Raffaella Cusmai

List of Publications by Year
in descending order

57	1,972	218677	254184
papers	citations	26 h-index	43 g-index

61	61	61	2209
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Characterization of mutations in the genedoublecortin in patients with double cortex syndrome. Annals of Neurology, 1999, 45, 146-153.	5.3	175
2	The ketogenic diet in children, adolescents and young adults with refractory epilepsy: an Italian multicentric experience. Epilepsy Research, 2002, 48, 221-227.	1.6	134
3	Long-term neurological outcome in children with early-onset epilepsy associated with tuberous sclerosis. Epilepsy and Behavior, 2011, 22, 735-739.	1.7	120
4	Genetic and neuroradiological heterogeneity of double cortex syndrome. Annals of Neurology, 2000, 47, 265-269.	5.3	94
5	The Idiopathic Form of West Syndrome. Epilepsia, 1993, 34, 743-746.	5.1	86
6	Spectrum of phenotypes in female patients with epilepsy due to protocadherin 19 mutations. Epilepsia, 2011, 52, 1251-1257.	5.1	74
7	Reflex Myoclonic Epilepsy in Infancy: A New Age-Dependent Idiopathic Epileptic Syndrome Related to Startle Reaction. Epilepsia, 1995, 36, 342-348.	5.1	65
8	Focal seizures with affective symptoms are a major feature of PCDH19 geneâ€“related epilepsy. Epilepsia, 2012, 53, 2111-2119.	5.1	63
9	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
10	<scp>PCDH</scp>19â€related epilepsy in two mosaic male patients. Epilepsia, 2016, 57, e51-5.	5.1	57
11	Telomere shortening and telomere position effect in mild ring 17 syndrome. Epigenetics and Chromatin, 2014, 7, 1.	3.9	56
12	West Syndrome Due to Perinatal Insults. Epilepsia, 1993, 34, 738-742.	5.1	49
13	Ketogenic diet in early myoclonic encephalopathy due to non ketotic hyperglycinemia. European Journal of Paediatric Neurology, 2012, 16, 509-513.	1.6	47
14	Levetiracetam in juvenile myoclonic epilepsy: longâ€term efficacy in newly diagnosed adolescents. Developmental Medicine and Child Neurology, 2008, 50, 29-32.	2.1	45
15	Neuroimaging Changes in Menkes Disease, Part 1. American Journal of Neuroradiology, 2017, 38, 1850-1857.	2.4	42
16	Reduced steroidogenesis in patients with <scp>PCDH</scp>19â€female limited epilepsy. Epilepsia, 2017, 58, e91-e95.	5.1	40
17	Defining the electroclinical phenotype and outcome of PCDH19â€related epilepsy: A multicenter study. Epilepsia, 2018, 59, 2260-2271.	5.1	39
18	The ketogenic diet for Dravet syndrome and other epileptic encephalopathies: An Italian consensus. Epilepsia, 2011, 52, 83-89.	5.1	37

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19	Missense mutations of CACNA1A are a frequent cause of autosomal dominant nonprogressive congenital ataxia. European Journal of Paediatric Neurology, 2017, 21, 450-456.	1.6	37
20	Tubulin-related cerebellar dysplasia: definition of a distinct pattern of cerebellar malformation. European Radiology, 2017, 27, 5080-5092.	4.5	36
21	Congenital disorders of glycosylation presenting as epileptic encephalopathy with migrating partial seizures in infancy. Developmental Medicine and Child Neurology, 2016, 58, 1085-1091.	2.1	33
22	Current role of rufinamide in the treatment of childhood epilepsy: Literature review and treatment guidelines. European Journal of Paediatric Neurology, 2014, 18, 685-690.	1.6	32
23	Cognitive development in females with PCDH19 gene-related epilepsy. Epilepsy and Behavior, 2015, 42, 36-40.	1.7	32
24	Bilateral, Reversible, Selective Thalamic Involvement Demonstrated by Brain MR and Acute Severe Neurological Dysfunction with Favorable Outcome*. Neuropediatrics, 1994, 25, 44-47.	0.6	30
25	Gelastic Epilepsy and True Precocious Puberty due to Hypothalamic Hamartoma. Developmental Medicine and Child Neurology, 1984, 26, 509-514.	2.1	30
26	Long-term follow-up in children with benign convulsions associated with gastroenteritis. European Journal of Paediatric Neurology, 2014, 18, 572-577.	1.6	30
27	Autism and Infantile Spasms in Children with Tuberous Sclerosis. Developmental Medicine and Child Neurology, 1987, 29, 551-551.	2.1	25
28	Electroclinical Features and Long-Term Outcome of Cryptogenic Epilepsy in Children with Down Syndrome. Journal of Pediatrics, 2013, 163, 1754-1758.	1.8	25
29	Current role of perampanel in pediatric epilepsy. Italian Journal of Pediatrics, 2017, 43, 51.	2.6	25
30	Short-term Nonhormonal and Nonsteroid Treatment in West Syndrome. Epilepsia, 2003, 44, 1085-1088.	5.1	24
31	Epilepsy in the setting of full trisomy 18: A multicenter study on 18 affected children with and without structural brain abnormalities. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2016, 172, 288-295.	1.6	24
32	ATP1A3 -related epileptic encephalopathy responding to ketogenic diet. Brain and Development, 2018, 40, 433-438.	1.1	23
33	PRRT2 is mutated in familial and non-familial benign infantile seizures. European Journal of Paediatric Neurology, 2013, 17, 77-81.	1.6	22
34	Convulsions associated with gastroenteritis in the spectrum of benign focal epilepsies in infancy: 30 cases including four cases with ictal EEG recording. Epileptic Disorders, 2010, 12, 255-261.	1.3	22
35	White matter disruption is associated with persistent seizures in tuberous sclerosis complex. Epilepsy and Behavior, 2016, 60, 63-67.	1.7	21
36	Neuroimaging Changes in Menkes Disease, Part 2. American Journal of Neuroradiology, 2017, 38, 1858-1865.	2.4	20

