Federico Zara

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/2271994/publications.pdf

Version: 2024-02-01

328 papers	17,900 citations	17405 63 h-index	19690 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.	6.0	2,642
2	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	6.0	1,085
3	Mutations in the caveolin-3 gene cause autosomal dominant limb-girdle muscular dystrophy. Nature Genetics, 1998, 18, 365-368.	9.4	555
4	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. Brain, 2017, 140, 1316-1336.	3.7	426
5	De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	2.6	388
6	The genetics of Dravet syndrome. Epilepsia, 2011, 52, 24-29.	2.6	287
7	Genetic determinants of common epilepsies: a meta-analysis of genome-wide association studies. Lancet Neurology, The, 2014, 13, 893-903.	4.9	264
8	Benign familial neonatal-infantile seizures: Characterization of a new sodium channelopathy. Annals of Neurology, 2004, 55, 550-557.	2.8	250
9	Spectrum of SCN1A mutations in severe myoclonic epilepsy of infancy. Neurology, 2003, 60, 1961-1967.	1.5	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.	2.6	237
11	De novo mutations in HCN1 cause early infantile epileptic encephalopathy. Nature Genetics, 2014, 46, 640-645.	9.4	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.	2.6	188
13	Genetic testing in the epilepsiesâ€"Report of the ILAE Genetics Commission. Epilepsia, 2010, 51, 655-670.	2.6	175
14	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. American Journal of Human Genetics, 2015, 96, 808-815.	2.6	173
15	Epilepsy, hippocampal sclerosis and febrile seizures linked by common genetic variation around SCN1A. Brain, 2013, 136, 3140-3150.	3.7	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.	1.5	158
17	SCN1A duplications and deletions detected in Dravet syndrome: Implications for molecular diagnosis. Epilepsia, 2009, 50, 1670-1678.	2.6	152
18	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	5.8	150

#	Article	IF	Citations
19	Genome search for susceptibility loci of common idiopathic generalised epilepsies. Human Molecular Genetics, 2000, 9, 1465-1472.	1.4	147
20	Clinical and molecular characterisation of 80 patients with 5p deletion: genotype-phenotype correlation. Journal of Medical Genetics, 2001, 38, 151-158.	1.5	147
21	TBC1D24, an ARF6-Interacting Protein, Is Mutated in Familial Infantile Myoclonic Epilepsy. American Journal of Human Genetics, 2010, 87, 365-370.	2.6	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.	1.4	134
23	PRRT2 Is a Key Component of the Ca 2+ -Dependent Neurotransmitter Release Machinery. Cell Reports, 2016, 15, 117-131.	2.9	121
24	Clinical and genetic heterogeneity of branching enzyme deficiency (glycogenosis type IV). Neurology, 2004, 63, 1053-1058.	1.5	120
25	Mutations in <scp>KCNT</scp>1 cause a spectrum of focal epilepsies. Epilepsia, 2015, 56, e114-20.	2.6	117
26	Microbiota-gut brain axis involvement in neuropsychiatric disorders. Expert Review of Neurotherapeutics, 2019, 19, 1037-1050.	1.4	116
27	An open-label trial of levetiracetam in severe myoclonic epilepsy of infancy. Neurology, 2007, 69, 250-254.	1.5	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.	1.7	110
29	Genetic testing in benign familial epilepsies of the first year of life: Clinical and diagnostic significance. Epilepsia, 2013, 54, 425-436.	2.6	110
30	Novel Compound Heterozygous Mutations inTBC1D24Cause Familial Malignant Migrating Partial Seizures of Infancy. Human Mutation, 2013, 34, 869-872.	1.1	110
31	Mapping of genes predisposing to idiopathic generalized epilepsy. Human Molecular Genetics, 1995, 4, 1201-1207.	1.4	109
32	Somatic and germline mosaicisms in Severe Myoclonic Epilepsy of Infancy. Biochemical and Biophysical Research Communications, 2006, 341, 489-493.	1.0	102
33	PRRT2 controls neuronal excitability by negatively modulating Na+ channel 1.2/1.6 activity. Brain, 2018, 141, 1000-1016.	3.7	99
34	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	5.8	99
35	TBC1D24 genotypeâ€“phenotype correlation. Neurology, 2016, 87, 77-85.	1.5	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.	3.7	96

#	Article	IF	Citations
37	HCN1mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. Brain, 2018, 141, 3160-3178.	3.7	96
38	Dravet syndrome: Early clinical manifestations and cognitive outcome in 37 Italian patients. Brain and Development, 2010, 32, 71-77.	0.6	94
39	PRRT2 Mutations are the major cause of benign familial infantile seizures. Human Mutation, 2012, 33, 1439-1443.	1.1	93
40	POMT2 gene mutation in limb-girdle muscular dystrophy with inflammatory changes. Biochemical and Biophysical Research Communications, 2007, 363, 1033-1037.	1.0	91
41	The leukodystrophy protein FAM126A (hyccin) regulates PtdIns(4)P synthesis at the plasmaÂmembrane. Nature Cell Biology, 2016, 18, 132-138.	4.6	91
42	Coexistence of epilepsy and Brugada syndrome in a family with SCN5A mutation. Epilepsy Research, 2013, 105, 415-418.	0.8	90
43	Brain MRI Findings in Severe Myoclonic Epilepsy in Infancy and Genotype?Phenotype Correlations. Epilepsia, 2007, 48, 1092-1096.	2.6	89
44	A pilot trial of levetiracetam in eyelid myoclonia with absences (Jeavons syndrome). Epilepsia, 2008, 49, 425-430.	2.6	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.	2.6	88
46	Progressive myoclonic epilepsies. Neurology, 2014, 82, 405-411.	1.5	87
47	GLUT1 mutations are a rare cause of familial idiopathic generalized epilepsy. Neurology, 2012, 78, 557-562.	1.5	86
48	Deficiency of hyccin, a newly identified membrane protein, causes hypomyelination and congenital cataract. Nature Genetics, 2006, 38, 1111-1113.	9.4	82
49	Cryptic chromosome deletions involving SCN1A in severe myoclonic epilepsy of infancy. Neurology, 2006, 67, 1230-1235.	1.5	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.	3.3	80
51	Impairment of ceramide synthesis causes a novel progressive myoclonus epilepsy. Annals of Neurology, 2014, 76, 206-212.	2.8	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.	2.6	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.	2.6	78
54	Benign adult familial myoclonic epilepsy. Neurology, 2003, 60, 1381-1385.	1.5	75

