

## Malonyl-CoA decarboxylase deficiency

### Description

Malonyl-CoA decarboxylase deficiency is a condition that prevents the body from converting certain fats to energy. The signs and symptoms of this disorder typically appear in early childhood. Almost all affected children have delayed development. Additional signs and symptoms can include weak muscle tone (hypotonia), seizures, diarrhea, vomiting, and low blood sugar (hypoglycemia). A heart condition called cardiomyopathy, which weakens and enlarges the heart muscle, is another common feature of malonyl-CoA decarboxylase deficiency.

### Frequency

This condition is very rare; fewer than 30 cases have been reported.

### Causes

Mutations in the *MLYCD* gene cause malonyl-CoA decarboxylase deficiency. The *MLYCD* gene provides instructions for making an enzyme called malonyl-CoA decarboxylase. Within cells, this enzyme helps regulate the formation and breakdown of a group of fats called fatty acids. Many tissues, including the heart muscle, use fatty acids as a major source of energy.

Mutations in the *MLYCD* gene reduce or eliminate the function of malonyl-CoA decarboxylase. A shortage of this enzyme disrupts the normal balance of fatty acid formation and breakdown in the body. As a result, fatty acids cannot be converted to energy, which can lead to characteristic features of this disorder including hypoglycemia and cardiomyopathy. Byproducts of fatty acid processing build up in tissues, which also contributes to the signs and symptoms of malonyl-CoA decarboxylase deficiency.

[Learn more about the gene associated with Malonyl-CoA decarboxylase deficiency](#)

- *MLYCD*

### Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal

recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

## **Other Names for This Condition**

- Deficiency of malonyl-CoA decarboxylase
- Malonic aciduria
- Malonyl-coenzyme A decarboxylase deficiency
- MCD deficiency

## **Additional Information & Resources**

### Genetic Testing Information

- Genetic Testing Registry: Deficiency of malonyl-CoA decarboxylase (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0342793/>)

### Genetic and Rare Diseases Information Center

- Malonic aciduria (<https://rarediseases.info.nih.gov/diseases/3371/index>)

### Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

### Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22Malonyl-CoA decarboxylase deficiency%22](https://clinicaltrials.gov/search?cond=%22Malonyl-CoA+decarboxylase+deficiency%22))

### Catalog of Genes and Diseases from OMIM

- MALONYL-CoA DECARBOXYLASE DEFICIENCY (<https://omim.org/entry/248360>)

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28malonyl-coa+decarboxylase+deficiency%5BTIAB%5D%29+OR+%28malonic+aciduria%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

## References

- de Wit MC, de Coo IF, Verbeek E, Schot R, Schoonderwoerd GC, Duran M, de Klerk JB, Huijmans JG, Lequin MH, Verheijen FW, Mancini GM. Brain abnormalities in a case of malonyl-CoA decarboxylase deficiency. *Mol Genet Metab*. 2006 Feb;87(2):102-6. doi: 10.1016/j.ymgme.2005.09.009. Epub 2005 Nov 4. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/16275149>)
- FitzPatrick DR, Hill A, Tolmie JL, Thorburn DR, Christodoulou J. The molecular basis of malonyl-CoA decarboxylase deficiency. *Am J Hum Genet*. 1999 Aug;65(2):318-26. doi: 10.1086/302492. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/10417274>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1377930/>)
- Sacksteder KA, Morrell JC, Wanders RJ, Matalon R, Gould SJ. MCD encodes peroxisomal 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. doi: 10.1074/jbc.274.35.24461. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/10455107>)
- 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 with malonyl-coenzyme A decarboxylase deficiency. *J Inher Metab Dis*. 2007 Feb;30(1):23-8. doi: 10.1007/s10545-006-0514-6. Epub 2006 Dec 20. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17186413>)

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