6. Turner Syndrome

DEFINITION AND INCIDENCE  (Ranke and Saenger, 2001)
- Turner syndrome (TS; also known as Ulrich-Turner syndrome): combination of characteristic physical features (short stature and gonadal dysgenesis) and complete or partial absence of 2nd X chromosome
- 1 in 2500 live births
- 1% of 45,X fetuses survive to term.
- 10% of spontaneous losses have 45,X karyotype (most common aneuploid in 1st trimester [TM] loss).
- Not associated with advanced maternal or paternal age
- http://www.turner-syndrome-us.org

DIAGNOSIS  (Ranke and Saenger, 2001)
- Ultrasound findings suggestive: ↑ nuchal translucency, cystic hygroma, coarctation of aorta ± left-sided cardiac defects, brachycephaly, renal anomalies, polyhydramnios, oligohydramnios, growth retardation
- Karyotype (chorionic villus sampling [CVS]/amniocentesis): necessary for diagnosis (confirm postnatally, if clinical suspicion is high and peripheral blood karyotype normal, then 2nd tissue should be checked)
- Potential mosaic karyotypes: 45,X/46,XX; 45,X/46,XY (mixed gonadal dysgenesis); 45,X/46,XX; 45,X/46,Xiq (phenotype, including stature, impossible to predict with mosaics, although there is clearly a ↓ fertility rate with ↑ risk of spontaneous loss and premature ovarian failure)

Indications for a Karyotype
- If virilization or fragment of sex chromosome of unknown origin (X or Y) is present, probe for Y chromosome (risk of gonadoblastoma → 7–10% [Gravholt et al., 2000]; if present, recommend gonadectomy).
- Any female patient with unexplained growth failure or pubertal delay
- Newborn/infant:
  - Edema of the hands/feet, nuchal folds, left-sided cardiac anomalies, coarctation of aorta, hypoplastic left heart, low hairline, low-set ears, small mandible
- Childhood:
  - Growth velocity <10th percentile for age
  - Markedly ↑ follicle-stimulating hormone (FSH)
  - Cubitus valgus, nail hypoplasia, hyperconvex uplifted nails, multiple pigmented nevi, characteristic facies, short 4th metacarpal, high arched palate