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	Assessing the landscape of STXBP1-related disorders in 534 individuals. Brain, 2022, 145, 1668-1683.	7.6	46
2	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. Brain, 2022, 145, 2991-3009.	7.6	69
3	An integrated approach to the evaluation of patients with asymptomatic or minimally symptomatic <scp>hyperCKemia</scp>. Muscle and Nerve, 2022, 65, 96-104.	2.2	10
4	Association of ultraâ€rare coding variants with genetic generalized epilepsy: A caseâ€“control whole exome sequencing study. Epilepsia, 2022, 63, 723-735.	5.1	8
5	Loss of Neuron Navigator 2 Impairs Brain and Cerebellar Development. Cerebellum, 2022, , 1.	2.5	5
6	Ocular phenotype and electroretinogram abnormalities in Lafora disease and correlation with disease stage. Journal of Neurology, 2022, 269, 3597-3604.	3.6	3
7	Structural mapping of GABRB3 variants reveals genotypeâ€“phenotype correlations. Genetics in Medicine, 2022, 24, 681-693.	2.4	10
8	Spectrum of Phenotypic, Genetic, and Functional Characteristics in Patients With Epilepsy With KCNC2 Pathogenic Variants. Neurology, 2022, 98, .	1.1	11
9	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.	4.1	19
10	Targeting of Ubiquitin E3 Ligase RNF5 as a Novel Therapeutic Strategy in Neuroectodermal Tumors. Cancers, 2022, 14, 1802.	3.7	4
11	A pharmacogenomic assessment of psychiatric adverse drug reactions to levetiracetam. Epilepsia, 2022, 63, 1563-1570.	5.1	11
12	Paroxysmal limb dystonias associated with GABBR2 pathogenic variant: A case-based literature review. Brain and Development, 2022, , .	1.1	2
13	A Phenotypic-Driven Approach for the Diagnosis of WOREE Syndrome. Frontiers in Pediatrics, 2022, 10, 847549.	1.9	3
14	Epilepsy Course and Developmental Trajectories in STXBP1 -DEE. Neurology: Genetics, 2022, 8, .	1.9	24
15	De novo truncating NOVA2 variants affect alternative splicing and lead to heterogeneous neurodevelopmental phenotypes. Human Mutation, 2022, 43, 1299-1313.	2.5	6
16	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.	4.1	6
17	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.	0.7	10
18	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.	1.4	7

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19	Basal Ganglia Dysmorphism in Patients With Aicardi Syndrome. Neurology, 2021, 96, e1319-e1333.	1.1	6
20	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.	1.9	1
21	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.	3.2	5
22	Climate change and epilepsy: Insights from clinical and basic science studies. Epilepsy and Behavior, 2021, 116, 107791.	1.7	30
23	Biallelic Variants in KIF17 Associated with Microphthalmia and Coloboma Spectrum. International Journal of Molecular Sciences, 2021, 22, 4471.	4.1	8
24	Genotype-Phenotype Correlations in Neurofibromatosis Type 1: A Single-Center Cohort Study. Cancers, 2021, 13, 1879.	3.7	21
25	Temporalâ€parietalâ€occipital epilepsy in GEFS+ associated with SCN1A mutation. Epileptic Disorders, 2021, 23, 397-401.	1.3	2
26	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
27	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.	4.1	19
28	Italian cohort of Lafora disease: Clinical features, disease evolution, and genotype-phenotype correlations. Journal of the Neurological Sciences, 2021, 424, 117409.	0.6	11
29	Assessing the role of rare genetic variants in drugâ€resistant, nonâ€lesional focal epilepsy. Annals of Clinical and Translational Neurology, 2021, 8, 1376-1387.	3.7	16
30	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
31	Molecular Genetics in Neuroblastoma Prognosis. Children, 2021, 8, 456.	1.5	10
32	Partial Rescue of F508del-CFTR Stability and Trafficking Defects by Double Corrector Treatment. International Journal of Molecular Sciences, 2021, 22, 5262.	4.1	45
33	RNF213 variant in a patient with Legius syndrome associated with moyamoya syndrome. Molecular Genetics & Genomic Medicine, 2021, 9, e1669.	1.2	3
34	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
35	Clinical and Genetic Features in Patients With Reflex Bathing Epilepsy. Neurology, 2021, 97, e577-e586.	1.1	11
36	KCNT1-related epilepsies and epileptic encephalopathies: phenotypic and mutational spectrum. Brain, 2021, 144, 3635-3650.	7.6	34