#	Article	IF	Citations
55	A Novel SCN2A Mutation in Family with Benign Familial Infantile Seizures. Epilepsia, 2006, 47, 218-220.	2.6	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.	1.1	74
57	The PRRT2 knockout mouse recapitulates the neurological diseases associated with PRRT2 mutations. Neurobiology of Disease, 2017, 99, 66-83.	2.1	72
58	Clinical and Genetic Findings in 26 Italian Patients with Lafora Disease. Epilepsia, 2006, 47, 640-643.	2.6	71
59	Therapeutic Potential of Proteasome Inhibition in Duchenne and Becker Muscular Dystrophies. American Journal of Pathology, 2010, 176, 1863-1877.	1.9	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.	2.6	71
61	The spectrum of intermediate <scp>SCN</scp>8Aâ€related epilepsy. Epilepsia, 2019, 60, 830-844.	2.6	70
62	Late-onset and Slow-progressing Lafora Disease in Four Siblings with EPM2B Mutation. Epilepsia, 2005, 46, 1695-1697.	2.6	69
63	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. Brain, 2022, 145, 2991-3009.	3.7	69
64	Addition of verapamil in the treatment of severe myoclonic epilepsy in infancy. Epilepsy Research, 2009, 85, 89-95.	0.8	68
65	PRRT2: from Paroxysmal Disorders to Regulation of Synaptic Function. Trends in Neurosciences, 2016, 39, 668-679.	4.2	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.	2.6	67
67	Pitfalls in genetic testing: the story of missed SCN1A mutations. Molecular Genetics & Genomic Medicine, 2016, 4, 457-464.	0.6	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.	4.9	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.	2.6	66
70	A novel KCNQ3 mutation in familial epilepsy with focal seizures and intellectual disability. Epilepsia, 2015, 56, e15-20.	2.6	66
71	Confirmation of mutations inPROSCas a novel cause of vitamin B_₆-dependent epilepsy. Journal of Medical Genetics, 2017, 54, 809-814.	1.5	66
72	Recent advances in epilepsy genetics. Neuroscience Letters, 2018, 667, 4-9.	1.0	66

#	Article	IF	Citations
73	PRRT2-related disorders: further PKD and ICCA cases and review of the literature. Journal of Neurology, 2013, 260, 1234-1244.	1.8	63
74	Autosomal dominant cortical tremor, myoclonus and epilepsy: many syndromes, one phenotype. Acta Neurologica Scandinavica, 2005, 111, 211-217.	1.0	61
75	Clinical Significance of Rare Copy Number Variations in Epilepsy. Archives of Neurology, 2012, 69, 322.	4.9	61
76	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	2.6	61
77	Genetics of reflex seizures and epilepsies in humans and animals. Epilepsy Research, 2016, 121, 47-54.	0.8	60
78	¹Hâ€MR spectroscopy indicates prominent cerebellar dysfunction in benign adult familial myoclonic epilepsy. Epilepsia, 2009, 50, 1491-1497.	2.6	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.	2.1	57
80	Efficacy of sodium channel blockers in SCN2A early infantile epileptic encephalopathy. Brain and Development, 2017, 39, 345-348.	0.6	57
81	Levetiracetam for cerebellar tremor in multiple sclerosis. Journal of Neurology, 2006, 253, 762-766.	1.8	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.	2.6	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.	2.1	56
84	Carbamazepineâ€•and oxcarbazepineâ€‐induced hyponatremia in people with epilepsy. Epilepsia, 2017, 58, 1227-1233.	2.6	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.	2.6	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.	1.4	53
87	NovelGNE mutations in Italian families with autosomal recessive hereditary inclusion-body myopathy. Human Mutation, 2004, 23, 632-632.	1.1	52
88	McArdle disease: the mutation spectrum ofPYGMin a large Italian cohort. Human Mutation, 2006, 27, 718-718.	1.1	52
89	Management of genetic epilepsies: From empirical treatment to precision medicine. Pharmacological Research, 2016, 107, 426-429.	3.1	52
90	Pontocerebellar hypoplasia. Neurology, 2010, 75, 1459-1464.	1.5	51

#	Article	IF	Citations
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.	2.6	50
92	West syndrome associated with 14q12 duplications harboring FOXG1. Neurology, 2011, 76, 1600-1602.	1.5	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.	2.6	48
94	Typical progression of myoclonic epilepsy of the Lafora type: a case report. Nature Clinical Practice Neurology, 2008, 4, 106-111.	2.7	47
95	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	3.7	47
96	The genetics of monogenic idiopathic epilepsies and epileptic encephalopathies. Seizure: the Journal of the British Epilepsy Association, 2012, 21, 3-11.	0.9	46
97	Homozygous STXBP1 variant causes encephalopathy and gain-of-function in synaptic transmission. Brain, 2020, 143, 441-451.	3.7	46
98	Assessing the landscape of STXBP1-related disorders in 534 individuals. Brain, 2022, 145, 1668-1683.	3.7	46
99	Exploration of the Genetic Architecture of Idiopathic Generalized Epilepsies. Epilepsia, 2006, 47, 1682-1690.	2.6	45
100	Natural history and long-term evolution in families with autosomal dominant cortical tremor, myoclonus, and epilepsy. Epilepsia, 2011, 52, 1245-1250.	2.6	45
101	Dramatic effect of levetiracetam in early-onset epileptic encephalopathy due to STXBP1 mutation. Brain and Development, 2016, 38, 128-131.	0.6	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.	1.4	45
103	Partial Rescue of F508del-CFTR Stability and Trafficking Defects by Double Corrector Treatment. International Journal of Molecular Sciences, 2021, 22, 5262.	1.8	45
104	6q Terminal Deletion Syndrome Associated with a Distinctive EEG and Clinical Pattern: A Report of Five Cases. Epilepsia, 2006, 47, 830-838.	2.6	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.	0.7	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.	0.7	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.	0.7	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.	0.7	43

