Maria Roberta Cilio

List of Publications by Year
in descending order

82 papers	6,170 citations	87401 40 h-index	81351 76 g-index
89 all docs	89 docs citations	89 times ranked	6683 citing authors

#	Article	IF	Citations
1	Characteristics of Neonates with Cardiopulmonary Disease Who Experience Seizures: A Multicenter Study. Journal of Pediatrics, 2022, 242, 63-73.	0.9	3
2	The ILAE classification of seizures and the epilepsies: Modification for seizures in the neonate. Position paper by the ILAE Task Force on Neonatal Seizures. Epilepsia, 2021, 62, 615-628.	2.6	158
3	Neonatal presentation of genetic epilepsies: Early differentiation from acute provoked seizures. Epilepsia, 2021, 62, 1907-1920.	2.6	32
4	Seizure Control in Neonates Undergoing Screening vs Confirmatory EEG Monitoring. Neurology, 2021, 97, e587-e596.	1.5	19
5	Disorders of Neuronal Migration/Organization Convey the Highest Risk of Neonatal Onset Epilepsy Compared to Other Congenital Brain Malformations. Pediatric Neurology, 2021, 127, 20-27.	1.0	2
6	Graph theory in paediatric epilepsy: A systematic review. Dialogues in Clinical Neuroscience, 2021, 23, 3-13.	1.8	15
7	Synthetic pharmaceutical grade cannabidiol for treatment of refractory infantile spasms: A multicenter phase-2 study. Epilepsy and Behavior, 2020, 102, 106826.	0.9	21
8	A Novel Kv7.3 Variant in the Voltage-Sensing S4 Segment in a Family With Benign Neonatal Epilepsy: Functional Characterization and in vitro Rescue by I²-Hydroxybutyrate. Frontiers in Physiology, 2020, 11, 1040.	1.3	7
9	Characterization of Death in Infants With Neonatal Seizures. Pediatric Neurology, 2020, 113, 21-25.	1.0	12
10	Response to cannabidiol in epilepsy of infancy with migrating focal seizures associated with KCNT1 mutations: An open-label, prospective, interventional study. European Journal of Paediatric Neurology, 2020, 25, 77-81.	0.7	13
11	Vomiting and retching as presenting signs of focal epilepsy in children. Epileptic Disorders, 2020, 22, 823-827.	0.7	1
12	Genetics of neonatal-onset epilepsies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2019, 162, 415-433.	1.0	23
13	Neonatal Developmental and Epileptic Encephalopathies. Seminars in Pediatric Neurology, 2019, 32, 100770.	1.0	24
14	Toward the elimination of bias in Pediatric Research. Pediatric Research, 2019, 86, 680-681.	1.1	0
15	Autism and developmental disability caused by KCNQ3 gainâ€ofâ€function variants. Annals of Neurology, 2019, 86, 181-192.	2.8	73
16	Pharmacokinetics and Tolerability of Multiple Doses of Pharmaceutical-Grade Synthetic Cannabidiol in Pediatric Patients with Treatment-Resistant Epilepsy. CNS Drugs, 2019, 33, 593-604.	2.7	57
17	Neonatal seizures: Is there a relationship between ictal electroclinical features and etiology? A critical appraisal based on a systematic literature review. Epilepsia Open, 2019, 4, 10-29.	1.3	42
18	The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. Epilepsia, 2019, 60, 797-806.	2.6	52

