Annapurna H Poduri

List of Publications by Year
in descending order

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178 papers	12,443 citations	36303 51 h-index	30922 102 g-index
185 all docs	185 docs citations	185 times ranked	15622 citing authors

#	Article	IF	Citations
1	De novo mutations in epileptic encephalopathies. Nature, 2013, 501, 217-221.	27.8	1,351
2	Somatic Mutation, Genomic Variation, and Neurological Disease. Science, 2013, 341, 1237758.	12.6	501
3	Single-Neuron Sequencing Analysis of L1 Retrotransposition and Somatic Mutation in the Human Brain. Cell, 2012, 151, 483-496.	28.9	500
4	Patient-Customized Oligonucleotide Therapy for a Rare Genetic Disease. New England Journal of Medicine, 2019, 381, 1644-1652.	27.0	481
5	Somatic Activation of AKT3 Causes Hemispheric Developmental Brain Malformations. Neuron, 2012, 74, 41-48.	8.1	413
6	De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	6.2	388
7	Using Whole-Exome Sequencing to Identify Inherited Causes of Autism. Neuron, 2013, 77, 259-273.	8.1	383
8	Somatic Mutations in Cerebral Cortical Malformations. New England Journal of Medicine, 2014, 371, 733-743.	27.0	326
9	Single-Cell, Genome-wide Sequencing Identifies Clonal Somatic Copy-Number Variation in the Human Brain. Cell Reports, 2014, 8, 1280-1289.	6.4	260
10	De Novo Mutations in SLC1A2 and CACNA1A Are Important Causes of Epileptic Encephalopathies. American Journal of Human Genetics, 2016, 99, 287-298.	6.2	247
11	Somatic Mutations Activating the mTOR Pathway in Dorsal Telencephalic Progenitors Cause a Continuum of Cortical Dysplasias. Cell Reports, 2017, 21, 3754-3766.	6.4	247
12	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.	6.2	237
13	Mammalian target of rapamycin pathway mutations cause hemimegalencephaly and focal cortical dysplasia. Annals of Neurology, 2015, 77, 720-725.	5.3	235
14	De novo variants in neurodevelopmental disorders with epilepsy. Nature Genetics, 2018, 50, 1048-1053.	21.4	230
15	SCN2A encephalopathy. Neurology, 2015, 85, 958-966.	1.1	211
16	Extending the KCNQ2 encephalopathy spectrum. Neurology, 2013, 81, 1697-1703.	1.1	198
17	Ultra-rare genetic variation in common epilepsies: a case-control sequencing study. Lancet Neurology, The, 2017, 16, 135-143.	10.2	190
18	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. Nature Genetics, 2017, 49, 1529-1538.	21.4	164

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19	Mutations in SLC12A5 in epilepsy of infancy with migrating focal seizures. Nature Communications, 2015, 6, 8038.	12.8	160
20	Copy number variation plays an important role in clinical epilepsy. Annals of Neurology, 2014, 75, 943-958.	5.3	147
21	Trends in Resource Utilization by Children with Neurological Impairment in the United States Inpatient Health Care System: A Repeat Cross-Sectional Study. PLoS Medicine, 2012, 9, e1001158.	8.4	145
22	Migrating partial seizures of infancy: expansion of the electroclinical, radiological and pathological disease spectrum. Brain, 2013, 136, 1578-1591.	7.6	144
23	The landscape of epilepsy-related GATOR1 variants. Genetics in Medicine, 2019, 21, 398-408.	2.4	137
24	Altered inhibition in tuberous sclerosis and type IIb cortical dysplasia. Annals of Neurology, 2012, 71, 539-551.	5.3	136
25	Epilepsy geneticsâ€”past, present, and future. Current Opinion in Genetics and Development, 2011, 21, 325-332.	3.3	124
26	Mutations in <scp>KCNT</scp>1 cause a spectrum of focal epilepsies. Epilepsia, 2015, 56, e114-20.	5.1	117
