Elena Parrini

List of Publications by Year
in descending order

68 papers	3,752 citations	172457 29 h-index	138484 58 g-index
70 all docs	70 docs citations	70 times ranked	5608 citing authors

#	Article	IF	Citations
1	Mutations in TUBG1, DYNC1H1, KIF5C and KIF2A cause malformations of cortical development and microcephaly. Nature Genetics, 2013, 45, 639-647.	21.4	399
2	Somatic Mutations in Cerebral Cortical Malformations. New England Journal of Medicine, 2014, 371, 733-743.	27.0	326
3	Periventricular heterotopia: phenotypic heterogeneity and correlation with Filamin A mutations. Brain, 2006, 129, 1892-1906.	7.6	315
4	Neuronal migration disorders. Neurobiology of Disease, 2010, 38, 154-166.	4.4	271
5	Increased Sensitivity of the Neuronal Nicotinic Receptor Î±2 Subunit Causes Familial Epilepsy with Nocturnal Wandering and Ictal Fear. American Journal of Human Genetics, 2006, 79, 342-350.	6.2	225
6	Genetic Basis of Brain Malformations. Molecular Syndromology, 2016, 7, 220-233.	0.8	156
7	Diagnostic Targeted Resequencing in 349 Patients with Drug-Resistant Pediatric Epilepsies Identifies Causative Mutations in 30 Different Genes. Human Mutation, 2017, 38, 216-225.	2.5	152
8	Epilepsy in Rett syndrome, and CDKL5â€•and FOXG1â€geneâ€“related encephalopathies. Epilepsia, 2012, 53, 2067-2078.	5.1	124
9	Lissencephaly: Expanded imaging and clinical classification. American Journal of Medical Genetics, Part A, 2017, 173, 1473-1488.	1.2	104
10	HCN1mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. Brain, 2018, 141, 3160-3178.	7.6	96
11	Clinical and genetic factors predicting Dravet syndrome in infants with SCN1A mutations. Neurology, 2017, 88, 1037-1044.	1.1	93
12	Analysis of 17 genes detects mutations in 81% of 811 patients with lissencephaly. Genetics in Medicine, 2018, 20, 1354-1364.	2.4	92
13	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. Brain, 2013, 136, 3378-3394.	7.6	85
14	Coâ€•occurring malformations of cortical development and <scp>SCN</scp>1A gene mutations. Epilepsia, 2014, 55, 1009-1019.	5.1	84
15	Delineating SPTAN1 associated phenotypes: from isolated epilepsy to encephalopathy with progressive brain atrophy. Brain, 2017, 140, 2322-2336.	7.6	82
16	Characterisation of mutations of the phosphoinositide-3-kinase regulatory subunit, PIK3R2, in perisylvian polymicrogyria: a next-generation sequencing study. Lancet Neurology, The, 2015, 14, 1182-1195.	10.2	74
17	Generalized Epilepsy with Febrile Seizures Plus (GEFS+): Clinical Spectrum in Seven Italian Families Unrelated to SCN1A, SCN1B, and GABRG2 Gene Mutations. Epilepsia, 2004, 45, 149-158.	5.1	67
18	Nonsyndromic mental retardation and cryptogenic epilepsy in women withDoublecortin gene mutations. Annals of Neurology, 2003, 54, 30-37.	5.3	65

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19	Symmetric polymicrogyria and pachygyria associated with TUBB2B gene mutations. European Journal of Human Genetics, 2012, 20, 995-998.	2.8	61
20	Xp22.3 genomic deletions involving the CDKL5 gene in girls with early onset epileptic encephalopathy. Epilepsia, 2010, 51, 647-654.	5.1	60
21	Clinical features and outcome of 6 new patients carrying de novo KCNB1 gene mutations. Neurology: Genetics, 2017, 3, e206.	1.9	53
22	International consensus recommendations on the diagnostic work-up for malformations of cortical development. Nature Reviews Neurology, 2020, 16, 618-635.	10.1	53
23	The Impact of Next-Generation Sequencing on the Diagnosis and Treatment of Epilepsy in Paediatric Patients. Molecular Diagnosis and Therapy, 2017, 21, 357-373.	3.8	49
24	Bilateral frontoparietal polymicrogyria, Lennoxâ€Gastaut syndrome, and GPR56 gene mutations. Epilepsia, 2009, 50, 1344-1353.	5.1	46
25	High frequency of genomic deletions--and a duplication--in the LIS1 gene in lissencephaly: implications for molecular diagnosis. Journal of Medical Genetics, 2008, 45, 355-361.	3.2	45
26	Mosaic mutations of the FLN1 gene cause a mild phenotype in patients with periventricular heterotopia. Neurogenetics, 2004, 5, 191-196.	1.4	44
27	TBC1D24-TLDc-related epilepsy exercise-induced dystonia: rescue by antioxidants in a disease model. Brain, 2019, 142, 2319-2335.	7.6	44
28	<scp>SCN3A</scp>â€Related Neurodevelopmental Disorder: A Spectrum of Epilepsy and Brain Malformation. Annals of Neurology, 2020, 88, 348-362.	5.3	42
29	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. Brain, 2021, 144, 1435-1450.	7.6	35
30	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	5.1	32
31	Peritrigonal and temporo-occipital heterotopia with corpus callosum and cerebellar dysgenesis. Neurology, 2012, 79, 1244-1251.	1.1	31
32	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	2.4	28
33	Autosomal Dominant Early-onset Cortical Myoclonus, Photic-induced Myoclonus, and Epilepsy in a Large Pedigree. Epilepsia, 2006, 47, 1643-1649.	5.1	26
34	Early infantile epileptic-dyskinetic encephalopathy due to biallelicPIGPmutations. Neurology: Genetics, 2020, 6, e387.	1.9	26
35	Optimizing the molecular diagnosis of <scp>CDKL</scp>5 geneâ€“related epileptic encephalopathy in boys. Epilepsia, 2014, 55, 1748-1753.	5.1	23
36	Multiple genomic copy number variants associated with periventricular nodular heterotopia indicate extreme genetic heterogeneity. European Journal of Human Genetics, 2019, 27, 909-918.	2.8	21

