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

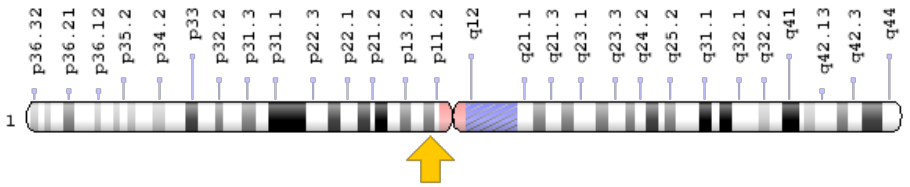
3-beta-hydroxysteroid dehydrogenase deficiency

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 109.20190607, 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

- Endocrinology: An Integrated Approach (first edition, 2001): The Major Steroidogenic Pathway in the Human Testis 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
  https://www.ncbi.nlm.nih.gov/clinvar?term=HSD3B2%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:5218
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:3284
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/3284
- 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
- Pang S, Carbunaru G, Haider A, Copeland KC, Chang YT, Lutfallah C, Mason JI. Carriers for type II 3beta-hydroxysteroid dehydrogenase (HSD3B2) deficiency can only be identified by HSD3B2 genotype study and not by hormone test. Clin Endocrinol (Oxf). 2003 Mar;58(3):323-31.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12608938
- Pang S, Wang W, Rich B, David R, Chang YT, Carbunaru G, Myers SE, Howie AF, Smillie KJ, Mason JI. A novel nonstop mutation in the stop codon and a novel missense mutation in the type II 3beta-hydroxysteroid dehydrogenase (3beta-HSD) gene causing, respectively, nonclassic and classic 3beta-HSD deficiency congenital adrenal hyperplasia. J Clin Endocrinol Metab. 2002 Jun; 87(6):2556-63.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12050213

- Pang S. Congenital adrenal hyperplasia owing to 3 beta-hydroxysteroid dehydrogenase deficiency. Endocrinol Metab Clin North Am. 2001 Mar;30(1):81-99, vi-vii. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11344940
- Simard J, Moisan AM, Morel Y. Congenital adrenal hyperplasia due to 3beta-hydroxysteroid dehydrogenase/Delta(5)-Delta(4) isomerase deficiency. Semin Reprod Med. 2002 Aug;20(3): 255-76. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12428206
- Simard J, Ricketts ML, Gingras S, Soucy P, Feltus FA, Melner MH. Molecular biology of the 3beta-hydroxysteroid dehydrogenase/delta5-delta4 isomerase gene family. Endocr Rev. 2005 Jun;26(4): 525-82. Epub 2005 Jan 4. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15632317
- Welzel M, Wüstemann N, Simic-Schleicher G, Dörr HG, Schulze E, Shaikh G, Clayton P, Grötzinger J, Holterhus PM, Riepe FG. Carboxyl-terminal mutations in 3beta-hydroxysteroid dehydrogenase type II cause severe salt-wasting congenital adrenal hyperplasia. J Clin Endocrinol Metab. 2008 Apr;93(4):1418-25. doi: 10.1210/jc.2007-1874. Epub 2008 Feb 5.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18252794

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/HSD3B2

Reviewed: February 2010
Published: September 10, 2019

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