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19	Response to antiseizure medications in neonates with acute symptomatic seizures. Epilepsia, 2019, 60, e20-e24.	2.6	33
20	Augmented Reticular Thalamic Bursting and Seizures in Scn1a-Dravet Syndrome. Cell Reports, 2019, 26, 54-64.e6.	2.9	44
21	Long-Term Safety, Tolerability, and Efficacy of Cannabidiol in Children with Refractory Epilepsy: Results from an Expanded Access Program in the US. CNS Drugs, 2019, 33, 47-60.	2.7	57
22	Neonatal-Onset Epilepsies. , 2019, , 131-140.		0
23	Neonatal epilepsies: Clinical management. Seminars in Fetal and Neonatal Medicine, 2018, 23, 204-212.	1.1	38
24	The ClinGen Epilepsy Gene Curation Expert Panelâ€"Bridging the divide between clinical domain knowledge and formal gene curation criteria. Human Mutation, 2018, 39, 1476-1484.	1.1	33
25	Lack of response to quinidine in <scp>KCNT</scp>1â€related neonatal epilepsy. Epilepsia, 2018, 59, 1889-1898.	2.6	53
26	Dynamic action potential clamp predicts functional separation in mild familial and severe de novo forms of SCN2A epilepsy. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E5516-E5525.	3.3	69
27	Predictive value of early EEG for seizures in neonates with hypoxicâ€"ischemic encephalopathy undergoing therapeutic hypothermia. Pediatric Research, 2018, 84, 399-402.	1.1	22
28	Neonatal nonepileptic myoclonus is a prominent clinical feature of <scp>KCNQ</scp>2 gainâ€ofâ€function variants R201C and R201H. Epilepsia, 2017, 58, 436-445.	2.6	80
29	Seizures in Preterm Neonates: A Multicenter Observational Cohort Study. Pediatric Neurology, 2017, 72, 19-24.	1.0	83
30	A Distinctive Ictal Amplitude-Integrated Electroencephalography Pattern in Newborns with Neonatal Epilepsy Associated with <i>KCNQ2</i> Mutations. Neonatology, 2017, 112, 387-393.	0.9	44
31	Profile of neonatal epilepsies. Neurology, 2017, 89, 893-899.	1.5	145
32	Electroencephalography in the Preterm and Term Infant. , 2017, , 1362-1389.e4.		0
33	Epilepsy Genetics. , 2017, , 513-518.		0
34	Mild hypothermia and hemorrhagic lesions in neonates with hypoxic-ischemic encephalopathy: experience in an outborn center. Journal of Maternal-Fetal and Neonatal Medicine, 2016, 29, 1-4.	0.7	1
35	Rapid and safe response to lowâ€dose carbamazepine in neonatal epilepsy. Epilepsia, 2016, 57, 2019-2030.	2.6	92
36	Current understanding and neurobiology of epileptic encephalopathies. Neurobiology of Disease, 2016, 92, 72-89.	2.1	71

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37	Cannabidiol in patients with treatment-resistant epilepsy: an open-label interventional trial. Lancet Neurology, The, 2016, 15, 270-278.	4.9	714
38	Evaluation of Presumably Disease Causing SCN1A Variants in a Cohort of Common Epilepsy Syndromes. PLoS ONE, 2016, 11, e0150426.	1.1	22
39	Transcriptional regulator PRDM12 is essential for human pain perception. Nature Genetics, 2015, 47, 803-808.	9.4	137
40	Dysregulation of locus coeruleus development in congenital central hypoventilation syndrome. Acta Neuropathologica, 2015, 130, 171-183.	3.9	45
41	Early and effective treatment of <scp>KCNQ</scp>2 encephalopathy. Epilepsia, 2015, 56, 685-691.	2.6	229
42	Symptomatic Neonatal Seizures Followed by Febrile Status Epilepticus. Journal of Child Neurology, 2015, 30, 615-618.	0.7	9
43	Cannabidiol: Pharmacology and potential therapeutic role in epilepsy and other neuropsychiatric disorders. Epilepsia, 2014, 55, 791-802.	2.6	766
44	The case for assessing cannabidiol in epilepsy. Epilepsia, 2014, 55, 787-790.	2.6	68
45	Pontocerebellar hypoplasia type 6 caused by mutations in RARS2: definition of the clinical spectrum and molecular findings in five patients. Journal of Inherited Metabolic Disease, 2013, 36, 43-53.	1.7	70
46	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. European Journal of Human Genetics, 2013, 21, 1074-1078.	1.4	64
47	Genotypeâ€“phenotype correlations in neonatal epilepsies caused by mutations in the voltage sensor of K _v 7.2 potassium channel subunits. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 4386-4391.	3.3	154
48	Sleep-Wake Cycling in a Neonate Admitted to the NICU. Journal of Perinatal and Neonatal Nursing, 2013, 27, 263-273.	0.5	9
49	Gating Currents from Neuronal KV7 Channels Carrying BFNS-Causing Mutations in the S4 Segment of the Voltage Sensing Domain. Biophysical Journal, 2011, 100, 426a.	0.2	0
50	The Voltage-Sensing Domain of Kv7.2 Channels as a Molecular Target for Epilepsy-Causing Mutations and Anticonvulsants. Frontiers in Pharmacology, 2011, 2, 2.	1.6	24
51	Etiology of Perinatal Stroke; A Role for Prothrombotic Coagulation Factors?. Pediatric Research, 2011, 70, 215-215.	1.1	0
52	Childhood refractory focal epilepsy following acute febrile encephalopathy. European Journal of Neurology, 2011, 18, 952-961.	1.7	19
53	Seizures and Magnetic Resonance Imagingâ€“Detected Brain Injury in Newborns Cooled for Hypoxic-Ischemic Encephalopathy. Journal of Pediatrics, 2011, 159, 731-735.e1.	0.9	103
54	Early-onset seizure variant of Rett syndrome: Definition of the clinical diagnostic criteria. Brain and Development, 2010, 32, 17-24.	0.6	62