27	Sodium Channel SCN3A (NaV1.3) Regulation of Human Cerebral Cortical Folding and Oral Motor Development. Neuron, 2018, 99, 905-913.e7.	8.1	109
28	Genetics and genotypeâ€“phenotype correlations in early onset epileptic encephalopathy with burst suppression. Annals of Neurology, 2017, 81, 419-429.	5.3	107
29	Mutations in QARS, Encoding Glutaminyl-tRNA Synthetase, Cause Progressive Microcephaly, Cerebral-Cerebellar Atrophy, and Intractable Seizures. American Journal of Human Genetics, 2014, 94, 547-558.	6.2	106
30	Homozygous PLCB1 deletion associated with malignant migrating partial seizures in infancy. Epilepsia, 2012, 53, e146-50.	5.1	104
31	SLC25A22 is a novel gene for migrating partial seizures in infancy. Annals of Neurology, 2013, 74, 873-882.	5.3	102
32	Diagnostic yield of genetic tests in epilepsy. Neurology, 2019, 92, .	1.1	102
33	Mutations in the HECT domain of NEDD4L lead to AKTâ€“mTOR pathway deregulation and cause periventricular nodular heterotopia. Nature Genetics, 2016, 48, 1349-1358.	21.4	101
34	Megalencephaly and hemimegalencephaly: Breakthroughs in molecular etiology. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 156-172.	1.6	96
35	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. Annals of Neurology, 2019, 86, 821-831.	5.3	96
36	Somatic SLC35A2 variants in the brain are associated with intractable neocortical epilepsy. Annals of Neurology, 2018, 83, 1133-1146.	5.3	95

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37	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.	6.2	87
38	Genetic diagnoses in epilepsy: The impact of dynamic exome analysis in a pediatric cohort. Epilepsia, 2020, 61, 249-258.	5.1	85
39	De novo GABRG2mutations associated with epileptic encephalopathies. Brain, 2017, 140, 49-67.	7.6	80
40	A mouse model of DEPDC5-related epilepsy: Neuronal loss of Depdc5 causes dysplastic and ectopic neurons, increased mTOR signaling, and seizure susceptibility. Neurobiology of Disease, 2018, 111, 91-101.	4.4	79
41	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.	6.2	78
42	Genetic and phenotypic diversity of <scp>NHE</scp>6 mutations in <scp>C</scp>hristianson syndrome. Annals of Neurology, 2014, 76, 581-593.	5.3	73
43	Mutations in epilepsy and intellectual disability genes in patients with features of Rett syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 2017-2025.	1.2	72
44	Ethnically diverse causes of Walker-Warburg syndrome (WWS):FCMDmutations are a more common cause of WWS outside of the Middle East. Human Mutation, 2008, 29, E231-E241.	2.5	67
45	Genetic testing in the epilepsiesâ€”developments and dilemmas. Nature Reviews Neurology, 2014, 10, 293-299.	10.1	66
46	<scp>PCDH</scp>19â€related epilepsy is associated with a broad neurodevelopmental spectrum. Epilepsia, 2018, 59, 679-689.	5.1	66
47	Biological concepts in human sodium channel epilepsies and their relevance in clinical practice. Epilepsia, 2020, 61, 387-399.	5.1	65
48	Mutations in PYCR2, Encoding Pyrroline-5-Carboxylate Reductase 2, Cause Microcephaly and Hypomyelination. American Journal of Human Genetics, 2015, 96, 709-719.	6.2	60
49	Copy number variant analysis from exome data in 349 patients with epileptic encephalopathy. Annals of Neurology, 2015, 78, 323-328.	5.3	59
50	Disorders of Microtubule Function in Neurons: Imaging Correlates. American Journal of Neuroradiology, 2016, 37, 528-535.	2.4	56
51	The KCC2 Cotransporter and Human Epilepsy. Neuroscientist, 2016, 22, 555-562.	3.5	56
52	Quantifying the Effects of 16p11.2 Copy Number Variants on Brain Structure: A Multisite Genetic-First Study. Biological Psychiatry, 2018, 84, 253-264.	1.3	56