#	Article	IF	Citations
109	Mild <scp>L</scp>afora disease: Clinical, neurophysiologic, and genetic findings. Epilepsia, 2014, 55, e129-33.	2.6	43
110	Genetic variation in CFH predicts phenytoin-induced maculopapular exanthema in European-descent patients. Neurology, 2018, 90, e332-e341.	1.5	43
111	The Î±_{2B}â€adrenergic receptor is mutant in cortical myoclonus and epilepsy. Annals of Neurology, 2014, 75, 77-87.	2.8	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.	2.6	41
113	Revelation of a Novel CLN5 Mutation in Early Juvenile Neuronal Ceroid Lipofuscinosis. Neuropediatrics, 2007, 38, 46-49.	0.3	41
114	Life-Threatening Status Epilepticus Following Gabapentin Administration in a Patient with Benign Adult Familial Myoclonic Epilepsy. Epilepsia, 2007, 48, 1995-1998.	2.6	41
115	Genetic and forensic implications in epilepsy and cardiac arrhythmias: a case series. International Journal of Legal Medicine, 2015, 129, 495-504.	1.2	40
116	Phenotypic characterization of hypomyelination and congenital cataract. Annals of Neurology, 2007, 62, 121-127.	2.8	39
117	Familial severe myoclonic epilepsy of infancy: truncation of Nav1.1 and genetic heterogeneity. Epileptic Disorders, 2003, 5, 21-5.	0.7	39
118	POMGnT1 Mutations in Congenital Muscular Dystrophy. Archives of Neurology, 2006, 63, 1491.	4.9	38
119	Genetic diagnosis in Lafora disease: Genotype-phenotype correlations and diagnostic pitfalls. Neurology, 2007, 68, 996-1001.	1.5	38
120	Severe Epilepsy in X-Linked Creatine Transporter Defect (CRTR-D). Epilepsia, 2007, 48, 1211-1213.	2.6	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.	2.6	38
122	Extending the phenotypic spectrum of <scp>RBFOX</scp>1 deletions: Sporadic focal epilepsy. Epilepsia, 2015, 56, e129-33.	2.6	38
123	Caveolin-3 T78M and T78K missense mutations lead to different phenotypes in vivo and in vitro. Laboratory Investigation, 2008, 88, 275-283.	1.7	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.	1.0	37
125	Expanding sialidosis spectrum by genome-wide screening. Neurology, 2014, 82, 2003-2006.	1.5	37
126	Genetic and Early Clinical Manifestations of Females Heterozygous for Duchenne/Becker Muscular Dystrophy. Pediatric Neurology, 2016, 55, 58-63.	1.0	37

#	Article	IF	Citations
127	No Evidence of a Major Locus for Benign Familial Infantile Convulsions on Chromosome 19q12-q13.1. Epilepsia, 1999, 40, 1799-1803.	2.6	36
128	Dramatic response to levetiracetam in post-ischaemic Holmes' tremor. Journal of Neurology, Neurosurgery and Psychiatry, 2006, 78, 438-439.	0.9	36
129	Multiplex real-time PCR for detection of deletions and duplications in dystrophin gene. Biochemical and Biophysical Research Communications, 2006, 339, 145-150.	1.0	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.	2.6	35
131	Comparative effectiveness of antiepileptic drugs in juvenile myoclonic epilepsy. Epilepsia Open, 2019, 4, 420-430.	1.3	34
132	KCNT1-related epilepsies and epileptic encephalopathies: phenotypic and mutational spectrum. Brain, 2021, 144, 3635-3650.	3.7	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.	1.0	33
134	Rare GABRA3 variants are associated with epileptic seizures, encephalopathy and dysmorphic features. Brain, 2017, 140, 2879-2894.	3.7	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.	1.6	33
136	The first three mosaic cri du chat syndrome patients with two rearranged cell lines. Journal of Medical Genetics, 2000, 37, 967-972.	1.5	32
137	Genomeâ€wide linkage metaâ€analysis identifies susceptibility loci at 2q34 and 13q31.3 for genetic generalized epilepsies. Epilepsia, 2012, 53, 308-318.	2.6	32
138	Clinical and molecular characterization of 112 single-center patients with Neurofibromatosis type 1. Italian Journal of Pediatrics, 2018, 44, 45.	1.0	32
139	Short and long interval cortical inhibition in patients with Unverricht-Lundborg and Lafora body disease. Epilepsy Research, 2010, 89, 232-237.	0.8	31
140	Brain Organoids as Model Systems for Genetic Neurodevelopmental Disorders. Frontiers in Cell and Developmental Biology, 2020, 8, 590119.	1.8	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.	0.9	30
142	Climate change and epilepsy: Insights from clinical and basic science studies. Epilepsy and Behavior, 2021, 116, 107791.	0.9	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.	0.9	30
144	Novel GABRG2 mutations cause familial febrile seizures. Neurology: Genetics, 2015, 1, e35.	0.9	29

