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

Normal Function

The HSD3B2 gene provides instructions for making the 3-beta-hydroxysteroid dehydrogenase (3β-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 3β-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 HSD3B2 gene have been found to cause 3β-HSD deficiency. Most of these mutations change single protein building blocks (amino acids) in the 3β-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 3β-HSD deficiency. Other mutations result in the production of an abnormally short, completely nonfunctional 3β-HSD enzyme, which causes the more severe, salt-wasting form of this condition. All types of 3β-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 109.20200228, 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/11344940

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12428206

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15632317

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18252794


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