

Annapurna H Poduri

List of Publications by Year in descending order

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Version: 2024-02-01

178
papers

12,443
citations

41627

51
h-index

35168

102
g-index

185
all docs

185
docs citations

185
times ranked

17008
citing authors

#	ARTICLE	IF	CITATIONS
1	De novo mutations in epileptic encephalopathies. <i>Nature</i> , 2013, 501, 217-221.	13.7	1,351
2	Somatic Mutation, Genomic Variation, and Neurological Disease. <i>Science</i> , 2013, 341, 1237758.	6.0	501
3	Single-Neuron Sequencing Analysis of L1 Retrotransposition and Somatic Mutation in the Human Brain. <i>Cell</i> , 2012, 151, 483-496.	13.5	500
4	Patient-Customized Oligonucleotide Therapy for a Rare Genetic Disease. <i>New England Journal of Medicine</i> , 2019, 381, 1644-1652.	13.9	481
5	Somatic Activation of AKT3 Causes Hemispheric Developmental Brain Malformations. <i>Neuron</i> , 2012, 74, 41-48.	3.8	413
6	De Novo Mutations in Synaptic Transmission Genes Including DNMT1 Cause Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2014, 95, 360-370.	2.6	388
7	Using Whole-Exome Sequencing to Identify Inherited Causes of Autism. <i>Neuron</i> , 2013, 77, 259-273.	3.8	383
8	Somatic Mutations in Cerebral Cortical Malformations. <i>New England Journal of Medicine</i> , 2014, 371, 733-743.	13.9	326
9	Single-Cell, Genome-wide Sequencing Identifies Clonal Somatic Copy-Number Variation in the Human Brain. <i>Cell Reports</i> , 2014, 8, 1280-1289.	2.9	260
10	De Novo Mutations in SLC1A2 and CACNA1A Are Important Causes of Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2016, 99, 287-298.	2.6	247
11	Somatic Mutations Activating the mTOR Pathway in Dorsal Telencephalic Progenitors Cause a Continuum of Cortical Dysplasias. <i>Cell Reports</i> , 2017, 21, 3754-3766.	2.9	247
12	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. <i>American Journal of Human Genetics</i> , 2019, 105, 267-282.	2.6	237
13	Mammalian target of rapamycin pathway mutations cause hemimegalencephaly and focal cortical dysplasia. <i>Annals of Neurology</i> , 2015, 77, 720-725.	2.8	235
14	De novo variants in neurodevelopmental disorders with epilepsy. <i>Nature Genetics</i> , 2018, 50, 1048-1053.	9.4	230
15	<i>SCN2A</i> encephalopathy. <i>Neurology</i> , 2015, 85, 958-966.	1.5	211
16	Extending the <i>KCNQ2</i> encephalopathy spectrum. <i>Neurology</i> , 2013, 81, 1697-1703.	1.5	198
17	Ultra-rare genetic variation in common epilepsies: a case-control sequencing study. <i>Lancet Neurology</i> , The, 2017, 16, 135-143.	4.9	190
18	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. <i>Nature Genetics</i> , 2017, 49, 1529-1538.	9.4	164

