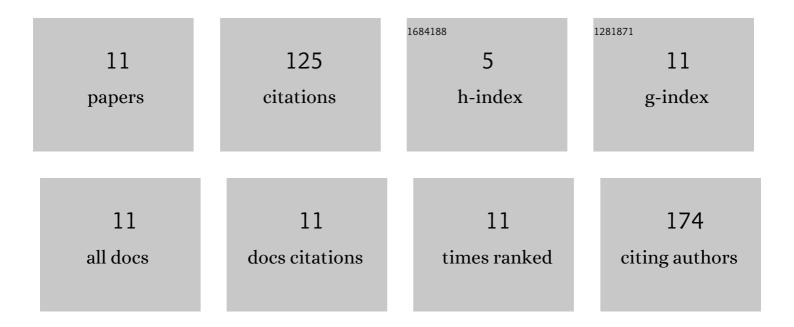
Yasuhiko Ago

List of Publications by Year in descending order

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Υλεμμικό Δοο

#	Article	IF	CITATIONS
1	Recent advances in understanding beta-ketothiolase (mitochondrial acetoacetyl-CoA thiolase, T2) deficiency. Journal of Human Genetics, 2019, 64, 99-111.	2.3	30
2	ATP6V0A1 encoding the a1-subunit of the V0 domain of vacuolar H+-ATPases is essential for brain development in humans and mice. Nature Communications, 2021, 12, 2107.	12.8	30
3	Deficiency of 3â€hydroxybutyrate dehydrogenase (BDH1) in mice causes low ketone body levels and fatty liver during fasting. Journal of Inherited Metabolic Disease, 2020, 43, 960-968.	3.6	21
4	Intronic antisense Alu elements have a negative splicing effect on the inclusion of adjacent downstream exons. Gene, 2018, 664, 84-89.	2.2	13
5	In vitro functional analysis of four variants of human asparagine synthetase. Journal of Inherited Metabolic Disease, 2021, 44, 1226-1234.	3.6	7
6	Diagnosis of Mucopolysaccharidoses and Mucolipidosis by Assaying Multiplex Enzymes and Glycosaminoglycans. Diagnostics, 2021, 11, 1347.	2.6	5
7	Glycosaminoglycans as Biomarkers for Mucopolysaccharidoses and Other Disorders. Diagnostics, 2021, 11, 1563.	2.6	5
8	Evading the AAV Immune Response in Mucopolysaccharidoses. International Journal of Molecular Sciences, 2020, 21, 3433.	4.1	4
9	Novel GYS2 mutations in a Japanese patient with glycogen storage disease type 0a. Molecular Genetics and Metabolism Reports, 2021, 26, 100702.	1.1	4
10	Japanese patients with mitochondrial 3‑hydroxy‑3‑methylglutaryl‑CoA synthase deficiency: ln vitro functional analysis of five novel HMGCS2 mutations. Experimental and Therapeutic Medicine, 2020, 20, 1-1.	1.8	3
11	Activity of daily living in mucopolysaccharidosis IVA patients: Evaluation of therapeutic efficacy. Molecular Genetics & Genomic Medicine, 2021, 9, e1806.	1.2	3