

AMACR gene

alpha-methylacyl-CoA racemase

Normal Function

The *AMACR* gene provides instructions for making an enzyme called alpha-methylacyl-CoA racemase (AMACR). This enzyme is found in the energy-producing centers in cells (mitochondria) and in cell structures called peroxisomes. Peroxisomes contain a variety of enzymes that break down many different substances, including fatty acids and certain toxic compounds. They are also important for the production (synthesis) of fats (lipids) used in digestion and in the nervous system.

In peroxisomes, the AMACR enzyme plays a role in the breakdown of a fatty acid called pristanic acid, which comes from meat and dairy foods in the diet. In mitochondria, AMACR is thought to help further break down the molecules derived from pristanic acid.

Health Conditions Related to Genetic Changes

Alpha-methylacyl-CoA racemase deficiency

Alpha-methylacyl-CoA racemase (AMACR) deficiency is caused by mutations in the *AMACR* gene. This disorder leads to a variety of neurological problems that begin in adulthood, including gradual loss in intellectual functioning (cognitive decline), seizures, and weakness and loss of sensation in the limbs due to nerve damage (sensorimotor neuropathy). Most individuals with AMACR deficiency have an *AMACR* gene mutation that replaces a protein building block (amino acid) called serine with an amino acid called proline at position 52 in the enzyme sequence, written as Ser52Pro or S52P. This mutation results in a lack (deficiency) of functional enzyme. The enzyme deficiency leads to accumulation of pristanic acid in the blood. However, it is unclear how this accumulation is related to the specific signs and symptoms of AMACR deficiency.

Other disorders

AMACR gene mutations that result in a lack of functional AMACR enzyme have also been identified in infants with a life-threatening disorder called congenital bile acid synthesis defect type 4. Babies with this disorder have cholestasis, which is a reduced ability to produce and release a digestive fluid called bile. Cholestasis leads to an enlarged liver (hepatomegaly) and irreversible liver disease (cirrhosis) in the first few months of life.

Some researchers consider congenital bile acid synthesis defect type 4 and AMACR deficiency (see above) to be variations of the same disorder. Because most individuals with congenital bile acid synthesis defect type 4 do not survive infancy, it is unclear whether they would have later developed the neurological symptoms seen in adults with AMACR deficiency.

Other Names for This Gene

- 2-methylacyl-CoA racemase
- AMACR_HUMAN
- AMACRD
- CBAS4
- RACE
- RM

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of AMACR ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23600\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23600[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28AMACR%5BTIAB%5D%29+OR+%28alpha-methylacyl-CoA+racemase%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+1800+days%22%5Bdp%5D%29%29>)

Catalog of Genes and Diseases from OMIM

- BILE ACID SYNTHESIS DEFECT, CONGENITAL, 4; CBAS4 (<https://omim.org/entry/214950>)
- ALPHA-METHYLACYL-CoA RACEMASE; AMACR (<https://omim.org/entry/604489>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/23600>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=AMACR\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=AMACR[gene]))

References

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Genomic Location

The *AMACR* gene is found on chromosome 5 (<https://medlineplus.gov/genetics/chromosome/5/>).

Last updated December 1, 2013