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19	Mutations in SLC12A5 in epilepsy of infancy with migrating focal seizures. <i>Nature Communications</i> , 2015, 6, 8038.	5.8	160
20	Copy number variation plays an important role in clinical epilepsy. <i>Annals of Neurology</i> , 2014, 75, 943-958.	2.8	147
21	Trends in Resource Utilization by Children with Neurological Impairment in the United States Inpatient Health Care System: A Repeat Cross-Sectional Study. <i>PLoS Medicine</i> , 2012, 9, e1001158.	3.9	145
22	Migrating partial seizures of infancy: expansion of the electroclinical, radiological and pathological disease spectrum. <i>Brain</i> , 2013, 136, 1578-1591.	3.7	144
23	The landscape of epilepsy-related GATOR1 variants. <i>Genetics in Medicine</i> , 2019, 21, 398-408.	1.1	137
24	Altered inhibition in tuberous sclerosis and type IIb cortical dysplasia. <i>Annals of Neurology</i> , 2012, 71, 539-551.	2.8	136
25	Epilepsy genetics—past, present, and future. <i>Current Opinion in Genetics and Development</i> , 2011, 21, 325-332.	1.5	124
26	Mutations in <i>KCNT1</i> cause a spectrum of focal epilepsies. <i>Epilepsia</i> , 2015, 56, e114-20.	2.6	117
27	Sodium Channel SCN3A (NaV1.3) Regulation of Human Cerebral Cortical Folding and Oral Motor Development. <i>Neuron</i> , 2018, 99, 905-913.e7.	3.8	109
28	Genetics and genotype–phenotype correlations in early onset epileptic encephalopathy with burst suppression. <i>Annals of Neurology</i> , 2017, 81, 419-429.	2.8	107
29	Mutations in QARS, Encoding Glutamyl-tRNA Synthetase, Cause Progressive Microcephaly, Cerebral-Cerebellar Atrophy, and Intractable Seizures. <i>American Journal of Human Genetics</i> , 2014, 94, 547-558.	2.6	106
30	Homozygous <i>PLCB1</i> deletion associated with malignant migrating partial seizures in infancy. <i>Epilepsia</i> , 2012, 53, e146-50.	2.6	104
31	<i>SLC25A22</i> is a novel gene for migrating partial seizures in infancy. <i>Annals of Neurology</i> , 2013, 74, 873-882.	2.8	102
32	Diagnostic yield of genetic tests in epilepsy. <i>Neurology</i> , 2019, 92, .	1.5	102
33	Mutations in the HECT domain of NEDD4L lead to AKT–mTOR pathway deregulation and cause periventricular nodular heterotopia. <i>Nature Genetics</i> , 2016, 48, 1349-1358.	9.4	101
34	Megalencephaly and hemimegalencephaly: Breakthroughs in molecular etiology. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2014, 166, 156-172.	0.7	96
35	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. <i>Annals of Neurology</i> , 2019, 86, 821-831.	2.8	96
36	Somatic <i>SLC35A2</i> variants in the brain are associated with intractable neocortical epilepsy. <i>Annals of Neurology</i> , 2018, 83, 1133-1146.	2.8	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.	2.6	87
38	Genetic diagnoses in epilepsy: The impact of dynamic exome analysis in a pediatric cohort. Epilepsia, 2020, 61, 249-258.	2.6	85
39	De novo GABRG2 mutations associated with epileptic encephalopathies. Brain, 2017, 140, 49-67.	3.7	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.	2.1	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.	2.6	78
42	Genetic and phenotypic diversity of NHE6 mutations in Christianson syndrome. Annals of Neurology, 2014, 76, 581-593.	2.8	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.	0.7	72
44	Ethnically diverse causes of Walker-Warburg syndrome (WWS): FCMD mutations are a more common cause of WWS outside of the Middle East. Human Mutation, 2008, 29, E231-E241.	1.1	67
45	Genetic testing in the epilepsies—developments and dilemmas. Nature Reviews Neurology, 2014, 10, 293-299.	4.9	66
46	PCDH19-related epilepsy is associated with a broad neurodevelopmental spectrum. Epilepsia, 2018, 59, 679-689.	2.6	66
47	Biological concepts in human sodium channel epilepsies and their relevance in clinical practice. Epilepsia, 2020, 61, 387-399.	2.6	65
48	Mutations in PYCR2, Encoding Pyrroline-5-Carboxylate Reductase 2, Cause Microcephaly and Hypomyelination. American Journal of Human Genetics, 2015, 96, 709-719.	2.6	60
49	Copy number variant analysis from exome data in 349 patients with epileptic encephalopathy. Annals of Neurology, 2015, 78, 323-328.	2.8	59
50	Disorders of Microtubule Function in Neurons: Imaging Correlates. American Journal of Neuroradiology, 2016, 37, 528-535.	1.2	56
51	The KCC2 Cotransporter and Human Epilepsy. Neuroscientist, 2016, 22, 555-562.	2.6	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.	0.7	56
53	Treatment of malignant migrating partial epilepsy of infancy with rufinamide: report of five cases. Epileptic Disorders, 2011, 13, 18-21.	0.7	55
54	Experience With Rufinamide in a Pediatric Population: A Single Center's Experience. Pediatric Neurology, 2010, 43, 155-158.	1.0	54

