Jorge Asin-Cayuela

List of Publications by Year
in descending order

8	230	1307594 7	1588992 8
papers	citations	h-index	g-index

8	8	8	683
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Î²-Mannosidosis caused by a novel homozygous intragenic inverted duplication in MANBA. Journal of Physical Education and Sports Management, 2019, 5, a003954.	1.2	16
2	Î³-glutamyl transpeptidase deficiency caused by a large homozygous intragenic deletion in GGT1. European Journal of Human Genetics, 2018, 26, 808-817.	2.8	16
3	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
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	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
6	Broad phenotypic variability in patients with complex I deficiency due to mutations in NDUFS1 and NDUFV1. Mitochondrion, 2015, 21, 33-40.	3.4	30
7	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
8	Whole exome sequencing reveals mutations inNARS2andPARS2, encoding the mitochondrial asparaginyl-tRNA synthetase and prolyl-tRNA synthetase, in patients with Alpers syndrome. Molecular Genetics & Genomic Medicine, 2015, 3, 59-68.	1.2	87