Renzo Guerrini

List of Publications by Year
in descending order

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573 papers	29,711 citations	4388 86 h-index	9103 144 g-index
618 all docs	618 docs citations	618 times ranked	23582 citing authors

#	Article	IF	Citations
1	The clinicopathologic spectrum of focal cortical dysplasias: A consensus classification proposed by an ad hoc Task Force of the ILAE Diagnostic Methods Commission1. Epilepsia, 2011, 52, 158-174.	5.1	1,454
2	A developmental and genetic classification for malformations of cortical development: update 2012. Brain, 2012, 135, 1348-1369.	7.6	849
3	Histopathological Findings in Brain Tissue Obtained during Epilepsy Surgery. New England Journal of Medicine, 2017, 377, 1648-1656.	27.0	621
4	Lamotrigine and Seizure Aggravation in Severe Myoclonic Epilepsy. Epilepsia, 1998, 39, 508-512.	5.1	505
5	Epilepsy in children. Lancet, The, 2006, 367, 499-524.	13.7	464
6	Proposed Criteria for Referral and Evaluation of Children for Epilepsy Surgery: Recommendations of the Subcommission for Pediatric Epilepsy Surgery. Epilepsia, 2006, 47, 952-959.	5.1	456
7	Lennox-Gastaut syndrome: a consensus approach on diagnosis, assessment, management, and trial methodology. Lancet Neurology, The, 2009, 8, 82-93.	10.2	412
8	Mutations in TUBG1, DYNC1H1, KIF5C and KIF2A cause malformations of cortical development and microcephaly. Nature Genetics, 2013, 45, 639-647.	21.4	399
9	De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	6.2	388
10	Malformations of cortical development: clinical features and genetic causes. Lancet Neurology, The, 2014, 13, 710-726.	10.2	382
11	Structural brain abnormalities in the common epilepsies assessed in a worldwide ENIGMA study. Brain, 2018, 141, 391-408.	7.6	352
12	Mutations in ARFGEF2 implicate vesicle trafficking in neural progenitor proliferation and migration in the human cerebral cortex. Nature Genetics, 2004, 36, 69-76.	21.4	340
13	Somatic Mutations in Cerebral Cortical Malformations. New England Journal of Medicine, 2014, 371, 733-743.	27.0	326
14	Mutations ofARX are associated with striking pleiotropy and consistent genotype-phenotype correlation. Human Mutation, 2004, 23, 147-159.	2.5	293
15	The genetics of Dravet syndrome. Epilepsia, 2011, 52, 24-29.	5.1	287
16	Neuronal migration disorders. Neurobiology of Disease, 2010, 38, 154-166.	4.4	271
17	Earlyâ€onset absence epilepsy caused by mutations in the glucose transporter GLUT1. Annals of Neurology, 2009, 66, 415-419.	5.3	266
18	Benign familial neonatal-infantile seizures: Characterization of a new sodium channelopathy. Annals of Neurology, 2004, 55, 550-557.	5.3	250

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19	The phenotypic spectrum of SCN8A encephalopathy. Neurology, 2015, 84, 480-489.	1.1	246
20	Definition and diagnostic criteria of sleep-related hypermotor epilepsy. Neurology, 2016, 86, 1834-1842.	1.1	245
21	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
22	Association ofMTORMutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. JAMA Neurology, 2016, 73, 836.	9.0	234
23	De novo variants in neurodevelopmental disorders with epilepsy. Nature Genetics, 2018, 50, 1048-1053.	21.4	230
24	Early and effective treatment of <scp>KCNQ</scp>2 encephalopathy. Epilepsia, 2015, 56, 685-691.	5.1	229
25	A versatile clearing agent for multi-modal brain imaging. Scientific Reports, 2015, 5, 9808.	3.3	228
26	Abnormal development of the human cerebral cortex: genetics, functional consequences and treatment options. Trends in Neurosciences, 2008, 31, 154-162.	8.6	227
27	Fenfluramine hydrochloride for the treatment of seizures in Dravet syndrome: a randomised, double-blind, placebo-controlled trial. Lancet, The, 2019, 394, 2243-2254.	13.7	227
28	Increased Sensitivity of the Neuronal Nicotinic Receptor Î±2 Subunit Causes Familial Epilepsy with Nocturnal Wandering and Ictal Fear. American Journal of Human Genetics, 2006, 79, 342-350.	6.2	225
29	Reversible Pseudoatrophy of the Brain and Mental Deterioration Associated with Valproate Treatment. Epilepsia, 1998, 39, 27-32.	5.1	219
30	Clinical and imaging heterogeneity of polymicrogyria: a study of 328 patients. Brain, 2010, 133, 1415-1427.	7.6	215
31	Extending the KCNQ2 encephalopathy spectrum. Neurology, 2013, 81, 1697-1703.	1.1	198
32	Idiopathic Photosensitive Occipital Lobe Epilepsy. Epilepsia, 1995, 36, 883-891.	5.1	191
33	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.	6.2	188
34	Antiepileptic Drugâ€Induced Worsening of Seizures in Children. Epilepsia, 1998, 39, S2-10.	5.1	183
35	Seizure outcome and use of antiepileptic drugs after epilepsy surgery according to histopathological diagnosis: a retrospective multicentre cohort study. Lancet Neurology, The, 2020, 19, 748-757.	10.2	177
36	Malformations of cortical development and epilepsy. Dialogues in Clinical Neuroscience, 2008, 10, 47-62.	3.7	176

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37	Characterization of mutations in the genedoublecortin in patients with double cortex syndrome. Annals of Neurology, 1999, 45, 146-153.	5.3	175
38	Angelman syndrome: Correlations between epilepsy phenotypes and genotypes. Annals of Neurology, 1998, 43, 485-493.	5.3	173
39	Disruption of neural progenitors along the ventricular and subventricular zones in periventricular heterotopia. Human Molecular Genetics, 2009, 18, 497-516.	2.9	169
40	Diagnostic methods and treatment options for focal cortical dysplasia. Epilepsia, 2015, 56, 1669-1686.	5.1	167
41	Genetic Basis of Brain Malformations. Molecular Syndromology, 2016, 7, 220-233.	0.8	156
42	Idiopathic Epilepsies with Seizures Precipitated by Fever and SCN1A Abnormalities. Epilepsia, 2007, 48, 1678-1685.	5.1	154
43	Autosomal recessive Rolandic epilepsy with paroxysmal exercise-induced dystonia and writer's cramp: Delineation of the syndrome and gene mapping to chromosome 16p12-11.2. Annals of Neurology, 1999, 45, 344-352.	5.3	153
44	SCN1A duplications and deletions detected in Dravet syndrome: Implications for molecular diagnosis. Epilepsia, 2009, 50, 1670-1678.	5.1	152
45	Diagnostic Targeted Resequencing in 349 Patients with Drug-Resistant Pediatric Epilepsies Identifies Causative Mutations in 30 Different Genes. Human Mutation, 2017, 38, 216-225.	2.5	152
46	Early Visual Seizures and Progressive Myoclonus Epilepsy in Neuronopathic Gaucher Disease Due to a Rare Compound Heterozygosity (N188S/S107L). Epilepsia, 2004, 45, 1154-1157.	5.1	148
47	Vagus nerve stimulation: Surgical technique of implantation and revision and related morbidity. Epilepsia, 2017, 58, 85-90.	5.1	145
48	Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918.	10.2	139
49	Lack of cortical contrast gain control in human photosensitive epilepsy. Nature Neuroscience, 2000, 3, 259-263.	14.8	138
50	Genetic malformations of cortical development. Experimental Brain Research, 2006, 173, 322-333.	1.5	138
51	Autosomal recessive cortical myoclonic tremor and epilepsy: association with a mutation in the potassium channel associated gene CNTN2. Brain, 2013, 136, 1155-1160.	7.6	137
52	The landscape of epilepsy-related GATOR1 variants. Genetics in Medicine, 2019, 21, 398-408.	2.4	137
53	Cortical myoclonus in angelman syndrome. Annals of Neurology, 1996, 40, 39-48.	5.3	136
54	Idiopathic Epilepsy and Paroxysmal Dyskinesia. Epilepsia, 2001, 42, 36-41.	5.1	136

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55	Visual Sensitivity and Epilepsy: A Proposed Terminology and Classification for Clinical and EEG Phenomenology. Epilepsia, 2001, 42, 692-701.	5.1	135
56	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.	2.9	134
57	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, .	5.0	134
58	Topical Review: Neuronal Migration Disorders, Genetics, and Epileptogenesis. Journal of Child Neurology, 2005, 20, 287-299.	1.4	131
59	SYNGAP1 encephalopathy. Neurology, 2019, 92, e96-e107.	1.1	131
60	7T <scp>MRI</scp> in focal epilepsy with unrevealing conventional field strength imaging. Epilepsia, 2016, 57, 445-454.	5.1	128
61	Electrophysiological characterization of spontaneous and carbamazepine-induced epileptic negative myoclonus in benign childhood epilepsy with centro-temporal spikes. Clinical Neurophysiology, 2004, 115, 50-58.	1.5	127
62	Polymicrogyria and deletion 22q11.2 syndrome: Window to the etiology of a common cortical malformation. American Journal of Medical Genetics, Part A, 2006, 140A, 2416-2425.	1.2	125
63	Germline and somatic mutations in the MTOR gene in focal cortical dysplasia and epilepsy. Neurology: Genetics, 2016, 2, e118.	1.9	125
64	Genotype-phenotype analysis of human frontoparietal polymicrogyria syndromes. Annals of Neurology, 2005, 58, 680-687.	5.3	124
65	Epilepsy in Rett syndrome, and CDKL5â€•and FOXG1â€geneâ€“related encephalopathies. Epilepsia, 2012, 53, 2067-2078.	5.1	124
66	White matter abnormalities across different epilepsy syndromes in adults: an ENIGMA-Epilepsy study. Brain, 2020, 143, 2454-2473.	7.6	123
67	Glucose Transporter 1 Deficiency as a Treatable Cause of Myoclonic Astatic Epilepsy. Archives of Neurology, 2011, 68, 1152.	4.5	121
68	Severe myoclonic epilepsy in infancy: A systematic review and a metaâ€•analysis of individual patient data. Epilepsia, 2008, 49, 343-348.	5.1	119
69	GM1 gangliosidosis and Morquio B disease: An update on genetic alterations and clinical findings. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2011, 1812, 782-790.	3.8	115
70	The phenotype of SCN8A developmental and epileptic encephalopathy. Neurology, 2018, 91, e1112-e1124.	1.1	114
71	Phenotypic spectrum of GABRA1. Neurology, 2016, 87, 1140-1151.	1.1	113
72	Genetic testing in benign familial epilepsies of the first year of life: Clinical and diagnostic significance. Epilepsia, 2013, 54, 425-436.	5.1	110

