Pasquale Striano

List of Publications by Year
in descending order

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618 papers	21,526 citations	15504 65 h-index	24258 110 g-index
645 all docs	645 docs citations	645 times ranked	22305 citing authors

#	Article	IF	Citations
1	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	12.6	1,085
2	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	27.0	663
3	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. Brain, 2017, 140, 1316-1336.	7.6	426
4	De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	6.2	388
5	Structural brain abnormalities in the common epilepsies assessed in a worldwide ENIGMA study. Brain, 2018, 141, 391-408.	7.6	352
6	Genetic determinants of common epilepsies: a meta-analysis of genome-wide association studies. Lancet Neurology, The, 2014, 13, 893-903.	10.2	264
7	STXBP1 encephalopathy. Neurology, 2016, 86, 954-962.	1.1	264
8	Mutations in XPR1 cause primary familial brain calcification associated with altered phosphate export. Nature Genetics, 2015, 47, 579-581.	21.4	237
9	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
10	De novo variants in neurodevelopmental disorders with epilepsy. Nature Genetics, 2018, 50, 1048-1053.	21.4	230
11	Extending the KCNQ2 encephalopathy spectrum. Neurology, 2013, 81, 1697-1703.	1.1	198
12	De novo mutations in HCN1 cause early infantile epileptic encephalopathy. Nature Genetics, 2014, 46, 640-645.	21.4	192
13	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
14	Posterior reversible encephalopathy syndrome in intensive care medicine. Intensive Care Medicine, 2007, 33, 230-236.	8.2	179
15	Characterization of a recurrent 15q24 microdeletion syndrome. Human Molecular Genetics, 2007, 16, 567-572.	2.9	173
16	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. American Journal of Human Genetics, 2015, 96, 808-815.	6.2	173
17	Epilepsy, hippocampal sclerosis and febrile seizures linked by common genetic variation around SCN1A. Brain, 2013, 136, 3140-3150.	7.6	168
18	Relationship between adverse effects of antiepileptic drugs, number of coprescribed drugs, and drug load in a large cohort of consecutive patients with drugâ€refractory epilepsy. Epilepsia, 2010, 51, 797-804.	5.1	160

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19	Infantile spasms syndrome, West syndrome and related phenotypes: What we know in 2013. Brain and Development, 2014, 36, 739-751.	1.1	159
20	Eyelid myoclonia with absences (Jeavons syndrome): A wellâ€defined idiopathic generalized epilepsy syndrome or a spectrum of photosensitive conditions?. Epilepsia, 2009, 50, 15-19.	5.1	156
21	LGI1mutations in autosomal dominant and sporadic lateral temporal epilepsy. Human Mutation, 2009, 30, 530-536.	2.5	155
22	SCN1A duplications and deletions detected in Dravet syndrome: Implications for molecular diagnosis. Epilepsia, 2009, 50, 1670-1678.	5.1	152
23	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
24	Consensus on diagnosis and management of JME: From founder's observations to current trends. Epilepsy and Behavior, 2013, 28, S87-S90.	1.7	142
25	The landscape of epilepsy-related GATOR1 variants. Genetics in Medicine, 2019, 21, 398-408.	2.4	137
26	TBC1D24, an ARF6-Interacting Protein, Is Mutated in Familial Infantile Myoclonic Epilepsy. American Journal of Human Genetics, 2010, 87, 365-370.	6.2	134
27	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
28	KCNQ2 encephalopathy: Delineation of the electroclinical phenotype and treatment response. Neurology, 2014, 82, 368-370.	1.1	130
29	Expert Opinion on the Management of Lennoxâ€“Gastaut Syndrome: Treatment Algorithms and Practical Considerations. Frontiers in Neurology, 2017, 8, 505.	2.4	129
30	Lafora disease. Epileptic Disorders, 2016, 18, 38-62.	1.3	127
31	White matter abnormalities across different epilepsy syndromes in adults: an ENIGMA-Epilepsy study. Brain, 2020, 143, 2454-2473.	7.6	123
32	Epilepsy in cerebrovascular diseases: Review of experimental and clinical data with metaâ€analysis of risk factors. Epilepsia, 2016, 57, 1205-1214.	5.1	122
33	Posterior reversible encephalopathy syndrome (PRES) in critically ill obstetric patients. Intensive Care Medicine, 2003, 29, 2323-2326.	8.2	118
34	Metabolic and endocrine effects of valproic acid chronic treatment. Epilepsy Research, 2013, 107, 1-8.	1.6	118
35	Mutations in <scp>KCNT</scp>1 cause a spectrum of focal epilepsies. Epilepsia, 2015, 56, e114-20.	5.1	117
36	Microbiota-gut brain axis involvement in neuropsychiatric disorders. Expert Review of Neurotherapeutics, 2019, 19, 1037-1050.	2.8	116

