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 <scp>ENIGMAâ€Epilepsy</scp> working group: Mapping disease from large data sets. Human Brain Mapping, 2022, 43, 113-128.	3.6	47
2	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. Brain, 2022, 145, 208-223.	7.6	15
3	Monoallelic KIF1A-related disorders: a multicenter cross sectional study and systematic literature review. Journal of Neurology, 2022, 269, 437-450.	3.6	12
4	A systemsâ€level analysis highlights microglial activation as a modifying factor in common epilepsies. Neuropathology and Applied Neurobiology, 2022, 48, .	3.2	22
5	Profiling PI3K-AKT-MTOR variants in focal brain malformations reveals new insights for diagnostic care. Brain, 2022, 145, 925-938.	7.6	25
6	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
7	De novo FZR1 loss-of-function variants cause developmental and epileptic encephalopathies. Brain, 2022, 145, 1684-1697.	7.6	5
8	Topographic divergence of atypical cortical asymmetry and atrophy patterns in temporal lobe epilepsy. Brain, 2022, 145, 1285-1298.	7.6	18
9	Atlas of lesion locations and postsurgical seizure freedom in focal cortical dysplasia: A MELD study. Epilepsia, 2022, 63, 61-74.	5.1	36
10	Distinct epilepsy phenotypes and response to drugs in KCNA1 gainâ€and lossâ€of function variants. Epilepsia, 2022, 63, .	5.1	20
11	GM3 synthase deficiency in non-Amish patients. Genetics in Medicine, 2022, 24, 492-498.	2.4	7
12	Development and Validation of a Prediction Model for Early Diagnosis of SCN1A -Related Epilepsies. Neurology, 2022, 98, .	1.1	24
13	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
14	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
15	Spectrum of Phenotypic, Genetic, and Functional Characteristics in Patients With Epilepsy With KCNC2 Pathogenic Variants. Neurology, 2022, 98, .	1.1	11
16	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
17	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
18	SLC7A3: In Silico Prediction of a Potential New Cause of Childhood Epilepsy. Neuropediatrics, 2022, 53, 046-051.	0.6	0

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19	Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis. Brain, 2022, 145, 2687-2703.	7.6	11
20	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
21	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
22	Monoallelic and biallelic mutations inRELNunderlie a graded series of neurodevelopmental disorders. Brain, 2022, 145, 3274-3287.	7.6	6
23	An Enhanced Distributed Computational Platform for Developmental and Epileptic Encephalopathies. , 2022, , .		0
24	Efficacy and Safety of Fenfluramine for the Treatment of Seizures Associated With Lennox-Gastaut Syndrome. JAMA Neurology, 2022, 79, 554.	9.0	43
25	Orphan Drug Use in Patients With Rare Diseases: A Population-Based Cohort Study. Frontiers in Pharmacology, 2022, 13, .	3.5	4
26	SEMA6B variants cause intellectual disability and alter dendritic spine density and axon guidance. Human Molecular Genetics, 2022, 31, 3325-3340.	2.9	5
27	Eventâ€based modeling in temporal lobe epilepsy demonstrates progressive atrophy from crossâ€sectional data. Epilepsia, 2022, 63, 2081-2095.	5.1	11
28	Natural History Study of STXBP1-Developmental and Epileptic Encephalopathy Into Adulthood. Neurology, 2022, 99, .	1.1	23
29	Spatial centrosome proteome of human neural cells uncovers disease-relevant heterogeneity. Science, 2022, 376, .	12.6	25
30	Networks Underlie Temporal Onset of Dysplasiaâ€Related Epilepsy: A <scp>MELD</scp> Study. Annals of Neurology, 2022, 92, 503-511.	5.3	7
31	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
32	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
33	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
34	The spectrum of brain malformations and disruptions in twins. American Journal of Medical Genetics, Part A, 2021, 185, 2690-2718.	1.2	13
35	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
36	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	2.4	28

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37	Temporal lobe epilepsy surgery in children and adults: A multicenter study. Epilepsia, 2021, 62, 128-142.	5.1	33
38	Basal Ganglia Dysmorphism in Patients With Aicardi Syndrome. Neurology, 2021, 96, e1319-e1333.	1.1	6
39	Deciphering the premature mortality in PIGA-CDG â€“ An untold story. Epilepsy Research, 2021, 170, 106530.	1.6	15
40	Thermal inactivation of SARS COVID-2 virus: Are steam inhalations a potential treatment?. Life Sciences, 2021, 265, 118801.	4.3	12
41	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
42	Is Focal Cortical Dysplasia/Epilepsy Caused by Somatic MTOR Mutations Always a Unilateral Disorder?. Neurology: Genetics, 2021, 7, e540.	1.9	26
43	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
44	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
45	A survey of the European Reference Network EpiCARE on clinical practice for selected rare epilepsies. Epilepsia Open, 2021, 6, 160-170.	2.4	3
46	SLC32A1. Neurology, 2021, 96, 831-832.	1.1	1
47	Phenotypic Spectrum of Seizure Disorders in MBD5-Associated Neurodevelopmental Disorder. Neurology: Genetics, 2021, 7, e579.	1.9	8
48	Morquio B disease: From pathophysiology towards diagnosis. Molecular Genetics and Metabolism, 2021, 132, 180-188.	1.1	7
49	Climate change and epilepsy: Insights from clinical and basic science studies. Epilepsy and Behavior, 2021, 116, 107791.	1.7	30
50	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
51	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
52	Diverse genetic causes of polymicrogyria with epilepsy. Epilepsia, 2021, 62, 973-983.	5.1	12
53	Focal cortical dysplasia: an update on diagnosis and treatment. Expert Review of Neurotherapeutics, 2021, 21, 1213-1224.	2.8	21
54	Consensus statements on the information to deliver after a febrile seizure. European Journal of Pediatrics, 2021, 180, 2993-2999.	2.7	7

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55	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
56	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
57	Large-scale, cell-resolution volumetric mapping allows layer-specific investigation of human brain cytoarchitecture. Biomedical Optics Express, 2021, 12, 3684.	2.9	18
58	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
59	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
60	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. Brain, 2021, 144, 1435-1450.	7.6	35
61	Migrating Focal Seizures and Myoclonic Status in ARV1- Related Encephalopathy. Neurology: Genetics, 2021, 7, e593.	1.9	6
62	Bi-allelic variants in MTMR5/SBF1 cause Charcot-Marie-Tooth type 4B3 featuring mitochondrial dysfunction. BMC Medical Genomics, 2021, 14, 157.	1.5	2
63	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
64	Whole Exome Sequencing Is the Minimal Technological Approach in Probands Born to Consanguineous Couples. Genes, 2021, 12, 962.	2.4	0
65	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
66	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
67	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
68	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
69	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
70	Clinical and molecular delineation of <scp>PUS3</scp>â€associated neurodevelopmental disorders. Clinical Genetics, 2021, 100, 628-633.	2.0	23
71	Multicenter Validation of a Deep Learning Detection Algorithm for Focal Cortical Dysplasia. Neurology, 2021, 97, e1571-e1582.	1.1	39
72	Monogenic Epilepsies. Neurology, 2021, 97, 817-831.	1.1	38