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73	Autism with Seizures and Intellectual Disability: Possible Causative Role of Gain-of-function of the Inwardly-Rectifying K+ Channel Kir4.1. Neurobiology of Disease, 2011, 43, 239-247.	4.4	108
74	PRRT2 mutations in familial infantile seizures, paroxysmal dyskinesia, and hemiplegic migraine. Neurology, 2012, 79, 2109-2114.	1.1	106
75	Subcortical band heterotopia (SBH) in males: clinical, imaging and genetic findings in comparison with females. Brain, 2002, 125, 2507-2522.	7.6	105
76	Malformations of Cortical Development and Epilepsy. Cold Spring Harbor Perspectives in Medicine, 2015, 5, a022392-a022392.	6.2	104
77	Lissencephaly: Expanded imaging and clinical classification. American Journal of Medical Genetics, Part A, 2017, 173, 1473-1488.	1.2	104
78	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain, 2017, 140, 2610-2622.	7.6	102
79	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
80	The role of the nicotinic acetylcholine receptors in sleep-related epilepsy. Biochemical Pharmacology, 2007, 74, 1308-1314.	4.4	99
81	Cognitive development in Dravet syndrome: A retrospective, multicenter study of 26 patients. Epilepsia, 2011, 52, 386-392.	5.1	99
82	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	12.8	99
83	Role of the phosphoinositide phosphatase FIG4 gene in familial epilepsy with polymicrogyria. Neurology, 2014, 82, 1068-1075.	1.1	97
84	Neurologic phenotypes associated with COL4A1 / 2 mutations. Neurology, 2018, 91, e2078-e2088.	1.1	97
85	Network-based atrophy modeling in the common epilepsies: A worldwide ENIGMA study. Science Advances, 2020, 6, .	10.3	97
86	HCN1mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. Brain, 2018, 141, 3160-3178.	7.6	96
87	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. Annals of Neurology, 2019, 86, 821-831.	5.3	96
88	Genetic and neuroradiological heterogeneity of double cortex syndrome. Annals of Neurology, 2000, 47, 265-269.	5.3	94
89	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. Human Molecular Genetics, 2015, 24, 5250-5259.	2.9	93
90	Clinical and genetic factors predicting Dravet syndrome in infants with SCN1A mutations. Neurology, 2017, 88, 1037-1044.	1.1	93

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91	Delayed appearance of interictal EEG abnormalities in early onset childhood epilepsy with occipital paroxysms. Brain and Development, 1997, 19, 343-346.	1.1	92
92	Continuous Spike-and-Wave Activity During Slow-Wave Sleep: Syndrome or EEG Pattern?. Epilepsia, 1999, 40, 1593-1601.	5.1	92
93	Analysis of 17 genes detects mutations in 81% of 811 patients with lissencephaly. Genetics in Medicine, 2018, 20, 1354-1364.	2.4	92
94	Epileptic Syndromes and Visually Induced Seizures. Epilepsia, 2004, 45, 14-18.	5.1	91
95	Myoclonic status epilepticus following high-dosage lamotrigine therapy. Brain and Development, 1999, 21, 420-424.	1.1	89
96	Brain MRI Findings in Severe Myoclonic Epilepsy in Infancy and Genotype?Phenotype Correlations. Epilepsia, 2007, 48, 1092-1096.	5.1	89
97	Epileptic Encephalopathies with Myoclonic Seizures in Infants and Children (Severe Myoclonic) Tj ETQq1 1 0.784314 rgBT /Overlock 10	1.7	88
98	The <scp>ILAE</scp> consensus classification of focal cortical dysplasia: An update proposed by an ad hoc task force of the <scp>ILAE</scp> diagnostic methods commission. Epilepsia, 2022, 63, 1899-1919.	5.1	88
99	Rapid assay of topiramate in dried blood spots by a new liquid chromatography-tandem mass spectrometric method. Journal of Pharmaceutical and Biomedical Analysis, 2008, 48, 1392-1396.	2.8	87
100	Efficacy and safety of ketamine in refractory status epilepticus in children. Neurology, 2012, 79, 2355-2358.	1.1	87
101	Mutations in GABRB3. Neurology, 2017, 88, 483-492.	1.1	87
102	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
103	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. Brain, 2013, 136, 3378-3394.	7.6	85
104	Bilateral periventricular nodular heterotopia due to filamin 1 gene mutation: widespread glomeruloid microvascular anomaly and dysplastic cytoarchitecture in the cerebral cortex. Acta Neuropathologica, 2002, 104, 649-657.	7.7	84
105	Coâ€occurring malformations of cortical development and <scp>SCN</scp>1A gene mutations. Epilepsia, 2014, 55, 1009-1019.	5.1	84
106	GNAO1 encephalopathy. Neurology: Genetics, 2017, 3, e143.	1.9	84
107	Benign childhood focal epilepsies. Epilepsia, 2012, 53, 9-18.	5.1	83
108	Targeted sequencing of 351 candidate genes for epileptic encephalopathy in a large cohort of patients. Molecular Genetics & Genomic Medicine, 2016, 4, 568-580.	1.2	83

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109	Genetic Malformations of the Cerebral Cortex and Epilepsy. Epilepsia, 2005, 46, 32-37.	5.1	82
110	Delineating SPTAN1 associated phenotypes: from isolated epilepsy to encephalopathy with progressive brain atrophy. Brain, 2017, 140, 2322-2336.	7.6	82
111	Topiramate and its clinical applications in epilepsy. Expert Opinion on Pharmacotherapy, 2006, 7, 811-823.	1.8	78
112	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
113	Mosaic SCN1A Mutation in Familial Severe Myoclonic Epilepsy of Infancy. Epilepsia, 2006, 47, 1737-1740.	5.1	76
114	Aberrant Inclusion of a Poison Exon Causes Dravet Syndrome and Related SCN1A-Associated Genetic Epilepsies. American Journal of Human Genetics, 2018, 103, 1022-1029.	6.2	76
115	Topiramate concentrations in neonates treated with prolonged whole body hypothermia for hypoxic ischemic encephalopathy. Epilepsia, 2009, 50, 2355-2361.	5.1	75
116	PRRT2 phenotypic spectrum includes sporadic and fever-related infantile seizures. Neurology, 2012, 79, 2104-2108.	1.1	75
117	Identification of a Duplication of Xq28 Associated with Bilateral Periventricular Nodular Heterotopia. American Journal of Human Genetics, 1997, 61, 379-387.	6.2	74
118	Epileptogenic brain malformations: clinical presentation, malformative patterns and indications for genetic testing. Seizure: the Journal of the British Epilepsy Association, 2001, 10, 532-547.	2.0	74
119	Spectrum of phenotypes in female patients with epilepsy due to protocadherin 19 mutations. Epilepsia, 2011, 52, 1251-1257.	5.1	74
120	Copy number variants and infantile spasms: evidence for abnormalities in ventral forebrain development and pathways of synaptic function. European Journal of Human Genetics, 2011, 19, 1238-1245.	2.8	74
121	Characterisation of mutations of the phosphoinositide-3-kinase regulatory subunit, PIK3R2, in perisylvian polymicrogyria: a next-generation sequencing study. Lancet Neurology, The, 2015, 14, 1182-1195.	10.2	74
122	Periventricular nodular heterotopia with overlying polymicrogyria. Brain, 2005, 128, 2811-2821.	7.6	73
123	A nonsense mutation of theATRX gene causing mild mental retardation and epilepsy. Annals of Neurology, 2000, 47, 117-121.	5.3	72
124	Angelman Syndrome. Paediatric Drugs, 2003, 5, 647-661.	3.1	72
125	Earlyâ€onset Absence Epilepsy and Paroxysmal Dyskinesia. Epilepsia, 2002, 43, 1224-1229.	5.1	71
126	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.	5.1	71

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127	Bilateral periventricular nodular heterotopia with mental retardation and frontonasal malformation. Neurology, 1998, 51, 499-503.	1.1	70
128	The spectrum of intermediate <scp>SCN</scp>8Aâ€related epilepsy. Epilepsia, 2019, 60, 830-844.	5.1	70
129	De novo mutations of the ATP6V1A gene cause developmental encephalopathy with epilepsy. Brain, 2018, 141, 1703-1718.	7.6	69
130	Pitfalls in genetic testing: the story of missed SCN1A mutations. Molecular Genetics & Genomic Medicine, 2016, 4, 457-464.	1.2	67
131	Nonsyndromic mental retardation and cryptogenic epilepsy in women withDoublecortin gene mutations. Annals of Neurology, 2003, 54, 30-37.	5.3	65
132	Tandem mass spectrometry, but not T-cell receptor excision circle analysis, identifies newborns with late-onset adenosine deaminase deficiency. Journal of Allergy and Clinical Immunology, 2013, 131, 1604-1610.	2.9	65
133	Genetically induced dysfunctions of Kir2.1 channels: implications for short QT3 syndrome and autismâ€“epilepsy phenotype. Human Molecular Genetics, 2014, 23, 4875-4886.	2.9	65
134	Idiopathic focal epilepsies: the â€œlost tribeâ€• Epileptic Disorders, 2016, 18, 252-288.	1.3	65
135	Oral Topiramate in Neonates with Hypoxic Ischemic Encephalopathy Treated with Hypothermia: A Safety Study. Journal of Pediatrics, 2010, 157, 361-366.	1.8	64
136	CDKL5 geneâ€related epileptic encephalopathy: electroclinical findings in the first year of life. Developmental Medicine and Child Neurology, 2011, 53, 354-360.	2.1	64
137	Next Generation Molecular Diagnosis of Hereditary Spastic Paraplegias: An Italian Cross-Sectional Study. Frontiers in Neurology, 2018, 9, 981.	2.4	64
138	Focal seizures with affective symptoms are a major feature of PCDH19 geneâ€“related epilepsy. Epilepsia, 2012, 53, 2111-2119.	5.1	63
139	Antiepileptic Drug Treatment in Children with Epilepsy. CNS Drugs, 2015, 29, 847-863.	5.9	63
140	Defective mitochondrial rRNA methyltransferase MRM2 causes MELAS-like clinical syndrome. Human Molecular Genetics, 2017, 26, 4257-4266.	2.9	63
141	Safety and efficacy of ganaxolone in patients with CDKL5 deficiency disorder: results from the double-blind phase of a randomised, placebo-controlled, phase 3 trial. Lancet Neurology, The, 2022, 21, 417-427.	10.2	63
142	A New Benign Adult Familial Myoclonic Epilepsy (BAFME) Pedigree Suggesting Linkage to Chromosome 2p11.1-q12.2. Epilepsia, 2004, 45, 190-192.	5.1	62
143	Symmetric polymicrogyria and pachygyria associated with TUBB2B gene mutations. European Journal of Human Genetics, 2012, 20, 995-998.	2.8	61
144	Effectiveness of antiepileptic therapy in patients with PCDH19 mutations. Seizure: the Journal of the British Epilepsy Association, 2016, 35, 106-110.	2.0	61

