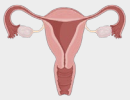
Turner Syndrome

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]

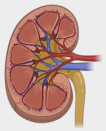
Short stature



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, years[6]	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 syndrome[2]:

- 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

[a]Based on epidemiological and newborn genetic screening data from Europe, Japan, and the United States. [b]Based on retrospective studies conducted in Europe, Japan, and the United States.

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

References: 1. Khan N, et al. *Orphanet J Rare Dis.* 2024;19(1):314. 2. Gravholt CH, et al. *Eur J Endocrinol.* 2024;190(6):G53-G151. 3. Stochholm K, et al. *J Clin Endocrinol Metab.* 2006;91(10):3897-3902. 4. Nielsen J, Wohlert M. *Hum Genet.* 1991;87(1):81-83. 5. Martin-Giacalone BA, et al. *Am J Med Genet A.* 2023;191(5):1339-1349. 6. Lin AE, et al. *Am J Med Genet A.* 2019;179(10):1987-2033. 7. Aversa T, et al. *Clin Ther.* 2024;46(2):146-153. 8. Aly J, Kruszka P. *Curr Opin Pediatr.* 2022;34(4):447-460. 9. Sybert VP, McCauley E. *N Engl J Med.* 2004;351(12):1227-1238. 10. Quigley CA, et al. *Horm Res Paediatr.* 2021;94(1-2):18-35. 11. Hindmarsh PC, Dattani MT. *Nat Clin Pract Endocrinol Metab.* 2006;2(5):260-268. 12. Marchini A, et al. *Endocr Rev.* 2016;37(4):417-448. 13. Hoyer-Kuhn H, et al. *J Pediatr Endocrinol Metab.* 2018;31(1):25-31. 14. Ellison JW, et al. *Hum Mol Genet.* 1997;6(8):1341-1347. 15. Rao E, et al. *Nat Genet.* 1997;16(1):54-63. 16. Rappold G, et al. *J Med Genet.* 2007;44(5):306-313.

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