2013, 21, 444-450.	1.4	14
155	Genome-wide rare copy number variation screening in ulcerative colitis identifies potential susceptibility loci. BMC Medical Genetics, 2016, 17, 26.	2.1	14
156	The role of SLC2A1 in early onset and childhood absence epilepsies. Epilepsy Research, 2013, 105, 229-233.	0.8	13
157	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
158	A duplication in 1q21.3 in a family with early onset and childhood absence epilepsy. Epilepsia, 2010, 51, 2453-2456.	2.6	12
159	Agammaglobulinemia with normal B-cell numbers in a patient lacking Bob1. Journal of Allergy and Clinical Immunology, 2021, 147, 1977-1980.	1.5	12
160	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
161	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
162	Spectrum of Phenotypic, Genetic, and Functional Characteristics in Patients With Epilepsy With KCNC2 Pathogenic Variants. Neurology, 2022, 98, .	1.5	11

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163	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
164	Early-onset genetic epilepsies reaching adult clinics. Brain, 2020, 143, e19-e19.	3.7	10
165	Variants in ATP6V0A1 cause progressive myoclonus epilepsy and developmental and epileptic encephalopathy. Brain Communications, 2021, 3, fcab245.	1.5	10
166	Computational analysis of neurodevelopmental phenotypes: Harmonization empowers clinical discovery. Human Mutation, 2022, 43, 1642-1658.	1.1	10
167	Common risk variants for epilepsy are enriched in families previously targeted for rare monogenic variant discovery. EBioMedicine, 2022, 81, 104079.	2.7	10
168	Epileptic Encephalopathies as Neurodegenerative Disorders. Advances in Neurobiology, 2017, 15, 295-315.	1.3	9
169	Using common genetic variants to find drugs for common epilepsies. Brain Communications, 2021, 3, fcab287.	1.5	9
170	Comprehensive analysis of candidate genes for photosensitivity using a complementary bioinformatic and experimental approach. Epilepsia, 2011, 52, e143-e147.	2.6	8
171	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
172	Child Neurology Telemedicine: Understanding the Data We Have and Finding the Patients We Do Not See. Pediatric Neurology, 2021, 116, 84.	1.0	8
173	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
174	Predicting the functional effects of voltage-gated potassium channel missense variants with multi-task learning. EBioMedicine, 2022, 81, 104115.	2.7	8
175	Commentary: Pathogenic EFHC1 mutations are tolerated in healthy individuals dependent on reported ancestry. Epilepsia, 2015, 56, 195-196.	2.6	7
176	Gene expression analysis in untreated absence epilepsy demonstrates an inconsistent pattern. Epilepsy Research, 2017, 132, 84-90.	0.8	7
177	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
178	Epileptic encephalopathy, movement disorder, and the yin and yang of GNAO1 function. Neurology, 2017, 89, 754-755.	1.5	7
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