

HSD17B3 gene

hydroxysteroid 17-beta dehydrogenase 3

Normal Function

The *HSD17B3* gene provides instructions for making an enzyme called 17-beta hydroxysteroid dehydrogenase 3. This enzyme is active in the male gonads (testes), where it helps to produce the male sex hormone testosterone from a precursor hormone called androstenedione.

Health Conditions Related to Genetic Changes

17-beta hydroxysteroid dehydrogenase 3 deficiency

More than 20 mutations that cause 17-beta hydroxysteroid dehydrogenase 3 deficiency have been identified in the *HSD17B3* gene. In the Arab population of Gaza, where the condition is most common, almost all affected individuals have two copies of the same mutation. This mutation replaces the protein building block (amino acid) arginine with the amino acid glutamine at protein position 80 (written as Arg80Gln or R80Q).

Mutations in the *HSD17B3* gene result in a 17-beta hydroxysteroid dehydrogenase 3 enzyme with little or no activity, reducing testosterone production. A shortage of testosterone affects the development of the reproductive tract in the male fetus, resulting in the abnormalities in the external sex organs that occur in 17-beta hydroxysteroid dehydrogenase 3 deficiency.

Other Names for This Gene

- 17-beta-HSD3
- DHB3_HUMAN
- EDH17B3
- estradiol 17 beta-dehydrogenase 3
- hydroxysteroid (17-beta) dehydrogenase 3
- SDR12C2

Tests Listed in the Genetic Testing Registry

- ## Scientific Articles on PubMed

- ## Catalog of Genes and Diseases from OMIM

- ## Gene and Variant Databases

- ## References

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- Lee YS, Kirk JM, Stanhope RG, Johnston DI, Harland S, Auchus RJ, Andersson S, Hughes IA. Phenotypic variability in 17beta-hydroxysteroid dehydrogenase-3 deficiency and diagnostic pitfalls. Clin Endocrinol (Oxf). 2007 Jul;67(1):20-8. doi: 10.1111/j.1365-2265.2007.02829.x. Epub 2007 Apr 27. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17466011>)
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- Moghrabi N, Hughes IA, Dunaif A, Andersson S. Deleterious missense mutations and silent polymorphism in the human 17beta-hydroxysteroid dehydrogenase 3 gene (HSD17B3). J Clin Endocrinol Metab. 1998 Aug;83(8):2855-60. doi:10.1210/jcem.83.8.5052. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/9709959>)

Genomic Location

The *HSD17B3* gene is found on chromosome 9 (<https://medlineplus.gov/genetics/chromosome/9/>).

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