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37	Phenotypic spectrum of GABRA1. Neurology, 2016, 87, 1140-1151.	1.1	113
38	Epidemiology of juvenile myoclonic epilepsy. Epilepsy and Behavior, 2013, 28, S15-S17.	1.7	111
39	The ACMSD gene, involved in tryptophan metabolism, is mutated in a family with cortical myoclonus, epilepsy, and parkinsonism. Journal of Molecular Medicine, 2013, 91, 1399-1406.	3.9	111
40	The Pharmacoresistant Epilepsy: An Overview on Existant and New Emerging Therapies. Frontiers in Neurology, 2021, 12, 674483.	2.4	111
41	Genetic testing in benign familial epilepsies of the first year of life: Clinical and diagnostic significance. Epilepsia, 2013, 54, 425-436.	5.1	110
42	Somatic and germline mosaicisms in Severe Myoclonic Epilepsy of Infancy. Biochemical and Biophysical Research Communications, 2006, 341, 489-493.	2.1	102
43	Defining the phenotypic spectrum of SLC6A1 mutations. Epilepsia, 2018, 59, 389-402.	5.1	99
44	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	12.8	99
45	TBC1D24 genotypeâ€“phenotype correlation. Neurology, 2016, 87, 77-85.	1.1	97
46	Neurologic phenotypes associated with COL4A1 / 2 mutations. Neurology, 2018, 91, e2078-e2088.	1.1	97
47	Network-based atrophy modeling in the common epilepsies: A worldwide ENIGMA study. Science Advances, 2020, 6, .	10.3	97
48	HCN1mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. Brain, 2018, 141, 3160-3178.	7.6	96
49	Heterozygous Reelin Mutations Cause Autosomal-Dominant Lateral Temporal Epilepsy. American Journal of Human Genetics, 2015, 96, 992-1000.	6.2	94
50	Migralepsy, hemicrania epileptica, post-ictal headache and â€œictal epileptic headacheâ€: a proposal for terminology and classification revision. Journal of Headache and Pain, 2011, 12, 289-294.	6.0	93
51	PRRT2 Mutations are the major cause of benign familial infantile seizures. Human Mutation, 2012, 33, 1439-1443.	2.5	93
52	Coexistence of epilepsy and Brugada syndrome in a family with SCN5A mutation. Epilepsy Research, 2013, 105, 415-418.	1.6	90
53	Brain MRI Findings in Severe Myoclonic Epilepsy in Infancy and Genotype?Phenotype Correlations. Epilepsia, 2007, 48, 1092-1096.	5.1	89
54	A pilot trial of levetiracetam in eyelid myoclonia with absences (Jeavons syndrome). Epilepsia, 2008, 49, 425-430.	5.1	88

