Stickler Syndrome

Genetics

-Gene: COL2A1 (AD), COL11A1 (AD), COL11A2 (AD), COL9A1 (AR), COL9A2 (AR), COL9A3 (AR)

Clinical findings/Dysmorphic features

-Connective tissue disorder; ocular findings (myopia, cataract, retinal detachment); HL (conductive and sensorineural); midfacial underdevelopment and cleft palate (either alone or as part of the Robin sequence); mild spondyloepiphyseal dysplasia and/or precocious arthritis

-Variable phenotypic expression both within and among families (locus/allelic heterogeneity)

Etiology

-Incidence among neonates is 1:7,500-1:9,000

Pathogenesis

-Haploinsufficiency of type II collagen --> vestigial gel forms in the retrolental space

Genetic testing/diagnosis

-COL2A1 (80-90%): Seq 99%; COL11A1 (10-20%): Seq 99%