

Turner Syndrome



Turner syndrome, a genetic condition resulting from the complete or partial loss of an X chromosome, is the most common congenital sex chromosomal condition in females^{1,2}

Turner syndrome affects multiple organ systems and is associated with a range of physical features and clinical manifestations, including:^{1,2}



Short stature



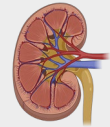
Ovarian
insufficiency



Cardiovascular
anomalies



Liver disease



Renal malformation

By the Numbers

Estimated
prevalence^{1,3-5,a}

1 in 2000 to 2500
live female births

Median age of
diagnosis, years⁶

15

Average deficit in final
adult height for
untreated individuals
compared to the
general female
population^{7-11,b}

20 cm

The Role of the *SHOX* Gene

- The *short stature homeobox (SHOX)* gene, which is located on the X and Y chromosomes, encodes a transcription factor that is expressed in developing skeletal tissue, and plays an important role in the differentiation and proliferation of chondrocytes^{12,13}
- Haploinsufficiency of the *SHOX* gene contributes to the short stature and other skeletal dysplasias observed in Turner syndrome^{1,14-16}

Management

According to international clinical guidelines for Turner syndrome²:

- Treatment goals include promotion of linear growth, maintenance of secondary sexual characteristics, and prevention of osteoporosis
- Growth hormone treatment is recommended as young as the age of 2 until epiphyseal closure to maximize the benefit of treatment

^aBased on epidemiological and newborn genetic screening data from Europe, Japan, and the United States. ^bBased on retrospective studies conducted in Europe, Japan, and the United States.

Abbreviations: cm, centimeter; SHOX, short stature homeobox.

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