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73	7T Epilepsy Task Force Consensus Recommendations on the Use of 7T MRI in Clinical Practice. Neurology, 2021, 96, 327-341.	1.1	52
74	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
75	Early-onset bradykinetic rigid syndrome and reflex seizures in a child with PURA syndrome. Epileptic Disorders, 2021, 23, 745-748.	1.3	3
76	How to Interpret Photoparoxysmal EEG Results?. , 2021, , 175-184.		0
77	Extracellular LGALS3BP regulates neural progenitor position and relates to human cortical complexity. Nature Communications, 2021, 12, 6298.	12.8	21
78	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
79	Meeting report: EpiXchange II brings together European epilepsy research projects to discuss latest advances. Epilepsy Research, 2021, 178, 106811.	1.6	1
80	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
81	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
82	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
83	Lesional and non-lesional epilepsies: A blurring genetic boundary. European Journal of Paediatric Neurology, 2020, 24, 24-29.	1.6	8
84	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
85	Infantile-Onset Syndromic Cerebellar Ataxia and CACNA1G Mutations. Pediatric Neurology, 2020, 104, 40-45.	2.1	17
86	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
87	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
88	KCNQ2 encephalopathy manifesting with Rett-like features. Neurology: Genetics, 2020, 6, e510.	1.9	3
89	Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918.	10.2	139
90	Network-based atrophy modeling in the common epilepsies: A worldwide ENIGMA study. Science Advances, 2020, 6, .	10.3	97

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91	SARS-CoV-2 infection in a patient with propionic acidemia. Orphanet Journal of Rare Diseases, 2020, 15, 306.	2.7	14
92	Reply:. American Journal of Neuroradiology, 2020, 41, E12-E12.	2.4	0
93	Mutations in the exocyst component EXOC2 cause severe defects in human brain development. Journal of Experimental Medicine, 2020, 217, .	8.5	17
94	Developmental and epilepsy spectrum of KCNB1 encephalopathy with longâ€term outcome. Epilepsia, 2020, 61, 2461-2473.	5.1	17
95	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
96	Breaking up genetic influences on seizure onset, spread, and termination. Neurology, 2020, 95, 667-668.	1.1	0
97	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
98	International consensus recommendations on the diagnostic work-up for malformations of cortical development. Nature Reviews Neurology, 2020, 16, 618-635.	10.1	53
99	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
100	White matter abnormalities across different epilepsy syndromes in adults: an ENIGMA-Epilepsy study. Brain, 2020, 143, 2454-2473.	7.6	123
101	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
102	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
103	Phenotypic and genetic spectrum of epilepsy with myoclonic atonic seizures. Epilepsia, 2020, 61, 995-1007.	5.1	30
104	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	5.1	32
105	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	7.6	47
106	<scp>SCN3A</scp>â€Related Neurodevelopmental Disorder: A Spectrum of Epilepsy and Brain Malformation. Annals of Neurology, 2020, 88, 348-362.	5.3	42
107	Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. Genome Medicine, 2020, 12, 28.	8.2	42
108	Emerging Role of the Autophagy/Lysosomal Degradative Pathway in Neurodevelopmental Disorders With Epilepsy. Frontiers in Cellular Neuroscience, 2020, 14, 39.	3.7	22

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109	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
110	Quantitative MRI-Based Analysis Identifies Developmental Limbic Abnormalities in PCDH19 Encephalopathy. Cerebral Cortex, 2020, 30, 6039-6050.	2.9	12
111	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
112	Bi-allelic GAD1 variants cause a neonatal onset syndromic developmental and epileptic encephalopathy. Brain, 2020, 143, 1447-1461.	7.6	18
113	Early infantile epileptic-dyskinetic encephalopathy due to biallelicPIGPmutations. Neurology: Genetics, 2020, 6, e387.	1.9	26
114	Cerebral malformations. , 2020, , 249-267.		1
115	The landscape of epilepsy-related GATOR1 variants. Genetics in Medicine, 2019, 21, 398-408.	2.4	137
116	Genetic heterogeneity in infantile spasms. Epilepsy Research, 2019, 156, 106181.	1.6	38
117	Somatic double-hit in MTOR and RPS6 in hemimegalencephaly with intractable epilepsy. Human Molecular Genetics, 2019, 28, 3755-3765.	2.9	42
118	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
119	Generalized epilepsies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2019, 161, 3-15.	1.8	22
120	What is the role of next generation sequencing in status epilepticus?. Epilepsy and Behavior, 2019, 101, 106373.	1.7	5
121	Automatic detection and sonification of nonmotor generalized onset epileptic seizures: Preliminary results. Brain Research, 2019, 1721, 146341.	2.2	16
122	TBC1D24-TLDc-related epilepsy exercise-induced dystonia: rescue by antioxidants in a disease model. Brain, 2019, 142, 2319-2335.	7.6	44
123	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
124	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
125	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	12.8	99
126	Biallelic DMXL2 mutations impair autophagy and cause Ohtahara syndrome with progressive course. Brain, 2019, 142, 3876-3891.	7.6	23

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127	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. Annals of Neurology, 2019, 86, 821-831.	5.3	96
128	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
129	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
130	Advancing research toward faster diagnosis, better treatment, and end of stigma in epilepsy. Epilepsia, 2019, 60, 1281-1292.	5.1	17
131	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
132	Genomic <scp>DNA</scp> methylation distinguishes subtypes of human focal cortical dysplasia. Epilepsia, 2019, 60, 1091-1103.	5.1	61
133	Focal Cortical Dysplasia. , 2019, , 455-565.		0
134	Agyriaâ€“pachygyria band spectrum. , 2019, , 466-474.		0
135	Polymicrogyria and Schizencephaly. , 2019, , 480-491.		0
136	Periventricular Nodular Heterotopia. , 2019, , 492-496.		0
137	Disorders Associated with Tubulinopathies and mTORopathies. , 2019, , 513-520.		0
138	Epilepsy Associated with Ganglioglioma, Dysembryoplastic Neuroepithelial Tumor, and Related Tumors. , 2019, , 570-580.		0
139	Sleep and Epilepsy. , 2019, , 821-829.		0
140	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
141	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	5.1	61
142	The spectrum of intermediate <scp>SCN</scp>8Aâ€related epilepsy. Epilepsia, 2019, 60, 830-844.	5.1	70
143	Epileptogenesis in Idiopathic Epilepsy. , 2019, , 8-23.		0
144	Epileptogenesis in Symptomatic Epilepsy. , 2019, , 35-45.		0

