HEB

STICKLERS SYNDROME



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ABSTRACT

Stickler syndrome is a group of hereditary conditions characterized by a distinctive facial appearance, eye abnormalities, hearing loss, and joint problems. These signs and symptoms vary widely among affected individuals. A characteristic feature of Stickler syndrome is a somewhat flattened facial appearance. Eye problems. In addition to severe near sightedness, children who have Stickler syndrome often experience cataracts, glaucoma and retinal detachments, hearing difficulties. The extent of hearing loss varies among people who have Stickler syndrome. Bone and joint abnormalities. Your child is more likely to be born with Stickler syndrome if either you or your partner has the disorder. Overall, the estimated prevalence of Stickler syndrome is about 1 in 10,000 people. Stickler syndrome affects 1 in 7,500 to 9,000 newborns.

Stickler syndrome is caused by mutations in certain genes involved in the formation of collagen — one of the building blocks of many types of connective tissues. The type of collagen most commonly affected is that used to produce joint cartilage and the vitreous jelly found within the eyes. Therapy: Speech therapy, Physical therapy, Hearing aids, Special education. Surgery: Tracheostomy, Jaw surgery, Cleft palate repair, Ear tubes, Eye surgeries, Joint replacement, Spinal bracing or fusion surgeries.