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37	The syndrome of polymicrogyria, thalamic hypoplasia, and epilepsy with CSWS. Neurology, 2016, 86, 1250-1259.	1.1	19
38	Developmental and epilepsy spectrum of KCNB1 encephalopathy with longâ€term outcome. Epilepsia, 2020, 61, 2461-2473.	5.1	17
39	Familial periventricular nodular heterotopia, epilepsy and Melnickâ€“Needles Syndrome caused by a singleFLNAmutation with combined gain-of-function and loss-of-function effects. Journal of Medical Genetics, 2015, 52, 405-412.	3.2	15
40	ATP1A3 spectrum disorders: A video-documented history of 7 genetically confirmed early onset cases. European Journal of Paediatric Neurology, 2018, 22, 264-271.	1.6	15
41	Deciphering the premature mortality in PIGA-CDG â€“ An untold story. Epilepsy Research, 2021, 170, 106530.	1.6	15
42	Periventricular nodular heterotopia in Smithâ€Magenis syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 3142-3147.	1.2	14
43	The spectrum of brain malformations and disruptions in twins. American Journal of Medical Genetics, Part A, 2021, 185, 2690-2718.	1.2	13
44	Expanding the genetic and phenotypic spectrum of <scp>CHD2</scp>â€related disease: From early neurodevelopmental disorders to adultâ€onset epilepsy. American Journal of Medical Genetics, Part A, 2022, 188, 522-533.	1.2	13
45	Inâ€frame deletion in FLNA causing familial periventricular heterotopia with skeletal dysplasia in males. American Journal of Medical Genetics, Part A, 2011, 155, 1140-1146.	1.2	12
46	PRICKLE1â€related early onset epileptic encephalopathy. American Journal of Medical Genetics, Part A, 2018, 176, 2841-2845.	1.2	12
47	Intrafamilial variability in SPTAN1-related disorder: From benign convulsions with mild gastroenteritis to developmental encephalopathy. European Journal of Paediatric Neurology, 2020, 28, 237-239.	1.6	11
48	Lesional and non-lesional epilepsies: A blurring genetic boundary. European Journal of Paediatric Neurology, 2020, 24, 24-29.	1.6	8
49	Severe presentation and complex brain malformations in an individual carrying a CCND2 variant. Molecular Genetics & Genomic Medicine, 2019, 7, e708.	1.2	7
50	A de novo KCNQ2 Gene Mutation Associated With Non-familial Early Onset Seizures: Case Report and Revision of Literature Data. Frontiers in Pediatrics, 2019, 7, 348.	1.9	6
51	Cerebral folate transporter deficiency syndrome in three siblings: Why genetic testing for developmental and epileptic encephalopathies should be performed early and include the <scp>FOLR1</scp> gene. American Journal of Medical Genetics, Part A, 2021, 185, 2526-2531.	1.2	6
52	Migrating Focal Seizures and Myoclonic Status in ARV1- Related Encephalopathy. Neurology: Genetics, 2021, 7, e593.	1.9	6
53	Monoallelic and biallelic mutations inRELNunderlie a graded series of neurodevelopmental disorders. Brain, 2022, 145, 3274-3287.	7.6	6
54	What is the role of next generation sequencing in status epilepticus?. Epilepsy and Behavior, 2019, 101, 106373.	1.7	5

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55	Shedding light on dark genes: enhanced targeted resequencing by optimizing the combination of enrichment technology and DNA fragment length. Scientific Reports, 2020, 10, 9424.	3.3	5
56	Familial dominant epilepsy and mild pachygyria associated with a constitutional LIS1 mutation. American Journal of Medical Genetics, Part A, 2018, 176, 2808-2812.	1.2	3
57	Early-onset bradykinetic rigid syndrome and reflex seizures in a child with PURA syndrome. Epileptic Disorders, 2021, 23, 745-748.	1.3	3
58	Posterior Lissencephaly Associated with Subcortical Band Heterotopia Due to a Variation in the CEP85L Gene: A Case Report and Refining of the Phenotypic Spectrum. Genes, 2021, 12, 1208.	2.4	2
59	Defining causal variants in rare epilepsies: an essential team effort between biomedical scientists, geneticists and epileptologists. European Journal of Medical Genetics, 2022, 65, 104531.	1.3	2
60	Agyriaâ€“pachygyria band spectrum. , 2011, , 298-304.		1
61	Cerebral malformations. , 2020, , 249-267.		1
62	A Novel Strategy Combining Array-CGH, Whole-exome Sequencing and In Utero Electroporation in Rodents to Identify Causative Genes for Brain Malformations. Journal of Visualized Experiments, 2017, , .	0.3	0
63	Malformations of Cortical Development in Newborns: Genetic Aspects. , 2018, , 2113-2124.		0
64	Longâ€term followâ€up of an individual with <scp>ITPR1</scp>â€related disorder. American Journal of Medical Genetics, Part A, 2020, 182, 1846-1847.	1.2	0
65	Reelin and Lissencephaly. , 2008, , 311-316.		0
66	Malformations of Cortical Development: Genetic Aspects. , 2012, , 1131-1136.		0
67	Malformations of Cortical Development in Newborns: Genetic Aspects. , 2016, , 1-12.		0
68	Malformations of Cortical Development in Newborns: Genetic Aspects. , 2018, , 1-13.		0