

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\[geneid\]](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%29>)

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\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ACAD8[gene]))

References

- Koeberl DD, Young SP, Gregersen NS, Vockley J, Smith WE, Benjamin DK Jr, An Y, Weavil SD, Chaing SH, Bali D, McDonald MT, Kishnani PS, Chen YT, Millington DS. Rare disorders of metabolism with elevated butyryl- and isobutyryl-carnitine detected by tandem mass spectrometry newborn screening. *Pediatr Res.* 2003 Aug;54(2):219-23. doi: 10.1203/01.PDR.0000074972.36356.89. Epub 2003 May 7. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12736383>)
- Nguyen TV, Andresen BS, Corydon TJ, Ghisla S, Abd-El Razik N, Mohsen AW, Cederbaum SD, Roe DS, Roe CR, Lench NJ, Vockley J. Identification of isobutyryl-CoA dehydrogenase and its deficiency in humans. *Mol Genet Metab.* 2002 Sep-Oct; 77(1-2):68-79. doi: 10.1016/s1096-7192(02)00152-x. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12359132>)
- Oglesbee D, He M, Majumder N, Vockley J, Ahmad A, Angle B, Burton B, Charrow J, Ensenuer R, Ficicioglu CH, Keppen LD, Marsden D, Tortorelli S, Hahn SH, Matern D. Development of a newborn screening follow-up algorithm for the diagnosis of isobutyryl-CoA dehydrogenase deficiency. *Genet Med.* 2007 Feb;9(2):108-16. doi: 10.1097/gim.0b013e31802f78d6. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17304052>)
- Pedersen CB, Bischoff C, Christensen E, Simonsen H, Lund AM, Young SP, Koeberl DD, Millington DS, Roe CR, Roe DS, Wanders RJ, Ruiter JP, Keppen LD, Stein Q, Knudsen I, Gregersen N, Andresen BS. Variations in IBD (ACAD8) in children with elevated C4-carnitine detected by tandem mass spectrometry newborn screening. *Pediatr Res.* 2006 Sep;60(3):315-20. doi: 10.1203/01.pdr.0000233085.

72522.04. Epub2006 Jul 20. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/16857760>)

- Roe CR, Cederbaum SD, Roe DS, Mardach R, Galindo A, Sweetman L. Isolated isobutyryl-CoA dehydrogenase deficiency: an unrecognized defect in human valine metabolism. *Mol Genet Metab*. 1998 Dec;65(4):264-71. doi: 10.1006/mgme.1998.2758. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/9889013>)
- Sass JO, Sander S, Zschocke J. Isobutyryl-CoA dehydrogenase deficiency: isobutyrylglycinuria and ACAD8 gene mutations in two infants. *J Inher Metab Dis*. 2004;27(6):741-5. doi: 10.1023/B:BOLI.0000045798.12425.1b. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15505379>)
- Yoo EH, Cho HJ, Ki CS, Lee SY. Isobutyryl-CoA dehydrogenase deficiency with a novel ACAD8 gene mutation detected by tandem mass spectrometry newborn screening. *Clin Chem Lab Med*. 2007;45(11):1495-7. doi: 10.1515/CCLM.2007.317. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17924841>)

Genomic Location

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

Last updated June 1, 2010