NR5A1 gene

nuclear receptor subfamily 5 group A member 1

Normal Function

The *NR5A1* gene provides instructions for producing a transcription factor called the steroidogenic factor 1. A transcription factor is a protein that attaches (binds) to specific regions of DNA and helps control the activity of particular genes. Steroidogenic factor 1 helps control the activity of several genes related to the development of the gonads (ovaries and testes) and the adrenal glands, which are small glands located on top of each kidney. The adrenal glands produce several hormones that regulate many essential functions in the body.

Health Conditions Related to Genetic Changes

Swyer syndrome

NR5A1 gene variants (also called mutations) have been identified in a small number of people with Swyer syndrome, also known as 46,XY complete gonadal dysgenesis or 46, XY pure gonadal dysgenesis. Swyer syndrome is a condition that affects sex development.

Sex development usually follows a particular pattern based on an individual's chromosomes. People usually have 46 chromosomes in each cell. Two of the 46 chromosomes, known as X and Y, are called sex chromosomes because they help determine whether a person will develop male or female sex characteristics. Girls and women typically have two X chromosomes (46,XX karyotype), while boys and men typically have one X chromosome and one Y chromosome (46,XY karyotype). However, individuals with Swyer syndrome have a male-typical chromosome pattern (46,XY karyotype), but they develop female-typical sex characteristics.

Variants in the *NR5A1* gene in people with Swyer syndrome impair the function of steroidogenic factor 1. These changes affect the process of sex development, preventing affected individuals with a 46,XY karyotype from developing testes and causing them to develop female reproductive structures (a uterus and fallopian tubes).

46,XX testicular difference of sex development

MedlinePlus Genetics provides information about 46,XX testicular difference of sex

development

Other disorders

NR5A1 gene variants have been identified in people with 46,XY disorder of sex development, also known as partial gonadal dysgenesis. Affected individuals may have external genitalia that do not look clearly male-typical or clearly female-typical or other abnormalities of the genitals and reproductive organs. Affected individuals may also have abnormalities of the adrenal glands, which may cause hormone deficiencies, resulting in a variety of health problems. *NR5A1* gene variants that cause 46,XY disorder of sex development impair the function of steroidogenic factor 1, though likely to a lesser extent than variants that cause Swyer syndrome (described above).

NR5A1 gene variants that affect gonadal development and function have also been identified in people whose gonads do not produce reproductive cells (eggs or sperm). These conditions, which are called spermatogenic failure in men and primary ovarian insufficiency in women, result in an inability to conceive children (infertility).

Other Names for This Gene

- AD4BP
- adrenal 4 binding protein
- ELP
- FTZ1
- FTZF1
- fushi tarazu factor homolog 1
- hSF-1
- nuclear receptor AdBP4
- nuclear receptor subfamily 5, group A, member 1
- SF-1
- SF1
- steroid hormone receptor Ad4BP
- steroidogenic factor 1
- STF1_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of NR5A1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2516[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28NR5A1%5BTIAB%5D%29+OR+%28%28ELP%5BTIAB%5D%29+OR+%28SF1%5BTIAB%5D%29+OR+%28FTZ1%5BTIAB%5D%29+OR+%28SF-1%5BTIAB%5D%29+OR+%28AD4BP%5BTIAB%5D%29+OR+%28FTZF1%5BTIAB%5D%29+OR+%28steroidogenic+factor+1%5BTIAB%5D%29+OR+%28adrenal+4+binding+protein%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+AND+%22last+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- NUCLEAR RECEPTOR SUBFAMILY 5, GROUP A, MEMBER 1; NR5A1 (https://omim.org/entry/184757)
- PREMATURE OVARIAN FAILURE 7; POF7 (https://omim.org/entry/612964)
- SPERMATOGENIC FAILURE 8; SPGF8 (https://omim.org/entry/613957)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2516)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=NR5A1[gene])

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Genomic Location

The *NR5A1* gene is found on chromosome 9 (https://medlineplus.gov/genetics/chromosome/9/).

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