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55	Spectrum of neurodevelopmental disease associated with the GNAO1 guanosine triphosphate-binding region. <i>Epilepsia</i> , 2019, 60, 406-418.	2.6	53
56	Genetic testing for the epilepsies: A systematic review. <i>Epilepsia</i> , 2022, 63, 375-387.	2.6	53
57	Spectrum of K _V 2.1 Dysfunction in <i>KCNB1</i> -Associated Neurodevelopmental Disorders. <i>Annals of Neurology</i> , 2019, 86, 899-912.	2.8	52
58	The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. <i>Epilepsia</i> , 2019, 60, 797-806.	2.6	52
59	Modelling and treating GRIN2A developmental and epileptic encephalopathy in mice. <i>Brain</i> , 2020, 143, 2039-2057.	3.7	51
60	Whole genome sequencing identifies <i>SCN2A</i> mutation in monozygotic twins with Ohtahara syndrome and unique neuropathologic findings. <i>Epilepsia</i> , 2013, 54, e81-5.	2.6	49
61	A Recurrent De Novo PACS2 Heterozygous Missense Variant Causes Neonatal-Onset Developmental Epileptic Encephalopathy, Facial Dysmorphism, and Cerebellar Dysgenesis. <i>American Journal of Human Genetics</i> , 2018, 102, 995-1007.	2.6	49
62	Missense Variants in RHOBTB2 Cause a Developmental and Epileptic Encephalopathy in Humans, and Altered Levels Cause Neurological Defects in <i>Drosophila</i> . <i>American Journal of Human Genetics</i> , 2018, 102, 44-57.	2.6	49
63	Heterogeneous clinical and functional features of GRIN2D-related developmental and epileptic encephalopathy. <i>Brain</i> , 2019, 142, 3009-3027.	3.7	49
64	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17,458 subjects. <i>Brain</i> , 2020, 143, 2106-2118.	3.7	47
65	Neocortical development and epilepsy: insights from focal cortical dysplasia and brain tumours. <i>Lancet Neurology</i> , The, 2021, 20, 943-955.	4.9	47
66	Assessing the landscape of <i>STXBP1</i> -related disorders in 534 individuals. <i>Brain</i> , 2022, 145, 1668-1683.	3.7	46
67	Somatic mutation: The hidden genetics of brain malformations and focal epilepsies. <i>Epilepsy Research</i> , 2019, 155, 106161.	0.8	45
68	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. <i>American Journal of Human Genetics</i> , 2019, 104, 948-956.	2.6	45
69	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. <i>American Journal of Human Genetics</i> , 2017, 101, 516-524.	2.6	43
70	Clinical application and evaluation of the <i>BR</i> diagnostic criteria for <i>Rasmussen</i> encephalitis. <i>Epilepsia</i> , 2013, 54, 1753-1760.	2.6	42
71	Hippocampal Formation Maldevelopment and Sudden Unexpected Death across the Pediatric Age Spectrum. <i>Journal of Neuropathology and Experimental Neurology</i> , 2016, 75, 981-997.	0.9	42
72	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

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73	The Epilepsy Phenome/Genome Project. <i>Clinical Trials</i> , 2013, 10, 568-586.	0.7	40
74	De novo and inherited private variants in MAP1B in periventricular nodular heterotopia. <i>PLoS Genetics</i> , 2018, 14, e1007281.	1.5	40
75	Infantile Spasms of Unknown Cause: Predictors of Outcome and Genotype-Phenotype Correlation. <i>Pediatric Neurology</i> , 2018, 87, 48-56.	1.0	39
76	Polymicrogyria-associated epilepsy: A multicenter phenotypic study from the Epilepsy Phenome/Genome Project. <i>Epilepsia</i> , 2013, 54, 1368-1375.	2.6	38
77	Children's rare disease cohorts: an integrative research and clinical genomics initiative. <i>Npj Genomic Medicine</i> , 2020, 5, 29.	1.7	38
78	Perfusion Imaging of Focal Cortical Dysplasia Using Arterial Spin Labeling. <i>Journal of Child Neurology</i> , 2013, 28, 1474-1482.	0.7	37
79	Sturge-Weber Syndrome: Brain Magnetic Resonance Imaging and Neuropathology Findings. <i>Pediatric Neurology</i> , 2016, 58, 25-30.	1.0	37
80	Surgery for Intractable Epilepsy Due to Unilateral Brain Disease: A Retrospective Study Comparing Hemispherectomy Techniques. <i>Pediatric Neurology</i> , 2014, 51, 336-343.	1.0	35
81	Chronic mTORC1 inhibition rescues behavioral and biochemical deficits resulting from neuronal <i>Depdc5</i> loss in mice. <i>Human Molecular Genetics</i> , 2019, 28, 2952-2964.	1.4	35
82	Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. <i>American Journal of Human Genetics</i> , 2021, 108, 965-982.	2.6	35
83	High-throughput brain activity mapping and machine learning as a foundation for systems neuropharmacology. <i>Nature Communications</i> , 2018, 9, 5142.	5.8	34
84	Oxcarbazepine in Children With Nocturnal Frontal-Lobe Epilepsy. <i>Pediatric Neurology</i> , 2007, 37, 345-349.	1.0	33
85	Megalencephaly and Macrocephaly. <i>Seminars in Neurology</i> , 2015, 35, 277-287.	0.5	33
86	The ClinGen Epilepsy Gene Curation Expert Panel "Bridging the divide between clinical domain knowledge and formal gene curation criteria. <i>Human Mutation</i> , 2018, 39, 1476-1484.	1.1	33
87	Somatic variants in diverse genes leads to a spectrum of focal cortical malformations. <i>Brain</i> , 2022, 145, 2704-2720.	3.7	33
88	When Should Genetic Testing be Performed in Epilepsy Patients?. <i>Epilepsy Currents</i> , 2017, 17, 16-22.	0.4	32
89	Dacrystic seizures: Demographic, semiologic, and etiologic insights from a multicenter study in long-term video-EEG monitoring units. <i>Epilepsia</i> , 2012, 53, 1810-1819.	2.6	31
90	<i>SCN1A</i> variants associated with sudden infant death syndrome. <i>Epilepsia</i> , 2018, 59, e56-e62.	2.6	30

