

Raffaella Cusmai

List of Publications by Year in descending order

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Version: 2024-02-01

57
papers

1,972
citations

218677

26
h-index

254184

43
g-index

61
all docs

61
docs citations

61
times ranked

2209
citing authors

#	ARTICLE	IF	CITATIONS
1	CASK related disorder: Epilepsy and developmental outcome. European Journal of Paediatric Neurology, 2021, 31, 61-69.	1.6	7
2	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
3	The Ketogenic Diet Increases In Vivo Glutathione Levels in Patients with Epilepsy. Metabolites, 2020, 10, 504.	2.9	15
4	ATP1A3 -related epileptic encephalopathy responding to ketogenic diet. Brain and Development, 2018, 40, 433-438.	1.1	23
5	Defining the electroclinical phenotype and outcome of PCDH19-related epilepsy: A multicenter study. Epilepsia, 2018, 59, 2260-2271.	5.1	39
6	Neuroimaging Changes in Menkes Disease, Part 1. American Journal of Neuroradiology, 2017, 38, 1850-1857.	2.4	42
7	Neuroimaging Changes in Menkes Disease, Part 2. American Journal of Neuroradiology, 2017, 38, 1858-1865.	2.4	20
8	Reduced steroidogenesis in patients with <sc>PCDH</sc>19-related female limited epilepsy. Epilepsia, 2017, 58, e91-e95.	5.1	40
9	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
10	Tubulin-related cerebellar dysplasia: definition of a distinct pattern of cerebellar malformation. European Radiology, 2017, 27, 5080-5092.	4.5	36
11	Current role of perampanel in pediatric epilepsy. Italian Journal of Pediatrics, 2017, 43, 51.	2.6	25
12	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
13	<i><sc>PCDH</sc>19</i>-related epilepsy in two mosaic male patients. Epilepsia, 2016, 57, e51-5.	5.1	57
14	White matter disruption is associated with persistent seizures in tuberous sclerosis complex. Epilepsy and Behavior, 2016, 60, 63-67.	1.7	21
15	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
16	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
17	Long-term outcome of epilepsy in patients with Prader-Willi syndrome. Journal of Neurology, 2015, 262, 116-123.	3.6	10
18	Cognitive development in females with PCDH19 gene-related epilepsy. Epilepsy and Behavior, 2015, 42, 36-40.	1.7	32

#	ARTICLE	IF	CITATIONS
19	Long-term follow-up in children with benign convulsions associated with gastroenteritis. <i>European Journal of Paediatric Neurology</i> , 2014, 18, 572-577.	1.6	30
20	Telomere shortening and telomere position effect in mild ring 17 syndrome. <i>Epigenetics and Chromatin</i> , 2014, 7, 1.	3.9	56
21	Epilepsy in Menkes disease: An electroclinical long-term study of 28 patients. <i>Epilepsy Research</i> , 2014, 108, 1597-1603.	1.6	11
22	Rufinamide for the treatment of refractory epilepsy secondary to neuronal migration disorders. <i>Epilepsy Research</i> , 2014, 108, 542-546.	1.6	18
23	Current role of rufinamide in the treatment of childhood epilepsy: Literature review and treatment guidelines. <i>European Journal of Paediatric Neurology</i> , 2014, 18, 685-690.	1.6	32
24	PRRT2 is mutated in familial and non-familial benign infantile seizures. <i>European Journal of Paediatric Neurology</i> , 2013, 17, 77-81.	1.6	22
25	Reflex myoclonic epilepsy in infancy: A multicenter clinical study. <i>Epilepsy Research</i> , 2013, 103, 237-244.	1.6	8
26	Electroclinical Features and Long-Term Outcome of Cryptogenic Epilepsy in Children with Down Syndrome. <i>Journal of Pediatrics</i> , 2013, 163, 1754-1758.	1.8	25
27	Lacosamide in pediatric and adult patients: Comparison of efficacy and safety. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2013, 22, 210-216.	2.0	60
28	Early onset absence epilepsy with onset in the first year of life: A multicenter cohort study. <i>Epilepsia</i> , 2013, 54, 66-69.	5.1	12
29	Clinical Reasoning: A girl presenting with stiffness episodes during sleep, café-au-lait spots, and flecked retina. <i>Neurology</i> , 2013, 80, e42-6.	1.1	0
30	Focal seizures with affective symptoms are a major feature of <i>PCDH19</i> gene-related epilepsy. <i>Epilepsia</i> , 2012, 53, 2111-2119.	5.1	63
31	Ketogenic diet in early myoclonic encephalopathy due to non ketotic hyperglycinemia. <i>European Journal of Paediatric Neurology</i> , 2012, 16, 509-513.	1.6	47
32	Rufinamide efficacy and safety as adjunctive treatment in children with focal drug resistant epilepsy: The first Italian prospective study. <i>Epilepsy Research</i> , 2012, 102, 94-99.	1.6	14
33	Long-term neurological outcome in children with early-onset epilepsy associated with tuberous sclerosis. <i>Epilepsy and Behavior</i> , 2011, 22, 735-739.	1.7	120
34	The ketogenic diet for Dravet syndrome and other epileptic encephalopathies: An Italian consensus. <i>Epilepsia</i> , 2011, 52, 83-89.	5.1	37
35	Spectrum of phenotypes in female patients with epilepsy due to protocadherin 19 mutations. <i>Epilepsia</i> , 2011, 52, 1251-1257.	5.1	74
36	Efficacy of levetiracetam in the treatment of drug-resistant Rett syndrome. <i>Epilepsy Research</i> , 2010, 88, 112-117.	1.6	18

