

Mario Mastrangelo

List of Publications by Year in descending order

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Version: 2024-02-01

64
papers

1,300
citations

471509

17
h-index

395702

33
g-index

68
all docs

68
docs citations

68
times ranked

1595
citing authors

#	ARTICLE	IF	CITATIONS
1	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
2	Genes of Early-Onset Epileptic Encephalopathies: From Genotype to Phenotype. Pediatric Neurology, 2012, 46, 24-31.	2.1	114
3	Consensus guideline for the diagnosis and treatment of tetrahydrobiopterin (BH4) deficiencies. Orphanet Journal of Rare Diseases, 2020, 15, 126.	2.7	85
4	New trends in neuronal migration disorders. European Journal of Paediatric Neurology, 2010, 14, 1-12.	1.6	70
5	<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
6	Inborn errors of creatine metabolism and epilepsy. Epilepsia, 2013, 54, 217-227.	5.1	54
7	Lennoxâ€Gastaut Syndrome: A State of the Art Review. Neuropediatrics, 2017, 48, 143-151.	0.6	54
8	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
9	Genetic background of febrile seizures. Reviews in the Neurosciences, 2014, 25, 129-61.	2.9	40
10	<i>ATP1A2</i> and <i>ATP1A3</i> associated early profound epileptic encephalopathy and polymicrogyria. Brain, 2021, 144, 1435-1450.	7.6	35
11	Epilepsy in <i>KCNH1</i>-related syndromes. Epileptic Disorders, 2016, 18, 123-136.	1.3	34
12	Novel Genes of Early-Onset Epileptic Encephalopathies: From Genotype to Phenotypes. Pediatric Neurology, 2015, 53, 119-129.	2.1	31
13	Pyridoxine-5â€²-phosphate oxidase (Pnpo) deficiency: Clinical and biochemical alterations associated with the C.347g > A (P.ÃArg116gln) mutation. Molecular Genetics and Metabolism, 2017, 122, 135-142.	1.1	30
14	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
15	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
16	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
17	Insights into the expanding phenotypic spectrum of inherited disorders of biogenic amines. Nature Communications, 2021, 12, 5529.	12.8	21
18	Eponym. European Journal of Pediatrics, 2010, 169, 919-924.	2.7	18

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19	Transdermal rotigotine in the treatment of aromatic <sc>L</sc>-amino acid decarboxylase deficiency. <i>Movement Disorders</i> , 2013, 28, 556-557.	3.9	18
20	Pyridoxine-dependent epilepsies: an observational study on clinical, diagnostic, therapeutic and prognostic features in a pediatric cohort. <i>Metabolic Brain Disease</i> , 2018, 33, 261-269.	2.9	18
21	Epilepsy in children with type 1 diabetes mellitus: Pathophysiological basis and clinical hallmarks. <i>European Journal of Paediatric Neurology</i> , 2019, 23, 240-247.	1.6	17
22	Early-onset Hereditary Neuropathy with Liability to Pressure Palsy. <i>Neuropediatrics</i> , 2007, 38, 50-54.	0.6	16
23	Progressive myoclonus epilepsy and ceroidlipofuscinosis 14: The multifaceted phenotypic spectrum of KCTD7-related disorders. <i>European Journal of Medical Genetics</i> , 2019, 62, 103591.	1.3	15
24	Acute ischemic stroke in childhood: a comprehensive review. <i>European Journal of Pediatrics</i> , 2022, 181, 45-58.	2.7	15
25	Diagnostic work-up and therapeutic options in management of pediatric status epilepticus. <i>World Journal of Pediatrics</i> , 2012, 8, 109-115.	1.8	14
26	Actual insights into the clinical management of febrile seizures. <i>European Journal of Pediatrics</i> , 2014, 173, 977-982.	2.7	14
27	Biochemical data from the characterization of a new pathogenic mutation of human pyridoxine-5'-phosphate oxidase (PNPO). <i>Data in Brief</i> , 2017, 15, 868-875.	1.0	14
28	Bilateral (opercular and paracentral lobular) polymicrogyria and neurofibromatosis type 1. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 582-585.	1.2	13
29	Mitochondrial Neurogastrointestinal Encephalomyopathy: Novel Pathogenic Mutations in Thymidine Phosphorylase Gene in Two Italian Brothers. <i>Neuropediatrics</i> , 2012, 43, 201-208.	0.6	13
30	Update on the treatment of vitamin B6 dependent epilepsies. <i>Expert Review of Neurotherapeutics</i> , 2019, 19, 1135-1147.	2.8	13
31	Expanding the genetic and phenotypic spectrum of <sc><i>CHD2</i></sc>-related disease: From early neurodevelopmental disorders to adult-onset epilepsy. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 522-533.	1.2	13
32	<i>PRICKLE1</i>-related early onset epileptic encephalopathy. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2841-2845.	1.2	12
33	Clinical approach to neurodegenerative disorders in childhood: an updated overview. <i>Acta Neurologica Belgica</i> , 2019, 119, 511-521.	1.1	11
34	Neuromotor and cognitive outcomes of early treatment in tyrosine hydroxylase deficiency type B. <i>Neurology</i> , 2017, 88, 501-502.	1.1	10
35	Epilepsy in inherited neurotransmitter disorders: Spotlights on pathophysiology and clinical management. <i>Metabolic Brain Disease</i> , 2021, 36, 29-43.	2.9	10
36	Compound heterozygosis in AADC deficiency: A complex phenotype dissected through comparison among heterodimeric and homodimeric AADC proteins. <i>Molecular Genetics and Metabolism</i> , 2021, 134, 147-155.	1.1	10

