Renzo Guerrini

List of Publications by Year
in descending order

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		5430	10399
573	29,711	85	144
papers	citations	h-index	g-index

618	618	618	25404
all docs	docs citations	times ranked	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.	1.9	47
2	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. Brain, 2022, 145, 208-223.	3.7	15
3	Monoallelic KIF1A-related disorders: a multicenter cross sectional study and systematic literature review. Journal of Neurology, 2022, 269, 437-450.	1.8	12
4	A systemsâ€level analysis highlights microglial activation as a modifying factor in common epilepsies. Neuropathology and Applied Neurobiology, 2022, 48, .	1.8	22
5	Profiling PI3K-AKT-MTOR variants in focal brain malformations reveals new insights for diagnostic care. Brain, 2022, 145, 925-938.	3.7	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.	0.7	13
7	De novo FZR1 loss-of-function variants cause developmental and epileptic encephalopathies. Brain, 2022, 145, 1684-1697.	3.7	5
8	Topographic divergence of atypical cortical asymmetry and atrophy patterns in temporal lobe epilepsy. Brain, 2022, 145, 1285-1298.	3.7	18
9	Atlas of lesion locations and postsurgical seizure freedom in focal cortical dysplasia: A MELD study. Epilepsia, 2022, 63, 61-74.	2.6	36
10	Distinct epilepsy phenotypes and response to drugs in KCNA1 gainâ€and lossâ€of function variants. Epilepsia, 2022, 63, .	2.6	20
11	GM3 synthase deficiency in non-Amish patients. Genetics in Medicine, 2022, 24, 492-498.	1.1	7
12	Development and Validation of a Prediction Model for Early Diagnosis of SCN1A -Related Epilepsies. Neurology, 2022, 98, .	1.5	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.	2.4	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.	2.9	24
15	Spectrum of Phenotypic, Genetic, and Functional Characteristics in Patients With Epilepsy With KCNC2 Pathogenic Variants. Neurology, 2022, 98, .	1.5	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.	1.1	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.	0.3	0
18	SLC7A3: In Silico Prediction of a Potential New Cause of Childhood Epilepsy. Neuropediatrics, 2022, 53, 046-051.	0.3	0

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19	Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis. Brain, 2022, 145, 2687-2703.	3.7	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.	1.8	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.	4.9	63
22	Monoallelic and biallelic mutations inRELNunderlie a graded series of neurodevelopmental disorders. Brain, 2022, 145, 3274-3287.	3.7	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.	4.5	43
25	Orphan Drug Use in Patients With Rare Diseases: A Population-Based Cohort Study. Frontiers in Pharmacology, 2022, 13, .	1.6	4
26	SEMA6B variants cause intellectual disability and alter dendritic spine density and axon guidance. Human Molecular Genetics, 2022, 31, 3325-3340.	1.4	5
27	Eventâ€based modeling in temporal lobe epilepsy demonstrates progressive atrophy from crossâ€sectional data. Epilepsia, 2022, 63, 2081-2095.	2.6	11
28	Natural History Study of STXBP1-Developmental and Epileptic Encephalopathy Into Adulthood. Neurology, 2022, 99, .	1.5	23
29	Spatial centrosome proteome of human neural cells uncovers disease-relevant heterogeneity. Science, 2022, 376, .	6.0	25
30	Networks Underlie Temporal Onset of Dysplasiaâ€Related Epilepsy: A <scp>MELD</scp> Study. Annals of Neurology, 2022, 92, 503-511.	2.8	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.	2.6	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.	0.7	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.	1.3	15
34	The spectrum of brain malformations and disruptions in twins. American Journal of Medical Genetics, Part A, 2021, 185, 2690-2718.	0.7	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.	0.6	7
36	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	1.1	28

