Saethre-Chotzen Syndrome

Genetics

-Gene: TWIST1 (Twist-related protein 1, 7p21)

-AD

Clinical findings/Dysmorphic features

-Coronal synostosis; facial asymmetry; ptosis; 2/3 hand syndactyly; mild-mod DD in minority; short; parietal foramina; vertebral fusions; radioulnar synostosis; CP; maxillary hypoplasia; CHD

-Characteristic appearance of the ear (small pinna with a prominent crus)

-Broad or duplicated great toes (eventually pointing away from each other)

Etiology

-Prevalence estimates range from 1:25,000 to 1:50,000

Pathogenesis

-Haploinsufficiency by gene deletion/rapid degradation of abnormal protein/altered subcellular localization --> disinhibition of RUNX2 and enhanced osteogenesis

Genetic testing/diagnosis

-TWIST1 (Seq 72%; InDel 23%)