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145	Animal Models of Causation of Epilepsy. , 2019, , 46-52.		0
146	Approach to the Genetic Diagnosis of Epileptic Encephalopathies and Developmental Encephalopathies with Epilepsy of Early Childhood. , 2019, , 60-68.		0
147	Approach to the Diagnosis Of Childhood-Onset Epilepsy Associated with Developmental Delay. , 2019, , 69-75.		0
148	Approach to the Diagnosis of Cortical Developmental Disorders and their Clinical Genetics. , 2019, , 76-85.		0
149	Idiopathic Generalized Epilepsies. , 2019, , 121-133.		19
150	Benign Partial Epilepsies of Childhood. , 2019, , 134-142.		0
151	Benign Familial Neonatal Epilepsy (BFNE). , 2019, , 143-153.		0
152	Sleep-Related Hypermotor Epilepsy (SHE). , 2019, , 147-153.		0
153	Dravet Syndrome and Other SCN1A Disorders. , 2019, , 158-165.		0
154	Familial Focal Epilepsy with Variable Foci. , 2019, , 171-174.		0
155	PCDH19 Mutations Related Epilepsy: Phenotype and Genotype. , 2019, , 175-187.		0
156	No evidence for a BRD 2 promoter hypermethylation inÂblood leukocytes of Europeans with juvenile myoclonicÂepilepsy. Epilepsia, 2019, 60, e31-e36.	5.1	4
157	Clinical spectrum of STX1B -related epileptic disorders. Neurology, 2019, 92, e1238-e1249.	1.1	43
158	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
159	Defining and expanding the phenotype of QARS-associated developmental epileptic encephalopathy. Neurology: Genetics, 2019, 5, e373.	1.9	5
160	Subcortical heterotopic gray matter brain malformations. Neurology, 2019, 93, e1360-e1373.	1.1	33
161	Drug Development for Rare Paediatric Epilepsies: Current State and Future Directions. Drugs, 2019, 79, 1917-1935.	10.9	13
162	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

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163	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
164	Next-generation sequencing approach to hyperCKemia. Neurology: Genetics, 2019, 5, e352.	1.9	31
165	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
166	Dravet syndrome as part of the clinical and genetic spectrum of sodium channel epilepsies and encephalopathies. Epilepsia, 2019, 60, S2-S7.	5.1	34
167	Dravet syndrome andÂotherÂsodiumÂchannelâ€related encephalopathies. Epilepsia, 2019, 60, S1.	5.1	1
168	Dravet syndrome: Treatment options and management of prolonged seizures. Epilepsia, 2019, 60, S39-S48.	5.1	56
169	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
170	SYNGAP1 encephalopathy. Neurology, 2019, 92, e96-e107.	1.1	131
171	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
172	SCN1A mutations in focal epilepsy with auditory features: widening the spectrum of GEFS plus. Epileptic Disorders, 2019, 21, 185-191.	1.3	5
173	Clinical and genetic spectrum of AMPD2-related pontocerebellar hypoplasia type 9. European Journal of Human Genetics, 2018, 26, 695-708.	2.8	22
174	Functional and pharmacological evaluation of novel GLA variants in Fabry disease identifies six (two) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5 2018, 481, 25-33.	1.1	13
175	Analysis of 17 genes detects mutations in 81% of 811 patients with lissencephaly. Genetics in Medicine, 2018, 20, 1354-1364.	2.4	92
176	De novo mutations of the ATP6V1A gene cause developmental encephalopathy with epilepsy. Brain, 2018, 141, 1703-1718.	7.6	69
177	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
178	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
179	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
180	Structural brain abnormalities in the common epilepsies assessed in a worldwide ENIGMA study. Brain, 2018, 141, 391-408.	7.6	352

#	Article	IF	Citations
181	Comparative efficacy of antiepileptic drugs in children and adolescents: A network metaâ€analysis. Epilepsia, 2018, 59, 297-314.	5.1	39
182	Genetic testing to prevent adverse reactions to antiepileptic drugs. Neurology, 2018, 90, 155-156.	1.1	3
183	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
184	SLC25A10 biallelic mutations in intractable epileptic encephalopathy with complex I deficiency. Human Molecular Genetics, 2018, 27, 499-504.	2.9	37
185	Tubulinopathies continued: refining the phenotypic spectrum associated with variants in TUBG1. European Journal of Human Genetics, 2018, 26, 1132-1142.	2.8	30
186	Relationships Between Morphologic and Functional Patterns in the Polymicrogyric Cortex. Cerebral Cortex, 2018, 28, 1076-1086.	2.9	6
187	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
188	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
189	Neurologic phenotypes associated with COL4A1 / 2 mutations. Neurology, 2018, 91, e2078-e2088.	1.1	97
190	Defining the electroclinical phenotype and outcome of PCDH19â€related epilepsy: A multicenter study. Epilepsia, 2018, 59, 2260-2271.	5.1	39
191	Next Generation Molecular Diagnosis of Hereditary Spastic Paraplegias: An Italian Cross-Sectional Study. Frontiers in Neurology, 2018, 9, 981.	2.4	64
192	A Primate-Specific Isoform of PLEKHG6 Regulates Neurogenesis and Neuronal Migration. Cell Reports, 2018, 25, 2729-2741.e6.	6.4	43
193	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
194	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
195	The application of artificial intelligence to understand the pathophysiological basis of psychogenic nonepileptic seizures. Epilepsy and Behavior, 2018, 87, 167-172.	1.7	29
196	Deep Convolutional Networks for Automated Detection of Epileptogenic Brain Malformations. Lecture Notes in Computer Science, 2018, , 490-497.	1.3	8
197	HCN1mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. Brain, 2018, 141, 3160-3178.	7.6	96
198	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
199	Malformations of Cortical Development in Newborns: Genetic Aspects. , 2018, , 2113-2124.		0
200	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
201	Ketamine for Refractory Status Epilepticus: A Systematic Review. CNS Drugs, 2018, 32, 997-1009.	5.9	49
202	The phenotype of SCN8A developmental and epileptic encephalopathy. Neurology, 2018, 91, e1112-e1124.	1.1	114
203	De novo variants in neurodevelopmental disorders with epilepsy. Nature Genetics, 2018, 50, 1048-1053.	21.4	230
204	Unstable non-coding pentanucleotide repeats destabilize cortical excitability. Brain, 2018, 141, 2232-2235.	7.6	4
205	Neuroimaging in mitochondrial disorders. Essays in Biochemistry, 2018, 62, 409-421.	4.7	32
206	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
207	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
208	De novo and inherited private variants in MAP1B in periventricular nodular heterotopia. PLoS Genetics, 2018, 14, e1007281.	3.5	40
209	Malformations of Cortical Development in Newborns: Genetic Aspects. , 2018, , 1-13.		0
210	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
211	Multimodal fiberâ€probe spectroscopy allows detecting epileptogenic focal cortical dysplasia in children. Journal of Biophotonics, 2017, 10, 896-904.	2.3	11
212	Fast Progression of Cerebellar Atrophy in PLA2G6-Associated Infantile Neuronal Axonal Dystrophy. Cerebellum, 2017, 16, 742-745.	2.5	6
213	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
214	Clinical and genetic factors predicting Dravet syndrome in infants with SCN1A mutations. Neurology, 2017, 88, 1037-1044.	1.1	93
215	Fiber-probe optical spectroscopy discriminates normal brain from focal cortical dysplasia in pediatric subjects. , 2017, , .		0
216	Leigh-like neuroimaging features associated with new biallelic mutations in OPA1. European Journal of Paediatric Neurology, 2017, 21, 671-677.	1.6	25

