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 methacrylyl-CoA. Other enzymes further break down methacrylyl-CoA into molecules that cells can use for energy.

Health Conditions Related to Genetic Changes

Isobutyryl-CoA dehydrogenase deficiency

Variants (also called mutations) in the *ACAD8* gene have been found to cause IBD deficiency. People with IBD deficiency cannot effectively break down valine. Normally, proteins from food are broken down into amino acids, which are further processed to provide energy for growth and development. IBD deficiency disrupts this process. Most people with IBD deficiency do not have any signs or symptoms of the condition. If signs or symptoms do appear, they typically show up early in life and only last for a short period of time.

The *ACAD8* gene variants that cause IBD deficiency reduce or eliminate the activity of the IBD enzyme. As a result, isobutyryl-CoA is not converted to methacrylyl-CoA and the breakdown of valine is impaired. An inability to process valine may reduce energy production, and some people may develop health problems related to 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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