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55	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
56	Eyelid myoclonia with absences: an overlooked epileptic syndrome?. Neurophysiologie Clinique, 2002, 32, 287-296.	2.2	87
57	Progressive myoclonic epilepsies. Neurology, 2014, 82, 405-411.	1.1	87
58	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. Brain, 2013, 136, 3378-3394.	7.6	85
59	Hyperhomocysteinemia in epileptic patients on new antiepileptic drugs. Epilepsia, 2010, 51, 274-279.	5.1	84
60	Coâ€occurring malformations of cortical development and <scp>SCN</scp>1A gene mutations. Epilepsia, 2014, 55, 1009-1019.	5.1	84
61	Gelastic Epilepsy: Symptomatic and Cryptogenic Cases. Epilepsia, 1999, 40, 294-302.	5.1	80
62	Impairment of ceramide synthesis causes a novel progressive myoclonus epilepsy. Annals of Neurology, 2014, 76, 206-212.	5.3	80
63	â€˜Ictal epileptic headacheâ€™: Recent concepts for new classifications criteria. Cephalalgia, 2012, 32, 723-724.	3.9	79
64	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
65	A Novel SCN2A Mutation in Family with Benign Familial Infantile Seizures. Epilepsia, 2006, 47, 218-220.	5.1	74
66	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
67	Clinical and Genetic Findings in 26 Italian Patients with Lafora Disease. Epilepsia, 2006, 47, 640-643.	5.1	71
68	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
69	The spectrum of intermediate <scp>SCN</scp>8Aâ€related epilepsy. Epilepsia, 2019, 60, 830-844.	5.1	70
70	Late-onset and Slow-progressing Lafora Disease in Four Siblings with EPM2B Mutation. Epilepsia, 2005, 46, 1695-1697.	5.1	69
71	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. Brain, 2022, 145, 2991-3009.	7.6	69
72	Pitfalls in genetic testing: the story of missed SCN1A mutations. Molecular Genetics & Genomic Medicine, 2016, 4, 457-464.	1.2	67

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73	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
74	Levetiracetam in patients with cortical myoclonus: A clinical and electrophysiological study. Movement Disorders, 2005, 20, 1610-1614.	3.9	66
75	A novel KCNQ3 mutation in familial epilepsy with focal seizures and intellectual disability. Epilepsia, 2015, 56, e15-20.	5.1	66
76	Confirmation of mutations inPROSCas a novel cause of vitamin B_₆-dependent epilepsy. Journal of Medical Genetics, 2017, 54, 809-814.	3.2	66
77	Recent advances in epilepsy genetics. Neuroscience Letters, 2018, 667, 4-9.	2.1	66
78	Cannabidiol efficacy and clobazam status: A systematic review and metaâ€analysis. Epilepsia, 2020, 61, 1090-1098.	5.1	66
79	Idiopathic focal epilepsies: the â€œlost tribeâ€• Epileptic Disorders, 2016, 18, 252-288.	1.3	65
80	Reflex seizures and reflex epilepsies: Old models for understanding mechanisms of epileptogenesis. Epilepsy Research, 2012, 100, 1-11.	1.6	64
81	Mutation inCPT1CAssociated With Pure Autosomal Dominant Spastic Paraplegia. JAMA Neurology, 2015, 72, 561.	9.0	64
82	PRRT2-related disorders: further PKD and ICCA cases and review of the literature. Journal of Neurology, 2013, 260, 1234-1244.	3.6	63
83	The epileptic and nonepileptic spectrum of paroxysmal dyskinesias: Channelopathies, synaptopathies, and transportopathies. Movement Disorders, 2017, 32, 310-318.	3.9	63
84	A New Benign Adult Familial Myoclonic Epilepsy (BAFME) Pedigree Suggesting Linkage to Chromosome 2p11.1-q12.2. Epilepsia, 2004, 45, 190-192.	5.1	62
85	The clinical spectrum and natural history of gelastic epilepsy-hypothalamic hamartoma syndrome. Seizure: the Journal of the British Epilepsy Association, 2005, 14, 232-239.	2.0	62
86	Biallelic Mutations in ADPRHL2, Encoding ADP-Ribosylhydrolase 3, Lead to a Degenerative Pediatric Stress-Induced Epileptic Ataxia Syndrome. American Journal of Human Genetics, 2018, 103, 431-439.	6.2	62
87	From Genetic Testing to Precision Medicine in Epilepsy. Neurotherapeutics, 2020, 17, 609-615.	4.4	62
88	Autosomal dominant cortical tremor, myoclonus and epilepsy: many syndromes, one phenotype. Acta Neurologica Scandinavica, 2005, 111, 211-217.	2.1	61
89	Migralepsy and related conditions: Advances in pathophysiology and classification. Seizure: the Journal of the British Epilepsy Association, 2011, 20, 271-275.	2.0	61
90	Clinical Significance of Rare Copy Number Variations in Epilepsy. Archives of Neurology, 2012, 69, 322.	4.5	61