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37	Temporal lobe epilepsy surgery in children and adults: A multicenter study. Epilepsia, 2021, 62, 128-142.	2.6	33
38	Basal Ganglia Dysmorphism in Patients With Aicardi Syndrome. Neurology, 2021, 96, e1319-e1333.	1.5	6
39	Deciphering the premature mortality in PIGA-CDG â€“ An untold story. Epilepsy Research, 2021, 170, 106530.	0.8	15
40	Thermal inactivation of SARS COVID-2 virus: Are steam inhalations a potential treatment?. Life Sciences, 2021, 265, 118801.	2.0	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.	0.9	7
42	Is Focal Cortical Dysplasia/Epilepsy Caused by Somatic MTOR Mutations Always a Unilateral Disorder?. Neurology: Genetics, 2021, 7, e540.	0.9	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.	2.3	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.	1.4	25
45	A survey of the European Reference Network EpiCARE on clinical practice for selected rare epilepsies. Epilepsia Open, 2021, 6, 160-170.	1.3	3
46	SLC32A1. Neurology, 2021, 96, 831-832.	1.5	1
47	Phenotypic Spectrum of Seizure Disorders in MBD5-Associated Neurodevelopmental Disorder. Neurology: Genetics, 2021, 7, e579.	0.9	8
48	Morquio B disease: From pathophysiology towards diagnosis. Molecular Genetics and Metabolism, 2021, 132, 180-188.	0.5	7
49	Climate change and epilepsy: Insights from clinical and basic science studies. Epilepsy and Behavior, 2021, 116, 107791.	0.9	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.	0.9	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.	0.6	7
52	Diverse genetic causes of polymicrogyria with epilepsy. Epilepsia, 2021, 62, 973-983.	2.6	12
53	Focal cortical dysplasia: an update on diagnosis and treatment. Expert Review of Neurotherapeutics, 2021, 21, 1213-1224.	1.4	21
54	Consensus statements on the information to deliver after a febrile seizure. European Journal of Pediatrics, 2021, 180, 2993-2999.	1.3	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.	0.9	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.	1.1	23
57	Large-scale, cell-resolution volumetric mapping allows layer-specific investigation of human brain cytoarchitecture. Biomedical Optics Express, 2021, 12, 3684.	1.5	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.	1.1	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.	0.7	6
60	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. Brain, 2021, 144, 1435-1450.	3.7	35
61	Migrating Focal Seizures and Myoclonic Status in ARV1- Related Encephalopathy. Neurology: Genetics, 2021, 7, e593.	0.9	6
62	Bi-allelic variants in MTMR5/SBF1 cause Charcot-Marie-Tooth type 4B3 featuring mitochondrial dysfunction. BMC Medical Genomics, 2021, 14, 157.	0.7	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.	2.6	35
64	Whole Exome Sequencing Is the Minimal Technological Approach in Probands Born to Consanguineous Couples. Genes, 2021, 12, 962.	1.0	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.	1.0	4
66	Pathogenic <scp>MAST3</scp> Variants in the <scp>STK</scp> Domain Are Associated with Epilepsy. Annals of Neurology, 2021, 90, 274-284.	2.8	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.	1.8	6
68	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	1.1	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.	1.0	2
70	Clinical and molecular delineation of <scp>PUS3</scp>â€associated neurodevelopmental disorders. Clinical Genetics, 2021, 100, 628-633.	1.0	23
71	Multicenter Validation of a Deep Learning Detection Algorithm for Focal Cortical Dysplasia. Neurology, 2021, 97, e1571-e1582.	1.5	39
72	Monogenic Epilepsies. Neurology, 2021, 97, 817-831.	1.5	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.	1.5	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.	1.4	4
75	Early-onset bradykinetic rigid syndrome and reflex seizures in a child with PURA syndrome. Epileptic Disorders, 2021, 23, 745-748.	0.7	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.	5.8	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.	2.0	4
79	Meeting report: EpiXchange II brings together European epilepsy research projects to discuss latest advances. Epilepsy Research, 2021, 178, 106811.	0.8	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.	0.5	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.	1.1	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.	0.6	14
83	Lesional and non-lesional epilepsies: A blurring genetic boundary. European Journal of Paediatric Neurology, 2020, 24, 24-29.	0.7	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.	1.1	15
85	Infantile-Onset Syndromic Cerebellar Ataxia and CACNA1G Mutations. Pediatric Neurology, 2020, 104, 40-45.	1.0	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.	1.1	2
87	Trends in pediatric epilepsy surgery in Europe between 2008 and 2015: Countryâ€, centerâ€, and ageâ€specific variation. Epilepsia, 2020, 61, 216-227.	2.6	44
88	KCNQ2 encephalopathy manifesting with Rett-like features. Neurology: Genetics, 2020, 6, e510.	0.9	3
89	Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918.	4.9	139
90	Network-based atrophy modeling in the common epilepsies: A worldwide ENIGMA study. Science Advances, 2020, 6, .	4.7	97

