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Stickler Syndrome

National Cancer Institute

Source

National Cancer Institute. Stickler Syndrome. NCI Thesaurus. Code C74984.

A rare autosomal dominant syndrome caused by mutations in the COL11A1, COL11A2, and COL2A1 genes which affect the production of type II and XI collagen. It is characterized by a range of signs and symptoms including cleft palate, large tongue, small lower jaw, hearing loss, myopia, glaucoma, retinal detachment, skeletal, and joint abnormalities.