Child With Suspected Turner Syndrome

Suggestive history and physical findings	Initial laboratory and/or radiologic work-up can include:	When to refer	Items useful for consultation	Additional information
Symptoms/Signs: • In utero cystic hygroma • Congenital lymphedema of hands and feet • Short stature • Absent/stalled puberty • High arched palate, micrognathia • Short neck, webbed neck, low posterior hair line • Low-set, posteriorly rotated ears • Cubitus valgus • Short 4th metacarpal • Nail hypoplasia • Bicuspid aortic valve, coarctation of aorta • Shield Chest • Multiple nevi • Emotional immaturity Differential Diagnosis	**Blood tests:** • Karyotype • LH, FSH Other tests to consider after consultation with Pediatric Endocrinologist: • Bone Age x-ray • Renal ultrasound • Echocardiogram • TSH, Free T4 • ALT, AST • Lipids • Audiology • Ophthalmology	**Routine:** Nearly always **Urgent:** Significant patient or family distress. Find a Pediatric Endocrinologist	Previous growth data/growth charts Pertinent medical records Recent laboratory and radiologic studies including the bone age film (not just the report)	Additional Information Link to Turner Syndrome Society Turner Syndrome: A Guide for Families References

Differential Diagnosis:

- Constitutional delay of growth and puberty: Usually height is < target height %
- Hypopituitarism with growth hormone and gonadotropin deficiency
- Isolated SHOX gene mutation
- Noonan's syndrome
- Hypogonadotropic hypogonadism
    - Possible associations: sensorineural hearing loss; anosmia/hyposomia; cleft palate; renal abnormalities
- Isolated Growth Hormone Deficiency
- Hypothyroidism
- Prolactinoma
- Chronic disease/ anorexia nervosa
- Other causes of ovarian failure (e.g. autoimmune)
- Mayer- Rokitansky- Kustner-Hauser syndrome
    - Mullerian duct agenesis/ congenital absence of the uterus and vaginal hypoplasia
    - May have associated renal and vertebral anomalies

Additional Information:

- TS may occur in as many as 3% of all fetuses and may cause up to 10% of all spontaneous fetal loss.
- Prenatal diagnosis is often incorrect, thus confirmatory peripheral blood from the baby is mandatory.
- Diagnosis: 20% at birth, 20% childhood short stature, 50% amenorrhea, 10% other.
- Up to 30% of girls have some degree of spontaneous puberty, especially high level mosaics.
- Elevated lifetime risk for: Hashimoto thyroiditis, Type 1 and Type 2 DM, celiac disease, significant hearing loss, reduced physical fitness, osteopenia, hepatic dysfunction, dyslipidemia, hypertension, aortic dilation and rupture.

Suggested References and Additional Reading:

- Levitsky, L. L., et al. (2015). Turner syndrome: update on biology and management across the life span. Curr Opin Endocrinol Diabetes Obes **22**(1): 65-72.
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- Gawlik, A. and E. Malecka-Tendera (2014). Transitions in endocrinology: treatment of Turner's syndrome during transition. Eur J Endocrinol **170**(2): R57-74.
- Pinsker, J. E. (2012). Clinical review: Turner syndrome: updating the paradigm of clinical care. J Clin Endocrinol Metab **97**(6): E994-1003.
- Bondy CA (2007). Care of girls and women with Turner syndrome: a guideline of the Turner Syndrome Study Group. J Clin Endocrinol Metab 92:10–25.
- Gravholt CH. (2004) Epidemiological, endocrine and metabolic features in Turner syndrome. *Eur J Endocrinol* **151:** 657–87.
- Gravholt CH. (2005) Clinical practice in Turner syndrome. *Nat Clin Pract Endocrinol Metab* **1:** 41–52.
- Stochholm K, Juul S, Juel K, Naeraa RW, Gravholt CH. (2006) Prevalence, incidence, diagnostic delay, and mortality in Turner syndrome. *J Clin Endocrinol Metab* **91:** 3897–902.

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