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55	Efficacy of levetiracetam in the treatment of drug-resistant Rett syndrome. Epilepsy Research, 2010, 88, 112-117.	0.8	18
56	Congenital central hypoventilation syndrome: genotypeâ€“phenotype correlation in parents of affected children carrying a PHOX2B expansion mutation. Clinical Genetics, 2010, 78, 289-293.	1.0	19
57	Neuronal potassium channel openers in the management of epilepsy: role and potential of retigabine. Clinical Pharmacology: Advances and Applications, 2010, 2, 225.	0.8	23
58	Dorsal Brain Stem Syndrome: MR Imaging Location of Brain Stem Tegmental Lesions in Neonates with Oral Motor Dysfunction. American Journal of Neuroradiology, 2010, 31, 1438-1442.	1.2	20
59	Synergistic neuroprotective therapies with hypothermia. Seminars in Fetal and Neonatal Medicine, 2010, 15, 293-298.	1.1	103
60	SCN1A duplications and deletions detected in Dravet syndrome: Implications for molecular diagnosis. Epilepsia, 2009, 50, 1670-1678.	2.6	152
61	Intravenous levetiracetam terminates refractory status epilepticus in two patients with migrating partial seizures in infancy. Epilepsy Research, 2009, 86, 66-71.	0.8	58
62	Variant late infantile ceroid lipofuscinoses associated with novel mutations in CLN6. Biochemical and Biophysical Research Communications, 2009, 379, 892-897.	1.0	45
63	The Usefulness of Near-Infrared Spectroscopy for Detecting and Monitoring Status Epilepticus After Pediatric Cardiac Surgery. Journal of Cardiothoracic and Vascular Anesthesia, 2009, 23, 668-671.	0.6	4
64	Gating currents from neuronal K(V)7.4 channels: general features and correlation with the ionic conductance. Channels, 2009, 3, 274-83.	1.5	22
65	Atypical Gating Of M-Type Potassium Channels Conferred by Mutations in Uncharged Residues in the S4 Region of KCNQ2 Causing Benign Familial Neonatal Convulsions. Journal of Neuroscience, 2007, 27, 4919-4928.	1.7	49
66	PHOX2B mutations and polyalanine expansions correlate with the severity of the respiratory phenotype and associated symptoms in both congenital and late onset Central Hypoventilation syndrome. Journal of Medical Genetics, 2004, 41, 373-380.	1.5	248
67	Long-term Effects of Status Epilepticus in the Immature Brain Are Specific for Age and Model. Epilepsia, 2003, 44, 518-528.	2.6	113
68	Seizure-Induced Changes in Place Cell Physiology: Relationship to Spatial Memory. Journal of Neuroscience, 2003, 23, 11505-11515.	1.7	111
69	Memory impairment following status epilepticus in immature rats: time-course and environmental effects. European Journal of Neuroscience, 2002, 16, 501-513.	1.2	101
70	hGFRÎ±-4: a new member of the GDNF receptor family and a candidate for NBIA. Pediatric Neurology, 2001, 25, 156-161.	1.0	8
71	Anticonvulsant action and long-term effects of gabapentin in the immature brain. Neuropharmacology, 2001, 40, 139-147.	2.0	75
72	Reduced Neurogenesis after Neonatal Seizures. Journal of Neuroscience, 2001, 21, 2094-2103.	1.7	224

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73	The long-term use of felbamate in children with severe refractory epilepsy. Epilepsy Research, 2001, 47, 1-7.	0.8	50
74	Timing of cognitive deficits following neonatal seizures: relationship to histological changes in the hippocampus. Developmental Brain Research, 2001, 131, 73-83.	2.1	102
75	Timing of ketogenic diet initiation in an experimental epilepsy model. Developmental Brain Research, 2000, 125, 131-138.	2.1	63
76	Long-term effects of neonatal seizures: a behavioral, electrophysiological, and histological study. Developmental Brain Research, 1999, 118, 99-107.	2.1	174
77	Vigabatrin Versus ACTH as First-Line Treatment for Infantile Spasms: A Randomized, Prospective Study. Epilepsia, 1997, 38, 1270-1274.	2.6	258
78	Homozygosity mapping of Hallervordenâ€“Spatz syndrome to chromosome 20p12.3â€“p13. Nature Genetics, 1996, 14, 479-481.	9.4	158
79	Familial White Matter Hypoplasia, Agenesis of the Corpus Callosum, Mental Retardation and Growth Deficiency: A New Distinctive Syndrome. Neuropediatrics, 1993, 24, 77-82.	0.3	12
80	Unusual cyclosporin related neurological complications in recipients of liver transplants.. Archives of Disease in Childhood, 1993, 68, 405-407.	1.0	34
81	136 SEIZURES ASSOCIATED WITH TOXIC LEVELS OF CYCLOSPORINE A IN LIVER TRANSPLANTED CHILDREN. Pediatric Research, 1991, 30, 650-650.	1.1	0
82	Adrenocorticotropic Hormone and Corticosteroids. , 0, , 411-419.		2