Isobutyryl-CoA dehydrogenase deficiency

Isobutyryl-CoA dehydrogenase (IBD) deficiency is a condition that disrupts the breakdown of certain proteins. Normally, proteins from food are broken down into parts called amino acids. Amino acids can be further processed to provide energy for growth and development. People with IBD deficiency have inadequate levels of an enzyme that helps break down a particular amino acid called valine.

Most people with IBD deficiency are asymptomatic, which means they do not have any signs or symptoms of the condition. A few children with IBD deficiency have developed features such as a weakened and enlarged heart (dilated cardiomyopathy), weak muscle tone (hypotonia), and developmental delay. This condition may also cause low numbers of red blood cells (anemia) and very low blood levels of carnitine, which is a natural substance that helps convert certain foods into energy. The range of signs and symptoms associated with IBD deficiency remains unclear because very few affected individuals have been reported.

Frequency

IBD deficiency is a rare disorder; approximately 22 cases have been reported in the medical literature.

Causes

Mutations in the *ACAD8* gene cause IBD deficiency. This gene provides instructions for making the IBD enzyme, which is involved in breaking down valine. *ACAD8* gene mutations reduce or eliminate the activity of the IBD enzyme. As a result, valine is not broken down properly. Impaired processing of valine may lead to reduced energy production and the features of IBD deficiency.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- deficiency of isobutyryl-CoA dehydrogenase
- IBD deficiency
- isobutyryl-coenzyme A dehydrogenase deficiency

Diagnosis & Management

Formal Diagnostic Criteria

- ACT Sheet: Elevated C4 acylcarnitine
  https://www.ncbi.nlm.nih.gov/books/NBK55827/bin/C4.pdf

Genetic Testing Information

- What is genetic testing?
  /primer/testing/genetictesting
- Genetic Testing Registry: Deficiency of isobutyryl-CoA dehydrogenase
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1969809/

Other Diagnosis and Management Resources

- Baby's First Test
  https://www.babysfirsttest.org/newborn-screening/conditions/isobutyrylglycinuria
- MedlinePlus Encyclopedia: Dilated Cardiomyopathy
  https://medlineplus.gov/ency/article/000168.htm

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Anemia
  https://medlineplus.gov/ency/article/000560.htm
- Encyclopedia: Dilated Cardiomyopathy
  https://medlineplus.gov/ency/article/000168.htm
- Health Topic: Amino Acid Metabolism Disorders
  https://medlineplus.gov/aminoacidmetabolismdisorders.html
- Health Topic: Cardiomyopathy
  https://medlineplus.gov/cardiomyopathy.html
- Health Topic: Newborn Screening
  https://medlineplus.gov/newbornscreening.html

Genetic and Rare Diseases Information Center

- Isobutyryl-CoA dehydrogenase deficiency
  https://rarediseases.info.nih.gov/diseases/10223/isobutyryl-coa-dehydrogenase-deficiency

Educational Resources

- MalaCards: isobutyryl-coa dehydrogenase deficiency
  https://www.malacards.org/card/isobutyryl_coa_dehydrogenase_deficiency
- Orphanet: Isobutyryl-CoA dehydrogenase deficiency
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=79159
- Screening, Technology and Research in Genetics
  http://www.newbornscreening.info/Parents/organicaciddisorders/ICoA.html

Patient Support and Advocacy Resources

- Children's Cardiomyopathy Foundation
  https://dev.childrenscardiomyopathy.org/
- Medical Home Portal
  https://www.medicalhomeportal.org/newborn/isobutyryl-coa-dehydrogenase-deficiency
- Metabolic Support UK
  https://www.metabolicsupportuk.org/
- Organic Acidemia Association
  https://www.oaanews.org/icbd.html

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28isobutyryl-coa+dehydrogenase+deficiency%5BTIAB%5D%29+OR+%28isobutyryl-coa+dehydrogenase%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

Catalog of Genes and Diseases from OMIM

- ISOBUTYRYL-CoA DEHYDROGENASE DEFICIENCY
  http://omim.org/entry/611283

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Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/condition/isobutyryl-coa-dehydrogenase-deficiency

Reviewed: June 2010
Published: August 17, 2020

Lister Hill National Center for Biomedical Communications
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National Institutes of Health
Department of Health & Human Services