53	Treatment of malignant migrating partial epilepsy of infancy with rufinamide: report of five cases. Epileptic Disorders, 2011, 13, 18-21.	1.3	55
54	Experience With Rufinamide in a Pediatric Population: A Single Center's Experience. Pediatric Neurology, 2010, 43, 155-158.	2.1	54

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55	Spectrum of neurodevelopmental disease associated with the GNAO1 guanosine triphosphateâ€“binding region. Epilepsia, 2019, 60, 406-418.	5.1	53
56	Genetic testing for the epilepsies: A systematic review. Epilepsia, 2022, 63, 375-387.	5.1	53
57	Spectrum of K_V2.1 Dysfunction in KCNB1â€Associated Neurodevelopmental Disorders. Annals of Neurology, 2019, 86, 899-912.	5.3	52
58	The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. Epilepsia, 2019, 60, 797-806.	5.1	52
59	Modelling and treating GRIN2A developmental and epileptic encephalopathy in mice. Brain, 2020, 143, 2039-2057.	7.6	51
60	Whole genome sequencing identifies <scp>SCN2A</scp> mutation in monozygotic twins with <scp>O</scp>htahara syndrome and unique neuropathologic findings. Epilepsia, 2013, 54, e81-5.	5.1	49
61	A Recurrent De Novo PACS2 Heterozygous Missense Variant Causes Neonatal-Onset Developmental Epileptic Encephalopathy, Facial Dysmorphism, and Cerebellar Dysgenesis. American Journal of Human Genetics, 2018, 102, 995-1007.	6.2	49
62	Missense Variants in RHOBTB2 Cause a Developmental and Epileptic Encephalopathy in Humans, and Altered Levels Cause Neurological Defects in Drosophila. American Journal of Human Genetics, 2018, 102, 44-57.	6.2	49
63	Heterogeneous clinical and functional features of GRIN2D-related developmental and epileptic encephalopathy. Brain, 2019, 142, 3009-3027.	7.6	49
64	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	7.6	47
65	Neocortical development and epilepsy: insights from focal cortical dysplasia and brain tumours. Lancet Neurology, The, 2021, 20, 943-955.	10.2	47
66	Assessing the landscape of STXBP1-related disorders in 534 individuals. Brain, 2022, 145, 1668-1683.	7.6	46
67	Somatic mutation: The hidden genetics of brain malformations and focal epilepsies. Epilepsy Research, 2019, 155, 106161.	1.6	45
68	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	6.2	45
69	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. American Journal of Human Genetics, 2017, 101, 516-524.	6.2	43
70	Clinical application and evaluation of the <scp>B</scp>ien diagnostic criteria for <scp>R</scp>asmussen encephalitis. Epilepsia, 2013, 54, 1753-1760.	5.1	42
71	Hippocampal Formation Maldevelopment and Sudden Unexpected Death across the Pediatric Age Spectrum. Journal of Neuropathology and Experimental Neurology, 2016, 75, 981-997.	1.7	42
72	Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. Genome Medicine, 2020, 12, 28.	8.2	42

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73	The Epilepsy Phenome/Genome Project. Clinical Trials, 2013, 10, 568-586.	1.6	40
74	De novo and inherited private variants in MAP1B in periventricular nodular heterotopia. PLoS Genetics, 2018, 14, e1007281.	3.5	40
75	Infantile Spasms of Unknown Cause: Predictors of Outcome and Genotype-Phenotype Correlation. Pediatric Neurology, 2018, 87, 48-56.	2.1	39
76	Polymicrogyriaâ€associated epilepsy: A multicenter phenotypic study from the Epilepsy Phenome/Genome Project. Epilepsia, 2013, 54, 1368-1375.	5.1	38
77	Childrenâ€™s rare disease cohorts: an integrative research and clinical genomics initiative. Npj Genomic Medicine, 2020, 5, 29.	3.8	38