#	Article	IF	Citations
217	Lissencephaly: Expanded imaging and clinical classification. American Journal of Medical Genetics, Part A, 2017, 173, 1473-1488.	1.2	104
218	Age-related differences in audiovisual interactions of semantically different stimuli.. Developmental Psychology, 2017, 53, 138-148.	1.6	3
219	Overview of Human Brain Malformations. , 2017, , 179-182.		0
220	Malformations of Cortical Development. , 2017, , 218-225.		2
221	Cognitive outcome after epilepsy surgery in children: A controlled longitudinal study. Epilepsy and Behavior, 2017, 73, 23-30.	1.7	24
222	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
223	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
224	Vagus nerve stimulation: Surgical technique of implantation and revision and related morbidity. Epilepsia, 2017, 58, 85-90.	5.1	145
225	GNAO1 encephalopathy. Neurology: Genetics, 2017, 3, e143.	1.9	84
226	Mutations in GABRB3. Neurology, 2017, 88, 483-492.	1.1	87
227	Histopathological Findings in Brain Tissue Obtained during Epilepsy Surgery. New England Journal of Medicine, 2017, 377, 1648-1656.	27.0	621
228	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
229	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
230	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain, 2017, 140, 2610-2622.	7.6	102
231	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
232	Delineating SPTAN1 associated phenotypes: from isolated epilepsy to encephalopathy with progressive brain atrophy. Brain, 2017, 140, 2322-2336.	7.6	82
233	Multimodal fiber-probe spectroscopy as a clinical tool for diagnosing and classifying biological tissues. , 2017, , .		0
234	Epilepsy surgery of â€œlow grade epilepsy associated neuroepithelial tumorsâ€: A retrospective nationwide Italian study. Epilepsia, 2017, 58, 1832-1841.	5.1	41

#	Article	IF	Citations
235	Clinical features and outcome of 6 new patients carrying de novo KCNB1 gene mutations. Neurology: Genetics, 2017, 3, e206.	1.9	53
236	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
237	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
238	Unilobar surgery for symptomatic epileptic spasms. Annals of Clinical and Translational Neurology, 2017, 4, 36-45.	3.7	25
239	Human Mutations Associated With Brain Malformations Resulting in Hyperexcitability in Rodents. , 2017, , 827-844.		1
240	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
241	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
242	Defective mitochondrial rRNA methyltransferase MRM2 causes MELAS-like clinical syndrome. Human Molecular Genetics, 2017, 26, 4257-4266.	2.9	63
243	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
244	Hemicerebellitis can drive handedness shift. Cerebellum and Ataxias, 2017, 4, 14.	1.9	4
245	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
246	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
247	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
248	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
249	7T <scp>MRI</scp> in focal epilepsy with unrevealing conventional field strength imaging. Epilepsia, 2016, 57, 445-454.	5.1	128
250	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
251	Effectiveness of antiepileptic therapy in patients with PCDH19 mutations. Seizure: the Journal of the British Epilepsy Association, 2016, 35, 106-110.	2.0	61
252	Definition and diagnostic criteria of sleep-related hypermotor epilepsy. Neurology, 2016, 86, 1834-1842.	1.1	245

#	Article	IF	Citations
253	Association ofMTORMutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. JAMA Neurology, 2016, 73, 836.	9.0	234
254	Idiopathic focal epilepsies: the â€œlost tribeâ€• Epileptic Disorders, 2016, 18, 252-288.	1.3	65
255	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
256	Pitfalls in genetic testing: the story of missed SCN1A mutations. Molecular Genetics & Genomic Medicine, 2016, 4, 457-464.	1.2	67
257	Phenotypic spectrum of GABRA1. Neurology, 2016, 87, 1140-1151.	1.1	113
258	Towards automated neuron tracing via global and local 3D image analysis. , 2016, , .		1
259	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
260	Genetic Basis of Brain Malformations. Molecular Syndromology, 2016, 7, 220-233.	0.8	156
261	Dravet syndrome. Neurology, 2016, 87, 245-246.	1.1	3
262	Symptomatic and presumed symptomatic focal epilepsies in childhood: An observational, prospective multicentre study. Epilepsia, 2016, 57, 1808-1816.	5.1	9
263	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
264	Germline and somatic mutations in the MTOR gene in focal cortical dysplasia and epilepsy. Neurology: Genetics, 2016, 2, e118.	1.9	125
265	Epilepsy in ring chromosome 20 syndrome. Epilepsy Research, 2016, 128, 83-93.	1.6	30
266	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
267	Pitfalls in the detection of gross gene rearrangements using MLPA in Fabry disease. Clinica Chimica Acta, 2016, 452, 82-86.	1.1	9
268	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
269	Testing HLA-B*15:02. Neurology, 2016, 86, 1080-1081.	1.1	4
270	The syndrome of polymicrogyria, thalamic hypoplasia, and epilepsy with CSWS. Neurology, 2016, 86, 1250-1259.	1.1	19

#	Article	IF	Citations
271	Probing focal cortical dysplasia in formalin fixed samples using tissue optical spectroscopy. , 2016, , .		0
272	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, .	5.0	134
273	Malformations of Cortical Development in Newborns: Genetic Aspects. , 2016, , 1-12.		0
274	Computer-based automatic identification of neurons in gigavoxel-sized 3D human brain images. , 2015, 2015, 7724-7.		2
275	PRIMA1 mutation: a new cause of nocturnal frontal lobe epilepsy. Annals of Clinical and Translational Neurology, 2015, 2, 821-830.	3.7	21
276	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
277	Diagnostic methods and treatment options for focal cortical dysplasia. Epilepsia, 2015, 56, 1669-1686.	5.1	167
278	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
279	A versatile clearing agent for multi-modal brain imaging. Scientific Reports, 2015, 5, 9808.	3.3	228
280	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
281	The phenotypic spectrum of SCN8A encephalopathy. Neurology, 2015, 84, 480-489.	1.1	246
282	Late-Onset Epileptic Spasms. Journal of Child Neurology, 2015, 30, 153-159.	1.4	15
283	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. Human Molecular Genetics, 2015, 24, 5250-5259.	2.9	93
284	Early and effective treatment of <scp>KCNQ</scp>2 encephalopathy. Epilepsia, 2015, 56, 685-691.	5.1	229
285	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
286	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
287	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
288	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

#	Article	IF	Citations
289	Malformations of Cortical Development and Epilepsy. Cold Spring Harbor Perspectives in Medicine, 2015, 5, a022392-a022392.	6.2	104
290	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
291	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
292	Ketamine in refractory convulsive status epilepticus in children avoids endotracheal intubation. Epilepsy and Behavior, 2015, 49, 343-346.	1.7	54
293	Antiepileptic Drug Treatment in Children with Epilepsy. CNS Drugs, 2015, 29, 847-863.	5.9	63
294	Nocturnal frontal lobe epilepsy with paroxysmal arousals due to CHRNA2 loss of function. Neurology, 2015, 84, 1520-1528.	1.1	32
295	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
296	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
297	Mitochondrial respiratory chain defects in skin fibroblasts from patients with Dravet syndrome. Neurological Sciences, 2015, 36, 2151-2155.	1.9	7
298	Dried blood spot assay for the quantification of phenytoin using Liquid Chromatography-Mass Spectrometry. Clinica Chimica Acta, 2015, 440, 31-35.	1.1	22
299	Dysgraphia as a Mild Expression of Dystonia in Children with Absence Epilepsy. PLoS ONE, 2015, 10, e0130883.	2.5	9
300	Expression of glutamine synthetase in balloon cells: a basis of their antiepileptic role?. , 2015, 34, 83-88.		6
301	Role of the phosphoinositide phosphatase FIG4 gene in familial epilepsy with polymicrogyria. Neurology, 2014, 82, 1068-1075.	1.1	97
302	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
303	The Î±_{2B}â€adrenergic receptor is mutant in cortical myoclonus and epilepsy. Annals of Neurology, 2014, 75, 77-87.	5.3	42
304	Optimizing the molecular diagnosis of <scp>CDKL</scp>5 geneâ€“related epileptic encephalopathy in boys. Epilepsia, 2014, 55, 1748-1753.	5.1	23
305	Coâ€occurring malformations of cortical development and <scp>SCN</scp>1A gene mutations. Epilepsia, 2014, 55, 1009-1019.	5.1	84
306	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

