

# Rocio Rius

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/7119444/publications.pdf>

Version: 2024-02-01

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	The diagnostic utility of genome sequencing in a pediatric cohort with suspected mitochondrial disease. <i>Genetics in Medicine</i> , 2020, 22, 1254-1261.	2.4	59
2	Biparental inheritance of mitochondrial DNA in humans is not a common phenomenon. <i>Genetics in Medicine</i> , 2019, 21, 2823-2826.	2.4	44
3	Novel diagnostic DNA methylation epigenatures expand and refine the epigenetic landscapes of Mendelian disorders. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100075.	1.7	42
4	Fatal Perinatal Mitochondrial Cardiac Failure Caused by Recurrent De Novo Duplications in the ATAD3 Locus. <i>Med</i> , 2021, 2, 49-73.e10.	4.4	33
5	Increased red cell distribution width in Fanconi anemia: a novel marker of stress erythropoiesis. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 102.	2.7	20
6	Leigh syndrome caused by mutations in <i>MTFMT</i> is associated with a better prognosis. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 515-524.	3.7	17
7	Clinical Spectrum and Functional Consequences Associated with Bi-Allelic Pathogenic PNPT1 Variants. <i>Journal of Clinical Medicine</i> , 2019, 8, 2020.	2.4	16
8	Cryptic intronic NBAS variant reveals the genetic basis of recurrent liver failure in a child. <i>Molecular Genetics and Metabolism</i> , 2019, 126, 77-82.	1.1	11
9	Genomic sequencing for the diagnosis of childhood mitochondrial disorders: a health economic evaluation. <i>European Journal of Human Genetics</i> , 2022, 30, 577-586.	2.8	9
10	Application of Genome Sequencing from Blood to Diagnose Mitochondrial Diseases. <i>Genes</i> , 2021, 12, 607.	2.4	8
11	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. <i>Neurology India</i> , 2018, 66, 1162.	0.4	4
12	Patient care standards for primary mitochondrial disease in Australia: an Australian adaptation of the Mitochondrial Medicine Society recommendations. <i>Internal Medicine Journal</i> , 2022, 52, 110-120.	0.8	3
13	Distinct diagnostic trajectories in NBAS-associated acute liver failure highlights the need for timely functional studies. <i>JIMD Reports</i> , 2022, 63, 240-249.	1.5	2
14	Any symptom, in any organ, at any age: A case report of multiple genetic diagnoses mimicking mitochondrial disease in an adult with kidney disease. <i>Nephrology</i> , 2022, , .	1.6	1
15	Further delineation of achondroplasia-hypochondroplasia complex with long-term survival. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1225-1231.	1.2	0