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91	Effectiveness of antiepileptic therapy in patients with PCDH19 mutations. Seizure: the Journal of the British Epilepsy Association, 2016, 35, 106-110.	2.0	61
92	Characterisation of CASPR2 deficiency disorder - a syndrome involving autism, epilepsy and language impairment. BMC Medical Genetics, 2016, 17, 8.	2.1	61
93	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	5.1	61
94	Long-term evolution of EEG in Unverricht-Lundborg disease. Epilepsy Research, 2007, 73, 219-227.	1.6	60
95	Familial mesial temporal lobe epilepsy (FMTLE). Journal of Neurology, 2008, 255, 16-23.	3.6	60
96	Lacosamide in pediatric and adult patients: Comparison of efficacy and safety. Seizure: the Journal of the British Epilepsy Association, 2013, 22, 210-216.	2.0	60
97	Genetics of reflex seizures and epilepsies in humans and animals. Epilepsy Research, 2016, 121, 47-54.	1.6	60
98	Can we â€˜seizeâ€™ the gut microbiota to treat epilepsy?. Neuroscience and Biobehavioral Reviews, 2019, 107, 750-764.	6.1	60
99	Challenges and management of neurological and psychiatric manifestations in SARS-CoV-2 (COVID-19) patients. Neurological Sciences, 2020, 41, 2353-2366.	1.9	60
100	The gelastic seizures-hypothalamic hamartoma syndrome: Facts, hypotheses, and perspectives. Epilepsy and Behavior, 2012, 24, 7-13.	1.7	59
101	¹Hâ€MR spectroscopy indicates prominent cerebellar dysfunction in benign adult familial myoclonic epilepsy. Epilepsia, 2009, 50, 1491-1497.	5.1	58
102	Further evidence of the association between LQT syndrome and epilepsy in a family with KCNQ1 pathogenic variant. Seizure: the Journal of the British Epilepsy Association, 2015, 25, 65-67.	2.0	58
103	Lateralizing Value of the Auditory Aura in Partial Seizures. Epilepsia, 2006, 47, 68-72.	5.1	57
104	Efficacy of sodium channel blockers in SCN2A early infantile epileptic encephalopathy. Brain and Development, 2017, 39, 345-348.	1.1	57
105	The best evidence for progressive myoclonic epilepsy: A pathway to precision therapy. Seizure: the Journal of the British Epilepsy Association, 2019, 71, 247-257.	2.0	57
106	Adjunctive Cannabidiol in Patients with Dravet Syndrome: A Systematic Review and Meta-Analysis of Efficacy and Safety. CNS Drugs, 2020, 34, 229-241.	5.9	57
107	Levetiracetam for cerebellar tremor in multiple sclerosis. Journal of Neurology, 2006, 253, 762-766.	3.6	56
108	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

