# AKR1D1 gene

aldo-keto reductase family 1 member D1

## Normal Function

The *AKR1D1* gene provides instructions for making an enzyme called 3-oxo-5-beta(β)-steroid 4-dehydrogenase. This enzyme is found in liver cells. It 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-oxo-5-β-steroid 4-dehydrogenase enzyme is responsible for the third step in that process, which converts 7alpha(α)-hydroxy-4-cholesten-3-one to 7α-hydroxy-5β-cholesten-3-one.

## Health Conditions Related to Genetic Changes

Congenital bile acid synthesis defect type 2

More than 10 mutations in the *AKR1D1* gene have been found to cause congenital bile acid synthesis defect type 2. 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 *AKR1D1* gene mutations replace single protein building blocks (amino acids) in the enzyme. These mutations result in production of a 3-oxo-5-β-steroid 4-dehydrogenase enzyme with severely reduced function. Without enough functional enzyme, the conversion of 7α-hydroxy-4-cholesten-3-one to 7α-hydroxy-5β-cholesten-3-one is impaired. The 7α-hydroxy-4-cholesten-3-one 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 digest fats. 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 2.

## Other Names for This Gene

- 3-oxo-5-beta-steroid 4-dehydrogenase
- 3o5bred
- 5-beta-reductase
- AK1D1_HUMAN
- aldo-keto reductase family 1, member D1

- delta 4-3-ketosteroid-5-beta-reductase
- delta(4)-3-ketosteroid 5-beta-reductase
- delta(4)-3-oxosteroid 5-beta-reductase
- SRD5B1
- steroid 5-beta-reductase

## Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of AKR1D1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6718[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28AKR1D1%5BTIAB%5D%29+OR+%28%28delta-4-3-ketosteroid-5-beta-reductase%5BTIAB%5D%29+OR+%285-beta-reductase%5BTIAB%5D%29+OR+%28steroid+5-beta-reductase%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)

Catalog of Genes and Diseases from OMIM

- ALDO-KETO REDUCTASE FAMILY 1, MEMBER D1; AKR1D1 (https://omim.org/entry/604741)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6718)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=AKR1D1[gene])

## References


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## Genomic Location

The *AKR1D1* gene is found on chromosome 7 (https://medlineplus.gov/genetics/chromosome/7/).

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