#	Article	IF	Citations
145	Identity by descent fine mapping of familial adult myoclonus epilepsy (FAME) to 2p11.2â€“2q11.2. Human Genetics, 2016, 135, 1117-1125.	1.8	29
146	Absence of mutations in major GEFS+ genes in myoclonic astatic epilepsy. Epilepsy Research, 2003, 56, 127-133.	0.8	28
147	Unfavourable outcome of Hashimoto encephalopathy due to status epilepticus. Journal of Neurology, 2006, 253, 248-249.	1.8	28
148	EXOSC3 mutations in isolated cerebellar hypoplasia and spinal anterior horn involvement. Journal of Neurology, 2013, 260, 1866-1870.	1.8	28
149	CHD2 mutations are a rare cause of generalized epilepsy with myoclonicâ€“atonic seizures. Epilepsy and Behavior, 2015, 51, 53-56.	0.9	28
150	Gain-of-functionHCN2variants in genetic epilepsy. Human Mutation, 2018, 39, 202-209.	1.1	28
151	Familial Infantile Myoclonic Epilepsy: Clinical Features in a Large Kindred with Autosomal Recessive Inheritance. Epilepsia, 2001, 42, 1541-1548.	2.6	27
152	Electroclinical and Genetic Findings in a Family with Cortical Tremor, Myoclonus, and Epilepsy. Epilepsia, 2005, 46, 1993-1995.	2.6	27
153	NF1 microdeletion syndrome: case report of two new patients. Italian Journal of Pediatrics, 2019, 45, 138.	1.0	27
154	Clinical and electrophysiological features of epilepsy in Italian patients with CLN8 mutations. Epilepsy and Behavior, 2007, 10, 187-191.	0.9	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.	0.9	26
156	Comparative effectiveness of antiepileptic drugs in patients with mesial temporal lobe epilepsy with hippocampal sclerosis. Epilepsia, 2017, 58, 1734-1741.	2.6	26
157	Hypomyelination and Congenital Cataract: Neuroimaging Features of a Novel Inherited White Matter Disorder. American Journal of Neuroradiology, 2008, 29, 301-305.	1.2	25
158	Refractory, life-threatening status epilepticus in a 3-year-old girl. Lancet Neurology, The, 2008, 7, 278-284.	4.9	24
159	A pilot open-label trial of zonisamide in Unverricht-Lundborg disease. Movement Disorders, 2011, 26, 341-343.	2.2	24
160	Genotype-phenotype correlations in patients with de novo KCNQ2 pathogenic variants. Neurology: Genetics, 2020, 6, e528.	0.9	24
161	Epilepsy Course and Developmental Trajectories in STXBP1 -DEE. Neurology: Genetics, 2022, 8, .	0.9	24
162	Linkage Analysis and Disease Models in Benign Familial Infantile Seizures: A Study of 16 Families. Epilepsia, 2006, 47, 1029-1034.	2.6	23

#	Article	IF	Citations
163	Pyridoxineâ€dependent epilepsy: An underâ€recognised cause of intractable seizures. Journal of Paediatrics and Child Health, 2012, 48, E113-5.	0.4	23
164	The ubiquitin ligase tripartite-motif-protein 32 is induced in Duchenne muscular dystrophy. Laboratory Investigation, 2016, 96, 862-871.	1.7	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.	5.0	23
166	Type 1 diabetes and epilepsy: More than a casual association?. Epilepsia, 2010, 51, 320-321.	2.6	22
167	Hypomyelination and Congenital Cataract. Archives of Neurology, 2011, 68, 1191.	4.9	22
168	PRRT2 is mutated in familial and non-familial benign infantile seizures. European Journal of Paediatric Neurology, 2013, 17, 77-81.	0.7	22
169	Heterogeneous contribution of microdeletions in the development of common generalised and focal epilepsies. Journal of Medical Genetics, 2017, 54, 598-606.	1.5	22
170	Novel AMPD2 mutation in pontocerebellar hypoplasia, dysmorphisms, and teeth abnormalities. Neurology: Genetics, 2017, 3, e179.	0.9	22
171	Testing association of rare genetic variants with resistance to three common antiseizure medications. Epilepsia, 2020, 61, 657-666.	2.6	22
172	Loss of Wwox Perturbs Neuronal Migration and Impairs Early Cortical Development. Frontiers in Neuroscience, 2020, 14, 644.	1.4	22
173	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	3.7	22
174	Evaluation of Presumably Disease Causing SCN1A Variants in a Cohort of Common Epilepsy Syndromes. PLoS ONE, 2016, 11, e0150426.	1.1	22
175	Lack of SCN1A Mutations in Familial Febrileâ€Seizures. Epilepsia, 2002, 43, 559-562.	2.6	21
176	Periodic Myoclonus Due to Cytomegalovirus Encephalitis in a Patient With Good Syndrome. Archives of Neurology, 2007, 64, 277.	4.9	21
177	Psychiatric comorbidities in patients from seven families with autosomal dominant cortical tremor, myoclonus, and epilepsy. Epilepsy and Behavior, 2016, 56, 38-43.	0.9	21
178	Pharmacoresponse in genetic generalized epilepsy: a genome-wide association study. Pharmacogenomics, 2020, 21, 325-335.	0.6	21
179	Biallelic MFSD2A variants associated with congenital microcephaly, developmental delay, and recognizable neuroimaging features. European Journal of Human Genetics, 2020, 28, 1509-1519.	1.4	21
180	Genotype-Phenotype Correlations in Neurofibromatosis Type 1: A Single-Center Cohort Study. Cancers, 2021, 13, 1879.	1.7	21