#	Article	IF	Citations
307	Language Regression Associated With Autistic Regression and Electroencephalographic (EEG) Abnormalities. Journal of Child Neurology, 2014, 29, 855-859.	1.4	3
308	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
309	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
310	Genetic Epilepsy Syndromes Without Structural Brain Abnormalities: Clinical Features and Experimental Models. Neurotherapeutics, 2014, 11, 269-285.	4.4	51
311	De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	6.2	388
312	Epilepsy phenotypes and genotype determinants. Neurology, 2014, 83, 1038-1039.	1.1	0
313	Tissue Border Enhancement by inversion recovery MRI at 7.0 Tesla. Neuroradiology, 2014, 56, 517-523.	2.2	14
314	Autism-epilepsy phenotype with macrocephaly suggests PTEN, but not GLIALCAM, genetic screening. BMC Medical Genetics, 2014, 15, 26.	2.1	55
315	Somatic Mutations in Cerebral Cortical Malformations. New England Journal of Medicine, 2014, 371, 733-743.	27.0	326
316	Malformations of cortical development: clinical features and genetic causes. Lancet Neurology, The, 2014, 13, 710-726.	10.2	382
317	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
318	Periventricular nodular heterotopia in Smithâ€Magenis syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 3142-3147.	1.2	14
319	New clinical and molecular insights on Barth syndrome. Orphanet Journal of Rare Diseases, 2013, 8, 27.	2.7	35
320	Mutations in TUBG1, DYNC1H1, KIF5C and KIF2A cause malformations of cortical development and microcephaly. Nature Genetics, 2013, 45, 639-647.	21.4	399
321	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
322	Do mutations in SCN1B cause Dravet syndrome?. Epilepsy Research, 2013, 103, 97-100.	1.6	11
323	Epilepsy surgery in Neurofibromatosis Type 1. Epilepsy Research, 2013, 105, 384-395.	1.6	44
324	Genetic testing in benign familial epilepsies of the first year of life: Clinical and diagnostic significance. Epilepsia, 2013, 54, 425-436.	5.1	110

#	Article	IF	Citations
325	A randomized phase <scp>III</scp> trial of adjunctive zonisamide in pediatric patients with partial epilepsy. Epilepsia, 2013, 54, 1473-1480.	5.1	48
326	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
327	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
328	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
329	The medical and surgical treatment of tumoral seizures: Current and future perspectives. Epilepsia, 2013, 54, 84-90.	5.1	30
330	Overview of presurgical assessment and surgical treatment of epilepsy from the Italian League Against Epilepsy. Epilepsia, 2013, 54, 35-48.	5.1	45
331	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. Brain, 2013, 136, 3378-3394.	7.6	85
332	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
333	Novel brain expression of ClC-1 chloride channels and enrichment of CLCN1 variants in epilepsy. Neurology, 2013, 80, 1078-1085.	1.1	43
334	The epileptic encephalopathies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 111, 619-626.	1.8	21
335	Extending the KCNQ2 encephalopathy spectrum. Neurology, 2013, 81, 1697-1703.	1.1	198
336	Myoclonus and epilepsy. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 111, 667-679.	1.8	11
337	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
338	Somatic Overgrowth Predisposes to Seizures in Autism Spectrum Disorders. PLoS ONE, 2013, 8, e75015.	2.5	18
339	PRRT2 mutations in familial infantile seizures, paroxysmal dyskinesia, and hemiplegic migraine. Neurology, 2012, 79, 2109-2114.	1.1	106
340	Efficacy and safety of ketamine in refractory status epilepticus in children. Neurology, 2012, 79, 2355-2358.	1.1	87
341	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
342	Symmetric polymicrogyria and pachygyria associated with TUBB2B gene mutations. European Journal of Human Genetics, 2012, 20, 995-998.	2.8	61

#	Article	IF	Citations
343	Paroxysmal disorders associated with PRRT2 mutations shake up expectations on ion channel genes. Neurology, 2012, 79, 2086-2088.	1.1	18
344	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
345	Benign childhood focal epilepsies. Epilepsia, 2012, 53, 9-18.	5.1	83
346	Age-related epileptic encephalopathies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2012, 107, 179-193.	1.8	12
347	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
348	Epilepsy in Rett syndrome, and CDKL5â€•and FOXG1â€geneâ€“related encephalopathies. Epilepsia, 2012, 53, 2067-2078.	5.1	124
349	Peritrigonal and temporo-occipital heterotopia with corpus callosum and cerebellar dysgenesis. Neurology, 2012, 79, 1244-1251.	1.1	31
350	PRRT2 phenotypic spectrum includes sporadic and fever-related infantile seizures. Neurology, 2012, 79, 2104-2108.	1.1	75
351	Focal seizures with affective symptoms are a major feature of PCDH19 geneâ€“related epilepsy. Epilepsia, 2012, 53, 2111-2119.	5.1	63
352	Safety and Tolerability of Antiepileptic Drug Treatment in Children with Epilepsy. Drug Safety, 2012, 35, 519-533.	3.2	41
353	Dravet syndrome: The main issues. European Journal of Paediatric Neurology, 2012, 16, S1-S4.	1.6	40
354	A developmental and genetic classification for malformations of cortical development: update 2012. Brain, 2012, 135, 1348-1369.	7.6	849
355	Safety and efficacy of topiramate in neonates with hypoxic ischemic encephalopathy treated with hypothermia (NeoNATI). BMC Pediatrics, 2012, 12, 144.	1.7	28
356	Periventricular heterotopia with white matter abnormalities associated with 6p25 deletion. American Journal of Medical Genetics, Part A, 2012, 158A, 1793-1797.	1.2	29
357	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
358	Early clinical features in Dravet syndrome patients with and without SCN1A mutations. Epilepsy Research, 2012, 99, 21-27.	1.6	24
359	Impaired object identification in idiopathic childhood occipital epilepsy. Epilepsia, 2012, 53, 686-694.	5.1	7
360	Malformations of Cortical Development. , 2012, , 202-231.		7

