TURNER SYNDROME

- **~1/2500 live births**
- Complete or partial loss of X chromosome → 45,X
- Can occur w/ mosaicism

RISK FACTORS

- No established risk factors
- No link to maternal age
- Recurrence is rare

DIAGNOSIS

PRE-NATAL

- Standard prenatal screening does not accurately detect
- Suggestive US findings
  - Cystic hygroma
  - Hydrops
  - Lymphedema
  - Aortic coarctation
- Karyotype via CVS/amnio

INFANCY / CHILDHOOD

- Karyotype if suspected
- Infancy
  - ≥ 2 dysmorphic features
- Childhood
  - Short stature
  - No breast development
  - Amenorrhea
  - Infertility

PRESENTATION

CLASSIC FEATURES

- **Neonatal:** redundant nuchal skin, hand/foot lymphedema
- **Childhood:** short stature, ↓ growth velocity
- **Adolescent**: delayed puberty, lack of breast development, primary amenorrhea (↑ FSH)
- **General**
  - **Head:** webbed neck, low posterior hairline
  - **Ears:** posteriorly-rotated, looped helices
  - **Mouth:** malocclusion, micrognathia
  - **Body:** broad chest, widely-spaced nipples
  - **Extremities:** cubitus valgus, hyperconvex nails, short 4th metacarpals

COMPLICATIONS

- **OPT:** strabismus, congenital glaucoma
- **HEENT:** SN hearing loss, recurrent AOM
- **DENT:** orthodontic anomalies (e.g. malocclusion)
- **CV:** cardiac anomaly (33%), hypertension
  - Bicuspid aortic valve > aortic coarctation > progressive aortic root dilatation/dissection
- **GI:** transaminitis, Celiac, IBD
- **GU:** gonadal dysgenesis, renal malformation
  - Horseshoe kidney > duplicated/cleft renal pelvis
- **MSK:** congenital hip dysplasia, scoliosis, JIA
- **CNS:** developmental delay, learning disabilities
- **ENDO:** short stature, hypothyroid, T2DM, obesity
- **DERM:** keloid formation, pigmented nevi

MANAGEMENT

SURVEILLANCE

- Consult genetics + peds endo, use TS growth charts
- Refer to relevant clinical practice guidelines for details
- **OPT:** consult ophtho by 2yo, then yearly f/u
- **HEENT:** hearing test in childhood + adolescence
- **CV:** newborn echo + peds cardio consult, monitor BP/pulses/murmurs, repeat consult in adolescence
- **GI:** celiac screening + liver enzymes
- **GU:** renal US at Dx, repeat if recurrent UTIs/HTN
- **MSK:** observe for hip dysplasia + scoliosis
- **ENDO:** monitor puberty, thyroid, HbA1C, vitamin D

TREATMENT

- **Short Stature:** Growth Hormone → start in early childhood if height < 5th %ile + poor growth velocity
- **Gonadal Dysgenesis:** if Y-chromosome material present → prophylactic gonadectomy to ↓ risk of gonadoblastoma
- **Ovarian Failure:** start Estrogen by 13yo if no pubertal development, add Cyclic Progesterone after 2y, counsel regarding fertility
- **Lymphedema:** garments, exercises, diuretics
  - May be exacerbated by Estrogen

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