#	Article	IF	Citations
181	No evidence of GABRG2 mutations in severe myoclonic epilepsy of infancy. Epilepsy Research, 2003, 53, 196-200.	0.8	20
182	Temporal lobe epilepsy and anti glutamic acid decarboxylase autoimmunity. Neurological Sciences, 2011, 32, 547-550.	0.9	20
183	Hyccin, the Molecule Mutated in the Leukodystrophy Hypomyelination and Congenital Cataract (HCC), Is a Neuronal Protein. PLoS ONE, 2012, 7, e32180.	1.1	20
184	Autosomal dominant cortical tremor, myoclonus and epilepsy. Epileptic Disorders, 2016, 18, 139-144.	0.7	20
185	Progressive Myoclonus Epilepsies. Neurology: Genetics, 2021, 7, e641.	0.9	20
186	Benign Infantile Familial Convulsions: Natural History of a Case and Clinical Characteristics of a Large Italian Family. Neuropediatrics, 1999, 30, 99-101.	0.3	19
187	Earlyâ€onset absence epilepsy: <scp>SLC</scp>2<scp>A</scp>1 gene analysis and treatment evolution. European Journal of Neurology, 2013, 20, 856-859.	1.7	19
188	White matter involvement in a family with a novel PDGFB mutation. Neurology: Genetics, 2016, 2, e77.	0.9	19
189	Clinical intrafamilial variability in lethal familial neonatal seizure disorder caused by TBC1D24 mutations. American Journal of Medical Genetics, Part A, 2016, 170, 3207-3214.	0.7	19
190	Exosomes from Plasma of Neuroblastoma Patients Contain Doublestranded DNA Reflecting the Mutational Status of Parental Tumor Cells. International Journal of Molecular Sciences, 2021, 22, 3667.	1.8	19
191	The L467F-F508del Complex Allele Hampers Pharmacological Rescue of Mutant CFTR by Elexacaftor/Tezacaftor/Ivacaftor in Cystic Fibrosis Patients: The Value of the Ex Vivo Nasal Epithelial Model to Address Non-Responders to CFTR-Modulating Drugs. International Journal of Molecular Sciences, 2022, 23, 3175.	1.8	19
192	Familial adult myoclonic epilepsy: A new expansion repeats disorder. Seizure: the Journal of the British Epilepsy Association, 2019, 67, 73-77.	0.9	18
193	Unusual EEG pattern linked to chromosome 3p in a family with idiopathic generalized epilepsy. Neurology, 1998, 51, 493-498.	1.5	17
194	Epileptic seizures can follow high doses of oral vardenafil. BMJ: British Medical Journal, 2006, 333, 785.	2.4	17
195	Genetic epileptic encephalopathies: Is all written into the <scp>DNA</scp>?. Epilepsia, 2013, 54, 22-26.	2.6	17
196	Biallelic PI4KA variants cause neurological, intestinal and immunological disease. Brain, 2021, 144, 3597-3610.	3.7	17
197	Diagnostic Approach to Macrocephaly in Children. Frontiers in Pediatrics, 2021, 9, 794069.	0.9	17
198	Inherited neuromyotonia: A clinical and genetic study of a family. Neuromuscular Disorders, 2007, 17, 23-27.	0.3	16

#	Article	IF	Citations
199	Glutamic acid decarboxylase antibodies in idiopathic generalized epilepsy and type 1 diabetes. Annals of Neurology, 2008, 63, 127-128.	2.8	16
200	A very fast and accurate method for calling aberrations in array-CGH data. Biostatistics, 2010, 11, 515-518.	0.9	16
201	Long-term follow-up in two siblings with pyridoxine-dependent seizures associated with a novel ALDH7A1 mutation. European Journal of Paediatric Neurology, 2011, 15, 547-550.	0.7	16
202	Mutations in mTOR pathway linked to megalencephaly syndromes. Nature Reviews Neurology, 2012, 8, 542-544.	4.9	16
203	Exome Sequencing Fails to Identify the Genetic Cause of Aicardi Syndrome. Molecular Syndromology, 2016, 7, 234-238.	0.3	16
204	Assessing the role of rare genetic variants in drugâ€resistant, nonâ€lesional focal epilepsy. Annals of Clinical and Translational Neurology, 2021, 8, 1376-1387.	1.7	16
205	The impact of genetics on the classification of epilepsy syndromes. Epilepsia, 2009, 50, 11-14.	2.6	15
206	TEMPORAL LOBE ABNORMALITIES ON BRAIN MRI IN HEALTHY VOLUNTEERS: A PROSPECTIVE CASE-CONTROL STUDY. Neurology, 2010, 75, 377-378.	1.5	15
207	A clinical and genetic study of 33 new cases with early-onset absence epilepsy. Epilepsy Research, 2011, 95, 221-226.	0.8	15
208	A further contribution to the delineation of epileptic phenotype in PACS2-related syndrome. Seizure: the Journal of the British Epilepsy Association, 2020, 79, 53-55.	0.9	15
209	22â€YEARâ€OLD GIRL WITH STATUS EPILEPTICUS AND PROGRESSIVE NEUROLOGICAL SYMPTOMS. Brain Pathology, 2009, 19, 727-730.	2.1	14
210	Different electroclinical picture of generalized epilepsy in two families with 15q13.3 microdeletion. Epilepsia, 2013, 54, e69-73.	2.6	14
211	Clinical dissection of early onset absence epilepsy in children and prognostic implications. Epilepsia, 2013, 54, 1761-1770.	2.6	14
212	Clinical spectrum and genotype-phenotype correlations in PRRT2 Italian patients. European Journal of Paediatric Neurology, 2020, 28, 193-197.	0.7	14
213	CHD2 mutations: Only epilepsy? Description of cognitive and behavioral profile in a case with a new mutation. Seizure: the Journal of the British Epilepsy Association, 2017, 51, 186-189.	0.9	13
214	Targeting Alternative Splicing as a Potential Therapy for Episodic Ataxia Type 2. Biomedicines, 2020, 8, 332.	1.4	13
215	Targeted re-sequencing in malformations of cortical development: genotype-phenotype correlations. Seizure: the Journal of the British Epilepsy Association, 2020, 80, 145-152.	0.9	13
216	Autosomal Recessive Idiopathic Epilepsy in an Inbred Family from Turkey: Identification of a Putative Locus on Chromosome 9q32-33. Epilepsia, 2004, 45, 479-487.	2.6	12

