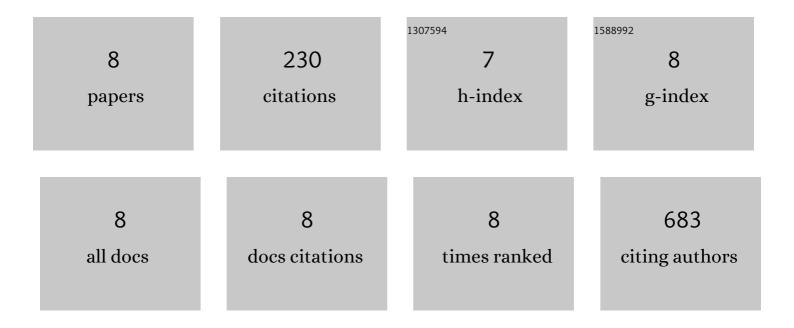
Jorge Asin-Cayuela

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/7472687/publications.pdf Version: 2024-02-01



#	Article	IF	CITATIONS
1	Whole exome sequencing reveals mutations in <i>NARS2</i> and <i>PARS2</i> , encoding the mitochondrial asparaginyl-tRNA synthetase and prolyl-tRNA synthetase, in patients with Alpers syndrome. Molecular Genetics & amp; Genomic Medicine, 2015, 3, 59-68.	1.2	87
2	Structural modeling of tissue-specific mitochondrial alanyl-tRNA synthetase (AARS2) defects predicts differential effects on aminoacylation. Frontiers in Genetics, 2015, 6, 21.	2.3	46
3	Broad phenotypic variability in patients with complex I deficiency due to mutations in NDUFS1 and NDUFV1. Mitochondrion, 2015, 21, 33-40.	3.4	30
4	Leukoencephalopathy due to Complex II Deficiency and Bi-Allelic SDHB Mutations: Further Cases and Implications for Genetic Counselling. JIMD Reports, 2016, 33, 69-77.	1.5	17
5	γ-glutamyl transpeptidase deficiency caused by a large homozygous intragenic deletion in GGT1. European Journal of Human Genetics, 2018, 26, 808-817.	2.8	16
6	β-Mannosidosis caused by a novel homozygous intragenic inverted duplication in <i>MANBA</i> . Journal of Physical Education and Sports Management, 2019, 5, a003954.	1.2	16
7	Identification of a novel mutation in PEX10 in a patient with attenuated Zellweger spectrum disorder: a case report. Journal of Medical Case Reports, 2017, 11, 218.	0.8	13
8	A novel mutation on the transferrin gene abolishes one N-glycosylation site and alters the pattern of transferrin isoforms, mimicking that observed after excessive alcohol consumption. Clinical Biochemistry, 2016, 49, 511-513.	1.9	5

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