

# Elena Parrini

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/1884851/publications.pdf>

Version: 2024-02-01

68  
papers

3,752  
citations

172386

29  
h-index

138417

58  
g-index

70  
all docs

70  
docs citations

70  
times ranked

5608  
citing authors

#	ARTICLE	IF	CITATIONS
1	Expanding the genetic and phenotypic spectrum of <i>CHD2</i> -related disease: From early neurodevelopmental disorders to adult-onset epilepsy. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 522-533.	0.7	13
2	Monoallelic and biallelic mutations in <i>RELN</i> underlie a graded series of neurodevelopmental disorders. <i>Brain</i> , 2022, 145, 3274-3287.	3.7	6
3	Defining causal variants in rare epilepsies: an essential team effort between biomedical scientists, geneticists and epileptologists. <i>European Journal of Medical Genetics</i> , 2022, 65, 104531.	0.7	2
4	The spectrum of brain malformations and disruptions in twins. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2690-2718.	0.7	13
5	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. <i>Genetics in Medicine</i> , 2021, 23, 363-373.	1.1	28
6	Deciphering the premature mortality in PIGA-CDG – An untold story. <i>Epilepsy Research</i> , 2021, 170, 106530.	0.8	15
7	Cerebral folate transporter deficiency syndrome in three siblings: Why genetic testing for developmental and epileptic encephalopathies should be performed early and include the <i>FOLR1</i> gene. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2526-2531.	0.7	6
8	<i>ATP1A2</i> and <i>ATP1A3</i> associated early profound epileptic encephalopathy and polymicrogyria. <i>Brain</i> , 2021, 144, 1435-1450.	3.7	35
9	Migrating Focal Seizures and Myoclonic Status in <i>ARV1</i> -Related Encephalopathy. <i>Neurology: Genetics</i> , 2021, 7, e593.	0.9	6
10	Posterior Lissencephaly Associated with Subcortical Band Heterotopia Due to a Variation in the <i>CEP85L</i> Gene: A Case Report and Refining of the Phenotypic Spectrum. <i>Genes</i> , 2021, 12, 1208.	1.0	2
11	Early-onset bradykinetic rigid syndrome and reflex seizures in a child with PURA syndrome. <i>Epileptic Disorders</i> , 2021, 23, 745-748.	0.7	3
12	Lesional and non-lesional epilepsies: A blurring genetic boundary. <i>European Journal of Paediatric Neurology</i> , 2020, 24, 24-29.	0.7	8
13	Intrafamilial variability in <i>SPTAN1</i> -related disorder: From benign convulsions with mild gastroenteritis to developmental encephalopathy. <i>European Journal of Paediatric Neurology</i> , 2020, 28, 237-239.	0.7	11
14	Developmental and epilepsy spectrum of <i>KCNB1</i> encephalopathy with long-term outcome. <i>Epilepsia</i> , 2020, 61, 2461-2473.	2.6	17
15	International consensus recommendations on the diagnostic work-up for malformations of cortical development. <i>Nature Reviews Neurology</i> , 2020, 16, 618-635.	4.9	53
16	Lessons learned from 40 novel <i>PIGA</i> patients and a review of the literature. <i>Epilepsia</i> , 2020, 61, 1142-1155.	2.6	32
17	Long-term follow-up of an individual with <i>ITPR1</i> -related disorder. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1846-1847.	0.7	0
18	<i>SCN3A</i> -Related Neurodevelopmental Disorder: A Spectrum of Epilepsy and Brain Malformation. <i>Annals of Neurology</i> , 2020, 88, 348-362.	2.8	42

