

# Maria Roberta Cilio

## List of Publications by Year in descending order

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Version: 2024-02-01

82  
papers

6,170  
citations

76326

40  
h-index

71685

76  
g-index

89  
all docs

89  
docs citations

89  
times ranked

6199  
citing authors



#	ARTICLE	IF	CITATIONS
1	Characteristics of Neonates with Cardiopulmonary Disease Who Experience Seizures: A Multicenter Study. <i>Journal of Pediatrics</i> , 2022, 242, 63-73.	1.8	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. <i>Epilepsia</i> , 2021, 62, 615-628.	5.1	158
3	Neonatal presentation of genetic epilepsies: Early differentiation from acute provoked seizures. <i>Epilepsia</i> , 2021, 62, 1907-1920.	5.1	32
4	Seizure Control in Neonates Undergoing Screening vs Confirmatory EEG Monitoring. <i>Neurology</i> , 2021, 97, e587-e596.	1.1	19
5	Disorders of Neuronal Migration/Organization Convey the Highest Risk of Neonatal Onset Epilepsy Compared to Other Congenital Brain Malformations. <i>Pediatric Neurology</i> , 2021, 127, 20-27.	2.1	2
6	Graph theory in paediatric epilepsy: A systematic review. <i>Dialogues in Clinical Neuroscience</i> , 2021, 23, 3-13.	3.7	15
7	Synthetic pharmaceutical grade cannabidiol for treatment of refractory infantile spasms: A multicenter phase-2 study. <i>Epilepsy and Behavior</i> , 2020, 102, 106826.	1.7	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 I <sup>2</sup> -Hydroxybutyrate. <i>Frontiers in Physiology</i> , 2020, 11, 1040.	2.8	7
9	Characterization of Death in Infants With Neonatal Seizures. <i>Pediatric Neurology</i> , 2020, 113, 21-25.	2.1	12
10	Response to cannabidiol in epilepsy of infancy with migrating focal seizures associated with KCNT1 mutations: An open-label, prospective, interventional study. <i>European Journal of Paediatric Neurology</i> , 2020, 25, 77-81.	1.6	13
11	Vomiting and retching as presenting signs of focal epilepsy in children. <i>Epileptic Disorders</i> , 2020, 22, 823-827.	1.3	1
12	Genetics of neonatal-onset epilepsies. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2019, 162, 415-433.	1.8	23
13	Neonatal Developmental and Epileptic Encephalopathies. <i>Seminars in Pediatric Neurology</i> , 2019, 32, 100770.	2.0	24
14	Toward the elimination of bias in Pediatric Research. <i>Pediatric Research</i> , 2019, 86, 680-681.	2.3	0
15	Autism and developmental disability caused by <i>KCNQ3</i> gain-of-function variants. <i>Annals of Neurology</i> , 2019, 86, 181-192.	5.3	73
16	Pharmacokinetics and Tolerability of Multiple Doses of Pharmaceutical-Grade Synthetic Cannabidiol in Pediatric Patients with Treatment-Resistant Epilepsy. <i>CNS Drugs</i> , 2019, 33, 593-604.	5.9	57
17	Neonatal seizures: Is there a relationship between ictal electroclinical features and etiology? A critical appraisal based on a systematic literature review. <i>Epilepsia Open</i> , 2019, 4, 10-29.	2.4	42
18	The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. <i>Epilepsia</i> , 2019, 60, 797-806.	5.1	52



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19	Response to antiseizure medications in neonates with acute symptomatic seizures. <i>Epilepsia</i> , 2019, 60, e20-e24.	5.1	33
20	Augmented Reticular Thalamic Bursting and Seizures in Scn1a-Dravet Syndrome. <i>Cell Reports</i> , 2019, 26, 54-64.e6.	6.4	44
21	Long-Term Safety, Tolerability, and Efficacy of Cannabidiol in Children with Refractory Epilepsy: Results from an Expanded Access Program in the US. <i>CNS Drugs</i> , 2019, 33, 47-60.	5.9	57
22	Neonatal-Onset Epilepsies. , 2019, , 131-140.		0
23	Neonatal epilepsies: Clinical management. <i>Seminars in Fetal and Neonatal Medicine</i> , 2018, 23, 204-212.	2.3	38
24	The ClinGen Epilepsy Gene Curation Expert Panel—Bridging the divide between clinical domain knowledge and formal gene curation criteria. <i>Human Mutation</i> , 2018, 39, 1476-1484.	2.5	33
25	Lack of response to quinidine in <i>KCNT1</i> -related neonatal epilepsy. <i>Epilepsia</i> , 2018, 59, 1889-1898.	5.1	53
26	Dynamic action potential clamp predicts functional separation in mild familial and severe de novo forms of <i>SCN2A</i> epilepsy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, E5516-E5525.	7.1	69
27	Predictive value of early EEG for seizures in neonates with hypoxic-ischemic encephalopathy undergoing therapeutic hypothermia. <i>Pediatric Research</i> , 2018, 84, 399-402.	2.3	22
28	Neonatal nonepileptic myoclonus is a prominent clinical feature of <i>KCNQ2</i> gain-of-function variants R201C and R201H. <i>Epilepsia</i> , 2017, 58, 436-445.	5.1	80
29	Seizures in Preterm Neonates: A Multicenter Observational Cohort Study. <i>Pediatric Neurology</i> , 2017, 72, 19-24.	2.1	83
30	A Distinctive Ictal Amplitude-Integrated Electroencephalography Pattern in Newborns with Neonatal Epilepsy Associated with <i>KCNQ2</i> Mutations. <i>Neonatology</i> , 2017, 112, 387-393.	2.0	44
31	Profile of neonatal epilepsies. <i>Neurology</i> , 2017, 89, 893-899.	1.1	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. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2016, 29, 1-4.	1.5	1
35	Rapid and safe response to low-dose carbamazepine in neonatal epilepsy. <i>Epilepsia</i> , 2016, 57, 2019-2030.	5.1	92
36	Current understanding and neurobiology of epileptic encephalopathies. <i>Neurobiology of Disease</i> , 2016, 92, 72-89.	4.4	71



