Federico Zara

List of Publications by Year
in descending order

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328 papers	17,900 citations	17440 63 h-index	19749 117 g-index
338 all docs	338 docs citations	338 times ranked	19888 citing authors

#	Article	IF	Citations
1	Friedreich's Ataxia: Autosomal Recessive Disease Caused by an Intronic GAA Triplet Repeat Expansion. Science, 1996, 271, 1423-1427.	12.6	2,642
2	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	12.6	1,085
3	Mutations in the caveolin-3 gene cause autosomal dominant limb-girdle muscular dystrophy. Nature Genetics, 1998, 18, 365-368.	21.4	555
4	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. Brain, 2017, 140, 1316-1336.	7.6	426
5	De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	6.2	388
6	The genetics of Dravet syndrome. Epilepsia, 2011, 52, 24-29.	5.1	287
7	Genetic determinants of common epilepsies: a meta-analysis of genome-wide association studies. Lancet Neurology, The, 2014, 13, 893-903.	10.2	264
8	Benign familial neonatal-infantile seizures: Characterization of a new sodium channelopathy. Annals of Neurology, 2004, 55, 550-557.	5.3	250
9	Spectrum of SCN1A mutations in severe myoclonic epilepsy of infancy. Neurology, 2003, 60, 1961-1967.	1.1	241
10	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
11	De novo mutations in HCN1 cause early infantile epileptic encephalopathy. Nature Genetics, 2014, 46, 640-645.	21.4	192
12	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
13	Genetic testing in the epilepsiesâ€"Report of the ILAE Genetics Commission. Epilepsia, 2010, 51, 655-670.	5.1	175
14	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. American Journal of Human Genetics, 2015, 96, 808-815.	6.2	173
15	Epilepsy, hippocampal sclerosis and febrile seizures linked by common genetic variation around SCN1A. Brain, 2013, 136, 3140-3150.	7.6	168
16	Mutation in the CAV3 gene causes partial caveolin-3 deficiency and persistent elevated levels of serum creatine kinase. Neurology, 2000, 54, 1373-1376.	1.1	158
17	SCN1A duplications and deletions detected in Dravet syndrome: Implications for molecular diagnosis. Epilepsia, 2009, 50, 1670-1678.	5.1	152
18	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150

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19	Genome search for susceptibility loci of common idiopathic generalised epilepsies. Human Molecular Genetics, 2000, 9, 1465-1472.	2.9	147
20	Clinical and molecular characterisation of 80 patients with 5p deletion: genotype-phenotype correlation. Journal of Medical Genetics, 2001, 38, 151-158.	3.2	147
21	TBC1D24, an ARF6-Interacting Protein, Is Mutated in Familial Infantile Myoclonic Epilepsy. American Journal of Human Genetics, 2010, 87, 365-370.	6.2	134
22	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
23	PRRT2 Is a Key Component of the Ca 2+ -Dependent Neurotransmitter Release Machinery. Cell Reports, 2016, 15, 117-131.	6.4	121
24	Clinical and genetic heterogeneity of branching enzyme deficiency (glycogenosis type IV). Neurology, 2004, 63, 1053-1058.	1.1	120
25	Mutations in <scp>KCNT</scp>1 cause a spectrum of focal epilepsies. Epilepsia, 2015, 56, e114-20.	5.1	117
26	Microbiota-gut brain axis involvement in neuropsychiatric disorders. Expert Review of Neurotherapeutics, 2019, 19, 1037-1050.	2.8	116
27	An open-label trial of levetiracetam in severe myoclonic epilepsy of infancy. Neurology, 2007, 69, 250-254.	1.1	115
28	Effects in Neocortical Neurons of Mutations of the Nav1.2 Na+ Channel causing Benign Familial Neonatal-Infantile Seizures. Journal of Neuroscience, 2006, 26, 10100-10109.	3.6	110
29	Genetic testing in benign familial epilepsies of the first year of life: Clinical and diagnostic significance. Epilepsia, 2013, 54, 425-436.	5.1	110
30	Novel Compound Heterozygous Mutations inTBC1D24Cause Familial Malignant Migrating Partial Seizures of Infancy. Human Mutation, 2013, 34, 869-872.	2.5	110
31	Mapping of genes predisposing to idiopathic generalized epilepsy. Human Molecular Genetics, 1995, 4, 1201-1207.	2.9	109
32	Somatic and germline mosaicisms in Severe Myoclonic Epilepsy of Infancy. Biochemical and Biophysical Research Communications, 2006, 341, 489-493.	2.1	102
33	PRRT2 controls neuronal excitability by negatively modulating Na+ channel 1.2/1.6 activity. Brain, 2018, 141, 1000-1016.	7.6	99
34	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	12.8	99
35	TBC1D24 genotypeâ€“phenotype correlation. Neurology, 2016, 87, 77-85.	1.1	97
36	Recessive mutations inSLC13A5result in a loss of citrate transport and cause neonatal epilepsy, developmental delay and teeth hypoplasia. Brain, 2015, 138, 3238-3250.	7.6	96

