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New case of mitochondrial HMG-CoA synthase deficiency. Functional analysis of eight mutations

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#	Paper	IF	Citations
22	Ketone body metabolism and its defects. <i>Journal of Inherited Metabolic Disease</i> , 2014 , 37, 541-51	5.4	130
21	Mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency: urinary organic acid profiles and expanded spectrum of mutations. <i>Journal of Inherited Metabolic Disease</i> , 2015 , 38, 459-66	5.4	31
20	Mitochondrial 3-Hydroxy-3-Methylglutaryl-CoA Synthase Deficiency: Unique Presenting Laboratory Values and a Review of Biochemical and Clinical Features. <i>JIMD Reports</i> , 2018 , 40, 63-69	1.9	20
19	Human Mitochondrial HMG-CoA Synthase Deficiency: Role of Enzyme Dimerization Surface and Characterization of Three New Patients. <i>International Journal of Molecular Sciences</i> , 2018 , 19,	6.3	15
18	A Japanese case of mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency who presented with severe metabolic acidosis and fatty liver without hypoglycemia. <i>JIMD Reports</i> , 2019 , 48, 19-25	1.9	10
17	Novel pathogenic variants in a Chinese family with mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency. <i>Pediatric Investigation</i> , 2019 , 3, 86-90	1.3	2
16	Severe clinical manifestation of mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency associated with two novel mutations: a case report. <i>BMC Pediatrics</i> , 2019 , 19, 344	2.6	7
15	Expanding phenotypic and mutational spectra of mitochondrial HMG-CoA synthase deficiency. <i>European Journal of Medical Genetics</i> , 2020 , 63, 104086	2.6	4
14	Hypoglycemia is not a defining feature of metabolic crisis in mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency: Further evidence of specific biochemical markers which may aid diagnosis. <i>JIMD Reports</i> , 2020 , 55, 26-31	1.9	3
13	Association of a novel homozygous mutation in the HMGCS2 gene with an HMGCSD in an Iranian patient. <i>Molecular Genetics & amp; Genomic Medicine</i> , 2020 , 8, e1507	2.3	2
12	Clinical, biochemical, molecular and therapeutic characteristics of four new patients of mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency. <i>Clinica Chimica Acta</i> , 2020 , 509, 83-5	96 ^{.2}	3
11	The gene encoding the ketogenic enzyme HMGCS2 displays a unique expression during gonad development in mice. <i>PLoS ONE</i> , 2020 , 15, e0227411	3.7	6
10	Expanding the clinical spectrum of mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency with Turkish cases harboring novel HMGCS2 gene mutations and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 1608-1614	2.5	1
9	Organic Acidemias and Disorders of Fatty Acid Oxidation. 2021 , 279-333		
8	Ketogenesis prevents diet-induced fatty liver injury and hyperglycemia. <i>Journal of Clinical Investigation</i> , 2014 , 124, 5175-90	15.9	115
7	Japanese patients with mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency: functional analysis of five novel mutations. <i>Experimental and Therapeutic Medicine</i> , 2020 , 20, 39	2.1	3
6	Recurrent loss of HMGCS2 shows that ketogenesis is not essential for the evolution of large mammalian brains. <i>ELife</i> , 2018 , 7,	8.9	22

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5	[Mitochondrial 3-hydroxy-3-methylglutaryl CoA synthase deficiency: a case report and literature review]. <i>Chinese Journal of Contemporary Pediatrics</i> , 2018 , 20, 930-933	0.8	3	
4	Metabolic Emergency in Flight Air Medical Journal, 2022 , 41, 141-146	1		
3	Clinical, Biochemical, Molecular, and Outcome Features of Mitochondrial 3-Hydroxy-3-Methylglutaryl-CoA Synthase Deficiency in 10 Chinese Patients <i>Frontiers in Genetics</i> , 2021 , 12, 816779	4.5		
2	Hypoglycaemia Metabolic Gene Panel Testing Frontiers in Endocrinology, 2022, 13, 826167	5.7	O	
1	Metabolic Changes Associated With Cardiomyocyte Dedifferentiation Enable Adult Mammalian Cardiac Regeneration. 2022 , 146, 1950-1967		1	