78	Perfusion Imaging of Focal Cortical Dysplasia Using Arterial Spin Labeling. Journal of Child Neurology, 2013, 28, 1474-1482.	1.4	37
79	Sturge-Weber Syndrome: Brain Magnetic Resonance Imaging andÂNeuropathology Findings. Pediatric Neurology, 2016, 58, 25-30.	2.1	37
80	Surgery for Intractable Epilepsy Due to Unilateral Brain Disease: A Retrospective Study Comparing Hemispherectomy Techniques. Pediatric Neurology, 2014, 51, 336-343.	2.1	35
81	Chronic mTORC1 inhibition rescues behavioral and biochemical deficits resulting from neuronal Depdc5 loss in mice. Human Molecular Genetics, 2019, 28, 2952-2964.	2.9	35
82	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.	6.2	35
83	High-throughput brain activity mapping and machine learning as a foundation for systems neuropharmacology. Nature Communications, 2018, 9, 5142.	12.8	34
84	Oxcarbazepine in Children With Nocturnal Frontal-Lobe Epilepsy. Pediatric Neurology, 2007, 37, 345-349.	2.1	33
85	Megalencephaly and Macrocephaly. Seminars in Neurology, 2015, 35, 277-287.	1.4	33
86	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.	2.5	33
87	Somatic variants in diverse genes leads to a spectrum of focal cortical malformations. Brain, 2022, 145, 2704-2720.	7.6	33
88	When Should Genetic Testing be Performed in Epilepsy Patients?. Epilepsy Currents, 2017, 17, 16-22.	0.8	32
89	Dacrystic seizures: Demographic, semiologic, and etiologic insights from a multicenter study in longâ€term videoâ€EEG monitoring units. Epilepsia, 2012, 53, 1810-1819.	5.1	31
90	SCN1A variants associated with sudden infant death syndrome. Epilepsia, 2018, 59, e56-e62.	5.1	30

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91	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	6.2	30
92	Altered white matter connectivity and network organization in polymicrogyria revealed by individual gyral topology-based analysis. NeuroImage, 2014, 86, 182-193.	4.2	29
93	Why West? Comparisons of clinical, genetic and molecular features of infants with and without spasms. PLoS ONE, 2018, 13, e0193599.	2.5	28
94	CA3 neuronal degeneration follows chronic entorhinal cortex lesions. Neuroscience Letters, 1995, 197, 1-4.	2.1	27
95	Gainâ€ofâ€function variants in the ODC1 gene cause a syndromic neurodevelopmental disorder associated with macrocephaly, alopecia, dysmorphic features, and neuroimaging abnormalities. American Journal of Medical Genetics, Part A, 2018, 176, 2554-2560.	1.2	26
96	A case-control collapsing analysis identifies epilepsy genes implicated in trio sequencing studies focused on de novo mutations. PLoS Genetics, 2017, 13, e1007104.	3.5	25
97	Hippocampal asymmetry and sudden unexpected death in infancy: a case report. Forensic Science, Medicine, and Pathology, 2012, 8, 441-446.	1.4	24
98	Microarray Noninvasive Neuronal Seizure Recordings from Intact Larval Zebrafish. PLoS ONE, 2016, 11, e0156498.	2.5	24
99	Semantic Similarity Analysis Reveals Robust Gene-Disease Relationships in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2020, 107, 683-697.	6.2	23
100	Mendelian etiologies identified with whole exome sequencing in cerebral palsy. Annals of Clinical and Translational Neurology, 2022, 9, 193-205.	3.7	23
101	Current neurologic treatment and emerging therapies in CDKL5 deficiency disorder. Journal of Neurodevelopmental Disorders, 2021, 13, 40.	3.1	22
102	Inheritance of Febrile Seizures in Sudden Unexplained Death in Toddlers. Pediatric Neurology, 2012, 46, 235-239.	2.1	21