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37	HCN1mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. Brain, 2018, 141, 3160-3178.	7.6	96
38	Dravet syndrome: Early clinical manifestations and cognitive outcome in 37 Italian patients. Brain and Development, 2010, 32, 71-77.	1.1	94
39	PRRT2 Mutations are the major cause of benign familial infantile seizures. Human Mutation, 2012, 33, 1439-1443.	2.5	93
40	POMT2 gene mutation in limb-girdle muscular dystrophy with inflammatory changes. Biochemical and Biophysical Research Communications, 2007, 363, 1033-1037.	2.1	91
41	The leukodystrophy protein FAM126A (hyccin) regulates PtdIns(4)P synthesis at the plasmaÂmembrane. Nature Cell Biology, 2016, 18, 132-138.	10.3	91
42	Coexistence of epilepsy and Brugada syndrome in a family with SCN5A mutation. Epilepsy Research, 2013, 105, 415-418.	1.6	90
43	Brain MRI Findings in Severe Myoclonic Epilepsy in Infancy and Genotype?Phenotype Correlations. Epilepsia, 2007, 48, 1092-1096.	5.1	89
44	A pilot trial of levetiracetam in eyelid myoclonia with absences (Jeavons syndrome). Epilepsia, 2008, 49, 425-430.	5.1	88
45	Mutations in the Neuronal Vesicular SNARE VAMP2 Affect Synaptic Membrane Fusion and Impair Human Neurodevelopment. American Journal of Human Genetics, 2019, 104, 721-730.	6.2	88
46	Progressive myoclonic epilepsies. Neurology, 2014, 82, 405-411.	1.1	87
47	GLUT1 mutations are a rare cause of familial idiopathic generalized epilepsy. Neurology, 2012, 78, 557-562.	1.1	86
48	Deficiency of hyccin, a newly identified membrane protein, causes hypomyelination and congenital cataract. Nature Genetics, 2006, 38, 1111-1113.	21.4	82
49	Cryptic chromosome deletions involving SCN1A in severe myoclonic epilepsy of infancy. Neurology, 2006, 67, 1230-1235.	1.1	80
50	TBC1D24 regulates neuronal migration and maturation through modulation of the ARF6-dependent pathway. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 2337-2342.	7.1	80
51	Impairment of ceramide synthesis causes a novel progressive myoclonus epilepsy. Annals of Neurology, 2014, 76, 206-212.	5.3	80
52	Homozygous c.649dupC mutation in PRRT2 worsens the BFIS/PKD phenotype with mental retardation, episodic ataxia, and absences. Epilepsia, 2012, 53, e196-9.	5.1	78
53	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
54	Benign adult familial myoclonic epilepsy. Neurology, 2003, 60, 1381-1385.	1.1	75

