Systematic literature review and meta-analysis on the epidemiology of methylmalonic acidemia (MMA) with a focus on MMA caused by methylmalonyl-CoA mutase (mut) deficiency

DOI: 10.1186/s13023-019-1063-z
Orphanet Journal of Rare Diseases, 2019, 14, 84.

#	Paper	IF	Citations
21	A New Era for Rare Genetic Diseases: Messenger RNA Therapy. *Human Gene Therapy*, **2019**, 30, 1180-1188	4.8	24
20	Second-Generation Pharmacological Chaperones: Beyond Inhibitors. *Molecules*, **2020**, 25,	4.8	11
19	Considering Proximal Urea Cycle Disorders in Expanded Newborn Screening. *International Journal of Neonatal Screening*, **2020**, 6,	2.6	5
18	Cardiometabolic risk factor clustering in patients with deficient branched-chain amino acid catabolism: A case-control study. *Journal of Inherited Metabolic Disease*, **2020**, 43, 981-993	5.4	1
17	A rare mutation c.1663G > A (p.A555T) in the MMUT gene associated with mild clinical and biochemical phenotypes of methylmalonic acidemia in 30 Chinese patients. *Orphanet Journal of Rare Diseases*, **2021**, 16, 22	4.2	0
16	Determination of Cytokines and Oxidative Stress Biomarkers in Cognitive Impairment Induced by Methylmalonic Acidemia. *NeuroImmunoModulation*, **2021**, 28, 178-186	2.5	0
15	Determining the pathogenicity of MUT gene variant by mini-gene splicing assay.		
14	Phenotypic and genotypic analysis of children with methylmalonic academia: A single-center study in China and a recent literature review. *Clinica Chimica Acta*, **2021**, 522, 14-22	6.2	1
13	Molecular and biochemical investigations of inborn errors of metabolism-altered redox homeostasis in branched-chain amino acid disorders, organic acidurias, and homocystinuria. *Free Radical Research*, **2021**, 55, 627-640	4	2
12	A False-Positive Case of Methylmalonic Aciduria by Tandem Mass Spectrometry Newborn Screening Dependent on Maternal Malnutrition in Pregnancy. *International Journal of Environmental Research and Public Health*, **2020**, 17,	4.6	6
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10	Prevalence of methylmalonic acidemia among newborns and the clinical-suspected population: a meta-analyse. *Journal of Maternal-Fetal and Neonatal Medicine*, **2021**, 1-16	2	0
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8	Application of a Novel Algorithm for Expanding Newborn Screening for Inherited Metabolic Disorders across Europe.. *International Journal of Neonatal Screening*, **2022**, 8,	2.6	3
7	Clinical application of NGS-based SNP haplotyping for PGT-M of methylmalonic acidemia.. *Systems Biology in Reproductive Medicine*, **2021**, 1-9	2.9	0
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5	Gene Therapy for Mitochondrial Diseases: Current Status and Future Perspective. *Pharmaceutics*, **2022**, 14, 1287	6.4	2

4	Renal outcome and plasma methylmalonic acid levels after isolated or combined liver or kidney transplantation in patients with methylmalonic acidemia: A multicenter analysis. **2022**,	0
3	mRNA-Based Approaches to Treating Liver Diseases. **2022**, 11, 3328	0
2	A case report of methylmalonic acidemia associated with CYBT gene mutation. **2022**, 7, 81	0
1	Methylmalonic acid levels in serum, exosomes, and urine and its association with cblC type methylmalonic acidemia-induced cognitive impairment. 13,	0