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37	Convulsions associated with gastroenteritis in the spectrum of benign focal epilepsies in infancy: 30 cases including four cases with ictal EEG recording. <i>Epileptic Disorders</i> , 2010, 12, 255-261.	1.3	22
38	Levetiracetam in juvenile myoclonic epilepsy: long-term efficacy in newly diagnosed adolescents. <i>Developmental Medicine and Child Neurology</i> , 2008, 50, 29-32.	2.1	45
39	Occurrence of a Prolonged Nonepileptic Motor Status after a Febrile Seizure. <i>Epilepsia</i> , 2006, 47, 1079-1081.	5.1	5
40	Short-term Nonhormonal and Nonsteroid Treatment in West Syndrome. <i>Epilepsia</i> , 2004, 45, 887-887.	5.1	0
41	Short-term Nonhormonal and Nonsteroid Treatment in West Syndrome. <i>Epilepsia</i> , 2003, 44, 1085-1088.	5.1	24
42	The ketogenic diet in children, adolescents and young adults with refractory epilepsy: an Italian multicentric experience. <i>Epilepsy Research</i> , 2002, 48, 221-227.	1.6	134
43	Genetic and neuroradiological heterogeneity of double cortex syndrome. <i>Annals of Neurology</i> , 2000, 47, 265-269.	5.3	94
44	Characterization of mutations in the gene doublecortin in patients with double cortex syndrome. <i>Annals of Neurology</i> , 1999, 45, 146-153.	5.3	175
45	Reflex Myoclonic Epilepsy in Infancy: A New Age-Dependent Idiopathic Epileptic Syndrome Related to Startle Reaction. <i>Epilepsia</i> , 1995, 36, 342-348.	5.1	65
46	Bilateral, Reversible, Selective Thalamic Involvement Demonstrated by Brain MR and Acute Severe Neurological Dysfunction with Favorable Outcome*. <i>Neuropediatrics</i> , 1994, 25, 44-47.	0.6	30
47	West Syndrome Due to Perinatal Insults. <i>Epilepsia</i> , 1993, 34, 738-742.	5.1	49
48	The Idiopathic Form of West Syndrome. <i>Epilepsia</i> , 1993, 34, 743-746.	5.1	86
49	Congenital X-linked ataxia, progressive myoclonic encephalopathy, macular degeneration and recurrent infections. <i>American Journal of Medical Genetics Part A</i> , 1992, 43, 443-451.	2.4	16
50	Polymicrogyria: A case detected by MRI. <i>Brain and Development</i> , 1989, 11, 257-259.	1.1	3
51	BAEPs in infantile spasms. <i>Brain and Development</i> , 1989, 11, 347-348.	1.1	4
52	Symmetrical bithalamic hyperdensities in asphyxiated full-term newborns: An early indicator of status marmoratus. <i>Brain and Development</i> , 1988, 10, 57-59.	1.1	8
53	OPTIC GLIOMA IN CHILDREN WITH NEUROFIBROMATOSIS. <i>Lancet, The</i> , 1987, 329, 1140.	13.7	3
54	Autism and Infantile Spasms in Children with Tuberous Sclerosis. <i>Developmental Medicine and Child Neurology</i> , 1987, 29, 551-551.	2.1	25

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55	Tuberous Sclerosis: Diagnostic and Prognostic Problems. <i>Pediatric Neurosurgery</i> , 1985, 12, 123-125.	0.7	13
56	DRUGS FOR ALTERNATING HEMIPLEGIC MIGRAINE. <i>Lancet, The</i> , 1984, 324, 980.	13.7	5
57	Gelastic Epilepsy and True Precocious Puberty due to Hypothalamic Hamartoma. <i>Developmental Medicine and Child Neurology</i> , 1984, 26, 509-514.	2.1	30