3-Methylglutaconic Aciduria

National Cancer Institute

Source

National Cancer Institute. 3-Methylglutaconic Aciduria. NCI Thesaurus. Code C98678.

A group of five inherited disorders caused by mutations in the AUH, DNAJC19, OPA3, and TAZ genes. The disorders are characterized by impairment in the function of mitochondria, resulting in the accumulation and excretion of 3-methylglutaconic acid, and the presence of 3-methylglutaric acid in the urine.