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91	SARS-CoV-2 infection in a patient with propionic acidemia. Orphanet Journal of Rare Diseases, 2020, 15, 306.	1.2	14
92	Reply:. American Journal of Neuroradiology, 2020, 41, E12-E12.	1.2	0
93	Mutations in the exocyst component EXOC2 cause severe defects in human brain development. Journal of Experimental Medicine, 2020, 217, .	4.2	17
94	Developmental and epilepsy spectrum of KCNB1 encephalopathy with longâ€term outcome. Epilepsia, 2020, 61, 2461-2473.	2.6	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.	2.6	37
96	Breaking up genetic influences on seizure onset, spread, and termination. Neurology, 2020, 95, 667-668.	1.5	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.	0.7	0
98	International consensus recommendations on the diagnostic work-up for malformations of cortical development. Nature Reviews Neurology, 2020, 16, 618-635.	4.9	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.	1.1	3
100	White matter abnormalities across different epilepsy syndromes in adults: an ENIGMA-Epilepsy study. Brain, 2020, 143, 2454-2473.	3.7	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.	4.9	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.	2.3	6
103	Phenotypic and genetic spectrum of epilepsy with myoclonic atonic seizures. Epilepsia, 2020, 61, 995-1007.	2.6	30
104	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	2.6	32
105	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	3.7	47
106	<scp>SCN3A</scp>â€Related Neurodevelopmental Disorder: A Spectrum of Epilepsy and Brain Malformation. Annals of Neurology, 2020, 88, 348-362.	2.8	42
107	Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. Genome Medicine, 2020, 12, 28.	3.6	42
108	Emerging Role of the Autophagy/Lysosomal Degradative Pathway in Neurodevelopmental Disorders With Epilepsy. Frontiers in Cellular Neuroscience, 2020, 14, 39.	1.8	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.	1.6	5
110	Quantitative MRI-Based Analysis Identifies Developmental Limbic Abnormalities in PCDH19 Encephalopathy. Cerebral Cortex, 2020, 30, 6039-6050.	1.6	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.	1.4	27
112	Bi-allelic GAD1 variants cause a neonatal onset syndromic developmental and epileptic encephalopathy. Brain, 2020, 143, 1447-1461.	3.7	18
113	Early infantile epileptic-dyskinetic encephalopathy due to biallelicPIGPmutations. Neurology: Genetics, 2020, 6, e387.	0.9	26
114	Cerebral malformations. , 2020, , 249-267.		1
115	The landscape of epilepsy-related GATOR1 variants. Genetics in Medicine, 2019, 21, 398-408.	1.1	137
116	Genetic heterogeneity in infantile spasms. Epilepsy Research, 2019, 156, 106181.	0.8	38
117	Somatic double-hit in MTOR and RPS6 in hemimegalencephaly with intractable epilepsy. Human Molecular Genetics, 2019, 28, 3755-3765.	1.4	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.	2.6	237
119	Generalized epilepsies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2019, 161, 3-15.	1.0	22
120	What is the role of next generation sequencing in status epilepticus?. Epilepsy and Behavior, 2019, 101, 106373.	0.9	5
121	Automatic detection and sonification of nonmotor generalized onset epileptic seizures: Preliminary results. Brain Research, 2019, 1721, 146341.	1.1	16
122	TBC1D24-TLDc-related epilepsy exercise-induced dystonia: rescue by antioxidants in a disease model. Brain, 2019, 142, 2319-2335.	3.7	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.	2.6	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.	1.1	2
125	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	5.8	99
126	Biallelic DMXL2 mutations impair autophagy and cause Ohtahara syndrome with progressive course. Brain, 2019, 142, 3876-3891.	3.7	23