#	Article	IF	Citations
217	Benign myoclonic epilepsy in infancy followed by childhood absence epilepsy. Seizure: the Journal of the British Epilepsy Association, 2011, 20, 727-730.	0.9	12
218	Novel FAM126A mutations in hypomyelination and congenital cataract disease. Biochemical and Biophysical Research Communications, 2013, 439, 369-372.	1.0	12
219	Chiari malformation type I: what information from the genetics?. Child's Nervous System, 2019, 35, 1665-1671.	0.6	12
220	Genomic and clinical predictors of lacosamide response in refractory epilepsies. Epilepsia Open, 2019, 4, 563-571.	1.3	12
221	Familial benign nonprogressive myoclonic epilepsies. Epilepsia, 2009, 50, 37-40.	2.6	11
222	AIMP1/p43 Mutation and PMLD. American Journal of Human Genetics, 2011, 88, 391.	2.6	11
223	Fever as a seizure precipitant factor in Panayiotopoulos syndrome: A clinical and genetic study. Seizure: the Journal of the British Epilepsy Association, 2012, 21, 141-143.	0.9	11
224	Functional Connectivity and Genetic Profile of a â€œDouble-Cortexâ€•Like Malformation. Frontiers in Integrative Neuroscience, 2018, 12, 22.	1.0	11
225	Emerging treatments for progressive myoclonus epilepsies. Expert Review of Neurotherapeutics, 2020, 20, 341-350.	1.4	11
226	Progress of Induced Pluripotent Stem Cell Technologies to Understand Genetic Epilepsy. International Journal of Molecular Sciences, 2020, 21, 482.	1.8	11
227	Italian cohort of Lafora disease: Clinical features, disease evolution, and genotype-phenotype correlations. Journal of the Neurological Sciences, 2021, 424, 117409.	0.3	11
228	Clinical and Genetic Features in Patients With Reflex Bathing Epilepsy. Neurology, 2021, 97, e577-e586.	1.5	11
229	Dramatic response to levetiracetam in post-ischaemic Holmes' tremor. BMJ Case Reports, 2009, 2009, bcr0820080643-bcr0820080643.	0.2	11
230	Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. American Journal of Human Genetics, 2021, 108, 2006-2016.	2.6	11
231	Spectrum of Phenotypic, Genetic, and Functional Characteristics in Patients With Epilepsy With KCNC2 Pathogenic Variants. Neurology, 2022, 98, .	1.5	11
232	A pharmacogenomic assessment of psychiatric adverse drug reactions to levetiracetam. Epilepsia, 2022, 63, 1563-1570.	2.6	11
233	Bioptically demonstrated Lafora disease without EPM2A mutation: a clinical and neurophysiological study of two sisters. Clinical Neurology and Neurosurgery, 2003, 106, 56-60.	0.6	10
234	Truncation of Caveolin-3 causes autosomal-recessive Rippling Muscle Disease. Journal of Neurology, Neurosurgery and Psychiatry, 2007, 79, 735-737.	0.9	10

#	Article	IF	Citations
235	Epileptic myoclonus as ciprofloxacin-associated adverse effect. Movement Disorders, 2007, 22, 1675-1676.	2.2	10
236	FAME 3: A NOVEL FORM OF PROGRESSIVE MYOCLONUS AND EPILEPSY. Neurology, 2008, 70, 85-86.	1.5	10
237	Genetic heterogeneity in malignant migrating partial seizures of infancy. Annals of Neurology, 2014, 75, 324-326.	2.8	10
238	Spinal motor neuron involvement in a patient with homozygous PRUNE mutation. European Journal of Paediatric Neurology, 2018, 22, 541-543.	0.7	10
239	Migrating focal seizures in Autosomal Dominant Sleep-related Hypermotor Epilepsy with KCNT1 mutation. Seizure: the Journal of the British Epilepsy Association, 2019, 67, 57-60.	0.9	10
240	Complex Neurological Phenotype Associated with a De Novo DHDDS Mutation in a Boy with Intellectual Disability, Refractory Epilepsy, and Movement Disorder. Journal of Pediatric Genetics, 2021, 10, 236-238.	0.3	10
241	Molecular Genetics in Neuroblastoma Prognosis. Children, 2021, 8, 456.	0.6	10
242	Prominent and Regressive Brain Developmental Disorders Associated with Nance-Horan Syndrome. Brain Sciences, 2021, 11, 1150.	1.1	10
243	An integrated approach to the evaluation of patients with asymptomatic or minimally symptomatic <scp>hyperCKemia</scp>. Muscle and Nerve, 2022, 65, 96-104.	1.0	10
244	Structural mapping of GABRB3 variants reveals genotypeâ€“phenotype correlations. Genetics in Medicine, 2022, 24, 681-693.	1.1	10
245	GDAP1 mutation in autosomal recessive Charcot-Marie-Tooth with pyramidal features. Journal of Neurology, 2006, 253, 1234-1235.	1.8	9
246	Topiramate-associated worsening symptoms in a patient with familial hemiplegic migraine. Journal of the Neurological Sciences, 2008, 272, 194-195.	0.3	9
247	Genotype-Phenotype Correlations in a Group of 15 SCN1A-Mutated Italian Patients with GEFS+ Spectrum (Seizures plus, Classical and Borderline Severe Myoclonic Epilepsy of Infancy). Journal of Child Neurology, 2010, 25, 1369-1376.	0.7	9
248	A proofâ€ofâ€concept trial of the whey protein alfaâ€lactalbumin in chronic cortical myoclonus. Movement Disorders, 2011, 26, 2573-2575.	2.2	9
249	Autosomal recessive progressive myoclonus epilepsy due to impaired ceramide synthesis. Epileptic Disorders, 2016, 18, 120-127.	0.7	9
250	Xp11.22 Microduplications Including HUWE1: Case Report and Literature Review. Neuropediatrics, 2016, 47, 051-056.	0.3	9
251	A genomeâ€wide association study of sodium levels and drug metabolism in an epilepsy cohort treated with carbamazepine and oxcarbazepine. Epilepsia Open, 2019, 4, 102-109.	1.3	9
252	Using common genetic variants to find drugs for common epilepsies. Brain Communications, 2021, 3, fcab287.	1.5	9