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37	Cannabidiol in patients with treatment-resistant epilepsy: an open-label interventional trial. <i>Lancet Neurology</i> , The, 2016, 15, 270-278.	10.2	714
38	Evaluation of Presumably Disease Causing SCN1A Variants in a Cohort of Common Epilepsy Syndromes. <i>PLoS ONE</i> , 2016, 11, e0150426.	2.5	22
39	Transcriptional regulator PRDM12 is essential for human pain perception. <i>Nature Genetics</i> , 2015, 47, 803-808.	21.4	137
40	Dysregulation of locus coeruleus development in congenital central hypoventilation syndrome. <i>Acta Neuropathologica</i> , 2015, 130, 171-183.	7.7	45
41	Early and effective treatment of <i>KCNQ2</i> encephalopathy. <i>Epilepsia</i> , 2015, 56, 685-691.	5.1	229
42	Symptomatic Neonatal Seizures Followed by Febrile Status Epilepticus. <i>Journal of Child Neurology</i> , 2015, 30, 615-618.	1.4	9
43	Cannabidiol: Pharmacology and potential therapeutic role in epilepsy and other neuropsychiatric disorders. <i>Epilepsia</i> , 2014, 55, 791-802.	5.1	766
44	The case for assessing cannabidiol in epilepsy. <i>Epilepsia</i> , 2014, 55, 787-790.	5.1	68
45	Pontocerebellar hypoplasia type 6 caused by mutations in <i>RARS2</i> : definition of the clinical spectrum and molecular findings in five patients. <i>Journal of Inherited Metabolic Disease</i> , 2013, 36, 43-53.	3.6	70
46	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. <i>European Journal of Human Genetics</i> , 2013, 21, 1074-1078.	2.8	64
47	Genotype-phenotype correlations in neonatal epilepsies caused by mutations in the voltage sensor of K <sub>v</sub> 7.2 potassium channel subunits. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 4386-4391.	7.1	154
48	Sleep-Wake Cycling in a Neonate Admitted to the NICU. <i>Journal of Perinatal and Neonatal Nursing</i> , 2013, 27, 263-273.	0.7	9
49	Gating Currents from Neuronal KV7 Channels Carrying BFNS-Causing Mutations in the S4 Segment of the Voltage Sensing Domain. <i>Biophysical Journal</i> , 2011, 100, 426a.	0.5	0
50	The Voltage-Sensing Domain of Kv7.2 Channels as a Molecular Target for Epilepsy-Causing Mutations and Anticonvulsants. <i>Frontiers in Pharmacology</i> , 2011, 2, 2.	3.5	24
51	Etiology of Perinatal Stroke; A Role for Prothrombotic Coagulation Factors?. <i>Pediatric Research</i> , 2011, 70, 215-215.	2.3	0
52	Childhood refractory focal epilepsy following acute febrile encephalopathy. <i>European Journal of Neurology</i> , 2011, 18, 952-961.	3.3	19
53	Seizures and Magnetic Resonance Imaging-Detected Brain Injury in Newborns Cooled for Hypoxic-Ischemic Encephalopathy. <i>Journal of Pediatrics</i> , 2011, 159, 731-735.e1.	1.8	103
54	Early-onset seizure variant of Rett syndrome: Definition of the clinical diagnostic criteria. <i>Brain and Development</i> , 2010, 32, 17-24.	1.1	62