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37	Role of Common Genetic Variants for Drug-Resistance to Specific Anti-Seizure Medications. Frontiers in Pharmacology, 2021, 12, 688386.	3.5	6
38	The first case of mosaic MNX1 mutation in an adult female with features of Currarino syndrome. Birth Defects Research, 2021, 113, 1161-1165.	1.5	0
39	Neuromuscular and Neuroendocrinological Features Associated With ZC4H2-Related Arthrogryposis Multiplex Congenita in a Sicilian Family: A Case Report. Frontiers in Neurology, 2021, 12, 704747.	2.4	6
40	New phenotype caused by POMGNT2 mutations. BMJ Case Reports, 2021, 14, e242358.	0.5	2
41	Biallelic PI4KA variants cause neurological, intestinal and immunological disease. Brain, 2021, 144, 3597-3610.	7.6	17
42	A case of Fibrodysplasia Ossificans Progressiva associated with a novel variant of the ACVR1 gene. Molecular Genetics & Genomic Medicine, 2021, 9, e1774.	1.2	4
43	Prominent and Regressive Brain Developmental Disorders Associated with Nance-Horan Syndrome. Brain Sciences, 2021, 11, 1150.	2.3	10
44	Cortical and Subcortical Network Dysfunction in a Female Patient With NEXMIF Encephalopathy. Frontiers in Neurology, 2021, 12, 722664.	2.4	3
45	L1CAM variants cause two distinct imaging phenotypes on fetal MRI. Annals of Clinical and Translational Neurology, 2021, 8, 2004-2012.	3.7	8
46	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.	2.0	0
47	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.	6.2	11
48	Hyperkinetic stereotyped movements in a boy with biallelic CNTNAP2 variants. Italian Journal of Pediatrics, 2021, 47, 208.	2.6	5
49	Using common genetic variants to find drugs for common epilepsies. Brain Communications, 2021, 3, fcab287.	3.3	9
50	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.	3.7	1
51	Progressive Myoclonus Epilepsies. Neurology: Genetics, 2021, 7, e641.	1.9	20
52	Diagnostic Approach to Macrocephaly in Children. Frontiers in Pediatrics, 2021, 9, 794069.	1.9	17
53	Homozygous STXBP1 variant causes encephalopathy and gain-of-function in synaptic transmission. Brain, 2020, 143, 441-451.	7.6	46
54	Targeting Alternative Splicing as a Potential Therapy for Episodic Ataxia Type 2. Biomedicines, 2020, 8, 332.	3.2	13

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55	Brain Organoids as Model Systems for Genetic Neurodevelopmental Disorders. Frontiers in Cell and Developmental Biology, 2020, 8, 590119.	3.7	31
56	Pharmacoresponse in genetic generalized epilepsy: a genome-wide association study. Pharmacogenomics, 2020, 21, 325-335.	1.3	21
57	Biallelic MFSD2A variants associated with congenital microcephaly, developmental delay, and recognizable neuroimaging features. European Journal of Human Genetics, 2020, 28, 1509-1519.	2.8	21
58	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	7.6	47
59	Targeted re-sequencing in malformations of cortical development: genotype-phenotype correlations. Seizure: the Journal of the British Epilepsy Association, 2020, 80, 145-152.	2.0	13
60	Testing association of rare genetic variants with resistance to three common antiseizure medications. Epilepsia, 2020, 61, 657-666.	5.1	22
61	Emerging treatments for progressive myoclonus epilepsies. Expert Review of Neurotherapeutics, 2020, 20, 341-350.	2.8	11
62	Loss of Wwox Perturbs Neuronal Migration and Impairs Early Cortical Development. Frontiers in Neuroscience, 2020, 14, 644.	2.8	22
63	Clinical spectrum and genotype-phenotype correlations in PRRT2 Italian patients. European Journal of Paediatric Neurology, 2020, 28, 193-197.	1.6	14
64	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
65	Distal motor neuropathy associated with novel EMILIN1 mutation. Neurobiology of Disease, 2020, 137, 104757.	4.4	6
66	Progress of Induced Pluripotent Stem Cell Technologies to Understand Genetic Epilepsy. International Journal of Molecular Sciences, 2020, 21, 482.	4.1	11
67	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.	2.0	15
68	Genotype-phenotype correlations in patients with de novo KCNQ2 pathogenic variants. Neurology: Genetics, 2020, 6, e528.	1.9	24
69	Chiari malformation type I: what information from the genetics?. Child's Nervous System, 2019, 35, 1665-1671.	1.1	12
70	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
71	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
72	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.	2.0	4