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91	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , 2019, 104, 530-541.	2.6	30
92	Altered white matter connectivity and network organization in polymicrogyria revealed by individual gyral topology-based analysis. <i>NeuroImage</i> , 2014, 86, 182-193.	2.1	29
93	Why West? Comparisons of clinical, genetic and molecular features of infants with and without spasms. <i>PLoS ONE</i> , 2018, 13, e0193599.	1.1	28
94	CA3 neuronal degeneration follows chronic entorhinal cortex lesions. <i>Neuroscience Letters</i> , 1995, 197, 1-4.	1.0	27
95	Gain-of-function variants in the <i>ODC1</i> gene cause a syndromic neurodevelopmental disorder associated with macrocephaly, alopecia, dysmorphic features, and neuroimaging abnormalities. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2554-2560.	0.7	26
96	A case-control collapsing analysis identifies epilepsy genes implicated in trio sequencing studies focused on de novo mutations. <i>PLoS Genetics</i> , 2017, 13, e1007104.	1.5	25
97	Hippocampal asymmetry and sudden unexpected death in infancy: a case report. <i>Forensic Science, Medicine, and Pathology</i> , 2012, 8, 441-446.	0.6	24
98	Microarray Noninvasive Neuronal Seizure Recordings from Intact Larval Zebrafish. <i>PLoS ONE</i> , 2016, 11, e0156498.	1.1	24
99	Semantic Similarity Analysis Reveals Robust Gene-Disease Relationships in Developmental and Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2020, 107, 683-697.	2.6	23
100	Mendelian etiologies identified with whole exome sequencing in cerebral palsy. <i>Annals of Clinical and Translational Neurology</i> , 2022, 9, 193-205.	1.7	23
101	Current neurologic treatment and emerging therapies in CDKL5 deficiency disorder. <i>Journal of Neurodevelopmental Disorders</i> , 2021, 13, 40.	1.5	22
102	Inheritance of Febrile Seizures in Sudden Unexplained Death in Toddlers. <i>Pediatric Neurology</i> , 2012, 46, 235-239.	1.0	21
103	Safety and retention rate of rufinamide in 300 patients: A single pediatric epilepsy center experience. <i>Epilepsia</i> , 2014, 55, 1235-1244.	2.6	21
104	Clinical Application of Epilepsy Genetics in Africa: Is Now the Time?. <i>Frontiers in Neurology</i> , 2018, 9, 276.	1.1	21
105	Mortality in infantile spasms: A hospital-based study. <i>Epilepsia</i> , 2020, 61, 702-713.	2.6	21
106	<i>KCNQ2</i> : developmental or epileptic encephalopathy?. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 666-676.	1.7	21
107	<i>DEPDC5</i> does it all: Shared genetics for diverse epilepsy syndromes. <i>Annals of Neurology</i> , 2014, 75, 631-633.	2.8	20
108	White matter spongiosis with vigabatrin therapy for infantile spasms. <i>Epilepsia</i> , 2018, 59, e40-e44.	2.6	20