#	Article	IF	Citations
361	Malformations of Cortical Development: Genetic Aspects. , 2012, , 1131-1136.		0
362	Hypothalamic hamartoma and gelastic epilepsy. , 2011, , 449-453.		3
363	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
364	Tuberous sclerosis complex. , 2011, , 177-182.		1
365	Urea cycle disorders. , 2011, , 246-248.		2
366	Inverted duplicated chromosome 15 (isodicentric chromosome 15). , 2011, , 281-284.		1
367	Polymicrogyria and schizencephaly. , 2011, , 311-321.		1
368	Uncommon causes of status epilepticus. , 2011, , 745-751.		1
369	Causes of non-convulsive status epilepticus in adults. , 2011, , 752-758.		2
370	Causes of epilepsia partialis continua. , 2011, , 759-766.		1
371	Agyriaâ€“pachygyria band spectrum. , 2011, , 298-304.		1
372	The etiological classification of epilepsy. , 2011, , 21-23.		3
373	Multiple sclerosis and other acquired demyelinating diseases. , 2011, , 607-611.		1
374	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
375	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
376	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
377	Cognitive development in Dravet syndrome: A retrospective, multicenter study of 26 patients. Epilepsia, 2011, 52, 386-392.	5.1	99
378	Severe myoclonic epilepsy in infancy (Dravet syndrome) 30 years later. Epilepsia, 2011, 52, 1-2.	5.1	47

#	Article	IF	Citations
379	Borderline Dravet syndrome: A useful diagnostic category?. Epilepsia, 2011, 52, 10-12.	5.1	28
380	The genetics of Dravet syndrome. Epilepsia, 2011, 52, 24-29.	5.1	287
381	Neuroimaging and neuropathology of Dravet syndrome. Epilepsia, 2011, 52, 30-34.	5.1	49
382	Spectrum of phenotypes in female patients with epilepsy due to protocadherin 19 mutations. Epilepsia, 2011, 52, 1251-1257.	5.1	74
383	Intrinsic epileptogenicity of gangliogliomas may be independent from co-occurring focal cortical dysplasia. Epilepsy Research, 2011, 97, 208-213.	1.6	31
384	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
385	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
386	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
387	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
388	Characterization of severe action myoclonus in sialidoses. Epilepsy Research, 2011, 94, 86-93.	1.6	24
389	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
390	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
391	Glucose Transporter 1 Deficiency as a Treatable Cause of Myoclonic Astatic Epilepsy. Archives of Neurology, 2011, 68, 1152.	4.5	121
392	Bilateral Perysilvian Polymicrogyria With Cerebellar Dysplasia and Ectopic Neurohypophysis. Journal of Child Neurology, 2011, 26, 361-365.	1.4	3
393	Malformations of Cortical Development and Aberrant Cortical Networks: Epileptogenesis and Functional Organization. Journal of Clinical Neurophysiology, 2010, 27, 372-379.	1.7	50
394	Oral Topiramate in Neonates with Hypoxic Ischemic Encephalopathy Treated with Hypothermia: A Safety Study. Journal of Pediatrics, 2010, 157, 361-366.	1.8	64
395	Neuronal migration disorders. Neurobiology of Disease, 2010, 38, 154-166.	4.4	271
396	Sydenham's chorea in a girl with juvenile idiopathic arthritis treated with antiâ€TNFÎ± therapy. Movement Disorders, 2010, 25, 511-514.	3.9	5

#	Article	IF	Citations
397	Xp22.3 genomic deletions involving the CDKL5 gene in girls with early onset epileptic encephalopathy. Epilepsia, 2010, 51, 647-654.	5.1	60
398	Polymicrogyria and epilepsy. Epilepsia, 2010, 51, 10-12.	5.1	8
399	Characteristics of a large population of patients with refractory epilepsy attending tertiary referral centers in Italy. Epilepsia, 2010, 51, 921-925.	5.1	35
400	Classification concepts and terminology: Is clinical description assertive and laboratory testing objective?. Epilepsia, 2010, 51, 718-720.	5.1	17
401	Familial Lennoxâ€Gastaut syndrome in male siblings with a novel DCX mutation and anterior pachygyria. Epilepsia, 2010, 51, 1902-1905.	5.1	9
402	Variable epilepsy phenotypes associated with a familial intragenic deletion of the SCN1A gene. Epilepsia, 2010, 51, 2474-2477.	5.1	56
403	Clinical and imaging heterogeneity of polymicrogyria: a study of 328 patients. Brain, 2010, 133, 1415-1427.	7.6	215
404	Nodular heterotopia is built upon layers. Neurology, 2009, 73, 742-743.	1.1	10
405	Disruption of neural progenitors along the ventricular and subventricular zones in periventricular heterotopia. Human Molecular Genetics, 2009, 18, 497-516.	2.9	169
406	Lennox-Gastaut syndrome: a consensus approach on diagnosis, assessment, management, and trial methodology. Lancet Neurology, The, 2009, 8, 82-93.	10.2	412
407	Earlyâ€onset absence epilepsy caused by mutations in the glucose transporter GLUT1. Annals of Neurology, 2009, 66, 415-419.	5.3	266
408	Bilateral frontoparietal polymicrogyria, Lennoxâ€Gastaut syndrome, and GPR56 gene mutations. Epilepsia, 2009, 50, 1344-1353.	5.1	46
409	SCN1A duplications and deletions detected in Dravet syndrome: Implications for molecular diagnosis. Epilepsia, 2009, 50, 1670-1678.	5.1	152
410	Topiramate concentrations in neonates treated with prolonged whole body hypothermia for hypoxic ischemic encephalopathy. Epilepsia, 2009, 50, 2355-2361.	5.1	75
411	Physiology of epilepsia partialis continua and subcortical mechanisms of status epilepticus. Epilepsia, 2009, 50, 7-9.	5.1	56
412	No major role for the EMX2 gene in schizencephaly. American Journal of Medical Genetics, Part A, 2008, 146A, 1142-1150.	1.2	51
413	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
414	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

#	Article	IF	Citations
415	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
416	Subcortical structures and infantile spasms. Developmental Medicine and Child Neurology, 2008, 50, 87-87.	2.1	0
417	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
418	Severe myoclonic epilepsy in infancy: A systematic review and a metaâ€analysis of individual patient data. Epilepsia, 2008, 49, 343-348.	5.1	119
419	Abnormal development of the human cerebral cortex: genetics, functional consequences and treatment options. Trends in Neurosciences, 2008, 31, 154-162.	8.6	227
420	Malformations of cortical development and epilepsy. Dialogues in Clinical Neuroscience, 2008, 10, 47-62.	3.7	176
421	Brain MRI Findings in Severe Myoclonic Epilepsy in Infancy and Genotype?Phenotype Correlations. Epilepsia, 2007, 48, 1092-1096.	5.1	89
422	Idiopathic Epilepsies with Seizures Precipitated by Fever and SCN1A Abnormalities. Epilepsia, 2007, 48, 1678-1685.	5.1	154
423	Mutational Analysis of EFHC1 Gene in Italian Families with Juvenile Myoclonic Epilepsy. Epilepsia, 2007, 48, 1686-1690.	5.1	44
424	The role of the nicotinic acetylcholine receptors in sleep-related epilepsy. Biochemical Pharmacology, 2007, 74, 1308-1314.	4.4	99
425	Valproate as a Mainstay of Therapy for Pediatric Epilepsy. Paediatric Drugs, 2006, 8, 113-129.	3.1	55
426	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
427	Epilepsy in children. Lancet, The, 2006, 367, 499-524.	13.7	464
428	Practitioner Review: Use of antiepileptic drugs in children. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2006, 47, 115-126.	5.2	8
429	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
430	Autosomal Dominant Early-onset Cortical Myoclonus, Photic-induced Myoclonus, and Epilepsy in a Large Pedigree. Epilepsia, 2006, 47, 1643-1649.	5.1	26
431	Mosaic SCN1A Mutation in Familial Severe Myoclonic Epilepsy of Infancy. Epilepsia, 2006, 47, 1737-1740.	5.1	76
432	Neuropsychological Findings in Idiopathic Occipital Lobe Epilepsies. Epilepsia, 2006, 47, 76-78.	5.1	28