#	ARTICLE	IF	CITATIONS
19	Shedding light on dark genes: enhanced targeted resequencing by optimizing the combination of enrichment technology and DNA fragment length. <i>Scientific Reports</i> , 2020, 10, 9424.	1.6	5
20	Early infantile epileptic-dyskinetic encephalopathy due to biallelic PIGP mutations. <i>Neurology: Genetics</i> , 2020, 6, e387.	0.9	26
21	Cerebral malformations. , 2020, , 249-267.		1
22	What is the role of next generation sequencing in status epilepticus?. <i>Epilepsy and Behavior</i> , 2019, 101, 106373.	0.9	5
23	TBC1D24-TLDC-related epilepsy exercise-induced dystonia: rescue by antioxidants in a disease model. <i>Brain</i> , 2019, 142, 2319-2335.	3.7	44
24	A de novo KCNQ2 Gene Mutation Associated With Non-familial Early Onset Seizures: Case Report and Revision of Literature Data. <i>Frontiers in Pediatrics</i> , 2019, 7, 348.	0.9	6
25	Multiple genomic copy number variants associated with periventricular nodular heterotopia indicate extreme genetic heterogeneity. <i>European Journal of Human Genetics</i> , 2019, 27, 909-918.	1.4	21
26	Severe presentation and complex brain malformations in an individual carrying a <i>CCND2</i> variant. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2019, 7, e708.	0.6	7
27	Analysis of 17 genes detects mutations in 81% of 811 patients with lissencephaly. <i>Genetics in Medicine</i> , 2018, 20, 1354-1364.	1.1	92
28	ATP1A3 spectrum disorders: A video-documented history of 7 genetically confirmed early onset cases. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 264-271.	0.7	15
29	<i>HCN1</i> mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. <i>Brain</i> , 2018, 141, 3160-3178.	3.7	96
30	Malformations of Cortical Development in Newborns: Genetic Aspects. , 2018, , 2113-2124.		0
31	<i>PRICKLE1</i> related early onset epileptic encephalopathy. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2841-2845.	0.7	12
32	Familial dominant epilepsy and mild pachygyria associated with a constitutional <i>LIS1</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2808-2812.	0.7	3
33	Malformations of Cortical Development in Newborns: Genetic Aspects. , 2018, , 1-13.		0
34	The Impact of Next-Generation Sequencing on the Diagnosis and Treatment of Epilepsy in Paediatric Patients. <i>Molecular Diagnosis and Therapy</i> , 2017, 21, 357-373.	1.6	49
35	Clinical and genetic factors predicting Dravet syndrome in infants with <i>SCN1A</i> mutations. <i>Neurology</i> , 2017, 88, 1037-1044.	1.5	93
36	Lissencephaly: Expanded imaging and clinical classification. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1473-1488.	0.7	104

