



MMAA gene

metabolism of cobalamin associated A

Normal Function

The *MMAA* gene provides instructions for making a protein that is involved in the formation of a compound called adenosylcobalamin (AdoCbl). AdoCbl, which is derived from vitamin B12 (also called cobalamin), is necessary for the normal function of an enzyme known as methylmalonyl CoA mutase. This enzyme helps break down certain proteins, fats (lipids), and cholesterol.

Research indicates that the MMAA protein may play a role in one of the last steps in AdoCbl formation, the transport of vitamin B12 into mitochondria (specialized structures inside cells that serve as energy-producing centers). Additional chemical reactions then convert vitamin B12 into AdoCbl. Other studies suggest that the MMAA protein may help stabilize methylmalonyl CoA mutase and protect the enzyme from being turned off (inactivated).

Health Conditions Related to Genetic Changes

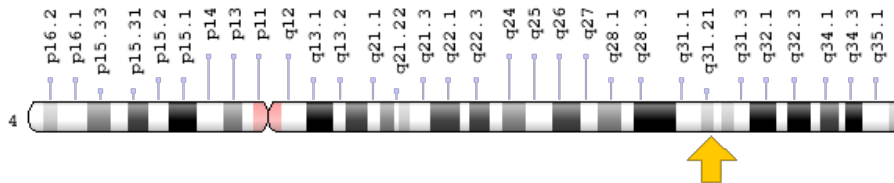
Methylmalonic acidemia

More than 25 mutations in the *MMAA* gene have been found to cause methylmalonic acidemia, a condition characterized by feeding difficulties, developmental delay, and long term health problems. Some of these mutations add, delete, or duplicate a small amount of genetic material in the gene. Other mutations change a single protein building block (amino acid) used to make the MMAA protein. These mutations can lead to the production of an unstable MMAA protein or an abnormally small, nonfunctional version of the protein. It is unclear how the abnormal MMAA protein leads to the serious medical problems associated with methylmalonic acidemia. Studies suggest that without the activity of this protein, AdoCbl may not be made properly. A lack of AdoCbl impairs the function of methylmalonyl CoA mutase, which results in the incomplete break down of certain proteins and lipids. This defect allows toxic compounds to build up in the body's organs and tissues. Research suggests that a lack of AdoCbl leading to impaired methylmalonyl CoA mutase function causes the signs and symptoms of methylmalonic acidemia.

Chromosomal Location

Cytogenetic Location: 4q31.21, which is the long (q) arm of chromosome 4 at position 31.21

Molecular Location: base pairs 145,619,385 to 145,660,033 on chromosome 4 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- cblA
- methylmalonic aciduria (cobalamin deficiency) cblA type
- methylmalonic aciduria (cobalamin deficiency) type A
- methylmalonic aciduria type A
- MMAA_HUMAN

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Methylmalonic aciduria may be secondary to defects of cobalamin metabolism
<https://www.ncbi.nlm.nih.gov/books/NBK27933/#A3118>

Clinical Information from GeneReviews

- Isolated Methylmalonic Acidemia
<https://www.ncbi.nlm.nih.gov/books/NBK1231>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28MMAA%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- MMAA GENE
<http://omim.org/entry/607481>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_MMAA.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=MMAA%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:18871
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:166785>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/166785>
- UniProt
<https://www.uniprot.org/uniprot/Q8IVH4>

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