103	Safety and retention rate of rufinamide in 300 patients: A single pediatric epilepsy center experience. Epilepsia, 2014, 55, 1235-1244.	5.1	21
104	Clinical Application of Epilepsy Genetics in Africa: Is Now the Time?. Frontiers in Neurology, 2018, 9, 276.	2.4	21
105	Mortality in infantile spasms: A hospitalâ€based study. Epilepsia, 2020, 61, 702-713.	5.1	21
106	KCNQ2â€DEE: developmental or epileptic encephalopathy?. Annals of Clinical and Translational Neurology, 2021, 8, 666-676.	3.7	21
107	DEPDC5 does it all: Shared genetics for diverse epilepsy syndromes. Annals of Neurology, 2014, 75, 631-633.	5.3	20
108	White matter spongiosis with vigabatrin therapy for infantile spasms. Epilepsia, 2018, 59, e40-e44.	5.1	20

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109	Mutations in NRXN1 and NRXN2 in a patient with early-onset epileptic encephalopathy and respiratory depression. Journal of Physical Education and Sports Management, 2019, 5, a003442.	1.2	20
110	Genetic Determinants of Sudden Unexpected Death in Pediatrics. Genetics in Medicine, 2022, 24, 839-850.	2.4	20
111	Genetic Forms of Epilepsies and Other Paroxysmal Disorders. Seminars in Neurology, 2014, 34, 266-279.	1.4	19
112	Focal Cortical Malformations Can Show Asymmetrically Higher Uptake on Interictal Fluorine-18 Fluorodeoxyglucose Positron Emission Tomography (PET). Journal of Child Neurology, 2007, 22, 232-237.	1.4	18
113	Clobazam: Effect on Frequency of Seizures and Safety Profile inÂDifferent Subgroups of Children With Epilepsy. Pediatric Neurology, 2014, 51, 60-66.	2.1	18
114	A randomized controlled trial of levodopa in patients with Angelman syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1099-1107.	1.2	18
115	Acute multi-sgRNA knockdown of KEOPS complex genes reproduces the microcephaly phenotype of the stable knockout zebrafish model. PLoS ONE, 2018, 13, e0191503.	2.5	18
116	<scp>BRAT</scp>1 encephalopathy: a recessive cause of epilepsy of infancy with migrating focal seizures. Developmental Medicine and Child Neurology, 2020, 62, 1096-1099.	2.1	18
117	Precision Therapy for Epilepsy Related to Brain Malformations. Neurotherapeutics, 2021, 18, 1548-1563.	4.4	18
118	Witnessed sleep-related seizure and sudden unexpected death in infancy: a case report. Forensic Science, Medicine, and Pathology, 2013, 9, 418-421.	1.4	17
119	Comparison of Drug Utilization Patterns in Observational Data: Antiepileptic Drugs in Pediatric Patients. Paediatric Drugs, 2015, 17, 401-410.	3.1	17
120	Detailed Magnetic Resonance Imaging (MRI) Analysis in Infantile Spasms. Journal of Child Neurology, 2018, 33, 405-412.	1.4	17
121	Epigenetics explained: a topic â€œprimerâ€•for the epilepsy community by the ILAE Genetics/Epigenetics Task Force. Epileptic Disorders, 2020, 22, 127-141.	1.3	17
122	Focal cortical dysplasia is more common in boys than in girls. Epilepsy and Behavior, 2013, 27, 121-123.	1.7	15
123	De Novo TUBB2A Variant Presenting With Anterior Temporal Pachygyria. Journal of Child Neurology, 2017, 32, 127-131.	1.4	15
124	Does Albendazole Affect Seizure Remission and Computed Tomography Response in Children With Neurocysticercosis? A Systematic Review and Meta-analysis. Journal of Child Neurology, 2007, 22, 135-142.	1.4	14
125	The syndrome of perisylvian polymicrogyria with congenital arthrogryposis. Brain and Development, 2010, 32, 550-555.	1.1	14
126	Polymicrogyria is Associated With Pathogenic Variants in PTEN. Annals of Neurology, 2020, 88, 1153-1164.	5.3	14