#	ARTICLE	IF	CITATIONS
37	Delineating SPTAN1 associated phenotypes: from isolated epilepsy to encephalopathy with progressive brain atrophy. <i>Brain</i> , 2017, 140, 2322-2336.	3.7	82
38	Clinical features and outcome of 6 new patients carrying de novo <i>KCNB1</i> gene mutations. <i>Neurology: Genetics</i> , 2017, 3, e206.	0.9	53
39	Diagnostic Targeted Resequencing in 349 Patients with Drug-Resistant Pediatric Epilepsies Identifies Causative Mutations in 30 Different Genes. <i>Human Mutation</i> , 2017, 38, 216-225.	1.1	152
40	A Novel Strategy Combining Array-CGH, Whole-exome Sequencing and <i>In Utero</i> Electroporation in Rodents to Identify Causative Genes for Brain Malformations. <i>Journal of Visualized Experiments</i> , 2017, , .	0.2	0
41	Genetic Basis of Brain Malformations. <i>Molecular Syndromology</i> , 2016, 7, 220-233.	0.3	156
42	The syndrome of polymicrogyria, thalamic hypoplasia, and epilepsy with CSWS. <i>Neurology</i> , 2016, 86, 1250-1259.	1.5	19
43	Malformations of Cortical Development in Newborns: Genetic Aspects. , 2016, , 1-12.		0
44	Familial periventricular nodular heterotopia, epilepsy and Melnickâ€œNeedles Syndrome caused by a single <i>FLN</i> mutation with combined gain-of-function and loss-of-function effects. <i>Journal of Medical Genetics</i> , 2015, 52, 405-412.	1.5	15
45	Characterisation of mutations of the phosphoinositide-3-kinase regulatory subunit, <i>PIK3R2</i> , in perisylvian polymicrogyria: a next-generation sequencing study. <i>Lancet Neurology</i> , The, 2015, 14, 1182-1195.	4.9	74
46	Optimizing the molecular diagnosis of <i>CDKL5</i> geneâ€œrelated epileptic encephalopathy in boys. <i>Epilepsia</i> , 2014, 55, 1748-1753.	2.6	23
47	Co-occurring malformations of cortical development and <i>SCN1A</i> gene mutations. <i>Epilepsia</i> , 2014, 55, 1009-1019.	2.6	84
48	Somatic Mutations in Cerebral Cortical Malformations. <i>New England Journal of Medicine</i> , 2014, 371, 733-743.	13.9	326
49	Periventricular nodular heterotopia in Smithâ€œMagenis syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 3142-3147.	0.7	14
50	Mutations in <i>TUBG1</i> , <i>DYNC1H1</i> , <i>KIF5C</i> and <i>KIF2A</i> cause malformations of cortical development and microcephaly. <i>Nature Genetics</i> , 2013, 45, 639-647.	9.4	399
51	Periventricular heterotopia in 6q terminal deletion syndrome: role of the <i>C6orf70</i> gene. <i>Brain</i> , 2013, 136, 3378-3394.	3.7	85
52	Symmetric polymicrogyria and pachygyria associated with <i>TUBB2B</i> gene mutations. <i>European Journal of Human Genetics</i> , 2012, 20, 995-998.	1.4	61
53	Epilepsy in Rett syndrome, and <i>CDKL5</i> and <i>FOXG1</i> geneâ€œrelated encephalopathies. <i>Epilepsia</i> , 2012, 53, 2067-2078.	2.6	124
54	Peritrigonal and temporo-occipital heterotopia with corpus callosum and cerebellar dysgenesis. <i>Neurology</i> , 2012, 79, 1244-1251.	1.5	31

#	ARTICLE	IF	CITATIONS
55	Malformations of Cortical Development: Genetic Aspects. , 2012, , 1131-1136.		0
56	Agyriaâ€“pachygyria band spectrum. , 2011, , 298-304.		1
57	Inâ€“frame deletion in <i>FLNA</i> causing familial periventricular heterotopia with skeletal dysplasia in males. American Journal of Medical Genetics, Part A, 2011, 155, 1140-1146.	0.7	12
58	Neuronal migration disorders. Neurobiology of Disease, 2010, 38, 154-166.	2.1	271
59	Xp22.3 genomic deletions involving the <i>CDKL5</i> gene in girls with early onset epileptic encephalopathy. Epilepsia, 2010, 51, 647-654.	2.6	60
60	Bilateral frontoparietal polymicrogyria, Lennoxâ€“Gastaut syndrome, and <i>GPR56</i> gene mutations. Epilepsia, 2009, 50, 1344-1353.	2.6	46
61	High frequency of genomic deletionsâ€“and a duplicationâ€“in the LIS1 gene in lissencephaly: implications for molecular diagnosis. Journal of Medical Genetics, 2008, 45, 355-361.	1.5	45
62	Reelin and Lissencephaly. , 2008, , 311-316.		0
63	Periventricular heterotopia: phenotypic heterogeneity and correlation with Filamin A mutations. Brain, 2006, 129, 1892-1906.	3.7	315
64	Increased Sensitivity of the Neuronal Nicotinic Receptor $\alpha 2$ Subunit Causes Familial Epilepsy with Nocturnal Wandering and Ictal Fear. American Journal of Human Genetics, 2006, 79, 342-350.	2.6	225
65	Autosomal Dominant Early-onset Cortical Myoclonus, Photic-induced Myoclonus, and Epilepsy in a Large Pedigree. Epilepsia, 2006, 47, 1643-1649.	2.6	26
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	Mosaic mutations of the FLN1 gene cause a mild phenotype in patients with periventricular heterotopia. Neurogenetics, 2004, 5, 191-196.	0.7	44
68	Nonsyndromic mental retardation and cryptogenic epilepsy in women with Doublecortin gene mutations. Annals of Neurology, 2003, 54, 30-37.	2.8	65