DBT gene

dihydrolipoamide branched chain transacylase E2

Normal Function

The *DBT* gene provides instructions for making part of a group of enzymes called the branched-chain alpha-keto acid dehydrogenase (BCKD) enzyme complex. Specifically, the protein produced from the *DBT* gene forms a critical piece of the enzyme complex called the E2 component.

The BCKD enzyme complex is responsible for one step in the normal breakdown of three protein building blocks (amino acids). These amino acids—leucine, isoleucine, and valine—are obtained from the diet. They are present in many kinds of food, particularly protein-rich foods such as milk, meat, and eggs. The BCKD enzyme complex is active in mitochondria, which are specialized structures inside cells that serve as energy-producing centers. The breakdown of leucine, isoleucine, and valine produces molecules that can be used for energy.

Health Conditions Related to Genetic Changes

Maple syrup urine disease

More than 70 mutations in the *DBT* gene have been identified in people with maple syrup urine disease, most often in individuals with mild variants of the disorder. These variant forms become apparent later in infancy or childhood, and they lead to delayed development and other health problems if not treated.

Mutations in the *DBT* gene include changes in single DNA building blocks (base pairs) and insertions or deletions of a small amount of DNA in the *DBT* gene. These changes disrupt the normal function of the E2 component, preventing the BCKD enzyme complex from effectively breaking down leucine, isoleucine, and valine. As a result, these amino acids and their byproducts build up in the body. This accumulation is toxic to cells and tissues, particularly in the nervous system. The buildup of these substances can lead to developmental delay and the other health problems associated with maple syrup urine disease.

Other Names for This Gene

- BCATE2

- dihydrolipoamide branched chain transacylase (E2 component of branched chain keto acid dehydrogenase complex; maple syrup urine disease)
- E2 component of branched chain keto acid dehydrogenase complex
- MSUD2
- ODB2_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of DBT (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1629[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28DBT+AND+maple+syrup+urine+disease%5BTIAB%5D%29+OR+%28dihydrolipoamide+branched+chain+transacylase%5BTIAB%5D%29%29+OR+%28%28dihydrolipoamide+branched+chain+transacylase%5BTIAB%5D%29+OR+%28MSUD2%5BTIAB%5D%29+OR+%28E2+%5Btiab%5D+AND+BCKD+%5Btiab%5D%29+OR+%28E2+%5Btiab%5D+AND+branched-chain+alpha-ketoacid+dehydrogenase+complex+%5Btiab%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

- DIHYDROLIPOAMIDE BRANCHED-CHAIN TRANSACYLASE; DBT (https://omim.org/entry/248610)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1629)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=DBT[gene])

References

- Chi CS, Tsai CR, Chen LH, Lee HF, Mak BS, Yang SH, Wang TY, Shu SG, Chen CH.Maple syrup urine disease in the Austronesian aboriginal tribe Paiwan of Taiwan: a novel DBT (E2) gene 4.7 kb founder deletion caused by a nonhomologousrecombination between LINE-1 and Alu and the carrier-frequency determination. EurJ Hum Genet. 2003 Dec;11(12):931-6. doi: 10.1038/sj.ejhg.5201069. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/14508502)
- Flaschker N, Feyen O, Fend S, Simon E, Schadewaldt P, Wendel U. Description

ofthe mutations in 15 subjects with variant forms of maple syrup urine disease. JInherit Metab Dis. 2007 Nov;30(6):903-9. doi: 10.1007/s10545-007-0579-x. Epub2007 Oct 8. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17922217)

- Henneke M, Flaschker N, Helbling C, Muller M, Schadewaldt P, Gartner J, WendelU. Identification of twelve novel mutations in patients with classic and variantforms of maple syrup urine disease. Hum Mutat. 2003 Nov;22(5):417. doi:10.1002/humu.9187. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/14517957)
- Nellis MM, Danner DJ. Gene preference in maple syrup urine disease. Am J HumGenet. 2001 Jan;68(1):232-7. doi: 10.1086/316950. Epub 2000 Dec 7. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/11112664) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1234918/)
- Quental S, Macedo-Ribeiro S, Matos R, Vilarinho L, Martins E, Teles EL,Rodrigues E, Diogo L, Garcia P, Eusebio F, Gaspar A, Sequeira S, Furtado F, LancaI, Amorim A, Prata MJ. Molecular and structural analyses of maple syrup urinedisease and identification of a founder mutation in a Portuguese Gypsy community.Mol Genet Metab. 2008 Jun;94(2):148-56. doi: 10.1016/j.ymgme.2008.02.008. Epub2008 Apr 2. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18378174)
- Rodriguez-Pombo P, Navarrete R, Merinero B, Gomez-Puertas P, Ugarte M. Mutational spectrum of maple syrup urine disease in Spain. Hum Mutat. 2006Jul;27(7):715. doi: 10.1002/humu.9428. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16786533)
- Strauss KA, Puffenberger EG, Carson VJ. Maple Syrup Urine Disease. 2006 Jan 30[updated 2020 Apr 23]. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Bean LJH, Gripp KW, Amemiya A, editors. GeneReviews(R) [Internet].Seattle (WA): University of Washington, Seattle; 1993-2024. Available fromhttp://www.ncbi.nlm.nih.gov/books/NBK1319/ Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20301495)

Genomic Location

The *DBT* gene is found on chromosome 1 (https://medlineplus.gov/genetics/chromosome/1/).

Last updated July 1, 2017