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55	A Novel SCN2A Mutation in Family with Benign Familial Infantile Seizures. Epilepsia, 2006, 47, 218-220.	5.1	74
56	Autoantibodies to glutamic acid decarboxylase (GAD) in focal and generalized epilepsy: A study on 233 patients. Journal of Neuroimmunology, 2009, 211, 120-123.	2.3	74
57	The PRRT2 knockout mouse recapitulates the neurological diseases associated with PRRT2 mutations. Neurobiology of Disease, 2017, 99, 66-83.	4.4	72
58	Clinical and Genetic Findings in 26 Italian Patients with Lafora Disease. Epilepsia, 2006, 47, 640-643.	5.1	71
59	Therapeutic Potential of Proteasome Inhibition in Duchenne and Becker Muscular Dystrophies. American Journal of Pathology, 2010, 176, 1863-1877.	3.8	71
60	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
61	The spectrum of intermediate <scp>SCN</scp>8Aâ€related epilepsy. Epilepsia, 2019, 60, 830-844.	5.1	70
62	Late-onset and Slow-progressing Lafora Disease in Four Siblings with EPM2B Mutation. Epilepsia, 2005, 46, 1695-1697.	5.1	69
63	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. Brain, 2022, 145, 2991-3009.	7.6	69
64	Addition of verapamil in the treatment of severe myoclonic epilepsy in infancy. Epilepsy Research, 2009, 85, 89-95.	1.6	68
65	PRRT2: from Paroxysmal Disorders to Regulation of Synaptic Function. Trends in Neurosciences, 2016, 39, 668-679.	8.6	68
66	Generalized Epilepsy with Febrile Seizures Plus (GEFS+): Clinical Spectrum in Seven Italian Families Unrelated to SCN1A, SCN1B, and GABRG2 Gene Mutations. Epilepsia, 2004, 45, 149-158.	5.1	67
67	Pitfalls in genetic testing: the story of missed SCN1A mutations. Molecular Genetics & Genomic Medicine, 2016, 4, 457-464.	1.2	67
68	Rare coding variants in genes encoding GABAA receptors in genetic generalised epilepsies: an exome-based case-control study. Lancet Neurology, The, 2018, 17, 699-708.	10.2	67
69	Benign Familial Infantile Convulsions: Mapping of a Novel Locus on Chromosome 2q24 and Evidence for Genetic Heterogeneity. American Journal of Human Genetics, 2001, 68, 1521-1526.	6.2	66
70	A novel KCNQ3 mutation in familial epilepsy with focal seizures and intellectual disability. Epilepsia, 2015, 56, e15-20.	5.1	66
71	Confirmation of mutations inPROSCas a novel cause of vitamin B_₆-dependent epilepsy. Journal of Medical Genetics, 2017, 54, 809-814.	3.2	66
72	Recent advances in epilepsy genetics. Neuroscience Letters, 2018, 667, 4-9.	2.1	66

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73	PRRT2-related disorders: further PKD and ICCA cases and review of the literature. Journal of Neurology, 2013, 260, 1234-1244.	3.6	63
74	Autosomal dominant cortical tremor, myoclonus and epilepsy: many syndromes, one phenotype. Acta Neurologica Scandinavica, 2005, 111, 211-217.	2.1	61
75	Clinical Significance of Rare Copy Number Variations in Epilepsy. Archives of Neurology, 2012, 69, 322.	4.5	61
76	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	5.1	61
77	Genetics of reflex seizures and epilepsies in humans and animals. Epilepsy Research, 2016, 121, 47-54.	1.6	60
78	¹Hâ€MR spectroscopy indicates prominent cerebellar dysfunction in benign adult familial myoclonic epilepsy. Epilepsia, 2009, 50, 1491-1497.	5.1	58
79	Pharmacological rescue of the dystrophin-glycoprotein complex in Duchenne and Becker skeletal muscle explants by proteasome inhibitor treatment. American Journal of Physiology - Cell Physiology, 2006, 290, C577-C582.	4.6	57
80	Efficacy of sodium channel blockers in SCN2A early infantile epileptic encephalopathy. Brain and Development, 2017, 39, 345-348.	1.1	57
81	Levetiracetam for cerebellar tremor in multiple sclerosis. Journal of Neurology, 2006, 253, 762-766.	3.6	56
82	Lossâ€ofâ€function <scp>KCNH</scp>2 mutation in a family with long <scp>QT</scp> syndrome, epilepsy, and sudden death. Epilepsia, 2013, 54, e112-6.	5.1	56
83	Early Treatment with Quinidine in 2 Patients with Epilepsy of Infancy with Migrating Focal Seizures (EIMFS) Due to Gain-of-Function KCNT1 Mutations: Functional Studies, Clinical Responses, and Critical Issues for Personalized Therapy. Neurotherapeutics, 2018, 15, 1112-1126.	4.4	56
84	Carbamazepineâ€and oxcarbazepineâ€induced hyponatremia in people with epilepsy. Epilepsia, 2017, 58, 1227-1233.	5.1	54
85	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
86	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
87	NovelGNE mutations in Italian families with autosomal recessive hereditary inclusion-body myopathy. Human Mutation, 2004, 23, 632-632.	2.5	52
88	McArdle disease: the mutation spectrum ofPYGMin a large Italian cohort. Human Mutation, 2006, 27, 718-718.	2.5	52
89	Management of genetic epilepsies: From empirical treatment to precision medicine. Pharmacological Research, 2016, 107, 426-429.	7.1	52
90	Pontocerebellar hypoplasia. Neurology, 2010, 75, 1459-1464.	1.1	51

