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	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
2	Diagnosis and management of mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. Genetics in Medicine, 2015, 17, 689-701.	2.4	414
3	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
4	Mitochondrial cardiomyopathy: pathophysiology, diagnosis, and management. Texas Heart Institute Journal, 2013, 40, 385-94.	0.3	138
5	Presentation and Diagnosis of Mitochondrial Disorders in Children. Pediatric Neurology, 2008, 38, 305-313.	2.1	133
6	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
7	Topical Rapamycin Therapy to Alleviate the Cutaneous Manifestations of Tuberous Sclerosis Complex. Drugs in R and D, 2012, 12, 121-126.	2.2	120
8	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
9	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
10	Regression of Subependymal Giant Cell Astrocytoma With Rapamycin in Tuberous Sclerosis Complex. Journal of Child Neurology, 2008, 23, 1238-1239.	1.4	80
11	Efficacy and Safety of Topical Rapamycin in Patients With Facial Angiofibromas Secondary to Tuberous Sclerosis Complex. JAMA Dermatology, 2018, 154, 773.	4.1	71
12	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
13	The neuroimaging of Leigh syndrome: case series and review of the literature. Pediatric Radiology, 2016, 46, 443-451.	2.0	64
14	Phenotype of GABA-transaminase deficiency. Neurology, 2017, 88, 1919-1924.	1.1	49
15	Diagnosis of â€˜possibleâ€™ mitochondrial disease: an existential crisis. Journal of Medical Genetics, 2019, 56, 123-130.	3.2	42
16	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
17	Practice patterns of mitochondrial disease physicians in North America. Part 2: treatment, care and management. Mitochondrion, 2013, 13, 681-687.	3.4	38
18	Safety and efficacy of omaveloxolone in patients with mitochondrial myopathy. Neurology, 2020, 94, e687-e698.	1.1	38

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19	Seizure semiology and <scp>EEG</scp> findings in mitochondrial diseases. Epilepsia, 2014, 55, 707-712.	5.1	37
20	Practice patterns of mitochondrial disease physicians in North America. Part 1: Diagnostic and clinical challenges. Mitochondrion, 2014, 14, 26-33.	3.4	36
21	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
22	Expanding the Phenotypic Spectrum of CACNA1H Mutations. Pediatric Neurology, 2019, 93, 50-55.	2.1	21
23	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
24	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
25	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
26	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
27	Genomic deletions upstream of lamin B1 lead to atypical autosomal dominant leukodystrophy. Neurology: Genetics, 2019, 5, e305.	1.9	16
28	Juvenile Onset Central Nervous System Folate Deficiency and Rheumatoid Arthritis. Journal of Child Neurology, 2008, 23, 106-107.	1.4	13
29	Sleep Disordered Breathing in Children with Mitochondrial Disease. Pulmonary Medicine, 2014, 2014, 1-8.	1.9	9
30	Involvement of Cerebellum in Leigh Syndrome: Case Report and Review of the Literature. Pediatric Neurology, 2017, 74, 97-99.	2.1	8
31	Hypoglycemia in mitochondrial disorders. Mitochondrion, 2021, 58, 179-183.	3.4	6
32	Mitochondrial Disorder Aggravated by Metoprolol. Case Reports in Pediatrics, 2016, 2016, 1-3.	0.4	5
33	Metabolomics Profile in ABAT Deficiency Pre- and Post-treatment. JIMD Reports, 2018, 43, 13-17.	1.5	5
34	Response to Newman et al.. Genetics in Medicine, 2017, 19, 1380-1380.	2.4	3
35	Two different genetic etiologies for tuberous sclerosis complex (TSC) in a single family. Molecular Genetics & Genomic Medicine, 2020, 8, e1296.	1.2	3
36	The Many Guises of Mitochondrial Disease. Neurographics, 2013, 3, 2-10.	0.1	0

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37	Nitric Oxide Deficiency Triggering Strokelike Episodesâ€"Reply. JAMA Neurology, 2016, 73, 1030.	9.0	0
38	Reply to the Letter to the Editor by Josef Finsterer and Sinda Zarrouk-Mahjoub. Pediatric Neurology, 2018, 78, e11.	2.1	0
39	Decreased Exhaled Nitric Oxide Levels in Patients with Mitochondrial Disorders. Open Respiratory Medicine Journal, 2013, 7, 67-70.	0.4	0