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145	Genomic <scp>DNA</scp> methylation distinguishes subtypes of human focal cortical dysplasia. Epilepsia, 2019, 60, 1091-1103.	5.1	61
146	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	5.1	61
147	Xp22.3 genomic deletions involving the CDKL5 gene in girls with early onset epileptic encephalopathy. Epilepsia, 2010, 51, 647-654.	5.1	60
148	Physiology of epilepsia partialis continua and subcortical mechanisms of status epilepticus. Epilepsia, 2009, 50, 7-9.	5.1	56
149	Variable epilepsy phenotypes associated with a familial intragenic deletion of the SCN1A gene. Epilepsia, 2010, 51, 2474-2477.	5.1	56
150	Diagnosis of immunodeficiency caused by a purine nucleoside phosphorylase defect by using tandem mass spectrometry on dried blood spots. Journal of Allergy and Clinical Immunology, 2014, 134, 155-159.e3.	2.9	56
151	Dravet syndrome: Treatment options and management of prolonged seizures. Epilepsia, 2019, 60, S39-S48.	5.1	56
152	Valproate as a Mainstay of Therapy for Pediatric Epilepsy. Paediatric Drugs, 2006, 8, 113-129.	3.1	55
153	Autism-epilepsy phenotype with macrocephaly suggests PTEN, but not GLIALCAM, genetic screening. BMC Medical Genetics, 2014, 15, 26.	2.1	55
154	Reciprocal translocations: a trap for cytogenetists?. Human Genetics, 2005, 117, 571-582.	3.8	54
155	Ketamine in refractory convulsive status epilepticus in children avoids endotracheal intubation. Epilepsy and Behavior, 2015, 49, 343-346.	1.7	54
156	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. American Journal of Human Genetics, 2017, 100, 676-688.	6.2	54
157	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.	2.9	53
158	Clinical features and outcome of 6 new patients carrying de novo KCNB1 gene mutations. Neurology: Genetics, 2017, 3, e206.	1.9	53
159	International consensus recommendations on the diagnostic work-up for malformations of cortical development. Nature Reviews Neurology, 2020, 16, 618-635.	10.1	53
160	Myoclonic Absence-Like Seizures and Chromosome Abnormality Syndromes. Epilepsia, 1998, 39, 660-663.	5.1	52
161	7T Epilepsy Task Force Consensus Recommendations on the Use of 7T MRI in Clinical Practice. Neurology, 2021, 96, 327-341.	1.1	52
162	No major role for the EMX2 gene in schizencephaly. American Journal of Medical Genetics, Part A, 2008, 146A, 1142-1150.	1.2	51

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163	Genetic Epilepsy Syndromes Without Structural Brain Abnormalities: Clinical Features and Experimental Models. Neurotherapeutics, 2014, 11, 269-285.	4.4	51
164	Malformations of Cortical Development and Aberrant Cortical Networks: Epileptogenesis and Functional Organization. Journal of Clinical Neurophysiology, 2010, 27, 372-379.	1.7	50
165	Safety and efficacy of topiramate in neonates with hypoxic ischemic encephalopathy treated with hypothermia (NeoNATI): a feasibility study. Journal of Maternal-Fetal and Neonatal Medicine, 2018, 31, 973-980.	1.5	50
166	Neuroimaging and neuropathology of Dravet syndrome. Epilepsia, 2011, 52, 30-34.	5.1	49
167	The Impact of Next-Generation Sequencing on the Diagnosis and Treatment of Epilepsy in Paediatric Patients. Molecular Diagnosis and Therapy, 2017, 21, 357-373.	3.8	49
168	Ketamine for Refractory Status Epilepticus: A Systematic Review. CNS Drugs, 2018, 32, 997-1009.	5.9	49
169	Cortical reflex myoclonus in rett syndrome. Annals of Neurology, 1998, 43, 472-479.	5.3	48
170	Clinical, MRI, and pathological features of polymicrogyria in chromosome 22q11 deletion syndrome. American Journal of Medical Genetics Part A, 2004, 127A, 313-317.	2.4	48
171	Rapid assay of rufinamide in dried blood spots by a new liquid chromatographyâ€“tandem mass spectrometric method. Journal of Pharmaceutical and Biomedical Analysis, 2011, 54, 192-197.	2.8	48
172	A randomized phase <scp>III</scp> trial of adjunctive zonisamide in pediatric patients with partial epilepsy. Epilepsia, 2013, 54, 1473-1480.	5.1	48
173	Severe myoclonic epilepsy in infancy (Dravet syndrome) 30 years later. Epilepsia, 2011, 52, 1-2.	5.1	47
174	The <scp>ENIGMAâ€Epilepsy</scp> working group: Mapping disease from large data sets. Human Brain Mapping, 2022, 43, 113-128.	3.6	47
175	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	7.6	47
176	Bilateral frontoparietal polymicrogyria, Lennoxâ€Gastaut syndrome, and GPR56 gene mutations. Epilepsia, 2009, 50, 1344-1353.	5.1	46
177	Overview of presurgical assessment and surgical treatment of epilepsy from the Italian League Against Epilepsy. Epilepsia, 2013, 54, 35-48.	5.1	45
178	Mutational Analysis of EFHC1 Gene in Italian Families with Juvenile Myoclonic Epilepsy. Epilepsia, 2007, 48, 1686-1690.	5.1	44
179	Epilepsy surgery in Neurofibromatosis Type 1. Epilepsy Research, 2013, 105, 384-395.	1.6	44
180	A prospective study of direct medical costs in a large cohort of consecutively enrolled patients with refractory epilepsy in Italy. Epilepsia, 2015, 56, 1162-1173.	5.1	44

#	Article	IF	Citations
181	TBC1D24-TLDc-related epilepsy exercise-induced dystonia: rescue by antioxidants in a disease model. Brain, 2019, 142, 2319-2335.	7.6	44
182	Trends in pediatric epilepsy surgery in Europe between 2008 and 2015: Countryâ€, centerâ€, and ageâ€specific variation. Epilepsia, 2020, 61, 216-227.	5.1	44
183	Novel brain expression of ClC-1 chloride channels and enrichment of CLCN1 variants in epilepsy. Neurology, 2013, 80, 1078-1085.	1.1	43
184	A Primate-Specific Isoform of PLEKHG6 Regulates Neurogenesis and Neuronal Migration. Cell Reports, 2018, 25, 2729-2741.e6.	6.4	43
185	Clinical spectrum of STX1B -related epileptic disorders. Neurology, 2019, 92, e1238-e1249.	1.1	43
186	Efficacy and Safety of Fenfluramine for the Treatment of Seizures Associated With Lennox-Gastaut Syndrome. JAMA Neurology, 2022, 79, 554.	9.0	43
187	The Î±_{2B}â€adrenergic receptor is mutant in cortical myoclonus and epilepsy. Annals of Neurology, 2014, 75, 77-87.	5.3	42
188	Somatic double-hit in MTOR and RPS6 in hemimegalencephaly with intractable epilepsy. Human Molecular Genetics, 2019, 28, 3755-3765.	2.9	42
189	<scp>SCN3A</scp>â€Related Neurodevelopmental Disorder: A Spectrum of Epilepsy and Brain Malformation. Annals of Neurology, 2020, 88, 348-362.	5.3	42
190	Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. Genome Medicine, 2020, 12, 28.	8.2	42
191	Safety and Tolerability of Antiepileptic Drug Treatment in Children with Epilepsy. Drug Safety, 2012, 35, 519-533.	3.2	41
192	Epilepsy surgery of â€œlow grade epilepsy associated neuroepithelial tumorsâ€: A retrospective nationwide Italian study. Epilepsia, 2017, 58, 1832-1841.	5.1	41
193	Dravet syndrome: The main issues. European Journal of Paediatric Neurology, 2012, 16, S1-S4.	1.6	40
194	The inclusion of ADA-SCID in expanded newborn screening by tandem mass spectrometry. Journal of Pharmaceutical and Biomedical Analysis, 2014, 88, 201-206.	2.8	40
195	De novo and inherited private variants in MAP1B in periventricular nodular heterotopia. PLoS Genetics, 2018, 14, e1007281.	3.5	40
196	Adolescent Onset of Idiopathic Photosensitive Occipital Epilepsy After Remission of Benign Rolandic Epilepsy. Epilepsia, 1997, 38, 777-781.	5.1	39
197	Therapeutic drug monitoring of carbamazepine and its metabolite in children from dried blood spots using liquid chromatography and tandem mass spectrometry. Journal of Pharmaceutical and Biomedical Analysis, 2015, 109, 164-170.	2.8	39
198	Comparative efficacy of antiepileptic drugs in children and adolescents: A network metaâ€analysis. Epilepsia, 2018, 59, 297-314.	5.1	39

