Shanti Balasubramaniam

List of Publications by Year
in descending order

23	706	759233 12	677142 22
papers	citations	h-index	g-index

23	23	23	1174
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Genomic sequencing for the diagnosis of childhood mitochondrial disorders: a health economic evaluation. European Journal of Human Genetics, 2022, 30, 577-586.	2.8	9
2	Patient care standards for primary mitochondrial disease in Australia: an Australian adaptation of the Mitochondrial Medicine Society recommendations. Internal Medicine Journal, 2022, 52, 110-120.	0.8	3
3	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. Genetics in Medicine, 2022, 24, 130-145.	2.4	45
4	FGF21 outperforms GDF15 as a diagnostic biomarker of mitochondrial disease in children. Molecular Genetics and Metabolism, 2022, 135, 63-71.	1.1	12
5	Deleterious variants in CRLS1 lead to cardiolipin deficiency and cause an autosomal recessive multi-system mitochondrial disease. Human Molecular Genetics, 2022, 31, 3597-3612.	2.9	11
6	Fatal Perinatal Mitochondrial Cardiac Failure Caused by Recurrent De Novo Duplications in the ATAD3 Locus. Med, 2021, 2, 49-73.e10.	4.4	33
7	A novel variant in COX16 causes cytochrome c oxidase deficiency, severe fatal neonatal lactic acidosis, encephalopathy, cardiomyopathy, and liver dysfunction. Human Mutation, 2021, 42, 135-141.	2.5	4
8	Barth syndrome with severe dilated cardiomyopathy and growth hormone resistance: a case report. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 951-955.	0.9	1
9	Challenges in Diagnosing Intermediate Maple Syrup Urine Disease by Newborn Screening and Functional Validation of Genomic Results Imperative for Reproductive Family Planning. International Journal of Neonatal Screening, 2021, 7, 25.	3.2	5
10	Teaching NeuroImages: Bilateral Nucleus Tractus Solitarius Lesions in Neurogenic Respiratory Failure. Neurology, 2021, , 10.1212/WNL.0000000000012614.	1.1	2
11	Beneficial outcome of early dietary lysine restriction as an adjunct to pyridoxine therapy in a child with pyridoxine dependant epilepsy due to Antiquitin deficiency. JIMD Reports, 2020, 54, 9-15.	1.5	7
12	The diagnostic utility of genome sequencing in a pediatric cohort with suspected mitochondrial disease. Genetics in Medicine, 2020, 22, 1254-1261.	2.4	59
13	Pathobiologic Mechanisms of Neurodegeneration in Osteopetrosis Derived From Structural and Functional Analysis of 14 ClC-7 Mutants. Journal of Bone and Mineral Research, 2020, 36, 531-545.	2.8	16
14	Disorders of riboflavin metabolism. Journal of Inherited Metabolic Disease, 2019, 42, 608-619.	3.6	82
15	Clinical Spectrum and Functional Consequences Associated with Bi-Allelic Pathogenic PNPT1 Variants. Journal of Clinical Medicine, 2019, 8, 2020.	2.4	16
16	Biochemical signatures mimicking multiple carboxylase deficiency in children with mutations in MT-ATP6. Mitochondrion, 2019, 44, 58-64.	3.4	19
17	A SLC39A8 variant causes manganese deficiency, and glycosylation and mitochondrial disorders. Journal of Inherited Metabolic Disease, 2017, 40, 261-269.	3.6	101
18	Progressive deafnessâ€“dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015.	5.3	63

#	Article	IF	Citations
19	Leigh-Like Syndrome Due to Homoplasmic m.8993T>G Variant with Hypocitrullinemia and Unusual Biochemical Features Suggestive of Multiple Carboxylase Deficiency (MCD). JIMD Reports, 2016, 33, 99-107.	1.5	17
20	MECR Mutations Cause Childhood-Onset Dystonia and Optic Atrophy, a Mitochondrial Fatty Acid Synthesis Disorder. American Journal of Human Genetics, 2016, 99, 1229-1244.	6.2	91
21	Phenotypic variation of TTC19â€deficient mitochondrial complex III deficiency: A case report and literature review. American Journal of Medical Genetics, Part A, 2015, 167, 1330-1336.	1.2	31
22	Perinatal hypophosphatasia presenting as neonatal epileptic encephalopathy with abnormal neurotransmitter metabolism secondary to reduced coâ€factor pyridoxalâ€5â€²â€phosphate availability. Journal of Inherited Metabolic Disease, 2010, 33, 25-33.	3.6	79
23	Safety and Efficacy of Elosulfase Alfa in Australian Patients with Morquio a Syndrome: A Phase 3b Study. Journal of Inborn Errors of Metabolism and Screening, 0, 8, .	0.3	0