

## SRD5A3 gene

steroid 5 alpha-reductase 3

### Normal Function

The *SRD5A3* gene provides instructions for making an enzyme called steroid 5-alpha reductase 3. This enzyme performs a chemical reaction that converts a compound called polyprenol into another compound called dolichol. Subsequently, another enzyme converts dolichol to dolichol phosphate. The production of dolichol phosphate is critical for a process called glycosylation, by which small chains of sugar molecules (oligosaccharides) are attached to proteins. Glycosylation changes proteins in ways that are important for their functions.

Dolichol phosphate is integral for the formation of the sugar chains that are attached to proteins during glycosylation. Individual sugars are added to dolichol phosphate to build the oligosaccharide chain. Once the chain is formed, it is transferred from dolichol phosphate to a specific site on the protein that needs to be glycosylated.

### Health Conditions Related to Genetic Changes

#### SRD5A3-congenital disorder of glycosylation

More than 15 variants (also known as mutations) in the *SRD5A3* gene have been found to cause *SRD5A3*-congenital disorder of glycosylation (*SRD5A3*-CDG). This condition primarily causes neurological and vision problems but can also cause other signs and symptoms.

Variants in the *SRD5A3* gene generally lead to the production of abnormally small steroid 5 alpha-reductase 3 enzyme. This abnormal enzyme either has no activity or is quickly broken down. The lack of steroid 5 alpha-reductase 3 enzyme activity impairs the formation of dolichol, which also reduces dolichol phosphate production. As a result, glycosylation cannot proceed normally. The signs and symptoms of *SRD5A3*-CDG are likely due to impaired glycosylation of proteins that are needed for the normal function of various organs and tissues.

## Other Names for This Gene

- FLJ13352
- SRD5A2L
- SRD5A2L1

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of SRD5A3 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=79644\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=79644[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=SRD5A3>)

### Catalog of Genes and Diseases from OMIM

- STEROID 5-ALPHA-REDUCTASE 3; SRD5A3 (<https://omim.org/entry/611715>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/79644>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SRD5A3\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SRD5A3[gene]))

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