Antonia Ribes Rubio

List of Publications by Year
in descending order

44 papers	1,466 citations	394421 19 h-index	361022 35 g-index
51 all docs	51 docs citations	51 times ranked	2355 citing authors

#	Article	IF	Citations
1	Clinicoâ€radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. Human Mutation, 2022, 43, 403-419.	2.5	9
2	Organic Acids. , 2022, , 51-64.		1
3	Over-Mutated Mitochondrial, Lysosomal and TFEB-Regulated Genes in Parkinsonâ€™s Disease. Journal of Clinical Medicine, 2022, 11, 1749.	2.4	3
4	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	8.2	85
5	Systematic Collaborative Reanalysis of Genomic Data Improves Diagnostic Yield in Neurologic Rare Diseases. Journal of Molecular Diagnostics, 2022, 24, 529-542.	2.8	6
6	Implementation of second-tier tests in newborn screening for the detection of vitamin B12 related acquired and genetic disorders: results on 258,637 newborns. Orphanet Journal of Rare Diseases, 2021, 16, 195.	2.7	17
7	Variants in the <scp>ethylmalonylâ€CoA</scp> decarboxylase (<scp>ECHDC1</scp>) gene: a novel player in ethylmalonic aciduria?. Journal of Inherited Metabolic Disease, 2021, 44, 1215-1225.	3.6	4
8	External quality assessment in the absence of proficiency testing: A split-sample testing program experience. Clinical Biochemistry, 2021, 97, 78-81.	1.9	1
9	Newborn Screening for SCID: Experience in Spain (Catalonia). International Journal of Neonatal Screening, 2021, 7, 46.	3.2	4
10	Active site variants in STT3A cause a dominant type I congenital disorder of glycosylation with neuromusculoskeletal findings. American Journal of Human Genetics, 2021, 108, 2130-2144.	6.2	5
11	Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308.	3.6	43
12	Biallelic mutations in NDUFA8 cause complex I deficiency in two siblings with favorable clinical evolution. Molecular Genetics and Metabolism, 2020, 131, 349-357.	1.1	6
13	Complex I deficiency, due to NDUFAF4 mutations, causes severe mitochondrial dysfunction and is associated to early death and dysmorphia. Mitochondrion, 2020, 55, 78-84.	3.4	4
14	Neuronal and Astrocytic Differentiation from Sanfilippo C Syndrome iPSCs for Disease Modeling and Drug Development. Journal of Clinical Medicine, 2020, 9, 644.	2.4	10
15	Physiopathological Bases of the Disease Caused by HACE1 Mutations: Alterations in Autophagy, Mitophagy and Oxidative Stress Response. Journal of Clinical Medicine, 2020, 9, 913.	2.4	11
16	First Universal Newborn Screening Program for Severe Combined Immunodeficiency in Europe. Two-Years' Experience in Catalonia (Spain). Frontiers in Immunology, 2019, 10, 2406.	4.8	45
17	ADCK2 Haploinsufficiency Reduces Mitochondrial Lipid Oxidation and Causes Myopathy Associated with CoQ Deficiency. Journal of Clinical Medicine, 2019, 8, 1374.	2.4	27
18	Mutations in TIMM50 cause severe mitochondrial dysfunction by targeting key aspects of mitochondrial physiology. Human Mutation, 2019, 40, 1700-1712.	2.5	16