#	Article	IF	Citations
253	No evidence of ATP1A2 involvement in 12 multiplex Italian families with benign familial infantile seizures. Neuroscience Letters, 2005, 388, 71-74.	1.0	8
254	Chemokine receptor CCR7 is expressed in muscle fibers in juvenile dermatomyositis. Biochemical and Biophysical Research Communications, 2005, 333, 540-543.	1.0	8
255	Alterations in the Î±₂Î´ ligand, thrombospondinâ€1, in a rat model of spontaneous absence epilepsy and in patients with idiopathic/genetic generalized epilepsies. Epilepsia, 2017, 58, 1993-2001.	2.6	8
256	Biallelic Variants in KIF17 Associated with Microphthalmia and Coloboma Spectrum. International Journal of Molecular Sciences, 2021, 22, 4471.	1.8	8
257	L1CAM variants cause two distinct imaging phenotypes on fetal MRI. Annals of Clinical and Translational Neurology, 2021, 8, 2004-2012.	1.7	8
258	Association of ultraâ€rare coding variants with genetic generalized epilepsy: A caseâ€“control whole exome sequencing study. Epilepsia, 2022, 63, 723-735.	2.6	8
259	Familial cortical tremor and epilepsy: A well-defined syndrome with genetic heterogeneity waiting for nosological placement in the ILAE classification. Epilepsy and Behavior, 2010, 19, 669.	0.9	7
260	Hypomyelination and congenital cataract: Identification of novel mutations in two unrelated families. European Journal of Paediatric Neurology, 2013, 17, 108-111.	0.7	7
261	Autosomal recessive epilepsy associated with contactin 2 mutation is different from familial cortical tremor, myoclonus and epilepsy. Brain, 2013, 136, e253-e253.	3.7	7
262	No evidence of a role for cystatin <scp>B</scp> gene in juvenile myoclonic epilepsy. Epilepsia, 2015, 56, e40-3.	2.6	7
263	Application of rare variant transmission disequilibrium tests to epileptic encephalopathy trio sequence data. European Journal of Human Genetics, 2017, 25, 894-899.	1.4	7
264	ARHGEF9 mutations cause a specific recognizable X-linked intellectual disability syndrome. Neurology: Genetics, 2017, 3, e159.	0.9	7
265	Clinical and molecular consequences of exon 78 deletion in DMD gene. Journal of Human Genetics, 2018, 63, 761-764.	1.1	7
266	De novo ARHGEF9 missense variants associated with neurodevelopmental disorder in females: expanding the genotypic and phenotypic spectrum of ARHGEF9 disease in females. Neurogenetics, 2021, 22, 87-94.	0.7	7
267	Exploration of a Putative Susceptibility Locus for Idiopathic Generalized Epilepsy on Chromosomeâ€ƒ8p12. Epilepsia, 2003, 44, 32-39.	2.6	6
268	Do regulatory regions matter in FOXG1 duplications?. European Journal of Human Genetics, 2013, 21, 365-366.	1.4	6
269	Lack of SLC2A1 (Glucose Transporter 1) Mutations in 30 Italian Patients With Alternating Hemiplegia of Childhood. Journal of Child Neurology, 2013, 28, 863-866.	0.7	6
270	Long incubation in imported human rabies. Annals of Neurology, 2014, 75, 324-325.	2.8	6

#	Article	IF	Citations
271	Rare gene deletions in genetic generalized and Rolandic epilepsies. PLoS ONE, 2018, 13, e0202022.	1.1	6
272	Distal motor neuropathy associated with novel EMILIN1 mutation. Neurobiology of Disease, 2020, 137, 104757.	2.1	6
273	Basal Ganglia Dysmorphism in Patients With Aicardi Syndrome. Neurology, 2021, 96, e1319-e1333.	1.5	6
274	Role of Common Genetic Variants for Drug-Resistance to Specific Anti-Seizure Medications. Frontiers in Pharmacology, 2021, 12, 688386.	1.6	6
275	Neuromuscular and Neuroendocrinological Features Associated With ZC4H2-Related Arthrogryposis Multiplex Congenita in a Sicilian Family: A Case Report. Frontiers in Neurology, 2021, 12, 704747.	1.1	6
276	Chitosan may decrease serum valproate and increase the risk of seizure reappearance. BMJ: British Medical Journal, 2009, 339, b3751-b3751.	2.4	6
277	De novo truncating NOVA2 variants affect alternative splicing and lead to heterogeneous neurodevelopmental phenotypes. Human Mutation, 2022, 43, 1299-1313.	1.1	6
278	Genomic Analysis Made It Possible to Identify Gene-Driver Alterations Covering the Time Window between Diagnosis of Neuroblastoma 4S and the Progression to Stage 4. International Journal of Molecular Sciences, 2022, 23, 6513.	1.8	6
279	No evidence for a susceptibility locus for idiopathic generalized epilepsy on chromosome 18q21.1. American Journal of Medical Genetics Part A, 2002, 114, 673-678.	2.4	5
280	No evidence for a susceptibility locus for idiopathic generalized epilepsy on chromosome 5 in families with typical absence seizures. Epilepsy Research, 2002, 51, 23-29.	0.8	5
281	Sudden death in Unverrichtâ€“Lundborg patients: is serotonin the key?. Neurological Sciences, 2010, 31, 115-116.	0.9	5
282	HLA alleles linked to carbamazepine hypersensitivity. Nature Reviews Neurology, 2011, 7, 365-366.	4.9	5
283	Neurological features and long-term follow-up in 15q11.2-13.1 duplication. European Journal of Medical Genetics, 2013, 56, 614-618.	0.7	5
284	17q21.31 microdeletion syndrome: Description of a case further contributing to the delineation of Koolen-de Vries syndrome. Brain and Development, 2016, 38, 663-668.	0.6	5
285	Common and rare epilepsies share genetic determinants. Nature Reviews Neurology, 2017, 13, 200-201.	4.9	5
286	Teaching Neuro Images : Figure of 8. Neurology, 2017, 89, e172-e173.	1.5	5
287	Musculoskeletal Features without Ataxia Associated with a Novel de novo Mutation in KCNA1 Impairing the Voltage Sensitivity of Kv1.1 Channel. Biomedicines, 2021, 9, 75.	1.4	5
288	Hyperkinetic stereotyped movements in a boy with biallelic CNTNAP2 variants. Italian Journal of Pediatrics, 2021, 47, 208.	1.0	5

