ACAD8 gene

acyl-CoA dehydrogenase family member 8

Normal Function

The *ACAD8* gene provides instructions for making an enzyme called isobutyryl-CoA dehydrogenase (IBD). This enzyme is found in mitochondria, the energy-producing centers inside cells. The IBD enzyme is involved in breaking down proteins from food. Specifically, this enzyme is responsible for the third step in the breakdown of a protein building block (amino acid) called valine. The IBD enzyme converts a molecule called isobutyryl-CoA into a molecule called methylacrylyl-CoA. Other enzymes further break down methylacrylyl-CoA into molecules that cells can use for energy.

Health Conditions Related to Genetic Changes

Isobutyryl-CoA dehydrogenase deficiency

At least 19 mutations in the *ACAD8* gene have been found to cause isobutyryl-CoA dehydrogenase (IBD) deficiency. Some of these mutations reduce the activity of the IBD enzyme, while other mutations prevent the gene from producing any functional enzyme. As a result, valine is not broken down properly. An inability to process valine may lead to reduced energy production and the features of IBD deficiency.

Other Names for This Gene

- ACAD-8
- ACAD8_HUMAN
- Activator-recruited cofactor 42 kDa component
- acyl-CoA dehydrogenase family, member 8
- acyl-coenzyme A dehydrogenase 8
- ARC42
- FLJ22590

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ACAD8 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=27034[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28ACAD8%5BTIAB%5D%29+OR+%28ACAD-8%5BTIAB%5D%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

- ACYL-CoA DEHYDROGENASE FAMILY, MEMBER 8; ACAD8 (https://omim.org/entry/604773)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/27034)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ACAD8[gene])

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Genomic Location

The *ACAD8* gene is found on chromosome 11 (https://medlineplus.gov/genetics/chromosome/11/).

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