

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 (3 β -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 β -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 (glucose) 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 3 β -HSD deficiency. Most of these mutations change single protein building blocks (amino acids) in the 3 β -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 β -HSD deficiency. Other mutations result in the production of an abnormally short, completely nonfunctional 3 β -HSD enzyme, which causes the more severe, salt-wasting form of this condition. All types of 3 β -HSD deficiency interfere with the production of a variety of hormones and lead to abnormalities of sexual development and maturation.

Other Names for This Gene

- 3 beta-HSD type II
- 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

- 3-beta-hydroxy-5-ene steroid dehydrogenase
- 3-beta-hydroxy-delta(5)-steroid dehydrogenase
- 3BHS2_HUMAN
- delta 5-delta 4-isomerase type II
- HSD3B
- HSDB

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of HSD3B2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3284\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3284[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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 (<https://omim.org/entry/201810>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3284>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=HSD3B2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=HSD3B2[gene]))

References

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Genomic Location

The *HSD3B2* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome/1/>).

Last updated February 1, 2010