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19	Newborn screening for homocystinurias: Recent recommendations versus current practice. Journal of Inherited Metabolic Disease, 2019, 42, 128-139.	3.6	37
20	Muscle Involvement in a Large Cohort of Pediatric Patients with Genetic Diagnosis of Mitochondrial Disease. Journal of Clinical Medicine, 2019, 8, 68.	2.4	14
21	Ghrelin Causes a Decline in GABA Release by Reducing Fatty Acid Oxidation in Cortex. Molecular Neurobiology, 2018, 55, 7216-7228.	4.0	10
22	FLAD1, encoding FAD synthase, is mutated in a patient with myopathy, scoliosis and cataracts. Clinical Genetics, 2018, 94, 592-593.	2.0	9
23	Cerebrospinal fluid monoamines, pterins, and folate in patients with mitochondrial diseases: systematic review and hospital experience. Journal of Inherited Metabolic Disease, 2018, 41, 1147-1158.	3.6	12
24	Medicina interna y enfermedades raras. TransiciÃ³n niÃ±o-adulto. Arbor, 2018, 194, 460.	0.3	2
25	Small molecules as therapeutic agents for inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2017, 40, 177-193.	3.6	18
26	Mutations inTRAPPC11are associated with a congenital disorder of glycosylation. Human Mutation, 2017, 38, 148-151.	2.5	34
27	Secondary coenzyme Q 10 deficiencies in oxidative phosphorylation (OXPHOS) and non-OXPHOS disorders. Mitochondrion, 2016, 30, 51-58.	3.4	70
28	A leaky splicing mutation in NFU1 is associated with a particular biochemical phenotype. Consequences for the diagnosis. Mitochondrion, 2016, 26, 72-80.	3.4	19
29	Differential diagnosis of lipoic acid synthesis defects. Journal of Inherited Metabolic Disease, 2016, 39, 781-793.	3.6	42
30	Lysine Restriction and Pyridoxal Phosphate Administration in a NADK2 Patient. Pediatrics, 2016, 138, .	2.1	23
31	Neonatal Screening for Inherited Metabolic Diseases in 2016. Seminars in Pediatric Neurology, 2016, 23, 257-272.	2.0	35
32	Free-thiamine is a potential biomarker of thiamine transporter-2 deficiency: a treatable cause of Leigh syndrome. Brain, 2016, 139, 31-38.	7.6	174
33	Targeted Next Generation Sequencing in Patients with Inborn Errors of Metabolism. PLoS ONE, 2016, 11, e0156359.	2.5	48
34	Cholestane-3Î²,5Î±,6Î²-triol: high levels in Niemann-Pick type C, cerebrotendinous xanthomatosis, and lysosomal acid lipase deficiency. Journal of Lipid Research, 2015, 56, 1926-1935.	4.2	62
35	Newborn screening for homocystinurias and methylation disorders: systematic review and proposed guidelines. Journal of Inherited Metabolic Disease, 2015, 38, 1007-1019.	3.6	100
36	Effect of Readthrough Treatment in Fibroblasts of Patients Affected by Lysosomal Diseases Caused by Premature Termination Codons. Neurotherapeutics, 2015, 12, 874-886.	4.4	17

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37	Fatty Acid Transport Protein 1 (FATP1) Localizes in Mitochondria in Mouse Skeletal Muscle and Regulates Lipid and Ketone Body Disposal. PLoS ONE, 2014, 9, e98109.	2.5	24
38	Protein expression profiles in patients carrying NFU1 mutations. Contribution to the pathophysiology of the disease. Journal of Inherited Metabolic Disease, 2013, 36, 841-847.	3.6	31
39	Mitochondrial DNA depletion syndrome: New descriptions and the use of citrate synthase as a helpful tool to better characterise the patients. Molecular Genetics and Metabolism, 2012, 107, 409-415.	1.1	31
40	Reply to He et al. European Journal of Human Genetics, 2011, 19, 124-124.	2.8	2
41	A Fatal Mitochondrial Disease Is Associated with Defective NFU1 Function in the Maturation of a Subset of Mitochondrial Fe-S Proteins. American Journal of Human Genetics, 2011, 89, 656-667.	6.2	262
42	Genetic and cellular studies of oxidative stress in methylmalonic aciduria (MMA) cobalamin deficiency type C (cblC) with homocystinuria (MMACHC). Human Mutation, 2009, 30, 1558-1566.	2.5	76
43	Hand tremor and orofacial dyskinesia: Clinical manifestations of glutaric aciduria type I in a young girl. Movement Disorders, 2003, 18, 1076-1079.	3.9	7
44	Calorie Restriction Rescues Mitochondrial Dysfunction in Adck2-Deficient Skeletal Muscle. Frontiers in Physiology, 0, 13, .	2.8	0