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73	Microbiota-gut brain axis involvement in neuropsychiatric disorders. Expert Review of Neurotherapeutics, 2019, 19, 1037-1050.	2.8	116
74	Comparative effectiveness of antiepileptic drugs in juvenile myoclonic epilepsy. Epilepsia Open, 2019, 4, 420-430.	2.4	34
75	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	12.8	99
76	Genomic and clinical predictors of lacosamide response in refractory epilepsies. Epilepsia Open, 2019, 4, 563-571.	2.4	12
77	NF1 microdeletion syndrome: case report of two new patients. Italian Journal of Pediatrics, 2019, 45, 138.	2.6	27
78	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
79	Pelizaeusâ€“Merzbacher Disease due to PLP1 Frameshift Mutation in a Female with Nonrandom Skewed X-Chromosome Inactivation. Neuropediatrics, 2019, 50, 268-270.	0.6	1
80	Epidemiology and familial clustering of pediatric epilepsy in the geographic isolate of Ischia. Epilepsy Research, 2019, 154, 86-89.	1.6	2
81	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
82	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
83	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.	2.0	10
84	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	5.1	61
85	The spectrum of intermediate <scp>SCN</scp>8Aâ€related epilepsy. Epilepsia, 2019, 60, 830-844.	5.1	70
86	Familial adult myoclonic epilepsy: A new expansion repeats disorder. Seizure: the Journal of the British Epilepsy Association, 2019, 67, 73-77.	2.0	18
87	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
88	No evidence for a BRD 2 promoter hypermethylation inÂblood leukocytes of Europeans with juvenile myoclonicÂepilepsy. Epilepsia, 2019, 60, e31-e36.	5.1	4
89	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.	2.4	9
90	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

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91	Spinal motor neuron involvement in a patient with homozygous PRUNE mutation. European Journal of Paediatric Neurology, 2018, 22, 541-543.	1.6	10
92	Genetic variation in CFH predicts phenytoin-induced maculopapular exanthema in European-descent patients. Neurology, 2018, 90, e332-e341.	1.1	43
93	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.	2.0	0
94	Clinical and molecular characterization of 112 single-center patients with Neurofibromatosis type 1. Italian Journal of Pediatrics, 2018, 44, 45.	2.6	32
95	Clinical and molecular consequences of exon 78 deletion in DMD gene. Journal of Human Genetics, 2018, 63, 761-764.	2.3	7
96	PRRT2 controls neuronal excitability by negatively modulating Na+ channel 1.2/1.6 activity. Brain, 2018, 141, 1000-1016.	7.6	99
97	De novo 12q22.q23.3 duplication associated with temporal lobe epilepsy. Seizure: the Journal of the British Epilepsy Association, 2018, 57, 63-65.	2.0	0
98	Recent advances in epilepsy genetics. Neuroscience Letters, 2018, 667, 4-9.	2.1	66
99	Gain-of-functionHCN2variants in genetic epilepsy. Human Mutation, 2018, 39, 202-209.	2.5	28
100	HCN1mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. Brain, 2018, 141, 3160-3178.	7.6	96
101	Rare gene deletions in genetic generalized and Rolandic epilepsies. PLoS ONE, 2018, 13, e0202022.	2.5	6
102	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	12.6	1,085
103	Functional Connectivity and Genetic Profile of a â€œDouble-Cortexâ€•Like Malformation. Frontiers in Integrative Neuroscience, 2018, 12, 22.	2.1	11
104	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
105	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
106	The PRRT2 knockout mouse recapitulates the neurological diseases associated with PRRT2 mutations. Neurobiology of Disease, 2017, 99, 66-83.	4.4	72
107	Common and rare epilepsies share genetic determinants. Nature Reviews Neurology, 2017, 13, 200-201.	10.1	5
108	Confirmation of mutations inPROSCas a novel cause of vitamin B_₆-dependent epilepsy. Journal of Medical Genetics, 2017, 54, 809-814.	3.2	66

