HSD3B2 gene
hydroxy-delta-5-steroid dehydrogenase, 3 beta- and steroid delta-isomerase 2

Normal Function

The \textit{HSD3B2} gene provides instructions for making the 3-beta-hydroxysteroid dehydrogenase (3\beta-HSD) enzyme. This enzyme is found in the gonads, which are the ovaries in females and testes in males, and in the adrenal glands, which are located on top of the kidneys. Within these hormone-producing tissues, the 3\beta-HSD enzyme is necessary for the production of many hormones, including cortisol, aldosterone, androgens, and estrogen. Cortisol has numerous functions such as maintaining energy and blood sugar levels, protecting the body from stress, and suppressing inflammation. Aldosterone is sometimes called the salt-retaining hormone because it regulates the amount of salt retained by the kidney. The retention of salt affects fluid levels and blood pressure. Androgens and estrogen are essential for normal sexual development and reproduction.

Health Conditions Related to Genetic Changes

3-beta-hydroxysteroid dehydrogenase deficiency

At least 37 mutations in the \textit{HSD3B2} gene have been found to cause 3\beta-HSD deficiency. Most of these mutations change single protein building blocks (amino acids) in the 3\beta-HSD enzyme, which typically reduces the activity of the enzyme. Mutations that allow the production of some functional enzyme, although at reduced levels, cause the less severe, non-salt-wasting or non-classic forms of 3\beta-HSD deficiency. Other mutations result in the production of an abnormally short, completely nonfunctional 3\beta-HSD enzyme, which causes the more severe, salt-wasting form of this condition. All types of 3\beta-HSD deficiency interfere with the production of a variety of hormones and lead to abnormalities of sexual development and maturation.
Chromosomal Location
Cytogenetic Location: 1p12, which is the short (p) arm of chromosome 1 at position 12
Molecular Location: base pairs 119,414,931 to 119,423,039 on chromosome 1 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene
- 3 beta-HSD type II
- 3-beta-hydroxy-5-ene steroid dehydrogenase
- 3-beta-hydroxy-delta(5)-steroid dehydrogenase
- 3 beta-hydroxysteroid dehydrogenase 2
- 3 beta-hydroxysteroid dehydrogenase type II, delta 5-delta 4-isomerase type II, 3 beta-HSD type II
- 3 beta-ol dehydrogenase
- 3BHS2_HUMAN
- delta 5-delta 4-isomerase type II
- HSD3B
- HSDB

Additional Information & Resources
Educational Resources
  https://www.ncbi.nlm.nih.gov/books/NBK29/?rendertype=box&id=A1174
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28HSD3B2%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+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 3-BETA-HYDROXYSTEROID DEHYDROGENASE 2 DEFICIENCY
  http://omim.org/entry/201810

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_HSD3B2.html
- ClinVar
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:3284
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P26439

Sources for This Summary

- OMIM: ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 3-BETA-HYDROXYSTEROID DEHYDROGENASE 2 DEFICIENCY
  http://omim.org/entry/201810
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12608938
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12050213


Reviewed: February 2010
Published: October 29, 2019

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services