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55	Efficacy of levetiracetam in the treatment of drug-resistant Rett syndrome. <i>Epilepsy Research</i> , 2010, 88, 112-117.	1.6	18
56	Congenital central hypoventilation syndrome: genotypeâ€“phenotype correlation in parents of affected children carrying a <i>PHOX2B</i> expansion mutation. <i>Clinical Genetics</i> , 2010, 78, 289-293.	2.0	19
57	Neuronal potassium channel openers in the management of epilepsy: role and potential of retigabine. <i>Clinical Pharmacology: Advances and Applications</i> , 2010, 2, 225.	1.2	23
58	Dorsal Brain Stem Syndrome: MR Imaging Location of Brain Stem Tegmental Lesions in Neonates with Oral Motor Dysfunction. <i>American Journal of Neuroradiology</i> , 2010, 31, 1438-1442.	2.4	20
59	Synergistic neuroprotective therapies with hypothermia. <i>Seminars in Fetal and Neonatal Medicine</i> , 2010, 15, 293-298.	2.3	103
60	<i>SCN1A</i> duplications and deletions detected in Dravet syndrome: Implications for molecular diagnosis. <i>Epilepsia</i> , 2009, 50, 1670-1678.	5.1	152
61	Intravenous levetiracetam terminates refractory status epilepticus in two patients with migrating partial seizures in infancy. <i>Epilepsy Research</i> , 2009, 86, 66-71.	1.6	58
62	Variant late infantile ceroid lipofuscinoses associated with novel mutations in <i>CLN6</i> . <i>Biochemical and Biophysical Research Communications</i> , 2009, 379, 892-897.	2.1	45
63	The Usefulness of Near-Infrared Spectroscopy for Detecting and Monitoring Status Epilepticus After Pediatric Cardiac Surgery. <i>Journal of Cardiothoracic and Vascular Anesthesia</i> , 2009, 23, 668-671.	1.3	4
64	Gating currents from neuronal K(V)7.4 channels: general features and correlation with the ionic conductance. <i>Channels</i> , 2009, 3, 274-83.	2.8	22
65	Atypical Gating Of M-Type Potassium Channels Conferred by Mutations in Uncharged Residues in the S4 Region of <i>KCNQ2</i> Causing Benign Familial Neonatal Convulsions. <i>Journal of Neuroscience</i> , 2007, 27, 4919-4928.	3.6	49
66	<i>PHOX2B</i> mutations and polyalanine expansions correlate with the severity of the respiratory phenotype and associated symptoms in both congenital and late onset Central Hypoventilation syndrome. <i>Journal of Medical Genetics</i> , 2004, 41, 373-380.	3.2	248
67	Long-term Effects of Status Epilepticus in the Immature Brain Are Specific for Age and Model. <i>Epilepsia</i> , 2003, 44, 518-528.	5.1	113
68	Seizure-Induced Changes in Place Cell Physiology: Relationship to Spatial Memory. <i>Journal of Neuroscience</i> , 2003, 23, 11505-11515.	3.6	111
69	Memory impairment following status epilepticus in immature rats: time-course and environmental effects. <i>European Journal of Neuroscience</i> , 2002, 16, 501-513.	2.6	101
70	hGFRÎ±4: a new member of the GDNF receptor family and a candidate for NBIA. <i>Pediatric Neurology</i> , 2001, 25, 156-161.	2.1	8
71	Anticonvulsant action and long-term effects of gabapentin in the immature brain. <i>Neuropharmacology</i> , 2001, 40, 139-147.	4.1	75
72	Reduced Neurogenesis after Neonatal Seizures. <i>Journal of Neuroscience</i> , 2001, 21, 2094-2103.	3.6	224



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73	The long-term use of felbamate in children with severe refractory epilepsy. Epilepsy Research, 2001, 47, 1-7.	1.6	50
74	Timing of cognitive deficits following neonatal seizures: relationship to histological changes in the hippocampus. Developmental Brain Research, 2001, 131, 73-83.	1.7	102
75	Timing of ketogenic diet initiation in an experimental epilepsy model. Developmental Brain Research, 2000, 125, 131-138.	1.7	63
76	Long-term effects of neonatal seizures: a behavioral, electrophysiological, and histological study. Developmental Brain Research, 1999, 118, 99-107.	1.7	174
77	Vigabatrin Versus ACTH as First-Line Treatment for Infantile Spasms: A Randomized, Prospective Study. Epilepsia, 1997, 38, 1270-1274.	5.1	258
78	Homozygosity mapping of Hallervordenâ€“Spatz syndrome to chromosome 20p12.3â€“p13. Nature Genetics, 1996, 14, 479-481.	21.4	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.	0.6	12
80	Unusual cyclosporin related neurological complications in recipients of liver transplants.. Archives of Disease in Childhood, 1993, 68, 405-407.	1.9	34
81	136 SEIZURES ASSOCIATED WITH TOXIC LEVELS OF CYCLOSPORINE A IN LIVER TRANSPLANTED CHILDREN. Pediatric Research, 1991, 30, 650-650.	2.3	0
82	Adrenocorticotrophic Hormone and Corticosteroids. , 0, , 411-419.		2