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109	De novo 12q22.q23.3 duplication associated with temporal lobe epilepsy. Seizure: the Journal of the British Epilepsy Association, 2017, 50, 80-82.	2.0	4
110	Application of rare variant transmission disequilibrium tests to epileptic encephalopathy trio sequence data. European Journal of Human Genetics, 2017, 25, 894-899.	2.8	7
111	Carbamazepineâ€•and oxcarbazepineâ€‐induced hyponatremia in people with epilepsy. Epilepsia, 2017, 58, 1227-1233.	5.1	54
112	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
113	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. Brain, 2017, 140, 1316-1336.	7.6	426
114	Teaching Neuro Images : Figure of 8. Neurology, 2017, 89, e172-e173.	1.1	5
115	Heterogeneous contribution of microdeletions in the development of common generalised and focal epilepsies. Journal of Medical Genetics, 2017, 54, 598-606.	3.2	22
116	Novel AMPD2 mutation in pontocerebellar hypoplasia, dysmorphisms, and teeth abnormalities. Neurology: Genetics, 2017, 3, e179.	1.9	22
117	Comparative effectiveness of antiepileptic drugs in patients with mesial temporal lobe epilepsy with hippocampal sclerosis. Epilepsia, 2017, 58, 1734-1741.	5.1	26
118	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.	5.1	8
119	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.	2.0	13
120	Confirmation of mutations in the PROSC gene as a novel cause of vitamin B6 dependent epilepsy. European Journal of Paediatric Neurology, 2017, 21, e1.	1.6	0
121	Efficacy of sodium channel blockers in SCN2A early infantile epileptic encephalopathy. Brain and Development, 2017, 39, 345-348.	1.1	57
122	ARHGEF9 mutations cause a specific recognizable X-linked intellectual disability syndrome. Neurology: Genetics, 2017, 3, e159.	1.9	7
123	Rare GABRA3 variants are associated with epileptic seizures, encephalopathy and dysmorphic features. Brain, 2017, 140, 2879-2894.	7.6	33
124	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
125	Autosomal dominant cortical tremor, myoclonus and epilepsy. Epileptic Disorders, 2016, 18, 139-144.	1.3	20
126	Autosomal recessive progressive myoclonus epilepsy due to impaired ceramide synthesis. Epileptic Disorders, 2016, 18, 120-127.	1.3	9

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127	White matter involvement in a family with a novel PDGFB mutation. Neurology: Genetics, 2016, 2, e77.	1.9	19
128	Management of genetic epilepsies: From empirical treatment to precision medicine. Pharmacological Research, 2016, 107, 426-429.	7.1	52
129	PRRT2 Is a Key Component of the Ca 2+ -Dependent Neurotransmitter Release Machinery. Cell Reports, 2016, 15, 117-131.	6.4	121
130	Pitfalls in genetic testing: the story of missed SCN1A mutations. Molecular Genetics & Genomic Medicine, 2016, 4, 457-464.	1.2	67
131	Clinical intrafamilial variability in lethal familial neonatal seizure disorder caused by TBC1D24 mutations. American Journal of Medical Genetics, Part A, 2016, 170, 3207-3214.	1.2	19
132	PRRT2: from Paroxysmal Disorders to Regulation of Synaptic Function. Trends in Neurosciences, 2016, 39, 668-679.	8.6	68
133	Exome Sequencing Fails to Identify the Genetic Cause of Aicardi Syndrome. Molecular Syndromology, 2016, 7, 234-238.	0.8	16
134	TBC1D24 genotypeâ€“phenotype correlation. Neurology, 2016, 87, 77-85.	1.1	97
135	Novel treatment perspectives from advances in understanding of genetic epilepsy syndromes. Expert Opinion on Orphan Drugs, 2016, 4, 485-490.	0.8	0
136	The ubiquitin ligase tripartite-motif-protein 32 is induced in Duchenne muscular dystrophy. Laboratory Investigation, 2016, 96, 862-871.	3.7	23
137	Genetic and Early Clinical Manifestations of Females Heterozygous for Duchenne/Becker Muscular Dystrophy. Pediatric Neurology, 2016, 55, 58-63.	2.1	37
138	Genetics of reflex seizures and epilepsies in humans and animals. Epilepsy Research, 2016, 121, 47-54.	1.6	60
139	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.	1.1	5
140	Psychiatric comorbidities in patients from seven families with autosomal dominant cortical tremor, myoclonus, and epilepsy. Epilepsy and Behavior, 2016, 56, 38-43.	1.7	21
141	Xp11.22 Microduplications Including HUWE1: Case Report and Literature Review. Neuropediatrics, 2016, 47, 051-056.	0.6	9
142	The leukodystrophy protein FAM126A (hyccin) regulates PtdIns(4)P synthesis at the plasmaÂmembrane. Nature Cell Biology, 2016, 18, 132-138.	10.3	91
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	Evaluation of Presumably Disease Causing SCN1A Variants in a Cohort of Common Epilepsy Syndromes. PLoS ONE, 2016, 11, e0150426.	2.5	22