#	Article	IF	Citations
199	Defining the electroclinical phenotype and outcome of PCDH19â€related epilepsy: A multicenter study. Epilepsia, 2018, 59, 2260-2271.	5.1	39
200	Perception of impact of Dravet syndrome on children and caregivers in multiple countries: looking beyond seizures. Developmental Medicine and Child Neurology, 2019, 61, 1229-1236.	2.1	39
201	Multicenter Validation of a Deep Learning Detection Algorithm for Focal Cortical Dysplasia. Neurology, 2021, 97, e1571-e1582.	1.1	39
202	Efficacy of ketamine in refractory convulsive status epilepticus in children: a protocol for a sequential design, multicentre, randomised, controlled, open-label, non-profit trial (KETASER01). BMJ Open, 2016, 6, e011565.	1.9	38
203	Genetic heterogeneity in infantile spasms. Epilepsy Research, 2019, 156, 106181.	1.6	38
204	Monogenic Epilepsies. Neurology, 2021, 97, 817-831.	1.1	38
205	Neuroprotective effects of topiramate and memantine in combination with hypothermia in hypoxic-ischemic brain injury in vitro and in vivo. Neuroscience Letters, 2018, 668, 103-107.	2.1	37
206	SLC25A10 biallelic mutations in intractable epileptic encephalopathy with complex I deficiency. Human Molecular Genetics, 2018, 27, 499-504.	2.9	37
207	Efficacy and safety of Fenfluramine hydrochloride for the treatment of seizures in Dravet syndrome: A realâ€world study. Epilepsia, 2020, 61, 2405-2414.	5.1	37
208	Atlas of lesion locations and postsurgical seizure freedom in focal cortical dysplasia: A MELD study. Epilepsia, 2022, 63, 61-74.	5.1	36
209	Characteristics of a large population of patients with refractory epilepsy attending tertiary referral centers in Italy. Epilepsia, 2010, 51, 921-925.	5.1	35
210	New clinical and molecular insights on Barth syndrome. Orphanet Journal of Rare Diseases, 2013, 8, 27.	2.7	35
211	Adjunctive zonisamide therapy in the longâ€term treatment of children with partial epilepsy: Results of an openâ€label extension study of a phase <scp>III</scp>, randomized, doubleâ€blind, placeboâ€controlled trial. Epilepsia, 2014, 55, 568-578.	5.1	35
212	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. Brain, 2021, 144, 1435-1450.	7.6	35
213	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
214	Familial Epilepsy with Unilateral and Bilateral Malformations of Cortical Development. Epilepsia, 1999, 40, 47-51.	5.1	34
215	Dravet syndrome as part of the clinical and genetic spectrum of sodium channel epilepsies and encephalopathies. Epilepsia, 2019, 60, S2-S7.	5.1	34
216	Different Neurophysiologic Patterns of Myoclonus Characterize Lennox-Gastaut Syndrome and Myoclonic Astaticâ€ƒEpilepsy. Epilepsia, 2002, 43, 609-615.	5.1	33

#	Article	IF	Citations
217	Unbalanced GLA mRNAs ratio quantified by real-time PCR in Fabry patients' fibroblasts results in Fabry disease. European Journal of Human Genetics, 2008, 16, 1311-1317.	2.8	33
218	Congenital disorders of glycosylation presenting as epileptic encephalopathy with migrating partial seizures in infancy. Developmental Medicine and Child Neurology, 2016, 58, 1085-1091.	2.1	33
219	De novo KCNA1 variants in the PVP motif cause infantile epileptic encephalopathy and cognitive impairment similar to recurrent KCNA2 variants. American Journal of Medical Genetics, Part A, 2018, 176, 1748-1752.	1.2	33
220	Subcortical heterotopic gray matter brain malformations. Neurology, 2019, 93, e1360-e1373.	1.1	33
221	Expanding the genetic and phenotypic relevance of KCNB1 variants in developmental and epileptic encephalopathies: 27 new patients and overview of the literature. Human Mutation, 2020, 41, 69-80.	2.5	33
222	Temporal lobe epilepsy surgery in children and adults: A multicenter study. Epilepsia, 2021, 62, 128-142.	5.1	33
223	Genomeâ€wide linkage metaâ€analysis identifies susceptibility loci at 2q34 and 13q31.3 for genetic generalized epilepsies. Epilepsia, 2012, 53, 308-318.	5.1	32
224	Nocturnal frontal lobe epilepsy with paroxysmal arousals due to CHRNA2 loss of function. Neurology, 2015, 84, 1520-1528.	1.1	32
225	The hyperkinetic movement disorder of <scp>FOXG</scp>1â€related epilepticâ€“dyskinetic encephalopathy. Developmental Medicine and Child Neurology, 2016, 58, 93-97.	2.1	32
226	Neuroimaging in mitochondrial disorders. Essays in Biochemistry, 2018, 62, 409-421.	4.7	32
227	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	5.1	32
228	Intrinsic epileptogenicity of gangliogliomas may be independent from co-occurring focal cortical dysplasia. Epilepsy Research, 2011, 97, 208-213.	1.6	31
229	Peritrigonal and temporo-occipital heterotopia with corpus callosum and cerebellar dysgenesis. Neurology, 2012, 79, 1244-1251.	1.1	31
230	Next-generation sequencing approach to hyperCKemia. Neurology: Genetics, 2019, 5, e352.	1.9	31
231	Epilepsy and malformations of the cerebral cortex. Epileptic Disorders, 2003, 5 Suppl 2, S9-26.	1.3	31
232	Dravet syndrome and SCN1A gene mutation relatedâ€epilepsies: cognitive impairment and its determinants. Developmental Medicine and Child Neurology, 2011, 53, 11-15.	2.1	30
233	The medical and surgical treatment of tumoral seizures: Current and future perspectives. Epilepsia, 2013, 54, 84-90.	5.1	30
234	Epilepsy in ring chromosome 20 syndrome. Epilepsy Research, 2016, 128, 83-93.	1.6	30

#	Article	IF	Citations
235	Pyridoxine-5â€²-phosphate oxidase (Pnpo) deficiency: Clinical and biochemical alterations associated with the C.347g > A (P.Â·Arg116gln) mutation. Molecular Genetics and Metabolism, 2017, 122, 135-142.	1.1	30
236	Tubulinopathies continued: refining the phenotypic spectrum associated with variants in TUBG1. European Journal of Human Genetics, 2018, 26, 1132-1142.	2.8	30
237	Outcome after hemispherotomy in patients with intractable epilepsy: Comparison of techniques in the Italian experience. Epilepsy and Behavior, 2019, 93, 22-28.	1.7	30
238	Phenotypic and genetic spectrum of epilepsy with myoclonic atonic seizures. Epilepsia, 2020, 61, 995-1007.	5.1	30
239	Climate change and epilepsy: Insights from clinical and basic science studies. Epilepsy and Behavior, 2021, 116, 107791.	1.7	30
240	Real-life survey of pitfalls and successes of precision medicine in genetic epilepsies. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 1044-1052.	1.9	30
241	Periventricular heterotopia with white matter abnormalities associated with 6p25 deletion. American Journal of Medical Genetics, Part A, 2012, 158A, 1793-1797.	1.2	29
242	Identity by descent fine mapping of familial adult myoclonus epilepsy (FAME) to 2p11.2â€“2q11.2. Human Genetics, 2016, 135, 1117-1125.	3.8	29
243	Clinical and neuroimaging features of autosomal recessive spastic paraplegia 35 (SPG35): case reports, new mutations, and brief literature review. Neurogenetics, 2018, 19, 123-130.	1.4	29
244	The application of artificial intelligence to understand the pathophysiological basis of psychogenic nonepileptic seizures. Epilepsy and Behavior, 2018, 87, 167-172.	1.7	29
245	Panic Attacks Mistaken for Relapse of Epilepsy. Epilepsia, 1995, 36, 48-51.	5.1	28
246	Neuropsychological Findings in Idiopathic Occipital Lobe Epilepsies. Epilepsia, 2006, 47, 76-78.	5.1	28
247	Borderline Dravet syndrome: A useful diagnostic category?. Epilepsia, 2011, 52, 10-12.	5.1	28
248	Safety and efficacy of topiramate in neonates with hypoxic ischemic encephalopathy treated with hypothermia (NeoNATI). BMC Pediatrics, 2012, 12, 144.	1.7	28
249	Symptoms of anxiety and depression and family's quality of life in children and adolescents with epilepsy. Epilepsy and Behavior, 2018, 79, 146-153.	1.7	28
250	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	2.4	28
251	Impaired surface Î±Î²Î³ GABAA receptor expression in familial epilepsy due to a GABRG2 frameshift mutation. Neurobiology of Disease, 2013, 50, 135-141.	4.4	27
252	Clinical relevance of short-chain acyl-CoA dehydrogenase (SCAD) deficiency: Exploring the role of new variants including the first SCAD-disease-causing allele carrying a synonymous mutation. BBA Clinical, 2016, 5, 114-119.	4.1	27

#	Article	IF	Citations
253	Increasing volume and complexity of pediatric epilepsy surgery with stable seizure outcome between 2008 and 2014: A nationwide multicenter study. Epilepsy and Behavior, 2017, 75, 151-157.	1.7	27
254	De novo TBR1 variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. European Journal of Human Genetics, 2020, 28, 770-782.	2.8	27
255	Autosomal Dominant Early-onset Cortical Myoclonus, Photic-induced Myoclonus, and Epilepsy in a Large Pedigree. Epilepsia, 2006, 47, 1643-1649.	5.1	26
256	Optimizing the Molecular Diagnosis of GALNS: Novel Methods to Define and Characterize Morquio-A Syndrome-Associated Mutations. Human Mutation, 2015, 36, 357-368.	2.5	26
257	Broad phenotypic spectrum and genotype-phenotype correlations in GMPPB-related dystroglycanopathies: an Italian cross-sectional study. Orphanet Journal of Rare Diseases, 2018, 13, 170.	2.7	26
258	Type I sialidosis, a normosomatic lysosomal disease, in the differential diagnosis of late-onset ataxia and myoclonus: An overview. Molecular Genetics and Metabolism, 2020, 129, 47-58.	1.1	26
259	Early infantile epileptic-dyskinetic encephalopathy due to biallelicPIGPmutations. Neurology: Genetics, 2020, 6, e387.	1.9	26
260	Is Focal Cortical Dysplasia/Epilepsy Caused by Somatic MTOR Mutations Always a Unilateral Disorder?. Neurology: Genetics, 2021, 7, e540.	1.9	26
261	Diffuse malformations of cortical development. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 111, 653-665.	1.8	25
262	Leigh-like neuroimaging features associated with new biallelic mutations in OPA1. European Journal of Paediatric Neurology, 2017, 21, 671-677.	1.6	25
263	Unilobar surgery for symptomatic epileptic spasms. Annals of Clinical and Translational Neurology, 2017, 4, 36-45.	3.7	25
264	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
265	Artificial intelligence for classification of temporal lobe epilepsy with ROI-level MRI data: A worldwide ENIGMA-Epilepsy study. NeuroImage: Clinical, 2021, 31, 102765.	2.7	25
266	Profiling PI3K-AKT-MTOR variants in focal brain malformations reveals new insights for diagnostic care. Brain, 2022, 145, 925-938.	7.6	25
267	Spatial centrosome proteome of human neural cells uncovers disease-relevant heterogeneity. Science, 2022, 376, .	12.6	25
268	Characterization of severe action myoclonus in sialidoses. Epilepsy Research, 2011, 94, 86-93.	1.6	24
269	Early clinical features in Dravet syndrome patients with and without SCN1A mutations. Epilepsy Research, 2012, 99, 21-27.	1.6	24
270	Cognitive outcome after epilepsy surgery in children: A controlled longitudinal study. Epilepsy and Behavior, 2017, 73, 23-30.	1.7	24