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109	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
110	Progressive Myoclonus Epilepsy: The Geneâ€Empowered Era. Epileptic Disorders, 2016, 18, 1-2.	1.3	55
111	PDXK mutations cause polyneuropathy responsive to pyridoxal 5â€²â€phosphate supplementation. Annals of Neurology, 2019, 86, 225-240.	5.3	54
112	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
113	Ictal epileptic headache: an old story with courses and appeals. Journal of Headache and Pain, 2012, 13, 607-613.	6.0	52
114	Management of genetic epilepsies: From empirical treatment to precision medicine. Pharmacological Research, 2016, 107, 426-429.	7.1	52
115	The pharmacological management of Lennox-Gastaut syndrome and critical literature review. Seizure: the Journal of the British Epilepsy Association, 2018, 63, 17-25.	2.0	52
116	No major role for the EMX2 gene in schizencephaly. American Journal of Medical Genetics, Part A, 2008, 146A, 1142-1150.	1.2	51
117	Intestinal inflammation increases convulsant activity and reduces antiepileptic drug efficacy in a mouse model of epilepsy. Scientific Reports, 2019, 9, 13983.	3.3	51
118	Hypertension, seizures, and epilepsy: a review on pathophysiology and management. Neurological Sciences, 2019, 40, 1775-1783.	1.9	51
119	Highly Purified Cannabidiol for Epilepsy Treatment: A Systematic Review of Epileptic Conditions Beyond Dravet Syndrome and Lennoxâ€“Gastaut Syndrome. CNS Drugs, 2021, 35, 265-281.	5.9	51
120	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
121	Clinical spectrum and critical care management of Posterior Reversible Encephalopathy Syndrome (PRES). Medical Science Monitor, 2005, 11, CR549-53.	1.1	50
122	Neuroimaging and neuropathology of Dravet syndrome. Epilepsia, 2011, 52, 30-34.	5.1	49
123	Antiepileptic drugs, hyperhomocysteinemia and B-vitamins supplementation in patients with epilepsy. Epilepsy Research, 2012, 102, 1-7.	1.6	49
124	Genetic investigation of sudden unexpected death in epilepsy cohort by panel target resequencing. International Journal of Legal Medicine, 2016, 130, 331-339.	2.2	49
125	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
126	Eyelid fluttering, typical EEG pattern, and impaired intellectual function: A homogeneous epileptic condition among the patients presenting with eyelid myoclonia. Epilepsia, 2009, 50, 1536-1541.	5.1	48

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127	The syndrome gelastic seizuresâ€“hypothalamic hamartoma: Severe, potentially reversible encephalopathy. Epilepsia, 2009, 50, 62-65.	5.1	48
128	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. American Journal of Human Genetics, 2019, 105, 689-705.	6.2	48
129	Relationship between serum mono-hydroxy-carbazepine concentrations and adverse effects in patients with epilepsy on high-dose oxcarbazepine therapy. Epilepsy Research, 2006, 69, 170-176.	1.6	47
130	Typical progression of myoclonic epilepsy of the Lafora type: a case report. Nature Clinical Practice Neurology, 2008, 4, 106-111.	2.5	47
131	What have we learned about ictal epileptic headache? A review of well-documented cases. Seizure: the Journal of the British Epilepsy Association, 2013, 22, 253-258.	2.0	47
132	De novo mutations ofKIAA2022in females cause intellectual disability and intractable epilepsy. Journal of Medical Genetics, 2016, 53, 850-858.	3.2	47
133	The <scp>ENIGMAâ€Epilepsy</scp> working group: Mapping disease from large data sets. Human Brain Mapping, 2022, 43, 113-128.	3.6	47
134	Bi-allelic JAM2 Variants Lead to Early-Onset Recessive Primary Familial Brain Calcification. American Journal of Human Genetics, 2020, 106, 412-421.	6.2	47
135	Consensus guidelines for the diagnosis and management of pyridoxineâ€dependent epilepsy due to Î±â€aminoadipic semialdehyde dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 178-192.	3.6	47
136	Third-Generation Antiseizure Medications for Adjunctive Treatment of Focal-Onset Seizures in Adults: A Systematic Review and Network Meta-analysis. Drugs, 2022, 82, 199-218.	10.9	47
137	The genetics of monogenic idiopathic epilepsies and epileptic encephalopathies. Seizure: the Journal of the British Epilepsy Association, 2012, 21, 3-11.	2.0	46
138	Homozygous STXBP1 variant causes encephalopathy and gain-of-function in synaptic transmission. Brain, 2020, 143, 441-451.	7.6	46
139	Assessing the landscape of STXBP1-related disorders in 534 individuals. Brain, 2022, 145, 1668-1683.	7.6	46
140	Temporal Lobe Epilepsy and Psychiatric Comorbidity. Frontiers in Neurology, 2021, 12, 775781.	2.4	46
141	Antiepileptic Drugs and MTHFR Polymorphisms Influence Hyper-Homocysteinemia Recurrence in Epileptic Patients. Epilepsia, 2007, 48, 1990-1994.	5.1	45
142	Natural history and long-term evolution in families with autosomal dominant cortical tremor, myoclonus, and epilepsy. Epilepsia, 2011, 52, 1245-1250.	5.1	45
143	Dramatic effect of levetiracetam in early-onset epileptic encephalopathy due to STXBP1 mutation. Brain and Development, 2016, 38, 128-131.	1.1	45
144	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

