# SRY gene

sex determining region Y

## Normal Function

The *SRY* gene provides instructions for making a protein called the sex-determining region Y protein. This protein is involved in male sexual development, which is usually determined by the chromosomes an individual has. 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).

The *SRY* gene is found on the Y chromosome. The sex-determining region Y protein produced from this gene acts as a transcription factor, which means it attaches (binds) to specific regions of DNA and helps control the activity of particular genes. This protein starts processes that cause a fetus to develop male gonads (testes) and prevent the development of female reproductive structures (uterus and fallopian tubes).

## Health Conditions Related to Genetic Changes

### 46,XX testicular disorder of sex development

In most individuals with 46,XX testicular disorder of sex development, the condition results from an abnormal exchange of genetic material between chromosomes (translocation). This exchange occurs as a random event during the formation of sperm cells in the affected person's father. The *SRY* gene is misplaced in this disorder, almost always onto an X chromosome. A fetus with an X chromosome that carries the *SRY* gene will develop male characteristics despite not having a Y chromosome.

### Swyer syndrome

Mutations in the *SRY* gene have been identified in approximately 15 percent of individuals with Swyer syndrome, also known as 46,XY complete gonadal dysgenesis or 46,XY pure gonadal dysgenesis. *SRY* gene mutations that cause Swyer syndrome prevent production of the sex-determining region Y protein or result in the production of a nonfunctioning protein. A fetus whose cells do not produce functional sex-determining region Y protein will not develop testes but will develop a uterus and fallopian tubes, despite having a typically male karyotype.

other disorders

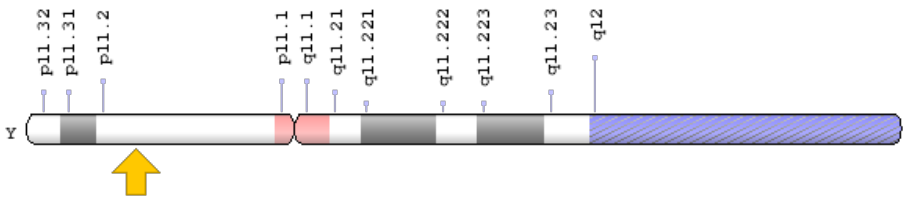
*SRY* gene mutations that impair but do not eliminate the function of the sex-determining region Y protein have been identified in a small number of people with 46,XY disorder of sex development, or partial gonadal dysgenesis. Affected individuals may have external genitalia that do not look clearly male or clearly female (ambiguous genitalia) or other abnormalities of the genitals and reproductive organs.

About 10 percent of individuals who have both testicular and ovarian tissue, a condition called ovotesticular disorder of sex development, have two X chromosomes with one carrying a misplaced copy of the *SRY* gene.

## Chromosomal Location

Cytogenetic Location: Yp11.2, which is the short (p) arm of the Y chromosome at position 11.2

Molecular Location: base pairs 2,786,855 to 2,787,741 on the Y chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)




Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- essential protein for sex determination in human males
- sex-determining region on Y
- sex determining region protein
- SRY_HUMAN
- TDF
- TDY
- testis-determining factor

## Additional Information & Resources

Educational Resources

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

GeneReviews

- Nonsyndromic 46,XX Testicular Disorders of Sex Development
  https://www.ncbi.nlm.nih.gov/books/NBK1416
- 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=%28%28SRY%5BTI%5D%29+OR+%28sex+determining+region+Y%5BTI%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+1080+days%22%5Bdp%5D

OMIM

- SEX-DETERMINING REGION Y
  http://omim.org/entry/480000

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SRY.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SRY%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11311
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/6736
- UniProt
  http://www.uniprot.org/uniprot/Q05066

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