#	Article	IF	Citations
271	Development and Validation of a Prediction Model for Early Diagnosis of SCN1A -Related Epilepsies. Neurology, 2022, 98, .	1.1	24
272	Drosophila functional screening of de novo variants in autism uncovers damaging variants and facilitates discovery of rare neurodevelopmental diseases. Cell Reports, 2022, 38, 110517.	6.4	24
273	Optimizing the molecular diagnosis of <scp>CDKL</scp>5 geneâ€“related epileptic encephalopathy in boys. Epilepsia, 2014, 55, 1748-1753.	5.1	23
274	Biallelic DMXL2 mutations impair autophagy and cause Ohtahara syndrome with progressive course. Brain, 2019, 142, 3876-3891.	7.6	23
275	Results From an Italian Expanded Access Program on Cannabidiol Treatment in Highly Refractory Dravet Syndrome and Lennoxâ€“Gastaut Syndrome. Frontiers in Neurology, 2021, 12, 673135.	2.4	23
276	Clinical and molecular delineation of <scp>PUS3</scp>â€associated neurodevelopmental disorders. Clinical Genetics, 2021, 100, 628-633.	2.0	23
277	Natural History Study of STXBP1-Developmental and Epileptic Encephalopathy Into Adulthood. Neurology, 2022, 99, .	1.1	23
278	A 7 Mb duplication at 22q13 in a girl with bipolar disorder and hippocampal malformation. American Journal of Medical Genetics, Part A, 2008, 146A, 1754-1760.	1.2	22
279	Focal cortical dysplasia type IIb in the rolandic cortex: Functional reorganization after early surgery documented by passive task functional MRI. Epilepsia, 2012, 53, e141-5.	5.1	22
280	Dried blood spot assay for the quantification of phenytoin using Liquid Chromatography-Mass Spectrometry. Clinica Chimica Acta, 2015, 440, 31-35.	1.1	22
281	Clinical and genetic spectrum of AMPD2-related pontocerebellar hypoplasia type 9. European Journal of Human Genetics, 2018, 26, 695-708.	2.8	22
282	Generalized epilepsies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2019, 161, 3-15.	1.8	22
283	Emerging Role of the Autophagy/Lysosomal Degradative Pathway in Neurodevelopmental Disorders With Epilepsy. Frontiers in Cellular Neuroscience, 2020, 14, 39.	3.7	22
284	Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. Genetics in Medicine, 2021, 23, 1715-1725.	2.4	22
285	A systemsâ€level analysis highlights microglial activation as a modifying factor in common epilepsies. Neuropathology and Applied Neurobiology, 2022, 48, .	3.2	22
286	The genetic and molecular basis of epilepsy. Trends in Molecular Medicine, 2003, 9, 300-306.	6.7	21
287	The epileptic encephalopathies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 111, 619-626.	1.8	21
288	PRIMA1 mutation: a new cause of nocturnal frontal lobe epilepsy. Annals of Clinical and Translational Neurology, 2015, 2, 821-830.	3.7	21

#	Article	IF	Citations
289	Multiple genomic copy number variants associated with periventricular nodular heterotopia indicate extreme genetic heterogeneity. European Journal of Human Genetics, 2019, 27, 909-918.	2.8	21
290	Focal cortical dysplasia: an update on diagnosis and treatment. Expert Review of Neurotherapeutics, 2021, 21, 1213-1224.	2.8	21
291	Extracellular LGALS3BP regulates neural progenitor position and relates to human cortical complexity. Nature Communications, 2021, 12, 6298.	12.8	21
292	How Can Advances in Epilepsy Genetics Lead to Better Treatments and Cures?. Advances in Experimental Medicine and Biology, 2014, 813, 309-317.	1.6	20
293	Distinct epilepsy phenotypes and response to drugs in KCNA1 gainâ€•and lossâ€•of function variants. Epilepsia, 2022, 63, .	5.1	20
294	Nonconvulsive Status Epilepticus Precipitated by Carbamazepine Presenting as Dissociative and Affective Disorders in Adolescents. Journal of Child Neurology, 2005, 20, 693-696.	1.4	19
295	The syndrome of polymicrogyria, thalamic hypoplasia, and epilepsy with CSWS. Neurology, 2016, 86, 1250-1259.	1.1	19
296	Idiopathic Generalized Epilepsies. , 2019, , 121-133.		19
297	Paroxysmal disorders associated with PRRT2 mutations shake up expectations on ion channel genes. Neurology, 2012, 79, 2086-2088.	1.1	18
298	Somatic Overgrowth Predisposes to Seizures in Autism Spectrum Disorders. PLoS ONE, 2013, 8, e75015.	2.5	18
299	A novel inherited SCN1A mutation associated with different neuropsychological phenotypes: Is there a common core deficit?. Epilepsy and Behavior, 2015, 43, 89-92.	1.7	18
300	Bi-allelic GAD1 variants cause a neonatal onset syndromic developmental and epileptic encephalopathy. Brain, 2020, 143, 1447-1461.	7.6	18
301	Large-scale, cell-resolution volumetric mapping allows layer-specific investigation of human brain cytoarchitecture. Biomedical Optics Express, 2021, 12, 3684.	2.9	18
302	Topographic divergence of atypical cortical asymmetry and atrophy patterns in temporal lobe epilepsy. Brain, 2022, 145, 1285-1298.	7.6	18
303	Classification concepts and terminology: Is clinical description assertive and laboratory testing objective?. Epilepsia, 2010, 51, 718-720.	5.1	17
304	Advancing research toward faster diagnosis, better treatment, and end of stigma in epilepsy. Epilepsia, 2019, 60, 1281-1292.	5.1	17
305	Infantile-Onset Syndromic Cerebellar Ataxia and CACNA1G Mutations. Pediatric Neurology, 2020, 104, 40-45.	2.1	17
306	Mutations in the exocyst component EXOC2 cause severe defects in human brain development. Journal of Experimental Medicine, 2020, 217, .	8.5	17

#	Article	IF	Citations
307	Developmental and epilepsy spectrum of KCNB1 encephalopathy with longâ€term outcome. Epilepsia, 2020, 61, 2461-2473.	5.1	17
308	Recessive mutations in SLC35A3 cause early onset epileptic encephalopathy with skeletal defects. American Journal of Medical Genetics, Part A, 2017, 173, 1119-1123.	1.2	16
309	Longâ€term efficacy of addâ€on lacosamide treatment in children and adolescents with refractory epilepsies: A singleâ€center observational study. Epilepsia, 2018, 59, 1004-1010.	5.1	16
310	Automatic detection and sonification of nonmotor generalized onset epileptic seizures: Preliminary results. Brain Research, 2019, 1721, 146341.	2.2	16
311	Ultra-High-Field Targeted Imaging of Focal Cortical Dysplasia: The Intracortical Black Line Sign in Type IIb. American Journal of Neuroradiology, 2019, 40, 2137-2142.	2.4	16
312	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
313	Late-Onset Epileptic Spasms. Journal of Child Neurology, 2015, 30, 153-159.	1.4	15
314	Familial periventricular nodular heterotopia, epilepsy and Melnickâ€“Needles Syndrome caused by a singleFLNAmutation with combined gain-of-function and loss-of-function effects. Journal of Medical Genetics, 2015, 52, 405-412.	3.2	15
315	De novo CLTC variants are associated with a variable phenotype from mild to severe intellectual disability, microcephaly, hypoplasia of the corpus callosum, and epilepsy. Genetics in Medicine, 2020, 22, 797-802.	2.4	15
316	Deciphering the premature mortality in PIGA-CDG â€“ An untold story. Epilepsy Research, 2021, 170, 106530.	1.6	15
317	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. Brain, 2022, 145, 208-223.	7.6	15
318	An examination of the efficacy and safety of fenfluramine in adults, children, and adolescents with Dravet syndrome in a <scp>realâ€world</scp> practice setting: A report from the Fenfluramine European Early Access Program. Epilepsia Open, 2022, 7, 578-587.	2.4	15
319	Segmental facial myoclonus in Moebius syndrome. Movement Disorders, 1999, 14, 1021-1024.	3.9	14
320	Tissue Border Enhancement by inversion recovery MRI at 7.0 Tesla. Neuroradiology, 2014, 56, 517-523.	2.2	14
321	Periventricular nodular heterotopia in Smithâ€Magenis syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 3142-3147.	1.2	14
322	Double-target Antisense U1snRNAs Correct Mis-splicing Due to c.639+861C>T and c.639+919G>A GLA Deep Intronic Mutations. Molecular Therapy - Nucleic Acids, 2016, 5, e380.	5.1	14
323	Biochemical data from the characterization of a new pathogenic mutation of human pyridoxine-5'-phosphate oxidase (PNPO). Data in Brief, 2017, 15, 868-875.	1.0	14
324	Mis-splicing of the GALNS gene resulting from deep intronic mutations as a cause of Morquio a disease. BMC Medical Genetics, 2018, 19, 183.	2.1	14