#	Article	IF	Citations
289	Loss of Neuron Navigator 2 Impairs Brain and Cerebellar Development. Cerebellum, 2022, , 1.	1.4	5
290	Comment to: Overlap cases of eyelid myoclonia with absences and juvenile myoclonic epilepsy. Seizure: the Journal of the British Epilepsy Association, 2007, 16, 557-558.	0.9	4
291	A 'going ape' model for SUDEP?. Nature Reviews Neurology, 2009, 5, 639-640.	4.9	4
292	Genetic epilepsies. European Journal of Paediatric Neurology, 2011, 15, 88-89.	0.7	4
293	Functional characterization of the c.<scp>462delA</scp> mutation in the <scp>NDUFS4</scp> subunit gene of mitochondrial complex I. Clinical Genetics, 2014, 86, 99-101.	1.0	4
294	De novo 12q22.q23.3 duplication associated with temporal lobe epilepsy. Seizure: the Journal of the British Epilepsy Association, 2017, 50, 80-82.	0.9	4
295	Severe early-onset developmental and epileptic encephalopathy (DEE) associated with novel compound heterozygous pathogenic variants in SLC25A22: Case report and literature review. Seizure: the Journal of the British Epilepsy Association, 2019, 70, 56-58.	0.9	4
296	No evidence for a BRD 2 promoter hypermethylation inÂblood leukocytes of Europeans with juvenile myoclonicÂepilepsy. Epilepsia, 2019, 60, e31-e36.	2.6	4
297	A case of Fibrodysplasia Ossificans Progressiva associated with a novel variant of the ACVR1 gene. Molecular Genetics & Genomic Medicine, 2021, 9, e1774.	0.6	4
298	Targeting of Ubiquitin E3 Ligase RNF5 as a Novel Therapeutic Strategy in Neuroectodermal Tumors. Cancers, 2022, 14, 1802.	1.7	4
299	A de novo 11p12-p15.4 duplication in a patient with pharmacoresistant epilepsy, mental retardation, and dysmorphisms. Brain and Development, 2010, 32, 248-252.	0.6	3
300	Suicide-Related Events in Patients Treated with Antiepileptic Drugs. New England Journal of Medicine, 2010, 363, 1873-1874.	13.9	3
301	RNF213 variant in a patient with Legius syndrome associated with moyamoya syndrome. Molecular Genetics & Genomic Medicine, 2021, 9, e1669.	0.6	3
302	Cortical and Subcortical Network Dysfunction in a Female Patient With NEXMIF Encephalopathy. Frontiers in Neurology, 2021, 12, 722664.	1.1	3
303	Ocular phenotype and electroretinogram abnormalities in Lafora disease and correlation with disease stage. Journal of Neurology, 2022, 269, 3597-3604.	1.8	3
304	A Phenotypic-Driven Approach for the Diagnosis of WOREE Syndrome. Frontiers in Pediatrics, 2022, 10, 847549.	0.9	3
305	Familial nonkinesigenic paroxysmal dyskinesia and intracranial calcifications: A new syndrome?. Movement Disorders, 2010, 25, 2468-2470.	2.2	2
306	Pathophysiology of Myoclonic Seizures. , 2010, , 235-242.		2

#	Article	IF	Citations
307	LESIONAL REFLEX EPILEPSY ASSOCIATED WITH THE THOUGHT OF FOOD. Neurology, 2010, 75, 288-289.	1.5	2
308	Epidemiology and familial clustering of pediatric epilepsy in the geographic isolate of Ischia. Epilepsy Research, 2019, 154, 86-89.	0.8	2
309	Temporalâ€parietalâ€occipital epilepsy in GEFS+ associated with SCN1A mutation. Epileptic Disorders, 2021, 23, 397-401.	0.7	2
310	New phenotype caused by POMGNT2 mutations. BMJ Case Reports, 2021, 14, e242358.	0.2	2
311	Autosomal Dominant Cortical Myoclonus and Epilepsy. , 2010, , 1051-1054.		2
312	Paroxysmal limb dystonias associated with GABBR2 pathogenic variant: A case-based literature review. Brain and Development, 2022, , .	0.6	2
313	Hemidystonia in Uncontrolled Type 2 Diabetes Mellitus. Archives of Neurology, 2011, 68, 674.	4.9	1
314	Importance of post-mortem genetic testing in SUDEP patients. Forensic Science International: Genetics Supplement Series, 2013, 4, e354-e355.	0.1	1
315	Pelizaeusâ€“Merzbacher Disease due to PLP1 Frameshift Mutation in a Female with Nonrandom Skewed X-Chromosome Inactivation. Neuropediatrics, 2019, 50, 268-270.	0.3	1
316	Dual diagnosis in a child with familial SCN8A-related encephalopathy complicated by a 1p13.2 deletion involving NRAS gene. Neurological Sciences, 2021, 42, 2115-2117.	0.9	1
317	Reply to â€œEpilepsies in childrenâ€”the power of making a syndrome diagnosisâ€• Nature Clinical Practice Neurology, 2008, 4, E3-E3.	2.7	1
318	Genetic Counselling in Epilepsy. , 0, , 341-360.		1
319	High Grade of Amplification of Six Regions on Chromosome 2p in a Neuroblastoma Patient with Very Poor Outcome: The Putative New Oncogene TSSC1. Cancers, 2021, 13, 5792.	1.7	1
320	Response to: 'Cortical tremor or cortical pseudotremor?'. Acta Neurologica Scandinavica, 2005, 112, 204-204.	1.0	0
321	Transient epileptic amnesia: a new epileptic syndrome in development?. Annals of Neurology, 2010, 67, 416-416.	2.8	0
322	Similar but not identical: Clinical implications for molecular studies in monozygotic discordant twins with epilepsy. Epilepsy and Behavior, 2011, 20, 419.	0.9	0
323	Novel treatment perspectives from advances in understanding of genetic epilepsy syndromes. Expert Opinion on Orphan Drugs, 2016, 4, 485-490.	0.5	0
324	Confirmation of mutations in the PROSC gene as a novel cause of vitamin B6 dependent epilepsy. European Journal of Paediatric Neurology, 2017, 21, e1.	0.7	0

#	Article	IF	Citations
325	Erratum to â€œDe novo 12q22.q23.3 duplication associated with temporal lobe epilepsyâ€•[Seizure 57 (2018) 63â€“65]. Seizure: the Journal of the British Epilepsy Association, 2018, 57, R1.	0.9	0
326	De novo 12q22.q23.3 duplication associated with temporal lobe epilepsy. Seizure: the Journal of the British Epilepsy Association, 2018, 57, 63-65.	0.9	0
327	The first case of mosaic MNX1 mutation in an adult female with features of Currarino syndrome. Birth Defects Research, 2021, 113, 1161-1165.	0.8	0
328	Reply to Braun etÂal. â€œNovel bathing epilepsy in a patient with 2q22.3q23.2 deletionâ€• Seizure: the Journal of the British Epilepsy Association, 2021, 91, 112-113.	0.9	0