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91	A functional polymorphism in the SCN1A gene does not influence antiepileptic drug responsiveness in Italian patients with focal epilepsy. Epilepsia, 2011, 52, e40-e44.	5.1	50
92	West syndrome associated with 14q12 duplications harboring FOXG1. Neurology, 2011, 76, 1600-1602.	1.1	49
93	Familial Occurrence of Febrile Seizures and Epilepsy in Severe Myoclonic Epilepsy of Infancy (SMEI) Patients with SCN1A Mutations. Epilepsia, 2006, 47, 1629-1635.	5.1	48
94	Typical progression of myoclonic epilepsy of the Lafora type: a case report. Nature Clinical Practice Neurology, 2008, 4, 106-111.	2.5	47
95	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	7.6	47
96	The genetics of monogenic idiopathic epilepsies and epileptic encephalopathies. Seizure: the Journal of the British Epilepsy Association, 2012, 21, 3-11.	2.0	46
97	Homozygous STXBP1 variant causes encephalopathy and gain-of-function in synaptic transmission. Brain, 2020, 143, 441-451.	7.6	46
98	Assessing the landscape of STXBP1-related disorders in 534 individuals. Brain, 2022, 145, 1668-1683.	7.6	46
99	Exploration of the Genetic Architecture of Idiopathic Generalized Epilepsies. Epilepsia, 2006, 47, 1682-1690.	5.1	45
100	Natural history and long-term evolution in families with autosomal dominant cortical tremor, myoclonus, and epilepsy. Epilepsia, 2011, 52, 1245-1250.	5.1	45
101	Dramatic effect of levetiracetam in early-onset epileptic encephalopathy due to STXBP1 mutation. Brain and Development, 2016, 38, 128-131.	1.1	45
102	Advances in genetic testing and optimization of clinical management in children and adults with epilepsy. Expert Review of Neurotherapeutics, 2020, 20, 251-269.	2.8	45
103	Partial Rescue of F508del-CFTR Stability and Trafficking Defects by Double Corrector Treatment. International Journal of Molecular Sciences, 2021, 22, 5262.	4.1	45
104	6q Terminal Deletion Syndrome Associated with a Distinctive EEG and Clinical Pattern: A Report of Five Cases. Epilepsia, 2006, 47, 830-838.	5.1	44
105	Novel mutations in CLN8 in Italian variant late infantile neuronal ceroid lipofuscinosis: another genetic hit in the Mediterranean. Neurogenetics, 2006, 7, 111-117.	1.4	43
106	Spastic paraplegia with thin corpus callosum: description of 20 new families, refinement of the SPG11 locus, candidate gene analysis and evidence of genetic heterogeneity. Neurogenetics, 2006, 7, 149-156.	1.4	43
107	Clinical phenotype and molecular characterization of 6q terminal deletion syndrome: Five new cases. American Journal of Medical Genetics, Part A, 2006, 140A, 1944-1949.	1.2	43
108	Benign adult familial myoclonic epilepsy (BAFME): evidence of an extended founder haplotype on chromosome 2p11.1-q12.2 in five Italian families. Neurogenetics, 2008, 9, 139-142.	1.4	43