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145	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
146	Mutational Analysis of EFHC1 Gene in Italian Families with Juvenile Myoclonic Epilepsy. Epilepsia, 2007, 48, 1686-1690.	5.1	44
147	A Distinctive Ictal Amplitude-Integrated Electroencephalography Pattern in Newborns with Neonatal Epilepsy Associated with <i>KCNQ2</i> Mutations. Neonatology, 2017, 112, 387-393.	2.0	44
148	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
149	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
150	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
151	Rescuable folding defective NaV1.1 (SCN1A) mutants in epilepsy: Properties, occurrence, and novel rescuing strategy with peptides targeted to the endoplasmic reticulum. Neurobiology of Disease, 2015, 75, 100-114.	4.4	43
152	Diagnostic criteria currently proposed for â€œictal epileptic headacheâ€• Perspectives on strengths, weaknesses and pitfalls. Seizure: the Journal of the British Epilepsy Association, 2015, 31, 56-63.	2.0	43
153	Genetic variation in CFH predicts phenytoin-induced maculopapular exanthema in European-descent patients. Neurology, 2018, 90, e332-e341.	1.1	43
154	The Î±_{2B}â€adrenergic receptor is mutant in cortical myoclonus and epilepsy. Annals of Neurology, 2014, 75, 77-87.	5.3	42
155	Revelation of a Novel CLN5 Mutation in Early Juvenile Neuronal Ceroid Lipofuscinosis. Neuropediatrics, 2007, 38, 46-49.	0.6	41
156	Life-Threatening Status Epilepticus Following Gabapentin Administration in a Patient with Benign Adult Familial Myoclonic Epilepsy. Epilepsia, 2007, 48, 1995-1998.	5.1	41
157	Intravenous lacosamide as treatment option in post-stroke non convulsive status epilepticus in the elderly: A proof-of-concept, observational study. Seizure: the Journal of the British Epilepsy Association, 2013, 22, 905-907.	2.0	41
158	Muscle and not neuronal biomarkers correlate with severity in spinal and bulbar muscular atrophy. Neurology, 2019, 92, e1205-e1211.	1.1	41
159	Adjunctive Cenobamate for Focal-Onset Seizures in Adults: A Systematic Review and Meta-Analysis. CNS Drugs, 2020, 34, 1105-1120.	5.9	41
160	Progressive myoclonus epilepsiesâ€”Residual unsolved cases have marked genetic heterogeneity including dolichol-dependent protein glycosylation pathway genes. American Journal of Human Genetics, 2021, 108, 722-738.	6.2	41
161	Gabapentin: A Ca2+ channel alpha2-delta ligand far beyond epilepsy therapy. Drugs of Today, 2008, 44, 353.	1.1	41
162	Lennoxâ€Gastaut syndrome with lateâ€onset and prominent reflex seizures in trisomy 21 patients. Epilepsia, 2009, 50, 1587-1595.	5.1	40

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163	Treatment and outcome of children with cerebral cavernomas: a survey on 32 patients. Neurological Sciences, 2010, 31, 117-123.	1.9	40
164	Genetic and forensic implications in epilepsy and cardiac arrhythmias: a case series. International Journal of Legal Medicine, 2015, 129, 495-504.	2.2	40
165	Faciobrachial dystonic attacks: Seizures or movement disorder?. Annals of Neurology, 2011, 70, 179-180.	5.3	39
166	Defining the electroclinical phenotype and outcome of PCDH19â€related epilepsy: A multicenter study. Epilepsia, 2018, 59, 2260-2271.	5.1	39
167	SLC35A2â€CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	2.5	39
168	Genetic heterogeneity in infantile spasms. Epilepsy Research, 2019, 156, 106181.	1.6	38
169	Ictal Epileptic Headache Mimicking Status Migrainosus: EEG and DWIâ€MRI Findings. Headache, 2011, 51, 160-162.	3.9	37
170	<scp>DEPDC</scp>5 mutations are not a frequent cause of familial temporal lobe epilepsy. Epilepsia, 2015, 56, e168-71.	5.1	37
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