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37	Actual insights into treatable inborn errors of metabolism causing epilepsy. <i>Journal of Pediatric Neurosciences</i> , 2018, 13, 13.	0.3	10
38	Stroke and stroke-like episodes in inborn errors of metabolism: Pathophysiological and clinical implications. <i>Molecular Genetics and Metabolism</i> , 2022, 135, 3-14.	1.1	10
39	Bilateral perysylvian polymicrogyria in Chiari I malformation. <i>Child's Nervous System</i> , 2006, 22, 1635-1637.	1.1	7
40	Successful Pregnancy in a Patient with Lâ€Amino Acid Decarboxylase Deficiency: Therapeutic Management and Clinical Outcome. <i>Movement Disorders Clinical Practice</i> , 2018, 5, 446-447.	1.5	7
41	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. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 1489-1502.	3.6	7
42	Complex epileptic (Foixâ€Chavanyâ€Marie like) syndrome in a child with neurofibromatosis type 1 (NF1) and bilateral (opercular and paracentral) polymicrogyria. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2009, 98, 760-762.	1.5	6
43	Management of Neurological Emergencies in Children: An Updated Overview. <i>Neuropediatrics</i> , 2021, 52, 242-251.	0.6	5
44	Minor Head Trauma in the Pediatric Emergency Department: Decision Making Nodes. <i>Current Pediatric Reviews</i> , 2018, 13, 92-99.	0.8	5
45	A new form of cerebral folate deficiency with severe selfâ€injurious behaviour. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2012, 101, e482-3.	1.5	4
46	Stupor and Fast Activity on Electroencephalography in a Child Treated With Valproate. <i>Pediatric Neurology</i> , 2009, 41, 53-56.	2.1	3
47	Acute diplopia in the pediatric Emergency Department. A cohort multicenter Italian study. <i>European Journal of Paediatric Neurology</i> , 2017, 21, 722-729.	1.6	3
48	Photosensitivity as an early marker of epileptic and developmental encephalopathies. <i>Epilepsia</i> , 2018, 59, 1086-1087.	5.1	3
49	Broadening phenotype of adenylosuccinate lyase deficiency: A novel clinical pattern resembling neuronal ceroid lipofuscinosis. <i>Molecular Genetics and Metabolism Reports</i> , 2019, 21, 100502.	1.1	3
50	KCNQ2 encephalopathy manifesting with Rett-like features. <i>Neurology: Genetics</i> , 2020, 6, e510.	1.9	3
51	Early post-cooling brain magnetic resonance for the prediction of neurodevelopmental outcome in newborns with hypoxicâ€ischemic encephalopathy. <i>Journal of Pediatric Neurosciences</i> , 2019, 14, 191.	0.3	3
52	Presenting Patterns of Genetically Determined Developmental Encephalopathies With Epilepsy and Movement Disorders: A Single Tertiary Center Retrospective Cohort Study. <i>Frontiers in Neurology</i> , 0, 13, .	2.4	3
53	Neurocardiogenic Syncope and Epilepsy in Pediatric Age. <i>Pediatric Emergency Care</i> , 2011, 27, 36-39.	0.9	2
54	The Spectrum of Early Movement Disorders in Congenital Defects of Biogenic Amine Metabolism. <i>Journal of Pediatric Neurology</i> , 2015, 13, 213-224.	0.2	2

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55	A novel developmental encephalopathy with epilepsy and hyperkinetic movement disorders associated with a deletion of the sodium channel gene cluster on chromosome 2q24.3. <i>Parkinsonism and Related Disorders</i> , 2019, 68, 1-3.	2.2	2
56	<i>AP1S2</i> truncating variant in a patient with severe neurodevelopmental disorder and cerebral folate deficiency. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2019, 108, 564-565.	1.5	2
57	TSC1 as a Novel Gene for Sleep-Related Hypermotor Epilepsy: A Child with a Mild Phenotype of Tuberous Sclerosis. <i>Neuropediatrics</i> , 2021, 52, 146-149.	0.6	2
58	Loss of Continuity of Care in Pediatric Neurology Services during COVID-19 Lockdown: An Additional Stressor for Parents. <i>Children</i> , 2022, 9, 867.	1.5	2
59	Laparoscopic patch repair of a Morgagni hernia in Menkes disease. <i>Asian Journal of Endoscopic Surgery</i> , 2021, 14, 553-556.	0.9	1
60	Teaching Video Neuro Images: Clinical course of infantile ascending hereditary spastic paralysis. <i>Neurology</i> , 2014, 82, e61.	1.1	0
61	Neurological "soft signs" in children and adolescents. <i>Journal of Pediatric Neurology</i> , 2015, 03, 123-125.	0.2	0
62	Towards an evidence-based treatment of pediatric status epilepticus: still a mountain to climb. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2020, 83, 143-144.	2.0	0
63	Neurodevelopmental Impairment As the Main Phenotypic Hallmark Associated with the Translocation t(7;10)(7p22.3;q26.11). <i>Journal of Pediatric Genetics</i> , 2020, 11, 68-73.	0.7	0
64	Contemporary onset of atypical chronic inflammatory demyelinating polyradiculoneuropathy and type 1 diabetes in an adolescent. <i>Diabetic Medicine</i> , 2021, 38, e14563.	2.3	0