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127	Characterization of the <scp>GABRB2</scp>â€Associated Neurodevelopmental Disorders. Annals of Neurology, 2021, 89, 573-586.	5.3	14
128	A pathogenic UFSP2 variant in an autosomal recessive form of pediatric neurodevelopmental anomalies and epilepsy. Genetics in Medicine, 2021, 23, 900-908.	2.4	14
129	The role of sodium channels in sudden unexpected death in pediatrics. Molecular Genetics & Genomic Medicine, 2020, 8, e1309.	1.2	14
130	CSNK2B: A broad spectrum of neurodevelopmental disability and epilepsy severity. Epilepsia, 2021, 62, e103-e109.	5.1	13
131	Phenotypic and imaging features of FLNA-negative patients with bilateral periventricular nodular heterotopia and epilepsy. Epilepsy and Behavior, 2015, 51, 321-327.	1.7	12
132	HLA-A*31:01 and Oxcarbazepine-Induced DRESS in a Patient With Seizures and Complete DCX Deletion. Pediatrics, 2018, 141, S434-S438.	2.1	12
133	Diverse genetic causes of polymicrogyria with epilepsy. Epilepsia, 2021, 62, 973-983.	5.1	12
134	Cerebral visual impairment in CDKL5 deficiency disorder: vision as an outcome measure. Developmental Medicine and Child Neurology, 2021, 63, 1308-1315.	2.1	12
135	A de novo BRPF1 variant in a case of Sudden Unexplained Death in Childhood. European Journal of Medical Genetics, 2020, 63, 104002.	1.3	11
136	Genetic Factors Underlying Sudden Infant Death Syndrome. The Application of Clinical Genetics, 2021, Volume 14, 61-76.	3.0	10
137	Juvenile myoclonic epilepsy and narcolepsy: A series of three cases. Epilepsy and Behavior, 2015, 51, 163-165.	1.7	9
138	Return of individual results in epilepsy genomic research: A view from the field. Epilepsia, 2018, 59, 1635-1642.	5.1	9
139	Compound heterozygosity with PRRT2: Pushing the phenotypic envelope in genetic epilepsies. Epilepsy & Behavior Case Reports, 2019, 11, 125-128.	1.5	9
140	Epilepsy Benchmarks Area II: Prevent Epilepsy and Its Progression. Epilepsy Currents, 2020, 20, 14S-22S.	0.8	9
141	Epilepsy Benchmarks Area III: Improved Treatment Options for Controlling Seizures and Epilepsy-Related Conditions Without Side Effects. Epilepsy Currents, 2020, 20, 23S-30S.	0.8	9
142	Epilepsy Benchmarks Area I: Understanding the Causes of the Epilepsies and Epilepsy-Related Neurologic, Psychiatric, and Somatic Conditions. Epilepsy Currents, 2020, 20, 5S-13S.	0.8	9
143	Using common genetic variants to find drugs for common epilepsies. Brain Communications, 2021, 3, fcab287.	3.3	9
144	Clinicoâ€radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. Human Mutation, 2022, 43, 403-419.	2.5	9

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145	Infantile spasms: Assessing the diagnostic yield of an institutional guideline and the impact of etiology on longâ€term treatment response. Epilepsia, 2022, 63, 1164-1176.	5.1	9
146	Posterior-onset Rasmussen's encephalitis with ipsilateral cerebellar atrophy and uveitis resistant to rituximab. Epilepsy and Behavior Reports, 2020, 14, 100360.	1.0	8
147	Effectiveness of once-daily high-dose ACTH for infantile spasms. Epilepsy and Behavior, 2016, 59, 4-8.	1.7	6
148	A Model Program for Translational Medicine in Epilepsy Genetics. Journal of Child Neurology, 2017, 32, 429-436.	1.4	6
149	Epilepsy Benchmarks Area IV: Limit or Prevent Adverse Consequence of Seizures and Their Treatment Across the Life Span. Epilepsy Currents, 2020, 20, 31S-39S.	0.8	6
150	Uridineâ€responsive epileptic encephalopathy due to inherited variants in CAD : A Tale of Two Siblings. Annals of Clinical and Translational Neurology, 2021, 8, 716-722.	3.7	6