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109	Mild <scp>L</scp>afora disease: Clinical, neurophysiologic, and genetic findings. Epilepsia, 2014, 55, e129-33.	5.1	43
110	Genetic variation in CFH predicts phenytoin-induced maculopapular exanthema in European-descent patients. Neurology, 2018, 90, e332-e341.	1.1	43
111	The Î±_{2B}â€adrenergic receptor is mutant in cortical myoclonus and epilepsy. Annals of Neurology, 2014, 75, 77-87.	5.3	42
112	Mapping of a Locus for a Familial Autosomal Recessive Idiopathic Myoclonic Epilepsy of Infancy to Chromosome 16p13. American Journal of Human Genetics, 2000, 66, 1552-1557.	6.2	41
113	Revelation of a Novel CLN5 Mutation in Early Juvenile Neuronal Ceroid Lipofuscinosis. Neuropediatrics, 2007, 38, 46-49.	0.6	41
114	Life-Threatening Status Epilepticus Following Gabapentin Administration in a Patient with Benign Adult Familial Myoclonic Epilepsy. Epilepsia, 2007, 48, 1995-1998.	5.1	41
115	Genetic and forensic implications in epilepsy and cardiac arrhythmias: a case series. International Journal of Legal Medicine, 2015, 129, 495-504.	2.2	40
116	Phenotypic characterization of hypomyelination and congenital cataract. Annals of Neurology, 2007, 62, 121-127.	5.3	39
117	Familial severe myoclonic epilepsy of infancy: truncation of Nav1.1 and genetic heterogeneity. Epileptic Disorders, 2003, 5, 21-5.	1.3	39
118	POMGnT1 Mutations in Congenital Muscular Dystrophy. Archives of Neurology, 2006, 63, 1491.	4.5	38
119	Genetic diagnosis in Lafora disease: Genotype-phenotype correlations and diagnostic pitfalls. Neurology, 2007, 68, 996-1001.	1.1	38
120	Severe Epilepsy in X-Linked Creatine Transporter Defect (CRTR-D). Epilepsia, 2007, 48, 1211-1213.	5.1	38
121	Electroclinical presentation and genotypeâ€“phenotype relationships in patients with Unverrichtâ€Lundborg disease carrying compound heterozygous CSTB point and indel mutations. Epilepsia, 2012, 53, 2120-2127.	5.1	38
122	Extending the phenotypic spectrum of <scp>RBFOX</scp>1 deletions: Sporadic focal epilepsy. Epilepsia, 2015, 56, e129-33.	5.1	38
123	Caveolin-3 T78M and T78K missense mutations lead to different phenotypes in vivo and in vitro. Laboratory Investigation, 2008, 88, 275-283.	3.7	37
124	Inclusion body myopathy, Pagetâ€™s disease of the bone and frontotemporal dementia: recurrence of the VCP R155H mutation in an Italian family and implications for genetic counselling. Clinical Genetics, 2008, 74, 54-60.	2.0	37
125	Expanding sialidosis spectrum by genome-wide screening. Neurology, 2014, 82, 2003-2006.	1.1	37
126	Genetic and Early Clinical Manifestations of Females Heterozygous for Duchenne/Becker Muscular Dystrophy. Pediatric Neurology, 2016, 55, 58-63.	2.1	37

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127	No Evidence of a Major Locus for Benign Familial Infantile Convulsions on Chromosome 19q12-q13.1. Epilepsia, 1999, 40, 1799-1803.	5.1	36
128	Dramatic response to levetiracetam in post-ischaemic Holmes' tremor. Journal of Neurology, Neurosurgery and Psychiatry, 2006, 78, 438-439.	1.9	36
129	Multiplex real-time PCR for detection of deletions and duplications in dystrophin gene. Biochemical and Biophysical Research Communications, 2006, 339, 145-150.	2.1	35
130	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
131	Comparative effectiveness of antiepileptic drugs in juvenile myoclonic epilepsy. Epilepsia Open, 2019, 4, 420-430.	2.4	34
132	KCNT1-related epilepsies and epileptic encephalopathies: phenotypic and mutational spectrum. Brain, 2021, 144, 3635-3650.	7.6	34
133	A new method for analysis of mitochondrial DNA point mutations and assess levels of heteroplasmy. Biochemical and Biophysical Research Communications, 2006, 342, 387-393.	2.1	33
134	Rare GABRA3 variants are associated with epileptic seizures, encephalopathy and dysmorphic features. Brain, 2017, 140, 2879-2894.	7.6	33
135	Constitutive Inactivation of the PRRT2 Gene Alters Short-Term Synaptic Plasticity and Promotes Network Hyperexcitability in Hippocampal Neurons. Cerebral Cortex, 2019, 29, 2010-2033.	2.9	33
136	The first three mosaic cri du chat syndrome patients with two rearranged cell lines. Journal of Medical Genetics, 2000, 37, 967-972.	3.2	32
137	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
138	Clinical and molecular characterization of 112 single-center patients with Neurofibromatosis type 1. Italian Journal of Pediatrics, 2018, 44, 45.	2.6	32
139	Short and long interval cortical inhibition in patients with Unverricht-Lundborg and Lafora body disease. Epilepsy Research, 2010, 89, 232-237.	1.6	31
140	Brain Organoids as Model Systems for Genetic Neurodevelopmental Disorders. Frontiers in Cell and Developmental Biology, 2020, 8, 590119.	3.7	31
141	Chewing induced reflex seizures (â€œeating epilepsyâ€) and eye closure sensitivity as a common feature in pediatric patients with SYNGAP1 mutations: Review of literature and report of 8 cases. Seizure: the Journal of the British Epilepsy Association, 2019, 65, 131-137.	2.0	30
142	Climate change and epilepsy: Insights from clinical and basic science studies. Epilepsy and Behavior, 2021, 116, 107791.	1.7	30
143	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
144	Novel GABRG2 mutations cause familial febrile seizures. Neurology: Genetics, 2015, 1, e35.	1.9	29

