Rocio Rius

List of Publications by Year
in descending order

15 papers	269 citations	1040056 9 h-index	1125743 13 g-index
15 all docs	15 docs citations	15 times ranked	522 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	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.7	42
4	Any symptom, in any organ, at any age: A case report of multiple genetic diagnoses mimicking mitochondrial disease in an adult with kidney disease. Nephrology, 2022, , .	1.6	1
5	Distinct diagnostic trajectories in <scp>NBAS</scp>â€associated acute liver failure highlights the need for timely functional studies. JIMD Reports, 2022, 63, 240-249.	1.5	2
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	Application of Genome Sequencing from Blood to Diagnose Mitochondrial Diseases. Genes, 2021, 12, 607.	2.4	8
8	The diagnostic utility of genome sequencing in a pediatric cohort with suspected mitochondrial disease. Genetics in Medicine, 2020, 22, 1254-1261.	2.4	59
9	Biparental inheritance of mitochondrial DNA in humans is not a common phenomenon. Genetics in Medicine, 2019, 21, 2823-2826.	2.4	44
10	Leigh syndrome caused by mutations in <scp>MTFMT</scp> is associated with a better prognosis. Annals of Clinical and Translational Neurology, 2019, 6, 515-524.	3.7	17
11	Clinical Spectrum and Functional Consequences Associated with Bi-Allelic Pathogenic PNPT1 Variants. Journal of Clinical Medicine, 2019, 8, 2020.	2.4	16
12	Cryptic intronic NBAS variant reveals the genetic basis of recurrent liver failure in a child. Molecular Genetics and Metabolism, 2019, 126, 77-82.	1.1	11
13	Further delineation of achondroplasiaâ€“hypochondroplasia complex with longâ€term survival. American Journal of Medical Genetics, Part A, 2018, 176, 1225-1231.	1.2	0
14	Identification of a novel SLC12A6 pathogenic variant associated with hereditary motor and sensory neuropathy with agenesis of the corpus callosum (HMSN/ACC) in a non-French-Canadian family. Neurology India, 2018, 66, 1162.	0.4	4
15	Increased red cell distribution width in Fanconi anemia: a novel marker of stress erythropoiesis. Orphanet Journal of Rare Diseases, 2016, 11, 102.	2.7	20