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109	Mutations in NRXN1 and NRXN2 in a patient with early-onset epileptic encephalopathy and respiratory depression. <i>Journal of Physical Education and Sports Management</i> , 2019, 5, a003442.	0.5	20
110	Genetic Determinants of Sudden Unexpected Death in Pediatrics. <i>Genetics in Medicine</i> , 2022, 24, 839-850.	1.1	20
111	Genetic Forms of Epilepsies and Other Paroxysmal Disorders. <i>Seminars in Neurology</i> , 2014, 34, 266-279.	0.5	19
112	Focal Cortical Malformations Can Show Asymmetrically Higher Uptake on Interictal Fluorine-18 Fluorodeoxyglucose Positron Emission Tomography (PET). <i>Journal of Child Neurology</i> , 2007, 22, 232-237.	0.7	18
113	Clobazam: Effect on Frequency of Seizures and Safety Profile in Different Subgroups of Children With Epilepsy. <i>Pediatric Neurology</i> , 2014, 51, 60-66.	1.0	18
114	A randomized controlled trial of levodopa in patients with Angelman syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1099-1107.	0.7	18
115	Acute multi-sgRNA knockdown of KEOPS complex genes reproduces the microcephaly phenotype of the stable knockout zebrafish model. <i>PLoS ONE</i> , 2018, 13, e0191503.	1.1	18
116	BRAT1 encephalopathy: a recessive cause of epilepsy of infancy with migrating focal seizures. <i>Developmental Medicine and Child Neurology</i> , 2020, 62, 1096-1099.	1.1	18
117	Precision Therapy for Epilepsy Related to Brain Malformations. <i>Neurotherapeutics</i> , 2021, 18, 1548-1563.	2.1	18
118	Witnessed sleep-related seizure and sudden unexpected death in infancy: a case report. <i>Forensic Science, Medicine, and Pathology</i> , 2013, 9, 418-421.	0.6	17
119	Comparison of Drug Utilization Patterns in Observational Data: Antiepileptic Drugs in Pediatric Patients. <i>Paediatric Drugs</i> , 2015, 17, 401-410.	1.3	17
120	Detailed Magnetic Resonance Imaging (MRI) Analysis in Infantile Spasms. <i>Journal of Child Neurology</i> , 2018, 33, 405-412.	0.7	17
121	Epigenetics explained: a topic primer for the epilepsy community by the ILAE Genetics/Epigenetics Task Force. <i>Epileptic Disorders</i> , 2020, 22, 127-141.	0.7	17
122	Focal cortical dysplasia is more common in boys than in girls. <i>Epilepsy and Behavior</i> , 2013, 27, 121-123.	0.9	15
123	De Novo TUBB2A Variant Presenting With Anterior Temporal Pachygyria. <i>Journal of Child Neurology</i> , 2017, 32, 127-131.	0.7	15
124	Does Albendazole Affect Seizure Remission and Computed Tomography Response in Children With Neurocysticercosis? A Systematic Review and Meta-analysis. <i>Journal of Child Neurology</i> , 2007, 22, 135-142.	0.7	14
125	The syndrome of perisylvian polymicrogyria with congenital arthrogryposis. <i>Brain and Development</i> , 2010, 32, 550-555.	0.6	14
126	Polymicrogyria is Associated With Pathogenic Variants in PTEN. <i>Annals of Neurology</i> , 2020, 88, 1153-1164.	2.8	14