#	Article	IF	Citations
433	Genetic malformations of cortical development. Experimental Brain Research, 2006, 173, 322-333.	1.5	138
434	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
435	Topiramate and its clinical applications in epilepsy. Expert Opinion on Pharmacotherapy, 2006, 7, 811-823.	1.8	78
436	Genetic Malformations of the Cerebral Cortex and Epilepsy. Epilepsia, 2005, 46, 32-37.	5.1	82
437	Genotype-phenotype analysis of human frontoparietal polymicrogyria syndromes. Annals of Neurology, 2005, 58, 680-687.	5.3	124
438	Reciprocal translocations: a trap for cytogenetists?. Human Genetics, 2005, 117, 571-582.	3.8	54
439	Topical Review: Neuronal Migration Disorders, Genetics, and Epileptogenesis. Journal of Child Neurology, 2005, 20, 287-299.	1.4	131
440	Periventricular nodular heterotopia with overlying polymicrogyria. Brain, 2005, 128, 2811-2821.	7.6	73
441	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
442	Pallisterâ€"Killian syndrome: an unusual cause of epileptic spasms. Developmental Medicine and Child Neurology, 2005, 47, 776-779.	2.1	0
443	Pathophysiology of myoclonic epilepsies. Advances in Neurology, 2005, 95, 23-46.	0.8	12
444	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
445	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
446	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
447	Epileptic Syndromes and Visually Induced Seizures. Epilepsia, 2004, 45, 14-18.	5.1	91
448	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
449	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
450	Benign familial neonatal-infantile seizures: Characterization of a new sodium channelopathy. Annals of Neurology, 2004, 55, 550-557.	5.3	250

#	Article	IF	Citations
451	Mutations ofARX are associated with striking pleiotropy and consistent genotype-phenotype correlation. Human Mutation, 2004, 23, 147-159.	2.5	293
452	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
453	Nonsyndromic mental retardation and cryptogenic epilepsy in women withDoublecortin gene mutations. Annals of Neurology, 2003, 54, 30-37.	5.3	65
454	The genetic and molecular basis of epilepsy. Trends in Molecular Medicine, 2003, 9, 300-306.	6.7	21
455	Angelman Syndrome. Paediatric Drugs, 2003, 5, 647-661.	3.1	72
456	Epileptic Encephalopathies with Myoclonic Seizures in Infants and Children (Severe Myoclonic) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 542	1.7	88
457	Epilepsy and malformations of the cerebral cortex. Epileptic Disorders, 2003, 5 Suppl 2, S9-26.	1.3	31
458	Subcortical band heterotopia (SBH) in males: clinical, imaging and genetic findings in comparison with females. Brain, 2002, 125, 2507-2522.	7.6	105
459	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
460	Earlyâ€onset Absence Epilepsy and Paroxysmal Dyskinesia. Epilepsia, 2002, 43, 1224-1229.	5.1	71
461	Different Neurophysiologic Patterns of Myoclonus Characterize Lennox-Gastaut Syndrome and Myoclonic Astaticâ€ƒEpilepsy. Epilepsia, 2002, 43, 609-615.	5.1	33
462	Epilepsy and paroxysmal dyskinesia: co-occurrence and differential diagnosis. Advances in Neurology, 2002, 89, 433-41.	0.8	4
463	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
464	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
465	Idiopathic Epilepsy and Paroxysmal Dyskinesia. Epilepsia, 2001, 42, 36-41.	5.1	136
466	Visual Sensitivity and Epilepsy: A Proposed Terminology and Classification for Clinical and EEG Phenomenology. Epilepsia, 2001, 42, 692-701.	5.1	135
467	A nonsense mutation of theATRX gene causing mild mental retardation and epilepsy. Annals of Neurology, 2000, 47, 117-121.	5.3	72
468	Genetic and neuroradiological heterogeneity of double cortex syndrome. Annals of Neurology, 2000, 47, 265-269.	5.3	94

#	Article	IF	Citations
469	Lack of cortical contrast gain control in human photosensitive epilepsy. Nature Neuroscience, 2000, 3, 259-263.	14.8	138
470	Genetic and neuroradiological heterogeneity of double cortex syndrome. Annals of Neurology, 2000, 47, 265-269.	5.3	4
471	Familial Epilepsy with Unilateral and Bilateral Malformations of Cortical Development. Epilepsia, 1999, 40, 47-51.	5.1	34
472	Continuous Spike-and-Wave Activity During Slow-Wave Sleep: Syndrome or EEG Pattern?. Epilepsia, 1999, 40, 1593-1601.	5.1	92
473	Characterization of mutations in the genedoublecortin in patients with double cortex syndrome. Annals of Neurology, 1999, 45, 146-153.	5.3	175
474	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
475	Segmental facial myoclonus in Moebius syndrome. Movement Disorders, 1999, 14, 1021-1024.	3.9	14
476	Myoclonic status epilepticus following high-dosage lamotrigine therapy. Brain and Development, 1999, 21, 420-424.	1.1	89
477	Reversible Pseudoatrophy of the Brain and Mental Deterioration Associated with Valproate Treatment. Epilepsia, 1998, 39, 27-32.	5.1	219
478	Lamotrigine and Seizure Aggravation in Severe Myoclonic Epilepsy. Epilepsia, 1998, 39, 508-512.	5.1	505
479	Myoclonic Absence-Like Seizures and Chromosome Abnormality Syndromes. Epilepsia, 1998, 39, 660-663.	5.1	52
480	Antiepileptic Drugâ€Induced Worsening of Seizures in Children. Epilepsia, 1998, 39, S2-10.	5.1	183
481	Cortical reflex myoclonus in rett syndrome. Annals of Neurology, 1998, 43, 472-479.	5.3	48
482	Angelman syndrome: Correlations between epilepsy phenotypes and genotypes. Annals of Neurology, 1998, 43, 485-493.	5.3	173
483	Bilateral periventricular nodular heterotopia with mental retardation and frontonasal malformation. Neurology, 1998, 51, 499-503.	1.1	70
484	Identification of a Duplication of Xq28 Associated with Bilateral Periventricular Nodular Heterotopia. American Journal of Human Genetics, 1997, 61, 379-387.	6.2	74
485	Delayed appearance of interictal EEG abnormalities in early onset childhood epilepsy with occipital paroxysms. Brain and Development, 1997, 19, 343-346.	1.1	92
486	Adolescent Onset of Idiopathic Photosensitive Occipital Epilepsy After Remission of Benign Rolandic Epilepsy. Epilepsia, 1997, 38, 777-781.	5.1	39