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37	Ketogenic diet in a patient with congenital hyperinsulinism: a novel approach to prevent brain damage. Orphanet Journal of Rare Diseases, 2015, 10, 120.	2.7	19
38	Efficacy of levetiracetam in the treatment of drug-resistant Rett syndrome. Epilepsy Research, 2010, 88, 112-117.	1.6	18
39	Rufinamide for the treatment of refractory epilepsy secondary to neuronal migration disorders. Epilepsy Research, 2014, 108, 542-546.	1.6	18
40	Congenital X-linked ataxia, progressive myoclonic encephalopathy, macular degeneration and recurrent infections. American Journal of Medical Genetics Part A, 1992, 43, 443-451.	2.4	16
41	The Ketogenic Diet Increases In Vivo Glutathione Levels in Patients with Epilepsy. Metabolites, 2020, 10, 504.	2.9	15
42	Rufinamide efficacy and safety as adjunctive treatment in children with focal drug resistant epilepsy: The first Italian prospective study. Epilepsy Research, 2012, 102, 94-99.	1.6	14
43	Tuberous Sclerosis: Diagnostic and Prognostic Problems. Pediatric Neurosurgery, 1985, 12, 123-125.	0.7	13
44	Early onset absence epilepsy with onset in the first year of life: A multicenter cohort study. Epilepsia, 2013, 54, 66-69.	5.1	12
45	Epilepsy in Menkes disease: An electroclinical long-term study of 28 patients. Epilepsy Research, 2014, 108, 1597-1603.	1.6	11
46	Long-term outcome of epilepsy in patients with Praderâ€“Willi syndrome. Journal of Neurology, 2015, 262, 116-123.	3.6	10
47	Symmetrical bithalamic hyperdensities in asphyxiated full-term newborns: An early indicator of status marmoratus. Brain and Development, 1988, 10, 57-59.	1.1	8
48	Reflex myoclonic epilepsy in infancy: A multicenter clinical study. Epilepsy Research, 2013, 103, 237-244.	1.6	8
49	CASK related disorder: Epilepsy and developmental outcome. European Journal of Paediatric Neurology, 2021, 31, 61-69.	1.6	7
50	DRUGS FOR ALTERNATING HEMIPLEGIC MIGRAINE. Lancet, The, 1984, 324, 980.	13.7	5
51	Occurrence of a Prolonged Nonepileptic Motor Status after a Febrile Seizure. Epilepsia, 2006, 47, 1079-1081.	5.1	5
52	Ketogenic diet as elective treatment in patients with drug-unresponsive hyperinsulinemic hypoglycemia caused by glucokinase mutations. Orphanet Journal of Rare Diseases, 2021, 16, 424.	2.7	5
53	BAEPs in infantile spasms. Brain and Development, 1989, 11, 347-348.	1.1	4
54	OPTIC GLIOMA IN CHILDREN WITH NEUROFIBROMATOSIS. Lancet, The, 1987, 329, 1140.	13.7	3

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55	Polymicrogyria: A case detected by MRI. Brain and Development, 1989, 11, 257-259.	1.1	3
56	Short-term Nonhormonal and Nonsteroid Treatment in West Syndrome. Epilepsia, 2004, 45, 887-887.	5.1	0
57	Clinical Reasoning: A girl presenting with stiffness episodes during sleep, cafÃ©-au-lait spots, and flecked retina. Neurology, 2013, 80, e42-6.	1.1	0