Mario Mastrangelo

List of Publications by Year
in descending order

64	1,300	471509 17	395702 33
papers	citations	h-index	g-index

68	68	68	1595
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Acute ischemic stroke in childhood: a comprehensive review. European Journal of Pediatrics, 2022, 181, 45-58.	2.7	15
2	Expanding the genetic and phenotypic spectrum of <scp>CHD2</scp>â€related disease: From early neurodevelopmental disorders to adultâ€onset epilepsy. American Journal of Medical Genetics, Part A, 2022, 188, 522-533.	1.2	13
3	Stroke and stroke-like episodes in inborn errors of metabolism: Pathophysiological and clinical implications. Molecular Genetics and Metabolism, 2022, 135, 3-14.	1.1	10
4	Loss of Continuity of Care in Pediatric Neurology Services during COVID-19 Lockdown: An Additional Stressor for Parents. Children, 2022, 9, 867.	1.5	2
5	Laparoscopic patch repair of a Morgagni hernia in Menkes disease. Asian Journal of Endoscopic Surgery, 2021, 14, 553-556.	0.9	1
6	Epilepsy in inherited neurotransmitter disorders: Spotlights on pathophysiology and clinical management. Metabolic Brain Disease, 2021, 36, 29-43.	2.9	10
7	TSC1 as a Novel Gene for Sleep-Related Hypermotor Epilepsy: A Child with a Mild Phenotype of Tuberous Sclerosis. Neuropediatrics, 2021, 52, 146-149.	0.6	2
8	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. Brain, 2021, 144, 1435-1450.	7.6	35
9	Contemporary onset of atypical chronic inflammatory demyelinating polyradiculoneuropathy and type 1 diabetes in an adolescent. Diabetic Medicine, 2021, 38, e14563.	2.3	0
10	Management of Neurological Emergencies in Children: An Updated Overview. Neuropediatrics, 2021, 52, 242-251.	0.6	5
11	Assessment of intellectual impairment, healthâ€related quality of life, and behavioral phenotype in patients with neurotransmitter related disorders: Data from the <scp>iNTD</scp> registry. Journal of Inherited Metabolic Disease, 2021, 44, 1489-1502.	3.6	7
12	Compound heterozygosis in AADC deficiency: A complex phenotype dissected through comparison among heterodimeric and homodimeric AADC proteins. Molecular Genetics and Metabolism, 2021, 134, 147-155.	1.1	10
13	Insights into the expanding phenotypic spectrum of inherited disorders of biogenic amines. Nature Communications, 2021, 12, 5529.	12.8	21
14	KCNQ2 encephalopathy manifesting with Rett-like features. Neurology: Genetics, 2020, 6, e510.	1.9	3
15	Towards an evidence-based treatment of pediatric status epilepticus: still a mountain to climb. Seizure: the Journal of the British Epilepsy Association, 2020, 83, 143-144.	2.0	0
16	Neurodevelopmental Impairment As the Main Phenotypic Hallmark Associated with the Translocation t(7;10)(7p22.3;q26.11). Journal of Pediatric Genetics, 2020, 11, 68-73.	0.7	0
17	<scp>AADC</scp> deficiency from infancy to adulthood: Symptoms and developmental outcome in an international cohort of 63 patients. Journal of Inherited Metabolic Disease, 2020, 43, 1121-1130.	3.6	59
18	Consensus guideline for the diagnosis and treatment of tetrahydrobiopterin (BH4) deficiencies. Orphanet Journal of Rare Diseases, 2020, 15, 126.	2.7	85