#	Article	IF	Citations
325	Neurosurgical treatment of subependymal giant cell astrocytomas in tuberous sclerosis complex: a series of 44 surgical procedures in 31 patients. Child's Nervous System, 2020, 36, 951-960.	1.1	14
326	SARS-CoV-2 infection in a patient with propionic acidemia. Orphanet Journal of Rare Diseases, 2020, 15, 306.	2.7	14
327	Functional and pharmacological evaluation of novel GLA variants in Fabry disease identifies six (two) Tj ETQq1 1 0.784314 rgBT /Over 2018, 481, 25-33.	1.1	13
328	Longâ€term efficacy of addâ€on stiripentol treatment in children, adolescents, and young adults with refractory epilepsies: A single center prospective observational study. Epilepsia, 2019, 60, 2255-2262.	5.1	13
329	Drug Development for Rare Paediatric Epilepsies: Current State and Future Directions. Drugs, 2019, 79, 1917-1935.	10.9	13
330	The spectrum of brain malformations and disruptions in twins. American Journal of Medical Genetics, Part A, 2021, 185, 2690-2718.	1.2	13
331	Expanding the genetic and phenotypic spectrum of <scp>CHD2</scp>â€related disease: From early neurodevelopmental disorders to adultâ€onset epilepsy. American Journal of Medical Genetics, Part A, 2022, 188, 522-533.	1.2	13
332	Inâ€frame deletion in FLNA causing familial periventricular heterotopia with skeletal dysplasia in males. American Journal of Medical Genetics, Part A, 2011, 155, 1140-1146.	1.2	12
333	Age-related epileptic encephalopathies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2012, 107, 179-193.	1.8	12
334	Vertical extraventricular functional hemispherotomy: a new variant for hemispheric disconnection. Technical notes and results in three patients. Child's Nervous System, 2015, 31, 2151-2160.	1.1	12
335	SAR prediction in adults and children by combining measured B1+ maps and simulations at 7.0 Tesla. Journal of Magnetic Resonance Imaging, 2016, 44, 1048-1055.	3.4	12
336	Mycophenolate mofetil as induction and long-term maintaining treatment in childhood: Primary angiitis of the central nervous system. Joint Bone Spine, 2017, 84, 353-356.	1.6	12
337	Left inferior frontal cortex can compensate the inhibitory functions of right inferior frontal cortex and preâ€supplementary motor area. Journal of Neuropsychology, 2019, 13, 503-508.	1.4	12
338	Quantitative MRI-Based Analysis Identifies Developmental Limbic Abnormalities in PCDH19 Encephalopathy. Cerebral Cortex, 2020, 30, 6039-6050.	2.9	12
339	Thermal inactivation of SARS COVID-2 virus: Are steam inhalations a potential treatment?. Life Sciences, 2021, 265, 118801.	4.3	12
340	Diverse genetic causes of polymicrogyria with epilepsy. Epilepsia, 2021, 62, 973-983.	5.1	12
341	Monoallelic KIF1A-related disorders: a multicenter cross sectional study and systematic literature review. Journal of Neurology, 2022, 269, 437-450.	3.6	12
342	Pathophysiology of myoclonic epilepsies. Advances in Neurology, 2005, 95, 23-46.	0.8	12

#	Article	IF	Citations
343	Corpus callosum agenesis, severe mental retardation, epilepsy, and dyskinetic quadriparesis due to a novel mutation in the homeodomain of ARX. , 2011, 155, 892-897.		11
344	Integration of PCR-Sequencing Analysis with Multiplex Ligation-Dependent Probe Amplification for Diagnosis of Hereditary Fructose Intolerance. JIMD Reports, 2012, 6, 31-37.	1.5	11
345	Do mutations in SCN1B cause Dravet syndrome?. Epilepsy Research, 2013, 103, 97-100.	1.6	11
346	Myoclonus and epilepsy. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 111, 667-679.	1.8	11
347	Multimodal fiberâ€probe spectroscopy allows detecting epileptogenic focal cortical dysplasia in children. Journal of Biophotonics, 2017, 10, 896-904.	2.3	11
348	Pre-diagnosing and managing patients with GM1 gangliosidosis and related disorders by the evaluation of GM1 ganglioside content. Scientific Reports, 2019, 9, 17684.	3.3	11
349	Spectrum of Phenotypic, Genetic, and Functional Characteristics in Patients With Epilepsy With KCNC2 Pathogenic Variants. Neurology, 2022, 98, .	1.1	11
350	Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis. Brain, 2022, 145, 2687-2703.	7.6	11
351	Eventâ€based modeling in temporal lobe epilepsy demonstrates progressive atrophy from crossâ€sectional data. Epilepsia, 2022, 63, 2081-2095.	5.1	11
352	Nodular heterotopia is built upon layers. Neurology, 2009, 73, 742-743.	1.1	10
353	Epimutations in both the TESK2 and MMACHC promoters in the Epi-cblC inherited disorder of intracellular metabolism of vitamin B12. Clinical Epigenetics, 2022, 14, 52.	4.1	10
354	Familial Lennoxâ€Gastaut syndrome in male siblings with a novel DCX mutation and anterior pachygyria. Epilepsia, 2010, 51, 1902-1905.	5.1	9
355	Symptomatic and presumed symptomatic focal epilepsies in childhood: An observational, prospective multicentre study. Epilepsia, 2016, 57, 1808-1816.	5.1	9
356	Pitfalls in the detection of gross gene rearrangements using MLPA in Fabry disease. Clinica Chimica Acta, 2016, 452, 82-86.	1.1	9
357	Progressive myoclonus epilepsy in Gaucher Disease due to a new Glyâ€“Gly mutation causing loss of an Exonic Splicing Enhancer. Journal of Neurology, 2019, 266, 92-101.	3.6	9
358	Dysgraphia as a Mild Expression of Dystonia in Children with Absence Epilepsy. PLoS ONE, 2015, 10, e0130883.	2.5	9
359	Practitioner Review: Use of antiepileptic drugs in children. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2006, 47, 115-126.	5.2	8
360	Polymicrogyria and epilepsy. Epilepsia, 2010, 51, 10-12.	5.1	8

#	Article	IF	Citations
361	Deep Convolutional Networks for Automated Detection of Epileptogenic Brain Malformations. Lecture Notes in Computer Science, 2018, , 490-497.	1.3	8
362	Lesional and non-lesional epilepsies: A blurring genetic boundary. European Journal of Paediatric Neurology, 2020, 24, 24-29.	1.6	8
363	Phenotypic Spectrum of Seizure Disorders in MBD5-Associated Neurodevelopmental Disorder. Neurology: Genetics, 2021, 7, e579.	1.9	8
364	Angiocentric glioma-associated seizures: The possible role of EATT2, pyruvate carboxylase and glutamine synthetase. Seizure: the Journal of the British Epilepsy Association, 2021, 86, 152-154.	2.0	8
365	Impaired object identification in idiopathic childhood occipital epilepsy. Epilepsia, 2012, 53, 686-694.	5.1	7
366	Mitochondrial respiratory chain defects in skin fibroblasts from patients with Dravet syndrome. Neurological Sciences, 2015, 36, 2151-2155.	1.9	7
367	Focal Cortical Dysplasia IIIa in Hippocampal Sclerosis-Associated Epilepsy: Anatomo-Electro-Clinical Profile and Surgical Results From a Multicentric Retrospective Study. Neurosurgery, 2021, 88, 384-393.	1.1	7
368	Somatic Focal Copy Number Gains of Noncoding Regions of Receptor Tyrosine Kinase Genes in Treatment-Resistant Epilepsy. Journal of Neuropathology and Experimental Neurology, 2021, 80, 160-168.	1.7	7
369	Morquio B disease: From pathophysiology towards diagnosis. Molecular Genetics and Metabolism, 2021, 132, 180-188.	1.1	7
370	Multicenter prospective longitudinal study in 34 patients with Dravet syndrome: Neuropsychological development in the first six years of life. Brain and Development, 2021, 43, 419-430.	1.1	7
371	Consensus statements on the information to deliver after a febrile seizure. European Journal of Pediatrics, 2021, 180, 2993-2999.	2.7	7
372	Pathogenic <scp>MAST3</scp> Variants in the <scp>STK</scp> Domain Are Associated with Epilepsy. Annals of Neurology, 2021, 90, 274-284.	5.3	7
373	Malformations of Cortical Development. , 2012, , 202-231.		7
374	GM3 synthase deficiency in non-Amish patients. Genetics in Medicine, 2022, 24, 492-498.	2.4	7
375	Prospective Evaluation of Ghrelin and Des-Acyl Ghrelin Plasma Levels in Children with Newly Diagnosed Epilepsy: Evidence for Reduced Ghrelin-to-Des-Acyl Ghrelin Ratio in Generalized Epilepsies. Journal of Personalized Medicine, 2022, 12, 527.	2.5	7
376	Networks Underlie Temporal Onset of Dysplasiaâ€Related Epilepsy: A <scp>MELD</scp> Study. Annals of Neurology, 2022, 92, 503-511.	5.3	7
377	Fast Progression of Cerebellar Atrophy in PLA2G6-Associated Infantile Neuronal Axonal Dystrophy. Cerebellum, 2017, 16, 742-745.	2.5	6
378	Relationships Between Morphologic and Functional Patterns in the Polymicrogyric Cortex. Cerebral Cortex, 2018, 28, 1076-1086.	2.9	6

#	Article	IF	Citations
379	Cortical formation abnormalities on foetal MR imaging: a proposed classification system trialled on 356 cases from Italian and UK centres. European Radiology, 2020, 30, 5250-5260.	4.5	6
380	Basal Ganglia Dysmorphism in Patients With Aicardi Syndrome. Neurology, 2021, 96, e1319-e1333.	1.1	6
381	Cerebral folate transporter deficiency syndrome in three siblings: Why genetic testing for developmental and epileptic encephalopathies should be performed early and include the <scp>FOLR1</scp> gene. American Journal of Medical Genetics, Part A, 2021, 185, 2526-2531.	1.2	6
382	Migrating Focal Seizures and Myoclonic Status in ARV1- Related Encephalopathy. Neurology: Genetics, 2021, 7, e593.	1.9	6
383	PRDX1 gene-related epi-cblC disease is a common type of inborn error of cobalamin metabolism with mono- or bi-allelic MMACHC epimutations. Clinical Epigenetics, 2021, 13, 137.	4.1	6
384	Expression of glutamine synthetase in balloon cells: a basis of their antiepileptic role?. , 2015, 34, 83-88.		6
385	Monoallelic and biallelic mutations inRELNunderlie a graded series of neurodevelopmental disorders. Brain, 2022, 145, 3274-3287.	7.6	6
386	Efficacy and Safety of Long-Term Treatment with Stiripentol in Children and Adults with Drug-Resistant Epilepsies: A Retrospective Cohort Study of 196 Patients. Drugs - Real World Outcomes, 2022, 9, 451-461.	1.6	6
387	Sydenham's chorea in a girl with juvenile idiopathic arthritis treated with antiâ€TNFÎ± therapy. Movement Disorders, 2010, 25, 511-514.	3.9	5
388	Screening of Lysosomal Storage Disorders: Application of the Online Trapping-and-Cleanup Liquid Chromatography/Mass Spectrometry Method for Mucopolysaccharidosis I. European Journal of Mass Spectrometry, 2013, 19, 497-503.	1.0	5
389	What is the role of next generation sequencing in status epilepticus?. Epilepsy and Behavior, 2019, 101, 106373.	1.7	5
390	Defining and expanding the phenotype of QARS-associated developmental epileptic encephalopathy. Neurology: Genetics, 2019, 5, e373.	1.9	5
391	Shedding light on dark genes: enhanced targeted resequencing by optimizing the combination of enrichment technology and DNA fragment length. Scientific Reports, 2020, 10, 9424.	3.3	5
392	De novo FZR1 loss-of-function variants cause developmental and epileptic encephalopathies. Brain, 2022, 145, 1684-1697.	7.6	5
393	SCN1A mutations in focal epilepsy with auditory features: widening the spectrum of GEFS plus. Epileptic Disorders, 2019, 21, 185-191.	1.3	5
394	Autosomal dominant cortical myoclonus and epilepsy (ADCME) with linkage to chromosome 2p11.1-q12.2. Advances in Neurology, 2005, 95, 273-9.	0.8	5
395	SEMA6B variants cause intellectual disability and alter dendritic spine density and axon guidance. Human Molecular Genetics, 2022, 31, 3325-3340.	2.9	5
396	Neurofibromatoses. , 0, , 183-188.		4