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145	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
146	Absence of mutations in major GEFS+ genes in myoclonic astatic epilepsy. Epilepsy Research, 2003, 56, 127-133.	1.6	28
147	Unfavourable outcome of Hashimoto encephalopathy due to status epilepticus. Journal of Neurology, 2006, 253, 248-249.	3.6	28
148	EXOSC3 mutations in isolated cerebellar hypoplasia and spinal anterior horn involvement. Journal of Neurology, 2013, 260, 1866-1870.	3.6	28
149	CHD2 mutations are a rare cause of generalized epilepsy with myoclonicâ€“atonic seizures. Epilepsy and Behavior, 2015, 51, 53-56.	1.7	28
150	Gain-of-functionHCN2variants in genetic epilepsy. Human Mutation, 2018, 39, 202-209.	2.5	28
151	Familial Infantile Myoclonic Epilepsy: Clinical Features in a Large Kindred with Autosomal Recessive Inheritance. Epilepsia, 2001, 42, 1541-1548.	5.1	27
152	Electroclinical and Genetic Findings in a Family with Cortical Tremor, Myoclonus, and Epilepsy. Epilepsia, 2005, 46, 1993-1995.	5.1	27
153	NF1 microdeletion syndrome: case report of two new patients. Italian Journal of Pediatrics, 2019, 45, 138.	2.6	27
154	Clinical and electrophysiological features of epilepsy in Italian patients with CLN8 mutations. Epilepsy and Behavior, 2007, 10, 187-191.	1.7	26
155	Gallowayâ€“Mowat syndrome: An early-onset progressive encephalopathy with intractable epilepsy associated to renal impairment. Two novel cases and review of literature. Seizure: the Journal of the British Epilepsy Association, 2010, 19, 132-135.	2.0	26
156	Comparative effectiveness of antiepileptic drugs in patients with mesial temporal lobe epilepsy with hippocampal sclerosis. Epilepsia, 2017, 58, 1734-1741.	5.1	26
157	Hypomyelination and Congenital Cataract: Neuroimaging Features of a Novel Inherited White Matter Disorder. American Journal of Neuroradiology, 2008, 29, 301-305.	2.4	25
158	Refractory, life-threatening status epilepticus in a 3-year-old girl. Lancet Neurology, The, 2008, 7, 278-284.	10.2	24
159	A pilot open-label trial of zonisamide in Unverricht-Lundborg disease. Movement Disorders, 2011, 26, 341-343.	3.9	24
160	Genotype-phenotype correlations in patients with de novo KCNQ2 pathogenic variants. Neurology: Genetics, 2020, 6, e528.	1.9	24
161	Epilepsy Course and Developmental Trajectories in STXBP1 -DEE. Neurology: Genetics, 2022, 8, .	1.9	24
162	Linkage Analysis and Disease Models in Benign Familial Infantile Seizures: A Study of 16 Families. Epilepsia, 2006, 47, 1029-1034.	5.1	23

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163	Pyridoxineâ€dependent epilepsy: An underâ€recognised cause of intractable seizures. Journal of Paediatrics and Child Health, 2012, 48, E113-5.	0.8	23
164	The ubiquitin ligase tripartite-motif-protein 32 is induced in Duchenne muscular dystrophy. Laboratory Investigation, 2016, 96, 862-871.	3.7	23
165	TBC1D24 regulates axonal outgrowth and membrane trafficking at the growth cone in rodent and human neurons. Cell Death and Differentiation, 2019, 26, 2464-2478.	11.2	23
166	Type 1 diabetes and epilepsy: More than a casual association?. Epilepsia, 2010, 51, 320-321.	5.1	22
167	Hypomyelination and Congenital Cataract. Archives of Neurology, 2011, 68, 1191.	4.5	22
168	PRRT2 is mutated in familial and non-familial benign infantile seizures. European Journal of Paediatric Neurology, 2013, 17, 77-81.	1.6	22
169	Heterogeneous contribution of microdeletions in the development of common generalised and focal epilepsies. Journal of Medical Genetics, 2017, 54, 598-606.	3.2	22
170	Novel AMPD2 mutation in pontocerebellar hypoplasia, dysmorphisms, and teeth abnormalities. Neurology: Genetics, 2017, 3, e179.	1.9	22
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