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19	Update on the treatment of vitamin B6 dependent epilepsies. Expert Review of Neurotherapeutics, 2019, 19, 1135-1147.	2.8	13
20	A novel developmental encephalopathy with epilepsy and hyperkinetic movement disorders associated with a deletion of the sodium channel gene cluster on chromosome 2q24.3. Parkinsonism and Related Disorders, 2019, 68, 1-3.	2.2	2
21	Broadening phenotype of adenylosuccinate lyase deficiency: A novel clinical pattern resembling neuronal ceroid lipofuscinosis. Molecular Genetics and Metabolism Reports, 2019, 21, 100502.	1.1	3
22	Clinical approach to neurodegenerative disorders in childhood: an updated overview. Acta Neurologica Belgica, 2019, 119, 511-521.	1.1	11
23	Epilepsy in children with type 1 diabetes mellitus: Pathophysiological basis and clinical hallmarks. European Journal of Paediatric Neurology, 2019, 23, 240-247.	1.6	17
24	<scp>AP</scp>1S2â€truncating variant in a patient with severe neurodevelopmental disorder and cerebral folate deficiency. Acta Paediatrica, International Journal of Paediatrics, 2019, 108, 564-565.	1.5	2
25	Progressive myoclonus epilepsy and ceroidolipofuscinosis 14: The multifaceted phenotypic spectrum of KCTD7-related disorders. European Journal of Medical Genetics, 2019, 62, 103591.	1.3	15
26	Early post-cooling brain magnetic resonance for the prediction of neurodevelopmental outcome in newborns with hypoxicâ€“ischemic encephalopathy. Journal of Pediatric Neurosciences, 2019, 14, 191.	0.3	3
27	Successful Pregnancy in a Patient with Lâ€Amino Acid Decarboxylase Deficiency: Therapeutic Management and Clinical Outcome. Movement Disorders Clinical Practice, 2018, 5, 446-447.	1.5	7
28	Photosensitivity as an early marker of epileptic and developmental encephalopathies. Epilepsia, 2018, 59, 1086-1087.	5.1	3
29	Pyridoxine-dependent epilepsies: an observational study on clinical, diagnostic, therapeutic and prognostic features in a pediatric cohort. Metabolic Brain Disease, 2018, 33, 261-269.	2.9	18
30	PRICKLE1â€related early onset epileptic encephalopathy. American Journal of Medical Genetics, Part A, 2018, 176, 2841-2845.	1.2	12
31	Actual insights into treatable inborn errors of metabolism causing epilepsy. Journal of Pediatric Neurosciences, 2018, 13, 13.	0.3	10
32	Minor Head Trauma in the Pediatric Emergency Department: Decision Making Nodes. Current Pediatric Reviews, 2018, 13, 92-99.	0.8	5
33	Consensus guideline for the diagnosis and treatment of aromatic l-amino acid decarboxylase (AADC) deficiency. Orphanet Journal of Rare Diseases, 2017, 12, 12.	2.7	172
34	Acute diplopia in the pediatric Emergency Department. A cohort multicenter Italian study. European Journal of Paediatric Neurology, 2017, 21, 722-729.	1.6	3
35	Lennoxâ€“Gastaut Syndrome: A State of the Art Review. Neuropediatrics, 2017, 48, 143-151.	0.6	54
36	Neuromotor and cognitive outcomes of early treatment in tyrosine hydroxylase deficiency type B. Neurology, 2017, 88, 501-502.	1.1	10