#	Article	IF	Citations
397	Biotinidase deficiency due to a de novo mutation or gonadal mosaicism in a first child. Clinica Chimica Acta, 2015, 445, 70-72.	1.1	4
398	Intra-individual plasticity of the TAZ gene leading to different heritable mutations in siblings with Barth syndrome. European Journal of Human Genetics, 2015, 23, 1708-1712.	2.8	4
399	Development and validation of a 2nd tier test for identification of purine nucleoside phosphorylase deficiency patients during expanded newborn screening by liquid chromatography-tandem mass spectrometry. Clinical Chemistry and Laboratory Medicine, 2016, 54, 627-32.	2.3	4
400	Testing HLA-B*15:02. Neurology, 2016, 86, 1080-1081.	1.1	4
401	Hemicerebellitis can drive handedness shift. Cerebellum and Ataxias, 2017, 4, 14.	1.9	4
402	Unstable non-coding pentanucleotide repeats destabilize cortical excitability. Brain, 2018, 141, 2232-2235.	7.6	4
403	No evidence for a BRD 2 promoter hypermethylation inÂblood leukocytes of Europeans with juvenile myoclonicÂepilepsy. Epilepsia, 2019, 60, e31-e36.	5.1	4
404	The Diagnostic Approach to Mitochondrial Disorders in Children in the Era of Next-Generation Sequencing: A 4-Year Cohort Study. Journal of Clinical Medicine, 2021, 10, 3222.	2.4	4
405	Genetic and neuroradiological heterogeneity of double cortex syndrome. Annals of Neurology, 2000, 47, 265-269.	5.3	4
406	A new strategy implementing mass spectrometry in the diagnosis of congenital disorders of N-glycosylation (CDG). Clinical Chemistry and Laboratory Medicine, 2021, 59, 165-171.	2.3	4
407	Rare and Complex Epilepsies from Childhood to Adulthood: Requirements for Separate Management or Scope for a Lifespan Holistic Approach?. Current Neurology and Neuroscience Reports, 2021, 21, 65.	4.2	4
408	Epilepsy and paroxysmal dyskinesia: co-occurrence and differential diagnosis. Advances in Neurology, 2002, 89, 433-41.	0.8	4
409	Epileptogenic brain malformations: clinical presentation, malformative patterns and indications for genetic testing. Seizure: the Journal of the British Epilepsy Association, 2002, 11 Suppl A, 532-43; quiz 544-7.	2.0	4
410	Orphan Drug Use in Patients With Rare Diseases: A Population-Based Cohort Study. Frontiers in Pharmacology, 2022, 13, .	3.5	4
411	Hypothalamic hamartoma and gelastic epilepsy. , 2011, , 449-453.		3
412	Progressive hemispheric shrinking in hemimegalencephaly: a possible role for seizureâ€related neuronal loss. Developmental Medicine and Child Neurology, 2008, 50, 553-557.	2.1	3
413	Focal cortical dysplasia and related variants. , 0, , 293-297.		3
414	The etiological classification of epilepsy. , 2011, , 21-23.		3

#	Article	IF	Citations
415	Bilateral Perysilvian Polymicrogyria With Cerebellar Dysplasia and Ectopic Neurohypophysis. Journal of Child Neurology, 2011, 26, 361-365.	1.4	3
416	Language Regression Associated With Autistic Regression and Electroencephalographic (EEG) Abnormalities. Journal of Child Neurology, 2014, 29, 855-859.	1.4	3
417	Dravet syndrome. Neurology, 2016, 87, 245-246.	1.1	3
418	Age-related differences in audiovisual interactions of semantically different stimuli.. Developmental Psychology, 2017, 53, 138-148.	1.6	3
419	Genetic testing to prevent adverse reactions to antiepileptic drugs. Neurology, 2018, 90, 155-156.	1.1	3
420	Familial dominant epilepsy and mild pachygyria associated with a constitutional LIS1 mutation. American Journal of Medical Genetics, Part A, 2018, 176, 2808-2812.	1.2	3
421	Broadening phenotype of adenylosuccinate lyase deficiency: A novel clinical pattern resembling neuronal ceroid lipofuscinosis. Molecular Genetics and Metabolism Reports, 2019, 21, 100502.	1.1	3
422	KCNQ2 encephalopathy manifesting with Rett-like features. Neurology: Genetics, 2020, 6, e510.	1.9	3
423	Patterns and predictors of language representation and the influence of epilepsy surgery on language reorganization in children and young adults with focal lesional epilepsy. PLoS ONE, 2020, 15, e0238389.	2.5	3
424	A survey of the European Reference Network EpiCARE on clinical practice for selected rare epilepsies. Epilepsia Open, 2021, 6, 160-170.	2.4	3
425	Early-onset bradykinetic rigid syndrome and reflex seizures in a child with PURA syndrome. Epileptic Disorders, 2021, 23, 745-748.	1.3	3
426	Defective IGF-1 prohormone N-glycosylation and reduced IGF-1 receptor signaling activation in congenital disorders of glycosylation. Cellular and Molecular Life Sciences, 2022, 79, 150.	5.4	3
427	The genetic contribution to epilepsy: the known and missing heritability. , 0, , 62-66.		2
428	Pyridoxine-dependent epilepsy. , 0, , 237-241.		2
429	Urea cycle disorders. , 2011, , 246-248.		2
430	Psychiatric disorders. , 0, , 593-606.		2
431	Causes of non-convulsive status epilepticus in adults. , 2011, , 752-758.		2
432	Non-accidental brain injury. , 0, , 425-432.		2

#	Article	IF	Citations
433	HIV infection. , 0, , 520-527.		2
434	Contractions in the second polyA tract of ARX are rare, nonâ€pathogenic polymorphisms. American Journal of Medical Genetics, Part A, 2011, 155, 164-167.	1.2	2
435	Computer-based automatic identification of neurons in gigavoxel-sized 3D human brain images. , 2015, 2015, 7724-7.		2
436	Malformations of Cortical Development. , 2017, , 218-225.		2
437	A novel developmental encephalopathy with epilepsy and hyperkinetic movement disorders associated with a deletion of the sodium channel gene cluster on chromosome 2q24.3. Parkinsonism and Related Disorders, 2019, 68, 1-3.	2.2	2
438	Epilepsy of infancy with migrating focal seizures or rigidity and multifocal seizure syndrome, lethal neonatal? Different emphases on a severe phenotype. Developmental Medicine and Child Neurology, 2020, 62, 1012-1012.	2.1	2
439	Bi-allelic variants in MTMR5/SBF1 cause Charcot-Marie-Tooth type 4B3 featuring mitochondrial dysfunction. BMC Medical Genomics, 2021, 14, 157.	1.5	2
440	Posterior Lissencephaly Associated with Subcortical Band Heterotopia Due to a Variation in the CEP85L Gene: A Case Report and Refining of the Phenotypic Spectrum. Genes, 2021, 12, 1208.	2.4	2
441	Autosomal dominant nocturnal frontal lobe epilepsy. , 0, , 70-73.		1
442	West syndrome and Lennoxâ€“Gastaut syndrome. , 0, , 119-134.		1
443	Dentato-rubro-pallido-luysian atrophy. , 0, , 139-142.		1
444	Mitochondrial cytopathies. , 0, , 147-157.		1
445	Tuberous sclerosis complex. , 2011, , 177-182.		1
446	Inverted duplicated chromosome 15 (isodicentric chromosome 15). , 2011, , 281-284.		1
447	Polymicrogyria and schizencephaly. , 2011, , 311-321.		1
448	Systemic lupus erythematosus and other collagen vascular diseases. , 0, , 579-584.		1
449	Benign adult familial myoclonic epilepsy. , 0, , 85-90.		1
450	Action myoclonusâ€“renal failure syndrome. , 0, , 169-171.		1

#	Article	IF	Citations
451	Drug-induced seizures. , 0, , 664-673.		1
452	Visual stimuli, photosensitivity, and photosensitive epilepsy. , 0, , 687-694.		1
453	Hot-water epilepsy. , 0, , 713-719.		1
454	Uncommon causes of status epilepticus. , 2011, , 745-751.		1
455	Causes of epilepsia partialis continua. , 2011, , 759-766.		1
456	Agyriaâ€“pachygyria band spectrum. , 2011, , 298-304.		1
457	Arachnoid cysts. , 0, , 341-345.		1
458	Epilepsy after epilepsy surgery. , 0, , 413-424.		1
459	Cavernous malformations. , 0, , 559-564.		1
460	Multiple sclerosis and other acquired demyelinating diseases. , 2011, , 607-611.		1
461	Towards automated neuron tracing via global and local 3D image analysis. , 2016, , .		1
462	Isolated recurrent myelitis in a 7-year-old child with serum aquaporin-4 IgG antibodies. Journal of Neurology, 2017, 264, 179-181.	3.6	1
463	Human Mutations Associated With Brain Malformations Resulting in Hyperexcitability in Rodents. , 2017, , 827-844.		1
464	Dravet syndrome andÂotherÂsodiumÂchannelâ€related encephalopathies. Epilepsia, 2019, 60, S1.	5.1	1
465	Changes in appearance of cortical formation abnormalities in the foetus detected on sequential in utero MR imaging. European Radiology, 2021, 31, 1367-1377.	4.5	1
466	SLC32A1. Neurology, 2021, 96, 831-832.	1.1	1
467	Cerebral malformations. , 2020, , 249-267.		1
468	Meeting report: EpiXchange II brings together European epilepsy research projects to discuss latest advances. Epilepsy Research, 2021, 178, 106811.	1.6	1

