

MMAA Gene

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metabolism of cobalamin associated A

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1. Introduction

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).

2. Health Conditions Related to Genetic Changes

2.1. 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.

3. Other Names for This Gene

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

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