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127	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. Annals of Neurology, 2019, 86, 821-831.	2.8	96
128	Broadening phenotype of adenylosuccinate lyase deficiency: A novel clinical pattern resembling neuronal ceroid lipofuscinosis. Molecular Genetics and Metabolism Reports, 2019, 21, 100502.	0.4	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.	1.4	21
130	Advancing research toward faster diagnosis, better treatment, and end of stigma in epilepsy. Epilepsia, 2019, 60, 1281-1292.	2.6	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.	2.6	78
132	Genomic <scp>DNA</scp> methylation distinguishes subtypes of human focal cortical dysplasia. Epilepsia, 2019, 60, 1091-1103.	2.6	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.	1.1	39
141	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	2.6	61
142	The spectrum of intermediate <scp>SCN</scp>8Aâ€related epilepsy. Epilepsia, 2019, 60, 830-844.	2.6	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.	2.6	4
157	Clinical spectrum of STX1B -related epileptic disorders. Neurology, 2019, 92, e1238-e1249.	1.5	43
158	Outcome after hemispherotomy in patients with intractable epilepsy: Comparison of techniques in the Italian experience. Epilepsy and Behavior, 2019, 93, 22-28.	0.9	30
159	Defining and expanding the phenotype of QARS-associated developmental epileptic encephalopathy. Neurology: Genetics, 2019, 5, e373.	0.9	5
160	Subcortical heterotopic gray matter brain malformations. Neurology, 2019, 93, e1360-e1373.	1.5	33
161	Drug Development for Rare Paediatric Epilepsies: Current State and Future Directions. Drugs, 2019, 79, 1917-1935.	4.9	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.	1.6	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.	1.2	16
164	Next-generation sequencing approach to hyperCKemia. Neurology: Genetics, 2019, 5, e352.	0.9	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.	6.3	227
166	Dravet syndrome as part of the clinical and genetic spectrum of sodium channel epilepsies and encephalopathies. Epilepsia, 2019, 60, S2-S7.	2.6	34
167	Dravet syndrome andÂotherÂsodiumÂchannelâ€related encephalopathies. Epilepsia, 2019, 60, S1.	2.6	1
168	Dravet syndrome: Treatment options and management of prolonged seizures. Epilepsia, 2019, 60, S39-S48.	2.6	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.	0.6	12
170	SYNGAP1 encephalopathy. Neurology, 2019, 92, e96-e107.	1.5	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.	1.8	9
172	SCN1A mutations in focal epilepsy with auditory features: widening the spectrum of GEFS plus. Epileptic Disorders, 2019, 21, 185-191.	0.7	5
173	Clinical and genetic spectrum of AMPD2-related pontocerebellar hypoplasia type 9. European Journal of Human Genetics, 2018, 26, 695-708.	1.4	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.	0.5	13
175	Analysis of 17 genes detects mutations in 81% of 811 patients with lissencephaly. Genetics in Medicine, 2018, 20, 1354-1364.	1.1	92
176	De novo mutations of the ATP6V1A gene cause developmental encephalopathy with epilepsy. Brain, 2018, 141, 1703-1718.	3.7	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.	2.6	16
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