

Turner Syndrome

Condition Description: Turner is caused by absence of all or part of one X chromosome. People who have Turner syndrome develop as females. The genes affected are involved in growth and sexual development, which is why girls with the disorder are shorter than normal and have abnormal sexual characteristics. Chromosome analysis is required to confirm the diagnosis.

Suspecting the diagnosis: The diagnosis can be considered at five different life stages:

1. Prenatal: Based on evidence of lymphedema or fetal hydrops. Turner syndrome is the most common chromosomal cause of miscarriage and fetal loss.
2. Neonatal: Evidence of lymphedema, congenital heart disease (40% of girls with coarctation of the aorta have Turner syndrome)
3. Childhood: Short stature
4. Adolescence: Delay or absence of puberty
5. Adulthood: Infertility

Clinical features¹:

1. Evidence of lymphedema (neck webbing, puffy extremities, narrow deep set nails)
2. Short stature (use Turner syndrome growth chart)
3. Prominent chest with wide-spaced nipples
4. Delayed or absent puberty
5. Normal cognition although specific patterns of learning disability may be present
6. Congenital heart disease (present in 17-45%, particularly coarctation of the aorta and bicuspid aortic valve. **Caveat—risk for aortic root dilation**)
7. Hypothyroidism (lifetime prevalence 15-30%)

Differential Diagnosis: [Noonan syndrome](#), any condition associated with short stature and/or delay or absence of puberty

Action: Chromosome analysis to confirm diagnosis. Echocardiogram at time of diagnosis. Consider referral for [clinical genetics evaluation](#). Follow management recommendations.¹⁻³

Reference & Resources:

1. [Eunice Kennedy Shriver National Institute for Child Health and Human Development: Turner syndrome.](http://turners.nichd.nih.gov/clinical.html) (<http://turners.nichd.nih.gov/clinical.html>)
2. AAP Committee on Genetics. [Health Supervision for Children with Turner syndrome](#) Pediatrics 111:691-702, 2003 (<http://aappolicy.aappublications.org/cgi/content/full/pediatrics;111/3/692>)
3. [Clinical Practice Guideline: Care of Girls and Women with Turner Syndrome: A guideline of the Turner syndrome study group.](#) J Clin Endocrinol Metab 92:10-25, 2007 (<http://jcem.endojournals.org/cgi/content/abstract/92/1/10>)
4. Genetics Home Reference – [Turner Syndrome](http://ghr.nlm.nih.gov/condition/turner-syndrome) (<http://ghr.nlm.nih.gov/condition/turner-syndrome>)

Patient Resources:

- [What is Turner Syndrome?](http://learn.genetics.utah.edu/content/disorders/whataregd/turner/) (learn.genetics.utah.edu/content/disorders/whataregd/turner/)
- [Turner Syndrome Society of the US](http://www.turnersyndrome.org/) (www.turnersyndrome.org/)