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145	Novel GABRG2 mutations cause familial febrile seizures. Neurology: Genetics, 2015, 1, e35.	1.9	29
146	Extending the phenotypic spectrum of <scp>RBFOX</scp>1 deletions: Sporadic focal epilepsy. Epilepsia, 2015, 56, e129-33.	5.1	38
147	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
148	A novel KCNQ3 mutation in familial epilepsy with focal seizures and intellectual disability. Epilepsia, 2015, 56, e15-20.	5.1	66
149	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. American Journal of Human Genetics, 2015, 96, 808-815.	6.2	173
150	No evidence of a role for cystatin <scp>B</scp> gene in juvenile myoclonic epilepsy. Epilepsia, 2015, 56, e40-3.	5.1	7
151	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
152	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
153	CHD2 mutations are a rare cause of generalized epilepsy with myoclonicâ€“atonic seizures. Epilepsy and Behavior, 2015, 51, 53-56.	1.7	28
154	Mutations in <scp>KCNT</scp>1 cause a spectrum of focal epilepsies. Epilepsia, 2015, 56, e114-20.	5.1	117
155	Genetic and forensic implications in epilepsy and cardiac arrhythmias: a case series. International Journal of Legal Medicine, 2015, 129, 495-504.	2.2	40
156	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
157	Mild <scp>L</scp>afora disease: Clinical, neurophysiologic, and genetic findings. Epilepsia, 2014, 55, e129-33.	5.1	43
158	Expanding sialidosis spectrum by genome-wide screening. Neurology, 2014, 82, 2003-2006.	1.1	37
159	The Î±_{2B}â€adrenergic receptor is mutant in cortical myoclonus and epilepsy. Annals of Neurology, 2014, 75, 77-87.	5.3	42
160	Genetic heterogeneity in malignant migrating partial seizures of infancy. Annals of Neurology, 2014, 75, 324-326.	5.3	10
161	Impairment of ceramide synthesis causes a novel progressive myoclonus epilepsy. Annals of Neurology, 2014, 76, 206-212.	5.3	80
162	De novo mutations in HCN1 cause early infantile epileptic encephalopathy. Nature Genetics, 2014, 46, 640-645.	21.4	192

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163	Long incubation in imported human rabies. Annals of Neurology, 2014, 75, 324-325.	5.3	6
164	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.	2.0	4
165	De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	6.2	388
166	Genetic determinants of common epilepsies: a meta-analysis of genome-wide association studies. Lancet Neurology, The, 2014, 13, 893-903.	10.2	264
167	Progressive myoclonic epilepsies. Neurology, 2014, 82, 405-411.	1.1	87
168	EXOSC3 mutations in isolated cerebellar hypoplasia and spinal anterior horn involvement. Journal of Neurology, 2013, 260, 1866-1870.	3.6	28
169	PRRT2-related disorders: further PKD and ICCA cases and review of the literature. Journal of Neurology, 2013, 260, 1234-1244.	3.6	63
170	Neurological features and long-term follow-up in 15q11.2-13.1 duplication. European Journal of Medical Genetics, 2013, 56, 614-618.	1.3	5
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