#	Article	IF	Citations
469	Pallisterâ€"Killian syndrome: an unusual cause of epileptic spasms. Developmental Medicine and Child Neurology, 2005, 47, 776-779.	2.1	0
470	Subcortical structures and infantile spasms. Developmental Medicine and Child Neurology, 2008, 50, 87-87.	2.1	0
471	Epileptogenesis in idiopathic epilepsy. , 0, , 24-34.		0
472	Mechanisms of epileptogenesis in symptomatic epilepsy. , 0, , 35-42.		0
473	Benign familial neonatal seizures. , 0, , 67-69.		0
474	Severe myoclonic epilepsy of infancy or Dravet syndrome. , 0, , 78-84.		0
475	Benign partial epilepsies of childhood. , 0, , 104-112.		0
476	Unverrichtâ€"Lundborg disease. , 0, , 135-138.		0
477	Neuronal ceroid lipofuscinoses. , 0, , 158-163.		0
478	Sialidosis and Gaucher disease. , 0, , 164-168.		0
479	Progressive myoclonus epilepsies: other rare causes. , 0, , 172-176.		0
480	Sturgeâ€"Weber syndrome. , 0, , 189-195.		0
481	Other neurocutaneous syndromes. , 0, , 196-200.		0
482	Angelman syndrome. , 0, , 201-205.		0
483	Lysosomal disorders and Menkes syndrome. , 0, , 206-211.		0
484	Neuroacanthocytosis. , 0, , 212-215.		0
485	Organic acid, amino acids, and peroxisomal disorders. , 0, , 216-230.		0
486	Porphyria. , 0, , 231-236.		0

#	Article	IF	Citations
487	Rett syndrome and MECP2 and CDKL5 genotypes. , 0, , 242-245.		0
488	Disorders of cobalamin and folate metabolism. , 0, , 252-257.		0
489	Other single-gene disorders. , 0, , 258-264.		0
490	4p (Wolfâ€“Hirschhorn) syndrome. , 0, , 277-280.		0
491	Ring chromosome 20. , 0, , 285-288.		0
492	Inflammatory and immunological diseases of the nervous system. , 0, , 585-592.		0
493	Hydrocephalus and porencephaly. , 0, , 612-617.		0
494	Alzheimer disease and other neurodegenerative diseases. , 0, , 618-624.		0
495	Genetic epilepsy with febrile seizures plus. , 0, , 74-77.		0
496	Lafora body disease. , 0, , 143-146.		0
497	The menstrual cycle and catamenial epilepsy. , 0, , 635-642.		0
498	Metabolic and endocrine-induced seizures. , 0, , 650-654.		0
499	Electrolyte and sugar disturbances. , 0, , 655-663.		0
500	Alcohol- and toxin-induced seizures. , 0, , 674-682.		0
501	How reflex mechanisms cause epilepsy. , 0, , 683-686.		0
502	Startle-induced (and other sensory-induced) epilepsy. , 0, , 695-699.		0
503	Primary reading epilepsy. , 0, , 700-703.		0
504	Auditory-induced epilepsy. , 0, , 704-708.		0

#	Article	IF	Citations
505	Focal reflex seizures â€“ with emphasis on seizures triggered by eating. , 0, , 709-712.		0
506	Reflex epilepsy with higher-level processing. , 0, , 720-722.		0
507	Causes of status epilepticus in children. , 0, , 730-734.		0
508	The causes of convulsive status epilepticus in adults. , 0, , 735-744.		0
509	Hemimegalencephaly. , 0, , 289-292.		0
510	Agenesis of the corpus callosum. , 0, , 305-310.		0
511	Periventricular nodular heterotopia. , 0, , 322-329.		0
512	Microcephaly. , 0, , 330-340.		0
513	Malformations of human cerebral cortex. , 0, , 346-362.		0
514	Hippocampal sclerosis. , 0, , 363-372.		0
515	Neonatal seizures and postneonatal epilepsy â€“ causes. , 0, , 373-381.		0
516	Vaccination and immunization. , 0, , 388-392.		0
517	Open head injury. , 0, , 393-399.		0
518	Closed head injury. , 0, , 400-406.		0
519	De novo epilepsy after neurosurgery. , 0, , 407-412.		0
520	Glioma. , 0, , 433-440.		0
521	Ganglioglioma, dysembryoplastic neuroepithelial tumor, and related tumors. , 0, , 441-448.		0
522	Metastatic disease. , 0, , 459-466.		0

#	Article	IF	Citations
523	Viral encephalitis. , 0, , 467-474.		0
524	Bacterial meningitis and focal suppurative intracranial infections in children. , 0, , 475-481.		0
525	Bacterial meningitis and pyogenic abscess in adults. , 0, , 482-491.		0
526	Other parasitic diseases. , 0, , 501-510.		0
527	Emerging and less common central nervous system viral encephalitides. , 0, , 528-536.		0
528	Cerebral hemorrhage. , 0, , 537-543.		0
529	Cerebral infarction and occult degenerative cerebrovascular disease. , 0, , 544-550.		0
530	Arteriovenous malformations. , 0, , 551-558.		0
531	Other vascular disorders. , 0, , 565-572.		0
532	Rasmussen encephalitis and related conditions. , 0, , 573-578.		0
533	Idiopathic generalized epilepsies. , 0, , 91-103.		0
534	Epilepsy phenotypes and genotype determinants. Neurology, 2014, 83, 1038-1039.	1.1	0
535	Probing focal cortical dysplasia in formalin fixed samples using tissue optical spectroscopy. , 2016, , .		0
536	Fiber-probe optical spectroscopy discriminates normal brain from focal cortical dysplasia in pediatric subjects. , 2017, , .		0
537	Overview of Human Brain Malformations. , 2017, , 179-182.		0
538	Response to the letter to the Editor regarding â€œLeigh-like neuroimaging features associated with new bi-allelic mutations in OPA1â€. European Journal of Paediatric Neurology, 2017, 21, 923.	1.6	0
539	Multimodal fiber-probe spectroscopy as a clinical tool for diagnosing and classifying biological tissues. , 2017, , .		0
540	A Novel Strategy Combining Array-CGH, Whole-exome Sequencing and In Utero Electroporation in Rodents to Identify Causative Genes for Brain Malformations. Journal of Visualized Experiments, 2017, , .	0.3	0

#	Article	IF	Citations
541	Le mycophÃ©nolate mofÃ©til en traitement dâ€™induction et traitement dâ€™entretien Ã long terme dans la prise en charge de la vascularite primitive du systÃ¨me nerveux central chez lâ€™enfant. Revue Du Rhumatisme (Edition Francaise), 2018, 85, 90-94.	0.0	0
542	Malformations of Cortical Development in Newborns: Genetic Aspects. , 2018, , 2113-2124.		0
543	Focal Cortical Dysplasia. , 2019, , 455-565.		0
544	Agyriaâ€“pachygyria band spectrum. , 2019, , 466-474.		0
545	Polymicrogyria and Schizencephaly. , 2019, , 480-491.		0
546	Periventricular Nodular Heterotopia. , 2019, , 492-496.		0
547	Disorders Associated with Tubulinopathies and mTORopathies. , 2019, , 513-520.		0
548	Epilepsy Associated with Ganglioglioma, Dysembryoplastic Neuroepithelial Tumor, and Related Tumors. , 2019, , 570-580.		0
549	Sleep and Epilepsy. , 2019, , 821-829.		0
550	Epileptogenesis in Idiopathic Epilepsy. , 2019, , 8-23.		0
551	Epileptogenesis in Symptomatic Epilepsy. , 2019, , 35-45.		0
552	Animal Models of Causation of Epilepsy. , 2019, , 46-52.		0
553	Approach to the Genetic Diagnosis of Epileptic Encephalopathies and Developmental Encephalopathies with Epilepsy of Early Childhood. , 2019, , 60-68.		0
554	Approach to the Diagnosis Of Childhood-Onset Epilepsy Associated with Developmental Delay. , 2019, , 69-75.		0
555	Approach to the Diagnosis of Cortical Developmental Disorders and their Clinical Genetics. , 2019, , 76-85.		0
556	Benign Partial Epilepsies of Childhood. , 2019, , 134-142.		0
557	Benign Familial Neonatal Epilepsy (BFNE). , 2019, , 143-153.		0
558	Sleep-Related Hypermotor Epilepsy (SHE). , 2019, , 147-153.		0

#	Article	IF	Citations
559	Dravet Syndrome and Other SCN1A Disorders. , 2019, , 158-165.		0
560	Familial Focal Epilepsy with Variable Foci. , 2019, , 171-174.		0
561	PCDH19 Mutations Related Epilepsy: Phenotype and Genotype. , 2019, , 175-187.		0
562	Reply:. American Journal of Neuroradiology, 2020, 41, E12-E12.	2.4	0
563	Breaking up genetic influences on seizure onset, spread, and termination. Neurology, 2020, 95, 667-668.	1.1	0
564	Mirror syndromes regarding <scp>AKT3</scp> mutations: Loss of function variant leading to microcephaly. American Journal of Medical Genetics, Part A, 2020, 182, 2800-2802.	1.2	0
565	Whole Exome Sequencing Is the Minimal Technological Approach in Probands Born to Consanguineous Couples. Genes, 2021, 12, 962.	2.4	0
566	Malformations of Cortical Development: Genetic Aspects. , 2012, , 1131-1136.		0
567	Malformations of Cortical Development in Newborns: Genetic Aspects. , 2016, , 1-12.		0
568	Malformations of Cortical Development in Newborns: Genetic Aspects. , 2018, , 1-13.		0
569	How to Interpret Photoparoxysmal EEG Results?. , 2021, , 175-184.		0
570	Generation of two hiPSC lines (UMILi027-A and UMILi028-A) from early and late-onset Congenital Central hypoventilation Syndrome (CCHS) patients carrying a polyalanine expansion mutation in the PHOX2B gene. Stem Cell Research, 2022, 61, 102781.	0.7	0
571	SLC7A3: In Silico Prediction of a Potential New Cause of Childhood Epilepsy. Neuropediatrics, 2022, 53, 046-051.	0.6	0
572	Epileptic spasms and abnormal neuronal migration. , 0, , 28-30.		0
573	An Enhanced Distributed Computational Platform for Developmental and Epileptic Encephalopathies. , 2022, , .		0