

Turner Syndrome

FAST FACTS

1/2,000–2,500

Turner syndrome affects 1 in every 2,000–2,500 live female births.

9 years

Signs may be evident during prenatal development, in infancy and the toddler years, but the average age of diagnosis is 9.

WHEN TO REFER MANAGEMENT/TREATMENT

For most girls and women, a karyotype should establish the diagnosis. Referral should follow for confirmed or suspected patients.

The Turner Syndrome Center at Cincinnati Children's provides multidisciplinary care, including appropriate and timely screening and interventions.

For more information or to discuss a case, contact the Division of Endocrinology at 513-636-4744.

Turner syndrome is a condition affecting girls and women in which all or part of one X chromosome is missing. These patients often have short stature, gonadal failure and cardiac defects, and are at risk for many other comorbidities.

With timely diagnosis and screening, specialists can identify and address comorbidities and limit their impact. Girls and women with Turner syndrome can live fulfilling and productive lives.

ASSESSMENT

Review medical, family and psychosocial history. Perform a physical exam.

Clinical Stature

A careful assessment of the patient's stature is critical. Does the patient have short stature with or without growth failure, particularly when compared to genetic expectations?

Stature may be "normal" for the general population but not normal for the particular family in the context of tall parents.

Health History

- Pubertal failure
- Frequent otitis media
- Classic left-sided heart defects (e.g. bicuspid aortic valve, coarctation)
- Renal defects
- Peripheral lymphedema (particularly at birth)

Physical Features

Typical dysmorphisms include low-set ears, pointed chin, down-slanting eyes, high palate, widely-spaced nipples and low hairline. Body proportions tend to be abnormal, with shorter legs and arms relative to trunk.

Psychosocial Features

- ADHD
- Difficulty with social skills
- Non-verbal learning disabilities

HPE (HISTORY AND PHYSICAL EXAM) RED FLAGS

If these red flags are present in a female patient, consider ordering a karyotype test (from blood) for Turner syndrome.

- Short stature that deviates from genetic context
- Typical dysmorphisms, particularly if accompanied by frequent otitis media or ADHD
- Pubertal failure
- Left-sided heart defects
- Renal defects

For urgent issues or to speak with the specialist on call 24/7, call the Physician Priority Link® at 1-888-987-7997.

If you would like additional copies of this tool, or would like more information, please contact the Physician Outreach and Engagement team at Cincinnati Children's.

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