#	Article	IF	Citations
487	Cortical myoclonus in angelman syndrome. Annals of Neurology, 1996, 40, 39-48.	5.3	136
488	Idiopathic Photosensitive Occipital Lobe Epilepsy. Epilepsia, 1995, 36, 883-891.	5.1	191
489	Panic Attacks Mistaken for Relapse of Epilepsy. Epilepsia, 1995, 36, 48-51.	5.1	28
490	Epileptogenesis in idiopathic epilepsy. , 0, , 24-34.		0
491	Mechanisms of epileptogenesis in symptomatic epilepsy. , 0, , 35-42.		0
492	The genetic contribution to epilepsy: the known and missing heritability. , 0, , 62-66.		2
493	Benign familial neonatal seizures. , 0, , 67-69.		0
494	Autosomal dominant nocturnal frontal lobe epilepsy. , 0, , 70-73.		1
495	Severe myoclonic epilepsy of infancy or Dravet syndrome. , 0, , 78-84.		0
496	Benign partial epilepsies of childhood. , 0, , 104-112.		0
497	West syndrome and Lennoxâ€“Gastaut syndrome. , 0, , 119-134.		1
498	Unverrichtâ€“Lundborg disease. , 0, , 135-138.		0
499	Dentato-rubro-pallido-luysian atrophy. , 0, , 139-142.		1
500	Mitochondrial cytopathies. , 0, , 147-157.		1
501	Neuronal ceroid lipofuscinoses. , 0, , 158-163.		0
502	Sialidosis and Gaucher disease. , 0, , 164-168.		0
503	Progressive myoclonus epilepsies: other rare causes. , 0, , 172-176.		0
504	Neurofibromatoses. , 0, , 183-188.		4

#	Article	IF	Citations
505	Sturgeâ€“Weber syndrome. , 0, , 189-195.		0
506	Other neurocutaneous syndromes. , 0, , 196-200.		0
507	Angelman syndrome. , 0, , 201-205.		0
508	Lysosomal disorders and Menkes syndrome. , 0, , 206-211.		0
509	Neuroacanthocytosis. , 0, , 212-215.		0
510	Organic acid, amino acids, and peroxisomal disorders. , 0, , 216-230.		0
511	Porphyria. , 0, , 231-236.		0
512	Pyridoxine-dependent epilepsy. , 0, , 237-241.		2
513	Rett syndrome and MECP2 and CDKL5 genotypes. , 0, , 242-245.		0
514	Disorders of cobalamin and folate metabolism. , 0, , 252-257.		0
515	Other single-gene disorders. , 0, , 258-264.		0
516	4p (Wolfâ€“Hirschhorn) syndrome. , 0, , 277-280.		0
517	Ring chromosome 20. , 0, , 285-288.		0
518	Systemic lupus erythematosus and other collagen vascular diseases. , 0, , 579-584.		1
519	Inflammatory and immunological diseases of the nervous system. , 0, , 585-592.		0
520	Psychiatric disorders. , 0, , 593-606.		2
521	Hydrocephalus and porencephaly. , 0, , 612-617.		0
522	Alzheimer disease and other neurodegenerative diseases. , 0, , 618-624.		0

#	Article	IF	Citations
523	Genetic epilepsy with febrile seizures plus. , 0, , 74-77.		0
524	Benign adult familial myoclonic epilepsy. , 0, , 85-90.		1
525	Action myoclonusâ€“renal failure syndrome. , 0, , 169-171.		1
526	Lafora body disease. , 0, , 143-146.		0
527	The menstrual cycle and catamenial epilepsy. , 0, , 635-642.		0
528	Metabolic and endocrine-induced seizures. , 0, , 650-654.		0
529	Electrolyte and sugar disturbances. , 0, , 655-663.		0
530	Drug-induced seizures. , 0, , 664-673.		1
531	Alcohol- and toxin-induced seizures. , 0, , 674-682.		0
532	How reflex mechanisms cause epilepsy. , 0, , 683-686.		0
533	Visual stimuli, photosensitivity, and photosensitive epilepsy. , 0, , 687-694.		1
534	Startle-induced (and other sensory-induced) epilepsy. , 0, , 695-699.		0
535	Primary reading epilepsy. , 0, , 700-703.		0
536	Auditory-induced epilepsy. , 0, , 704-708.		0
537	Focal reflex seizures â€“ with emphasis on seizures triggered by eating. , 0, , 709-712.		0
538	Hot-water epilepsy. , 0, , 713-719.		1
539	Reflex epilepsy with higher-level processing. , 0, , 720-722.		0
540	Causes of status epilepticus in children. , 0, , 730-734.		0

#	Article	IF	Citations
541	The causes of convulsive status epilepticus in adults. , 0, , 735-744.		0
542	Hemimegalencephaly. , 0, , 289-292.		0
543	Focal cortical dysplasia and related variants. , 0, , 293-297.		3
544	Agenesis of the corpus callosum. , 0, , 305-310.		0
545	Periventricular nodular heterotopia. , 0, , 322-329.		0
546	Microcephaly. , 0, , 330-340.		0
547	Arachnoid cysts. , 0, , 341-345.		1
548	Malformations of human cerebral cortex. , 0, , 346-362.		0
549	Hippocampal sclerosis. , 0, , 363-372.		0
550	Neonatal seizures and postneonatal epilepsy â€“ causes. , 0, , 373-381.		0
551	Vaccination and immunization. , 0, , 388-392.		0
552	Open head injury. , 0, , 393-399.		0
553	Closed head injury. , 0, , 400-406.		0
554	De novo epilepsy after neurosurgery. , 0, , 407-412.		0
555	Epilepsy after epilepsy surgery. , 0, , 413-424.		1
556	Non-accidental brain injury. , 0, , 425-432.		2
557	Glioma. , 0, , 433-440.		0
558	Ganglioglioma, dysembryoplastic neuroepithelial tumor, and related tumors. , 0, , 441-448.		0

#	Article	IF	Citations
559	Metastatic disease. , 0, , 459-466.		0
560	Viral encephalitis. , 0, , 467-474.		0
561	Bacterial meningitis and focal suppurative intracranial infections in children. , 0, , 475-481.		0
562	Bacterial meningitis and pyogenic abscess in adults. , 0, , 482-491.		0
563	Other parasitic diseases. , 0, , 501-510.		0
564	HIV infection. , 0, , 520-527.		2
565	Emerging and less common central nervous system viral encephalitides. , 0, , 528-536.		0
566	Cerebral hemorrhage. , 0, , 537-543.		0
567	Cerebral infarction and occult degenerative cerebrovascular disease. , 0, , 544-550.		0
568	Arteriovenous malformations. , 0, , 551-558.		0
569	Cavernous malformations. , 0, , 559-564.		1
570	Other vascular disorders. , 0, , 565-572.		0
571	Rasmussen encephalitis and related conditions. , 0, , 573-578.		0
572	Idiopathic generalized epilepsies. , 0, , 91-103.		0
573	Epileptic spasms and abnormal neuronal migration. , 0, , 28-30.		0