Turner syndrome

Genetics

-SHOX haploinsufficiency because of numeric or structural aberration of the sex chromosome

-SHOX (Xpter-p22.32)

Clinical findings/Dysmorphic features

-Short stature; gonadal dysgenesis; webbed neck; low posterior hairline; broad chest; widely spaced nipples; renal anomalies; cardiovascular anomalies (dilated aortic root, coarctation of the aorta, bicuspid aortic valve [30%]); hypertelorism and low set ears; lymphedema; lack of secondary sex characteristics; amenorrhea; usually normal intelligence; SNHL; Crohn’s disease; renal malformation; osteoporosis

Etiology

-TS occurs in 1:2,500 to 1:3,000 live female births

-99% of 45,X pregnancies lead to spontaneous abortions (Trisomy 21, only 80%)

Genetic testing/diagnosis

-45,X (50%), 46,X,i(Xq) (15%), 45,X/46,XX mosaic (15%), 45,X/46,X,i(Xq) mosaic (5%)

-Karyotype

Others

-Lifelong cardiac follow-up, at risk for aortic dilation and dissection with bicuspid aortic valve

-Cystic hygroma on