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37	Pyridoxine-5â€²-phosphate oxidase (Pnpo) deficiency: Clinical and biochemical alterations associated with the C.347g > A (P.Â·Arg116gln) mutation. Molecular Genetics and Metabolism, 2017, 122, 135-142.	1.1	30
38	Biochemical data from the characterization of a new pathogenic mutation of human pyridoxine-5'-phosphate oxidase (PNPO). Data in Brief, 2017, 15, 868-875.	1.0	14
39	Epilepsy in KCNH1â€related syndromes. Epileptic Disorders, 2016, 18, 123-136.	1.3	34
40	The International Working Group on Neurotransmitter related Disorders (iNTD): A worldwide research project focused on primary and secondary neurotransmitter disorders. Molecular Genetics and Metabolism Reports, 2016, 9, 61-66.	1.1	48
41	The Spectrum of Early Movement Disorders in Congenital Defects of Biogenic Amine Metabolism. Journal of Pediatric Neurology, 2015, 13, 213-224.	0.2	2
42	Novel Genes of Early-Onset Epileptic Encephalopathies: From Genotype to Phenotypes. Pediatric Neurology, 2015, 53, 119-129.	2.1	31
43	Neurological â€œsoft signsâ€•in children and adolescents. Journal of Pediatric Neurology, 2015, 03, 123-125.	0.2	0
44	The outcome of white matter abnormalities in early treated phenylketonuric patients: A retrospective longitudinal long-term study. Molecular Genetics and Metabolism, 2015, 116, 171-177.	1.1	27
45	Report of Two Never Treated Adult Sisters with Aromatic l-Amino Acid Decarboxylase Deficiency: A Portrait of the Natural History of the Disease or an Expanding Phenotype?. JIMD Reports, 2014, 15, 39-45.	1.5	29
46	Teaching Video Neuro Images : Clinical course of infantile ascending hereditary spastic paralysis. Neurology, 2014, 82, e61.	1.1	0
47	Actual insights into the clinical management of febrile seizures. European Journal of Pediatrics, 2014, 173, 977-982.	2.7	14
48	Genetic background of febrile seizures. Reviews in the Neurosciences, 2014, 25, 129-61.	2.9	40
49	Inborn errors of creatine metabolism and epilepsy. Epilepsia, 2013, 54, 217-227.	5.1	54
50	Transdermal rotigotine in the treatment of aromatic <scp>L</scp>â€amino acid decarboxylase deficiency. Movement Disorders, 2013, 28, 556-557.	3.9	18
51	Mitochondrial Neurogastrointestinal Encephalomyopathy: Novel Pathogenic Mutations in Thymidine Phosphorylase Gene in Two Italian Brothers. Neuropediatrics, 2012, 43, 201-208.	0.6	13
52	A new form of cerebral folate deficiency with severe selfâ€injurious behaviour. Acta Paediatrica, International Journal of Paediatrics, 2012, 101, e482-3.	1.5	4
53	Genes of Early-Onset Epileptic Encephalopathies: From Genotype to Phenotype. Pediatric Neurology, 2012, 46, 24-31.	2.1	114
54	Diagnostic work-up and therapeutic options in management of pediatric status epilepticus. World Journal of Pediatrics, 2012, 8, 109-115.	1.8	14

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55	A diagnostic algorithm for the evaluation of early onset genetic-metabolic epileptic encephalopathies. European Journal of Paediatric Neurology, 2012, 16, 179-191.	1.6	26
56	Neurocardiogenic Syncope and Epilepsy in Pediatric Age. Pediatric Emergency Care, 2011, 27, 36-39.	0.9	2
57	Bilateral (opercular and paracentral lobular) polymicrogyria and neurofibromatosis type 1. American Journal of Medical Genetics, Part A, 2011, 155, 582-585.	1.2	13
58	Eponym. European Journal of Pediatrics, 2010, 169, 919-924.	2.7	18
59	New trends in neuronal migration disorders. European Journal of Paediatric Neurology, 2010, 14, 1-12.	1.6	70
60	Complex epileptic (Foixâ€“Chavanyâ€“Marie like) syndrome in a child with neurofibromatosis type 1 (NF1) and bilateral (opercular and paracentral) polymicrogyria. Acta Paediatrica, International Journal of Paediatrics, 2009, 98, 760-762.	1.5	6
61	Stupor and Fast Activity on Electroencephalography in a Child Treated With Valproate. Pediatric Neurology, 2009, 41, 53-56.	2.1	3
62	Early-onset Hereditary Neuropathy with Liability to Pressure Palsy. Neuropediatrics, 2007, 38, 50-54.	0.6	16
63	Bilateral perysilvian polymicrogyria in Chiari I malformation. Child's Nervous System, 2006, 22, 1635-1637.	1.1	7
64	Presenting Patterns of Genetically Determined Developmental Encephalopathies With Epilepsy and Movement Disorders: A Single Tertiary Center Retrospective Cohort Study. Frontiers in Neurology, 0, 13, .	2.4	3