Stickler Syndrome

WHAT IS STICKLER SYNDROME?

Stickler syndrome is a progressive genetic disorder of connective tissue throughout the body. The condition was first described by Dr. Gunnar B. Stickler in 1965 and was originally called "Hereditary Progressive Arthro-ophthalmopathy" because of its tendency to affect the joints and the eyes.

WHAT ARE THE FEATURES OF STICKLER SYNDROME?

Stickler Syndrome is associated with problems of vision (severe nearsightedness and retinal detachments), hearing (hearing loss and frequent ear infections), craniofacial abnormalities (small noses and chins, cleft palates) musculoskeletal abnormalities (arthritis, neck and back problems and loose joints) as well as other problems caused by abnormal collagen. A particular group of physical features, called Pierre Robin sequence, is also common in people with Stickler Syndrome. Pierre Robin sequence includes cleft palate, a large tongue (macroglossia), and a small lower jaw (micrognathia).

WHAT CAUSES STICKLER SYNDROME?

Stickler Syndrome is usually caused by a mutation in the Type II pro-collagen (COL2A1) gene, although several other COL genes mutations have also been identified. These mutations cause abnormalities in the formation of connective tissues (collagen) throughout the body and give rise to the various features of Stickler Syndrome.

HOW IS STICKLER SYNDROME INHERITED?

Extreme myopia (nearsightedness) is one of the earliest and most characteristic signs of Stickler Syndrome. The associated thin peripheral retina can lead to retinal breaks, holes, and retinal detachment and scarring which can permanently reduce vision. Cataracts (clouding of the lens in the eye) can reduce vision, and typically occur at a younger age in individuals with Stickler Syndrome.

HOW IS STICKLER SYNDROME DIAGNOSED?

A 9-point system is used to diagnose Stickler syndrome based on the number of oral-facial, ocular, auditory and skeletal abnormalities detected. In addition, points are given for family history or the presence of a mutation in one of the genes known to be associated with Stickler Syndrome. Diagnosis requires 5 of the 9 points.

HOW IS STICKLER SYNDROME TREATED?

Early evaluation with regular, long term follow-up is essential. Glasses and/or contact lenses are utilized for myopia (near-sightedness). Laser or cryotherapy may be applied to areas of thin retina to reduce the risk of detachment. Additional retina surgery may be necessary to repair retinal detachments when or if they occur. Significant cataracts may also require surgery.

WHERE IS THERE MORE INFORMATION ABOUT STICKLER SYNDROME?

- Stickler Involved People

Updated 02/2018