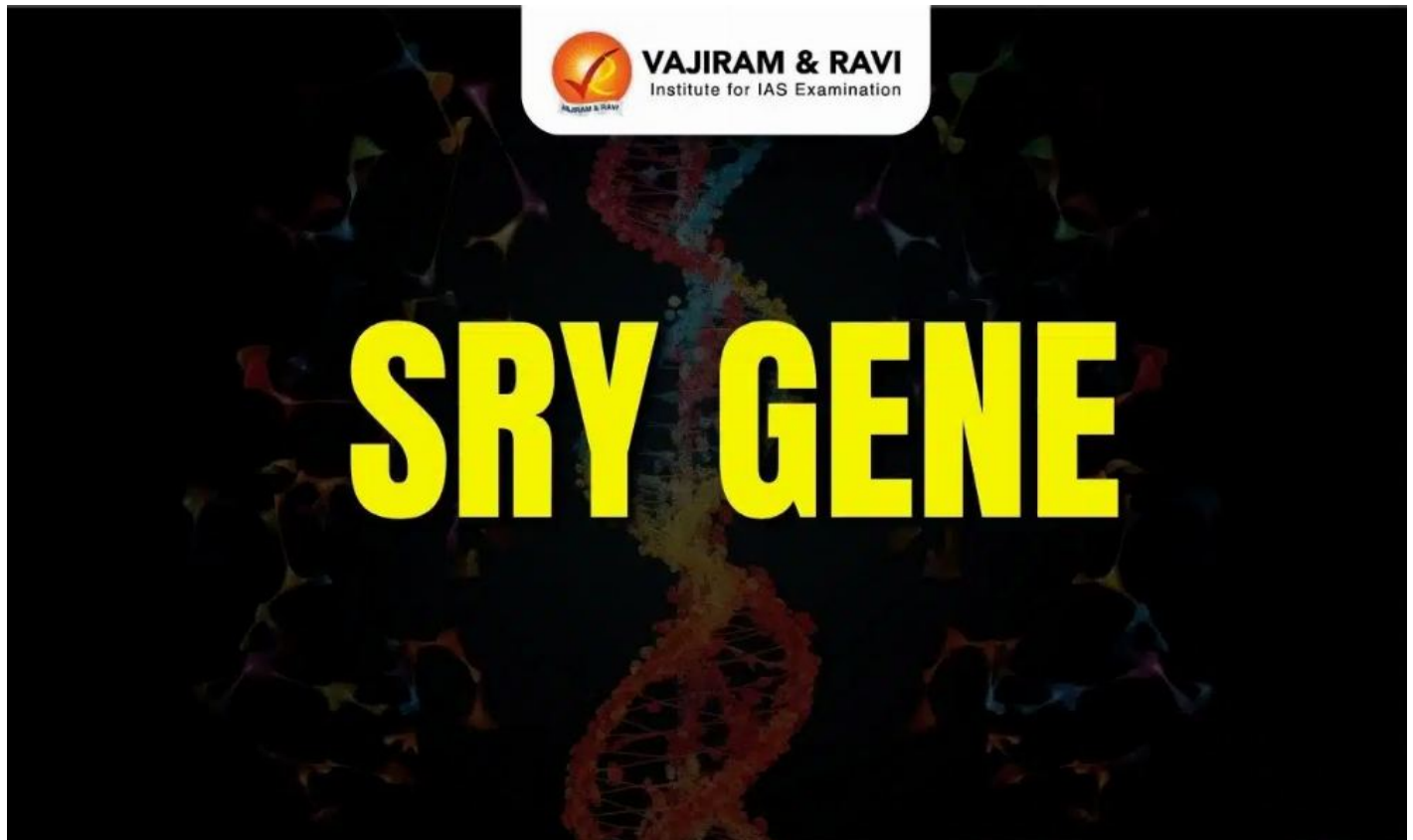


SRY (Sex-determining Region Y) Gene, Meaning, Role, Mechanism

October 1, 2025 • vajiramandravi.com



The SRY gene remains central to male sex determination, yet rare genetic anomalies can lead to unexpected outcomes. These exceptional cases underscore the complexity of sex differentiation and the need for careful genetic examination, especially of translocated chromosomes. Understanding such variations not only enhances scientific knowledge but also has practical implications for medical counseling, reproductive planning, and the management of DSDs. By studying these genetic anomalies, researchers and clinicians can better comprehend how chromosomal translocations, gene mutations, and X chromosome inactivation interact to influence sex development. This knowledge ultimately contributes to improved diagnostic accuracy, patient care, and guidance for families navigating these rare but significant conditions. In this article, we are going to cover SRY Gene, its mechanisms and its implications.

The SRY Gene (Sex-determining Region Y)

The SRY gene, located on the **Y chromosome**, plays an important role in determining whether a fetus develops as male or female. The presence of a functional SRY gene generally triggers male development, while its absence or mutation leads to female development. The gene produces the SRY protein, which binds to **DNA** and regulates the

activity of other genes essential for male differentiation. Specifically, the SRY protein initiates processes that form male gonads (testes) and inhibit the development of female reproductive structures.

Recent research has revealed extraordinary cases where individuals carrying the SRY gene, typically associated with male development, have developed as females. These cases show the intricate nature of genetic influence on sex determination and underscore the need for deeper understanding of chromosomal anomalies.

SRY Gene Mechanism of Sex Determination

- **Chromosomes:** Human cells contain 46 chromosomes arranged in 23 pairs. One of these pairs is the sex chromosomes, responsible for determining biological sex.
 - **Female sex:** Females possess two X chromosomes (XX).
 - **Male sex:** Males possess one **X chromosome** and one Y chromosome (XY).
- **Fertilization:** The sex of the zygote is determined at fertilization. If a sperm carrying an X chromosome fertilizes the egg, the zygote will be female (XX). If a sperm carrying a Y chromosome fertilizes the egg, the zygote will be male (XY), and the SRY gene will guide male differentiation.

SRY Gene Unusual Sex Development

- Although the SRY gene typically ensures male development, rare chromosomal anomalies can produce atypical outcomes. In some cases, the SRY gene can translocate from the Y chromosome to the X chromosome.
 - When a zygote inherits two X chromosomes but one carries the SRY gene, the individual generally develops male characteristics. However, such individuals are often infertile due to incomplete male reproductive development.
 - Remarkably, there are exceptional cases where individuals with two X chromosomes and the SRY gene develop as females.
- **X Chromosome Inactivation:** In these rare instances, the X chromosome carrying the SRY gene is often inactivated during early embryonic development. This process occurs because the SRY-carrying X chromosome may lack other essential genes. Turning it off helps prevent developmental complications, allowing the individual to develop as female despite the presence of the “male” gene.

Implications for Disorders of Sex Development (DSDs)

The study of the SRY gene is important for understanding Disorders of Sex Development (DSDs), a group of conditions where chromosomal, gonadal, or anatomical sex is atypical. Analysis of SRY translocation events and associated deletions can aid in the accurate diagnosis of DSDs.

- **Genetic counseling:** Families can receive informed guidance regarding potential reproductive issues and the likelihood of atypical sex development in offspring.
- **Medical management:** Identification of SRY anomalies can help healthcare providers design appropriate interventions, monitor hormonal development, and provide psychosocial support.