

Open Peer Review on Qeios

MMADHC wt Allele

National Cancer Institute

Source

National Cancer Institute. MMADHC wt Allele. NCI Thesaurus. Code C74466.

Human MMADHC wild-type allele is located in the vicinity of 2q23.2 and is approximately 18 kb in length. This allele, which encodes Methylmalonic aciduria and homocystinuria type D protein, mitochondrial, plays a role in the mediation of vitamin B12 metabolism. Mutation of the gene is associated with some cases of homocystinuria, and methylmalonic aciduria.

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