MMUT gene

methylmalonyl-CoA mutase

Normal Function

The *MMUT* gene provides instructions for making an enzyme called methylmalonyl CoA mutase. This enzyme is active in mitochondria, which are specialized structures inside cells that serve as energy-producing centers.

Methylmalonyl CoA mutase is responsible for a particular step in the breakdown of several protein building blocks (amino acids), specifically isoleucine, methionine, threonine, and valine. The enzyme also helps break down certain types of fats (lipids) and cholesterol. First, several chemical reactions convert the amino acids, lipids, or cholesterol to a molecule called methylmalonyl CoA. Then, working with a compound called adenosylcobalamin (AdoCbl), which is a form of vitamin B12, methylmalonyl CoA mutase converts methylmalonyl CoA to a compound called succinyl-CoA. Other enzymes break down succinyl-CoA into molecules that are later used for energy.

Health Conditions Related to Genetic Changes

Methylmalonic acidemia

More than 200 mutations in the *MMUT* gene have been identified in people with methylmalonic acidemia, a condition characterized by feeding difficulties, developmental delay, and long-term health problems. These genetic changes prevent the production of functional methylmalonyl CoA mutase or reduce the activity of the enzyme. As a result, certain proteins and lipids are not broken down properly. This defect allows methylmalonyl CoA and other toxic compounds to build up in the body's organs and tissues, causing the signs and symptoms of methylmalonic acidemia.

Mutations that prevent the production of any functional methylmalonyl CoA mutase lead to a form of methylmalonic acidemia designated mut^0. Mut^0 is the most severe form of this disorder and has the poorest outcome. Mutations that alter the structure of the enzyme but do not completely eliminate its activity lead to a form of the condition designated mut^-. The mut^- form is typically less severe, with more variable symptoms than the mut^0 form.

Other Names for This Gene

- MCM
- methylalonyl-CoA mutase
- methylamlony-CoA isomerase
- methylmalonyl CoA mutase
- methylmalonyl Coenzyme A mutase
- methylmalonyl Coenzyme A mutase precursor
- MUT
- MUTA_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MMUT (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4594[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28MUT%5BTI%5D%29+OR+%28methylmalonyl+Coenzyme+A+mutase%5BTIAB%5D%29+OR+%28methylmalonyl+CoA+mutase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- METHYLMALONYL-CoA MUTASE; MMUT (https://omim.org/entry/609058)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/4594)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MMUT[gene])

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Genomic Location

The *MMUT* gene is found on chromosome 6 (https://medlineplus.gov/genetics/chromos ome/6/).

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