151	Mosaic and non-mosaic protocadherin 19 mutation leads to neuronal hyperexcitability in zebrafish. Neurobiology of Disease, 2022, 169, 105738.	4.4	6
152	A Distinct Asymmetrical Pattern of Cortical Malformation: Large Unilateral Malformation of Cortical Development with Contralateral Periventricular Nodular Heterotopia in Three Pediatric Cases. Epilepsia, 2005, 46, 1317-1321.	5.1	5
153	The Expanding SCN8A-Related Epilepsy Phenotype. Epilepsy Currents, 2015, 15, 333-334.	0.8	5
154	Intellectual and developmental disabilities research centers: Fifty years of scientific accomplishments. Annals of Neurology, 2019, 86, 332-343.	5.3	5
155	Biallelic variants in ZNF526 cause a severe neurodevelopmental disorder with microcephaly, bilateral cataract, epilepsy and simplified gyration. Journal of Medical Genetics, 2021, , jmedgenet-2020-107430.	3.2	5
156	Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations. Epilepsia, 2021, 62, 1518-1527.	5.1	5
157	A channel for precision diagnosis and treatment in genetic epilepsy. Annals of Neurology, 2014, 76, 323-324.	5.3	4
158	Meta-Analysis Revives Genome-Wide Association Studies in Epilepsy. Epilepsy Currents, 2015, 15, 122-123.	0.8	4
159	The 2021 Epilepsy Research Benchmarksâ€”Respecting Core Principles, Reflecting Evolving Community Priorities. Epilepsy Currents, 2021, 21, 389-393.	0.8	4
160	PIGNencephalopathy: Characterizing the epileptology. Epilepsia, 2022, 63, 974-991.	5.1	4
161	An Open-Source Husbandry Repository. Zebrafish, 2018, 15, 656-658.	1.1	3
162	HCN1 Gain-Of-Function Mutations â€“ a New Cause of Epileptic Encephalopathy. Epilepsy Currents, 2014, 14, 348-349.	0.8	2

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163	Two Studies, One Message: High Yield of Genetic Testing in Infants and Young Children with Severe Epilepsies. Epilepsy Currents, 2018, 18, 24-26.	0.8	2
164	Variability Among Next-Generation Sequencing Panels for Early-Life Epilepsies. JAMA Pediatrics, 2018, 172, 779.	6.2	2
165	Gene tests in adults with epilepsy and intellectual disability. Nature Reviews Neurology, 2020, 16, 527-528.	10.1	2
166	Nocturnal Choking Episodes: Under-Recognized and Misdiagnosed. Pediatric Neurology, 2010, 43, 355-358.	2.1	1
167	Cerebral Volumetric Analysis Over Time in Children With Malformations of Cortical Development: A Quantitative Investigation. Journal of Child Neurology, 2011, 26, 171-178.	1.4	1
168	Expect the unexpected in epilepsy genetics: Mutations in an epilepsy gene considered to be benign result in a severe phenotype. Annals of Neurology, 2012, 71, 1-2.	5.3	1
169	Tracking the Fate of Cells in Health and Disease. New England Journal of Medicine, 2016, 375, 2494-2496.	27.0	1
170	Experiencing Positive Health, as a Family, While Living With a Rare Complex Disease: Bringing Participatory Medicine Through Collaborative Decision Making Into the Real World. Journal of Participatory Medicine, 2020, 12, e17602.	1.3	1
171	Response to â€œThe Role of Cytomegalovirus in Schizencephalyâ€•by Spalice et al.. American Journal of Medical Genetics, Part A, 2011, 155, 1769-1769.	1.2	0
172	Reply. Annals of Neurology, 2014, 75, 326-326.	5.3	0
173	Migrating partial seizures of infancy: delineation of the clinical and genetic features in a national patient cohort. Lancet, The, 2014, 383, S14.	13.7	0
174	Genetics in clinical epilepsy: Issues in genetic testing and counseling. Journal of Pediatric Epilepsy, 2015, 01, 135-142.	0.2	0
175	Comments from the Editor(s). Epilepsia, 2016, 57, 336-337.	5.1	0
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