Makiko Yasuda

List of Publications by Year
in descending order

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27 papers	1,079 citations	567281 15 h-index	526287 27 g-index
27 all docs	27 docs citations	27 times ranked	1474 citing authors

#	Article	IF	Citations
1	Fabry disease: Characterization of ?-galactosidase A double mutations and the D313Y plasma enzyme pseudodeficiency allele. Human Mutation, 2003, 22, 486-492.	2.5	133
2	Acute Intermittent Porphyria: Predicted Pathogenicity ofHMBSVariants Indicates Extremely Low Penetrance of the Autosomal Dominant Disease. Human Mutation, 2016, 37, 1215-1222.	2.5	129
3	Preclinical Development of a Subcutaneous ALAS1 RNAi Therapeutic for Treatment of Hepatic Porphyrias Using Circulating RNA Quantification. Molecular Therapy - Nucleic Acids, 2015, 4, e263.	5.1	107
4	RNAi-mediated silencing of hepatic Alas1 effectively prevents and treats the induced acute attacks in acute intermittent porphyria mice. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 7777-7782.	7.1	99
5	Fabry Disease: prevalence of affected males and heterozygotes with pathogenic GLA mutations identified by screening renal, cardiac and stroke clinics, 1995â€“2017. Journal of Medical Genetics, 2018, 55, 261-268.	3.2	91
6	Recent advances on porphyria genetics: Inheritance, penetrance & molecular heterogeneity, including new modifying/causative genes. Molecular Genetics and Metabolism, 2019, 128, 320-331.	1.1	59
7	Insight into GATA1 transcriptional activity through interrogation of cis elements disrupted in human erythroid disorders. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 4434-4439.	7.1	56
8	AAV8-mediated Gene Therapy Prevents Induced Biochemical Attacks of Acute Intermittent Porphyria and Improves Neuromotor Function. Molecular Therapy, 2010, 18, 17-22.	8.2	52
9	Liver Transplantation for Acute Intermittent Porphyria: Biochemical and Pathologic Studies of the Explanted Liver. Molecular Medicine, 2015, 21, 487-495.	4.4	51
10	Fabry Disease: Novel Î±-Galactosidase A 3â€²-Terminal Mutations Result in Multiple Transcripts Due to Aberrant 3â€²-End Formation. American Journal of Human Genetics, 2003, 73, 162-173.	6.2	39
11	A LCâ€“MS/MS method for the specific, sensitive, and simultaneous quantification of 5-aminolevulinic acid and porphobilinogen. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2011, 879, 2389-2396.	2.3	37
12	Human hydroxymethylbilane synthase: Molecular dynamics of the pyrrole chain elongation identifies step-specific residues that cause AIP. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E4071-E4080.	7.1	32
13	Acute Intermittent Porphyria in children: A case report and review of the literature. Molecular Genetics and Metabolism, 2016, 119, 295-299.	1.1	31
14	AAV2/6 Gene Therapy in a Murine Model of Fabry Disease Results in Supraphysiological Enzyme Activity and Effective Substrate Reduction. Molecular Therapy - Methods and Clinical Development, 2020, 18, 607-619.	4.1	29
15	Identification and characterization of 40 novel hydroxymethylbilane synthase mutations that cause acute intermittent porphyria. Journal of Inherited Metabolic Disease, 2019, 42, 186-194.	3.6	17
16	Homozygous hydroxymethylbilane synthase knock-in mice provide pathogenic insights into the severe neurological impairments present in human homozygous dominant acute intermittent porphyria. Human Molecular Genetics, 2019, 28, 1755-1767.	2.9	17
17	International Porphyria Molecular Diagnostic Collaborative: an evidence-based database of verified pathogenic and benign variants for the porphyrias. Genetics in Medicine, 2019, 21, 2605-2613.	2.4	16
18	Sex differences in vascular reactivity in mesenteric arteries from a mouse model of acute intermittent porphyria. Molecular Genetics and Metabolism, 2019, 128, 376-381.	1.1	16

#	Article	IF	Citations
19	Murine models of the human porphyrias: Contributions toward understanding disease pathogenesis and the development of new therapies. Molecular Genetics and Metabolism, 2019, 128, 332-341.	1.1	12
20	Identification and characterization of 40 novel hydroxymethylbilane synthase mutations that cause acute intermittent porphyria. Journal of Inherited Metabolic Disease, 2018, 42, 186.	3.6	9
21	Congenital erythropoietic porphyria and erythropoietic protoporphyria: Identification of 7 uroporphyrinogen III synthase and 20 ferrochelatase novel mutations. Molecular Genetics and Metabolism, 2019, 128, 358-362.	1.1	9
22	Porphyria cutanea tarda and hepatoerythropoietic porphyria: Identification of 19 novel uroporphyrinogen III decarboxylase mutations. Molecular Genetics and Metabolism, 2019, 128, 363-366.	1.1	9
23	ZFN-mediated inÂvivo gene editing in hepatocytes leads to supraphysiologic Î±-Gal A activity and effective substrate reduction in Fabry mice. Molecular Therapy, 2021, 29, 3230-3242.	8.2	9
24	Characterization of the hepatic transcriptome following phenobarbital induction in mice with AIP. Molecular Genetics and Metabolism, 2019, 128, 382-390.	1.1	7
25	Acute intermittent porphyria: vector optimization for gene therapy. Journal of Gene Medicine, 2007, 9, 806-811.	2.8	6
26	Severe hydroxymethylbilane synthase deficiency causes depression-like behavior and mitochondrial dysfunction in a mouse model of homozygous dominant acute intermittent porphyria. Acta Neuropathologica Communications, 2020, 8, 38.	5.2	5
27	Acute hepatic porphyrias: Identification of 46 hydroxymethylbilane synthase, 11 coproporphyrinogen oxidase, and 20 protoporphyrinogen oxidase novel mutations. Molecular Genetics and Metabolism, 2019, 128, 352-357.	1.1	2