MLYCD Gene

Subjects: Genetics & Heredity
Contributor: Lily Guo

malonyl-CoA decarboxylase

genes

1. Introduction

The *MLYCD* gene provides instructions for making an enzyme called malonyl-CoA decarboxylase. This enzyme helps regulate the formation and breakdown of a group of fats called fatty acids. Many tissues, including heart (cardiac) muscle, use fatty acids as a major source of energy. The body also uses fatty acids to build cell membranes, produce hormones, and carry out many other important processes.

Malonyl-CoA decarboxylase is responsible for the chemical reaction that converts a molecule called malonyl-CoA to a molecule called acetyl-CoA. This reaction is an important step in the breakdown of fatty acids. Acetyl-CoA is then used to make new fatty acids and can also be used to produce energy.

Malonyl-CoA decarboxylase is most active in cardiac muscle and in muscles used for movement (skeletal muscles). It is also found in other organs and tissues, including the brain, small intestine, liver, kidney, and pancreas. This enzyme probably functions in several parts of the cell, including mitochondria, which are cells' energy-producing centers, and peroxisomes, which are small sacs that process fatty acids and other molecules. Malonyl-CoA decarboxylase also functions in the fluid that surrounds these cell structures (the cytoplasm).

2. Health Conditions Related to Genetic Changes

2.1. Malonyl-CoA decarboxylase deficiency

More than 20 mutations in the *MLYCD* gene have been identified in people with malonyl-CoA decarboxylase deficiency. Some of these mutations lead to the production of an abnormally short, nonfunctional version of malonyl-CoA decarboxylase or prevent the gene from producing any of this enzyme. Other mutations change the structure of the enzyme so it cannot be delivered to the parts of the cell where it is needed (such as mitochondria and peroxisomes).

A lack of malonyl-CoA decarboxylase disrupts the normal balance of fatty acid formation and breakdown in the body. As a result, fatty acids cannot be converted to energy, which leads to characteristic features of this disorder

including low blood sugar (hypoglycemia) and a heart condition called cardiomyopathy. Byproducts of fatty acid processing build up in tissues, which also contributes to the signs and symptoms of malonyl-CoA decarboxylase deficiency.

3. Other Names for This Gene

- DCMC_HUMAN
- hMCD
- malonyl coenzyme A decarboxylase
- MCD

References

1. FitzPatrick DR, Hill A, Tolmie JL, Thorburn DR, Christodoulou J. The molecularbasis of malonyl-CoA decarboxylase deficiency. Am J Hum Genet. 1999Aug;65(2):318-26.
2. Gao J, Waber L, Bennett MJ, Gibson KM, Cohen JC. Cloning and mutationalanalysis of human malonyl-coenzyme A decarboxylase. J Lipid Res. 1999Jan;40(1):178-82.
3. Sacksteder KA, Morrell JC, Wanders RJ, Matalon R, Gould SJ. MCD encodesperoxisomal and cytoplasmic forms of malonyl-CoA decarboxylase and is mutated in malonyl-CoA decarboxylase deficiency. J Biol Chem. 1999 Aug 27;274(35):24461-8.
4. Saggerson D. Malonyl-CoA, a key signaling molecule in mammalian cells. AnnuRev Nutr. 2008;28:253-72. doi: 10.1146/annurev.nutr.28.061807.155434. Review.
5. Salomons GS, Jakobs C, Pope LL, Errami A, Potter M, Nowaczyk M, Olpin S,Manning N, Raiman JA, Slade T, Champion MP, Peck D, Gavrilov D, Hillman R,Hoganson GE, Donaldson K, Shield JP, Ketteridge D, Wasserstein M, Gibson KM.Clinical, enzymatic and molecular characterization of nine new patients withmalonyl-coenzyme A decarboxylase deficiency. J Inherit Metab Dis. 2007Feb;30(1):23-8.
6. Sambandam N, Steinmetz M, Chu A, Altarejos JY, Dyck JR, Lopaschuk GD.Malonyl-CoA decarboxylase (MCD) is differentially regulated in subcellularcompartments by 5'AMP-activated protein kinase (AMPK). Studies using H9c2 cellsoverexpressing MCD and AMPK by adenoviral gene transfer technique. Eur J Biochem.2004 Jul;271(13):2831-40.
7. Surendran S, Sacksteder KA, Gould SJ, Coldwell JG, Rady PL, Tyring SK, MatalonR. Malonyl CoA decarboxylase deficiency: C to T transition in intron 2 of the MCDgene. J Neurosci Res. 2001 Sep 15;65(6):591-4.
8. Wightman PJ, Santer R, Ribes A, Dougherty F, McGill N, Thorburn DR,FitzPatrick DR. MLYCD mutation analysis: evidence for protein mistargeting as acause of MLYCD deficiency. Hum Mutat. 2003 Oct;22(4):288-300.