NR0B1 gene

nuclear receptor subfamily 0 group B member 1

Normal Function

The *NR0B1* gene provides instructions for making a protein called DAX1. This protein plays an important role in the development and function of several hormone-producing (endocrine) tissues in the body. These tissues include the small glands located on top of each kidney (the adrenal glands), two hormone-secreting glands in the brain (the hypothalamus and pituitary), and the gonads (ovaries in females and testes in males). Before birth, the DAX1 protein helps regulate genes that direct the formation of these tissues. DAX1 also helps regulate hormone production in endocrine tissues after they have been formed.

Health Conditions Related to Genetic Changes

X-linked adrenal hypoplasia congenita

Several variants (also called mutations) in the *NR0B1* gene have been found to cause X-linked adrenal hypoplasia congenita. This condition affects the development of the adrenal glands, which are endocrine organs (glands) located on top of each kidney. Some of the genetic changes are deletions of all or part of the *NR0B1* gene. Other variants lead to the production of an abnormally short version of the DAX1 protein. Still other variants change single protein building blocks (amino acids) in a critical region of DAX1.

Most of the variants responsible for X-linked adrenal hypoplasia congenita prevent the *NR0B1* gene from producing any active DAX1 protein. A shortage of DAX1 disrupts the normal development and function of endocrine tissues in the body. The main characteristics of this condition result when endocrine glands such as the adrenals, hypothalamus, pituitary, and gonads do not produce the right amounts of specific hormones.

Swyer syndrome

MedlinePlus Genetics provides information about Swyer syndrome

Other disorders

In some cases, genetic material is deleted from a region of the X chromosome that contains several genes, including *NR0B1*. This deletion results in a condition called adrenal hypoplasia congenita with complex glycerol kinase deficiency. In addition to the signs and symptoms of X-linked adrenal hypoplasia congenita (described above), individuals with this condition may have delayed development and problems regulating their blood sugar (glucose) levels. In rare cases, the deletion also includes the gene associated with Duchenne and Becker muscular dystrophy. People with this larger deletion have progressive muscle weakness and wasting in addition to the other features of adrenal hypoplasia congenita with complex glycerol kinase deficiency.

Other Names for This Gene

- AHC
- AHCH
- AHX
- DAX-1
- DAX1
- DSS
- gonadotropin deficiency
- GTD
- HHG
- NR0B1_HUMAN
- nuclear hormone receptor
- nuclear receptor DAX-1
- nuclear receptor subfamily 0, group B, member 1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of NR0B1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=190[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=((NR0B1%5BTI%5D)+OR+(DAX1%5BTI%5D))+AND+english%5Bla%5D+AND+human%5Bmh%5D)

Catalog of Genes and Diseases from OMIM

- NUCLEAR RECEPTOR SUBFAMILY 0, GROUP B, MEMBER 1; NR0B1 (https://omim.org/entry/300473)

- GLYCEROL KINASE DEFICIENCY; GKD (https://omim.org/entry/307030)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/190)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=NR0B1[gene])

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Genomic Location

The *NR0B1* gene is found on the X chromosome (https://medlineplus.gov/genetics/chromosome/x/).

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