

## HSD3B7 gene

hydroxy-delta-5-steroid dehydrogenase, 3 beta- and steroid delta-isomerase 7

### Normal Function

The *HSD3B7* gene provides instructions for making an enzyme called 3 beta-hydroxysteroid dehydrogenase type 7 (3 $\beta$ -HSD7). This enzyme is found in liver cells. It is embedded in the membrane of a cell structure called the endoplasmic reticulum, which is involved in protein processing and distribution. The 3 $\beta$ -HSD7 enzyme participates in the production of bile acids, which are a component of a digestive fluid called bile. Bile acids stimulate bile flow and helps absorb fats and fat-soluble vitamins. Bile acids are produced from cholesterol in a multi-step process. The 3 $\beta$ -HSD7 enzyme is responsible for the second step in that process, which converts 7 $\alpha$ ( $\alpha$ )-hydroxycholesterol to 7 $\alpha$ -hydroxy-4-cholesten-3-one.

### Health Conditions Related to Genetic Changes

#### Congenital bile acid synthesis defect type 1

At least 17 mutations in the *HSD3B7* gene have been found to cause congenital bile acid synthesis defect type 1. This condition is characterized by cholestasis, a condition that impairs the production and release of a digestive fluid called bile from liver cells. Most of the *HSD3B7* gene mutations delete one or two DNA building blocks (base pairs) from the gene or replace single protein building blocks (amino acids) in the enzyme. These mutations result in production of a 3 $\beta$ -HSD7 enzyme with little or no function. Without enough functional 3 $\beta$ -HSD7 enzyme, the conversion of 7 $\alpha$ -hydroxycholesterol to 7 $\alpha$ -hydroxy-4-cholesten-3-one is impaired. The 7 $\alpha$ -hydroxycholesterol instead gets converted into abnormal bile acid compounds that cannot be transported out of the liver into the intestine, where the bile acids are needed to absorb fats and fat-soluble vitamins. This impaired production and release of bile acids leads to cholestasis. As a result, cholesterol and abnormal bile acids build up in the liver and fat-soluble vitamins are not absorbed, leading to the signs and symptoms of congenital bile acid synthesis defect type 1.

### Other Names for This Gene

- 3 beta-hydroxy-delta 5-C27-steroid oxidoreductase
- 3 beta-hydroxysteroid dehydrogenase type 7

- 3 beta-hydroxysteroid dehydrogenase type VII
- 3-beta-HSD VII
- 3-beta-hydroxy-Delta(5)-C27 steroid oxidoreductase
- 3BHS7\_HUMAN
- c(27) 3-beta-HSD
- C(27)-3BETA-HSD
- cholest-5-ene-3-beta,7-alpha-diol 3-beta-dehydrogenase
- SDR11E3
- short chain dehydrogenase/reductase family 11E, member 3

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of HSD3B7 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=80270\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=80270[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28HSD3B7%5BTIAB%5D%29+OR+%283+beta-hydroxy-delta+5-C27-steroid+oxidoreductase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D%29%29>)

### Catalog of Genes and Diseases from OMIM

- 3-BETA-HYDROXY-DELTA-5-C27-STEROID OXIDOREDUCTASE; HSD3B7 (<https://omim.org/entry/607764>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/80270>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=HSD3B7\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=HSD3B7[gene]))

## **References**

- Cheng JB, Jacquemin E, Gerhardt M, Nazer H, Cresteil D, Heubi JE, Setchell KD, Russell DW. Molecular genetics of 3beta-hydroxy-Delta5-C27-steroid oxidoreductase deficiency in 16 patients with loss of bile acid synthesis and liver disease. *J Clin Endocrinol Metab*. 2003 Apr;88(4):1833-41. doi: 10.1210/jc.2002-021580. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12679481>)
- Gioiello A, Cerra B, Zhang W, Vallerini GP, Costantino G, De Franco F, Passeri D,

Pellicciari R, Setchell KD. Synthesis of atypical bile acids for use as investigative tools for the genetic defect of 3beta-hydroxy-Delta(5)-C27-steroid oxidoreductase deficiency. *J Steroid Biochem Mol Biol*. 2014 Oct;144 Pt B:348-60. doi: 10.1016/j.jsbmb.2014.06.008. Epub 2014 Jun 19. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/24954360>)

- Subramaniam P, Clayton PT, Portmann BC, Mieli-Vergani G, Hadzic N. Variable clinical spectrum of the most common inborn error of bile acid metabolism--3beta-hydroxy-Delta 5-C27-steroid dehydrogenase deficiency. *J Pediatr Gastroenterol Nutr*. 2010 Jan;50(1):61-6. doi: 10.1097/MPG.0b013e3181b47b34. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19915491>)

## Genomic Location

The *HSD3B7* gene is found on chromosome 16 (<https://medlineplus.gov/genetics/chromosome/16/>).

**Last updated April 1, 2015**