

SEX DETERMINING REGION GENE: SCIENCE & TECHNOLOGY

NEWS: *Will the baby be a boy or a girl? SRY gene claims to have the answer*

WHAT'S IN THE NEWS?

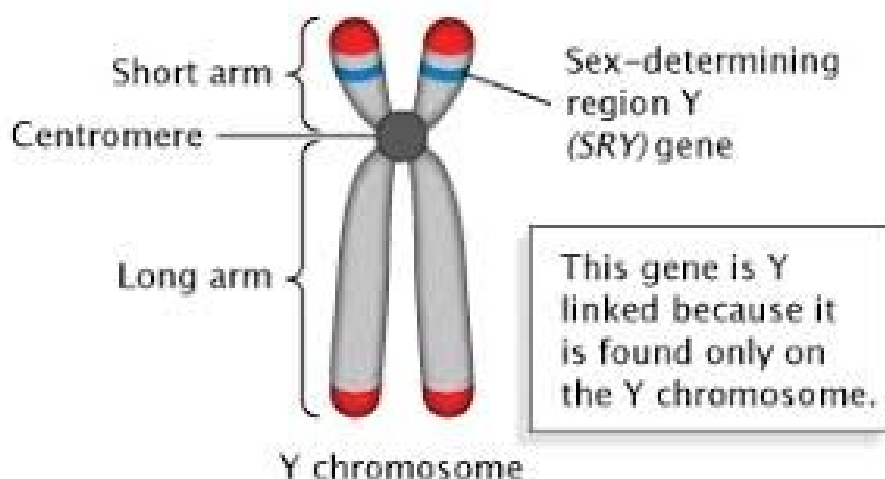
Recent studies from **Renato Dulbecco University Hospital, Italy, and Cincinnati Children's Hospital Medical Center, USA**, have reported rare cases of females possessing the **SRY gene**, which is normally present on the **Y chromosome**. These cases provide crucial insights into sex determination mechanisms, genetic mutations, and disorders of sex development. Additionally, research suggests that the **Y chromosome is undergoing gradual degeneration**, raising questions about the future of human sex determination.

1. What is the SRY Gene?

- **Full Form:** Sex-determining Region Y (SRY) gene.
- **Location:** Found on the **Y chromosome** in normal male development.
- **Function:** Acts as a **transcription factor**, triggering the development of **testes** from bipotential gonads.
- **Mechanism:**
 - The **SRY protein** binds to DNA and alters its structure.
 - This binding activates key genes like **SOX9**, which promotes **testis formation**.
 - The **testes then produce testosterone**, leading to **male secondary sexual characteristics**.

2. Role of SRY in Human Sex Determination

- **Genetic Basis of Sex Determination:**
 - Humans have **23 pairs of chromosomes**, including **one pair of sex chromosomes (XX or XY)**.
 - **Egg cells** always carry an **X chromosome**.
 - **Sperm cells** may carry either **X or Y**, determining the baby's sex at fertilization.
 - **XY = Male; XX = Female**.
- **SRY Gene Activation:**
 - In **normal males (46,XY)**, the **SRY gene is present and functional**, leading to male differentiation.
 - In **normal females (46,XX)**, the **SRY gene is absent**, leading to female development.

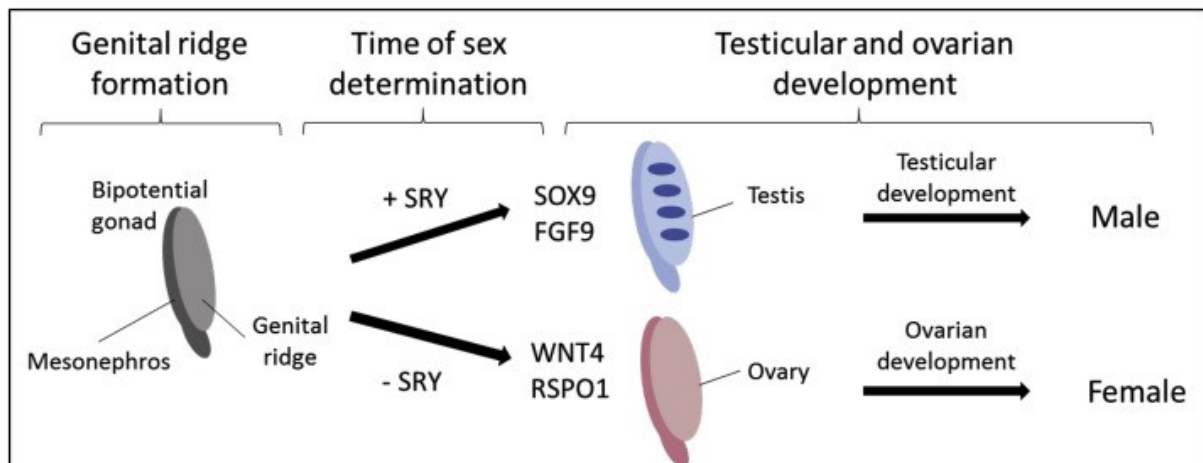


3. When the SRY Gene is Present in Females: 46,XX Testicular Disorder of Sex Development (DSD)

- **Cause:**
 - A **mutation causes the SRY gene to transfer (translocate) to an X chromosome.**
 - If a sperm carrying an **X chromosome with the SRY gene** fertilizes an egg, the resulting individual will have **two X chromosomes but still develop male characteristics.**
- **Features:**
 - **Male phenotype (appearance)** despite having an **XX chromosome pair.**
 - **Testes develop instead of ovaries**, but sperm production does not occur.
 - **Sterility** due to the absence of other Y chromosome genes essential for spermatogenesis.

4. When the SRY Gene Fails to Function: Swyer Syndrome (46,XY Gonadal Dysgenesis)

- **Cause:**
 - The **SRY gene is mutated or deleted**, preventing testis formation.
- **Features:**
 - Individuals have an **XY karyotype but develop female characteristics.**
 - **Gonads remain underdeveloped ("streak gonads")** and do not produce sex hormones.
 - **No puberty unless treated with hormone replacement therapy (HRT).**
 - Individuals have **female external genitalia but are infertile.**



5. Exceptional Cases: Females with the SRY Gene

- **Rare Occurrence:**
 - The **SRY gene can sometimes be translocated from the Y chromosome to an X chromosome.**
 - This happens due to a rare genetic mutation.
- **Genetic Mechanism:**
 - If a **sperm carrying an X chromosome with the SRY gene** fertilizes an egg, the resulting individual will have **XX chromosomes but develop testes instead of ovaries.**

- **Biological Impact:**
 - These individuals often appear **male at birth** but remain **sterile** due to the lack of other Y chromosome genes needed for sperm production.
 - The presence of **SRY alone is not sufficient for complete male reproductive function.**

6. Y Chromosome Degeneration: Future of Human Sex Determination

- **Scientific Findings:**
 - Studies indicate that the **Y chromosome is shrinking over evolutionary time.**
 - Over **millions of years, the Y chromosome may disappear**, leading to alternative mechanisms for sex determination.
- **Possible Future Adaptations:**
 - Some species have already lost their Y chromosomes but continue male differentiation through **alternative genes.**
 - Scientists are exploring whether the **SRY gene could relocate to another chromosome** to maintain sex differentiation in humans.

Source: <https://www.thehindu.com/sci-tech/science/will-the-baby-be-a-boy-or-a-girl-sry-gene-claims-to-have-the-answer/article69196574.ece>