Mary Kay Koenig

List of Publications by Year
in descending order

39 papers	2,747 citations	331670 21 h-index	361022 35 g-index
41 all docs	41 docs citations	41 times ranked	4510 citing authors

#	Article	IF	Citations
1	Hypoglycemia in mitochondrial disorders. Mitochondrion, 2021, 58, 179-183.	3.4	6
2	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
3	Two different genetic etiologies for tuberous sclerosis complex (TSC) in a single family. Molecular Genetics & Genomic Medicine, 2020, 8, e1296.	1.2	3
4	Safety and efficacy of omaveloxolone in patients with mitochondrial myopathy. Neurology, 2020, 94, e687-e698.	1.1	38
5	Vulnerability of pediatric patients with mitochondrial disease to vaccine-preventable diseases. Journal of Allergy and Clinical Immunology: in Practice, 2019, 7, 2415-2418.e3.	3.8	16
6	Diagnosis of â€˜possibleâ€™ mitochondrial disease: an existential crisis. Journal of Medical Genetics, 2019, 56, 123-130.	3.2	42
7	Genomic deletions upstream of lamin B1 lead to atypical autosomal dominant leukodystrophy. Neurology: Genetics, 2019, 5, e305.	1.9	16
8	Expanding the Phenotypic Spectrum of CACNA1H Mutations. Pediatric Neurology, 2019, 93, 50-55.	2.1	21
9	Metabolomics Profile in ABAT Deficiency Pre- and Post-treatment. JIMD Reports, 2018, 43, 13-17.	1.5	5
10	Reply to the Letter to the Editor by Josef Finsterer and Sinda Zarrouk-Mahjoub. Pediatric Neurology, 2018, 78, e11.	2.1	0
11	Treatment of Disfiguring Cutaneous Lesions in Neurofibromatosis-1 with Everolimus: A Phase II, Open-Label, Single-Arm Trial. Drugs in R and D, 2018, 18, 295-302.	2.2	17
12	Efficacy and Safety of Topical Rapamycin in Patients With Facial Angiofibromas Secondary to Tuberous Sclerosis Complex. JAMA Dermatology, 2018, 154, 773.	4.1	71
13	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
14	Phenotype of GABA-transaminase deficiency. Neurology, 2017, 88, 1919-1924.	1.1	49
15	Involvement of Cerebellum in Leigh Syndrome: Case Report and Review of the Literature. Pediatric Neurology, 2017, 74, 97-99.	2.1	8
16	Cytochrome c Oxidase Activity Is a Metabolic Checkpoint that Regulates Cell Fate Decisions During T Cell Activation and Differentiation. Cell Metabolism, 2017, 25, 1254-1268.e7.	16.2	125
17	Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. Genetics in Medicine, 2017, 19, 1380-1397.	2.4	173
18	Response to Newman et al.. Genetics in Medicine, 2017, 19, 1380-1380.	2.4	3

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19	Mitochondrial Disorder Aggravated by Metoprolol. Case Reports in Pediatrics, 2016, 2016, 1-3.	0.4	5
20	Nitric Oxide Deficiency Triggering Strokelike Episodesâ€"Reply. JAMA Neurology, 2016, 73, 1030.	9.0	0
21	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. American Journal of Human Genetics, 2016, 98, 347-357.	6.2	98
22	Recommendations for the Management of Strokelike Episodes in Patients With Mitochondrial Encephalomyopathy, Lactic Acidosis, and Strokelike Episodes. JAMA Neurology, 2016, 73, 591.	9.0	94
23	The neuroimaging of Leigh syndrome: case series and review of the literature. Pediatric Radiology, 2016, 46, 443-451.	2.0	64
24	Diagnosis and management of mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. Genetics in Medicine, 2015, 17, 689-701.	2.4	414
25	Identification of HIBCH Gene Mutations Causing Autosomal Recessive Leigh Syndrome: A Gene Involved in Valine Metabolism. Pediatric Neurology, 2015, 52, 361-365.	2.1	28
26	Sleep Disordered Breathing in Children with Mitochondrial Disease. Pulmonary Medicine, 2014, 2014, 1-8.	1.9	9
27	Practice patterns of mitochondrial disease physicians in North America. Part 1: Diagnostic and clinical challenges. Mitochondrion, 2014, 14, 26-33.	3.4	36
28	Seizure semiology and <scp>EEG</scp> findings in mitochondrial diseases. Epilepsia, 2014, 55, 707-712.	5.1	37
29	Tuberous Sclerosis Complex Surveillance and Management: Recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. Pediatric Neurology, 2013, 49, 255-265.	2.1	693
30	Practice patterns of mitochondrial disease physicians in North America. Part 2: treatment, care and management. Mitochondrion, 2013, 13, 681-687.	3.4	38
31	The Many Guises of Mitochondrial Disease. Neurographics, 2013, 3, 2-10.	0.1	0
32	Decreased Exhaled Nitric Oxide Levels in Patients with Mitochondrial Disorders. Open Respiratory Medicine Journal, 2013, 7, 67-70.	0.4	0
33	Mitochondrial cardiomyopathy: pathophysiology, diagnosis, and management. Texas Heart Institute Journal, 2013, 40, 385-94.	0.3	138
34	Impaired Gastric Emptying and Small Bowel Transit in Children With Mitochondrial Disorders. Journal of Pediatric Gastroenterology and Nutrition, 2012, 55, 194-199.	1.8	20
35	Topical Rapamycin Therapy to Alleviate the Cutaneous Manifestations of Tuberous Sclerosis Complex. Drugs in R and D, 2012, 12, 121-126.	2.2	120
36	Presentation and Diagnosis of Mitochondrial Disorders in Children. Pediatric Neurology, 2008, 38, 305-313.	2.1	133

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37	Regression of Subependymal Giant Cell Astrocytoma With Rapamycin in Tuberous Sclerosis Complex. Journal of Child Neurology, 2008, 23, 1238-1239.	1.4	80
38	Juvenile Onset Central Nervous System Folate Deficiency and Rheumatoid Arthritis. Journal of Child Neurology, 2008, 23, 106-107.	1.4	13
39	Central Nervous System Complications of Blastic Hyperleukocytosis in Childhood Acute Lymphoblastic Leukemia: Diagnostic and Prognostic Implications. Journal of Child Neurology, 2008, 23, 1347-1352.	1.4	17