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.

Health Conditions Related to Genetic Changes

Swyer syndrome

NR5A1 gene mutations have been identified in a small number of people with Swyer syndrome, a condition affecting sexual development also known as 46,XY complete gonadal dysgenesis or 46,XY pure gonadal dysgenesis.

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 usually have one X chromosome and one Y chromosome (46,XY karyotype).

Mutations in the *NR5A1* gene in people with Swyer syndrome affect the process of sexual differentiation, preventing affected individuals with a 46,XY karyotype from developing male gonads (testes) and causing them to develop female reproductive structures (a uterus and fallopian tubes).

Other disorders

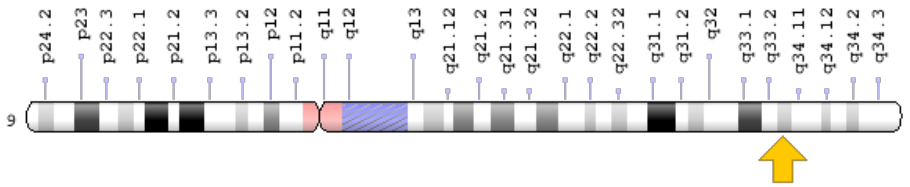
NR5A1 gene mutations have been identified in people with 46,XY disorder of sex development, also known as partial gonadal dysgenesis. Affected individuals may have external genitalia that do not look clearly male or clearly female (ambiguous genitalia) or other abnormalities of the genitals and reproductive organs. They may also have abnormalities of the adrenal glands, which produce several hormones that regulate many essential functions in the body. Adrenal gland abnormalities may cause a deficiency of these hormones, resulting in a variety of health problems.

NR5A1 gene mutations 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 premature ovarian failure in women, result in an inability to conceive children (infertility).

Chromosomal Location

Cytogenetic Location: 9q33.3, which is the long (q) arm of chromosome 9 at position 33.3

Molecular Location: base pairs 124,481,236 to 124,507,399 on chromosome 9 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

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

Educational Resources

- Developmental Biology (sixth edition, 2000): Chromosomal Sex Determination in Mammals
  https://www.ncbi.nlm.nih.gov/books/NBK9967/

Clinical Information from GeneReviews

- Nonsyndromic Disorders of Testicular Development
  https://www.ncbi.nlm.nih.gov/books/NBK1547

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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
  http://omim.org/entry/184757
- PREMATURE OVARIAN FAILURE 7
  http://omim.org/entry/612964
- SPERMATOGENIC FAILURE 8
  http://omim.org/entry/613957

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/NR5A1ID45858ch9q33.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=NR5A1%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:7983
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:2516
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2516
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