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127	Characterization of the <i>GABRB2</i> Associated Neurodevelopmental Disorders. <i>Annals of Neurology</i> , 2021, 89, 573-586.	2.8	14
128	A pathogenic <i>UFSP2</i> variant in an autosomal recessive form of pediatric neurodevelopmental anomalies and epilepsy. <i>Genetics in Medicine</i> , 2021, 23, 900-908.	1.1	14
129	The role of sodium channels in sudden unexpected death in pediatrics. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1309.	0.6	14
130	<i>CSNK2B</i> : A broad spectrum of neurodevelopmental disability and epilepsy severity. <i>Epilepsia</i> , 2021, 62, e103-e109.	2.6	13
131	Phenotypic and imaging features of <i>FLNA</i> -negative patients with bilateral periventricular nodular heterotopia and epilepsy. <i>Epilepsy and Behavior</i> , 2015, 51, 321-327.	0.9	12
132	<i>HLA-A*31:01</i> and Oxcarbazepine-Induced DRESS in a Patient With Seizures and Complete <i>DCX</i> Deletion. <i>Pediatrics</i> , 2018, 141, S434-S438.	1.0	12
133	Diverse genetic causes of polymicrogyria with epilepsy. <i>Epilepsia</i> , 2021, 62, 973-983.	2.6	12
134	Cerebral visual impairment in <i>CDKL5</i> deficiency disorder: vision as an outcome measure. <i>Developmental Medicine and Child Neurology</i> , 2021, 63, 1308-1315.	1.1	12
135	A de novo <i>BRPF1</i> variant in a case of Sudden Unexplained Death in Childhood. <i>European Journal of Medical Genetics</i> , 2020, 63, 104002.	0.7	11
136	Genetic Factors Underlying Sudden Infant Death Syndrome. <i>The Application of Clinical Genetics</i> , 2021, Volume 14, 61-76.	1.4	10
137	Juvenile myoclonic epilepsy and narcolepsy: A series of three cases. <i>Epilepsy and Behavior</i> , 2015, 51, 163-165.	0.9	9
138	Return of individual results in epilepsy genomic research: A view from the field. <i>Epilepsia</i> , 2018, 59, 1635-1642.	2.6	9
139	Compound heterozygosity with <i>PRRT2</i> : Pushing the phenotypic envelope in genetic epilepsies. <i>Epilepsy & Behavior Case Reports</i> , 2019, 11, 125-128.	1.5	9
140	Epilepsy Benchmarks Area II: Prevent Epilepsy and Its Progression. <i>Epilepsy Currents</i> , 2020, 20, 14S-22S.	0.4	9
141	Epilepsy Benchmarks Area III: Improved Treatment Options for Controlling Seizures and Epilepsy-Related Conditions Without Side Effects. <i>Epilepsy Currents</i> , 2020, 20, 23S-30S.	0.4	9
142	Epilepsy Benchmarks Area I: Understanding the Causes of the Epilepsies and Epilepsy-Related Neurologic, Psychiatric, and Somatic Conditions. <i>Epilepsy Currents</i> , 2020, 20, 5S-13S.	0.4	9
143	Using common genetic variants to find drugs for common epilepsies. <i>Brain Communications</i> , 2021, 3, fcab287.	1.5	9
144	Clinico-radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (<i>ITPase</i>) deficiency. <i>Human Mutation</i> , 2022, 43, 403-419.	1.1	9

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145	Infantile spasms: Assessing the diagnostic yield of an institutional guideline and the impact of etiology on long-term treatment response. <i>Epilepsia</i> , 2022, 63, 1164-1176.	2.6	9
146	Posterior-onset Rasmussen's encephalitis with ipsilateral cerebellar atrophy and uveitis resistant to rituximab. <i>Epilepsy and Behavior Reports</i> , 2020, 14, 100360.	0.5	8
147	Effectiveness of once-daily high-dose ACTH for infantile spasms. <i>Epilepsy and Behavior</i> , 2016, 59, 4-8.	0.9	6
148	A Model Program for Translational Medicine in Epilepsy Genetics. <i>Journal of Child Neurology</i> , 2017, 32, 429-436.	0.7	6
149	Epilepsy Benchmarks Area IV: Limit or Prevent Adverse Consequence of Seizures and Their Treatment Across the Life Span. <i>Epilepsy Currents</i> , 2020, 20, 31S-39S.	0.4	6
150	Uridine-responsive epileptic encephalopathy due to inherited variants in CAD : A Tale of Two Siblings. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 716-722.	1.7	6
151	Mosaic and non-mosaic protocadherin 19 mutation leads to neuronal hyperexcitability in zebrafish. <i>Neurobiology of Disease</i> , 2022, 169, 105738.	2.1	6
152	A Distinct Asymmetrical Pattern of Cortical Malformation: Large Unilateral Malformation of Cortical Development with Contralateral Periventricular Nodular Heterotopia in Three Pediatric Cases. <i>Epilepsia</i> , 2005, 46, 1317-1321.	2.6	5
153	The Expanding SCN8A-Related Epilepsy Phenotype. <i>Epilepsy Currents</i> , 2015, 15, 333-334.	0.4	5
154	Intellectual and developmental disabilities research centers: Fifty years of scientific accomplishments. <i>Annals of Neurology</i> , 2019, 86, 332-343.	2.8	5
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