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.

Chromosomal Location
Cytogenetic Location: 9q22.32, which is the long (q) arm of chromosome 9 at position 22.32
Molecular Location: base pairs 96,235,306 to 96,313,987 on chromosome 9 (Homo sapiens Updated Annotation Release 109.20200228, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI
Other Names for This Gene

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

Additional Information & Resources

Scientific Articles on PubMed

PubMed
https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28HSD17B3%5BTIAB%5D%29+OR+%28hydroxysteroid++++dehydrogenase+3%5BTIAB%5D%29%29+OR+%28%28estradiol+17+beta-dehydrogenase+3%5BTIAB%5D%29+OR+%2817-beta-HSD3%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- 17-BETA HYDROXYSTEROID DEHYDROGENASE III
  http://omim.org/entry/605573

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_HSD17B3.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=HSD17B3%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:3293
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P37058
Sources for This Summary

- OMIM: 17-BETA HYDROXYSTEROID DEHYDROGENASE III
  http://omim.org/entry/605573

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/8550739

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10599740

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18296911

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17466011

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12210481

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9709959

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