Stickler Syndrome

WHAT IS STICKLER SYNDROME?

Stickler syndrome is an inherited medical problem (runs in families) with the connective tissue throughout the body. It is present since birth and gets worse throughout life. This condition was first described by Dr. Gunnar B. Stickler in 1965 and was originally called "Hereditary Progressive Arthro-ophthalmopathy" because often affects the joints and the eyes.

WHAT ARE THE FEATURES OF STICKLER SYNDROME?

Stickler Syndrome causes 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 weak collagen (connective tissue).

Very large amounts of myopia (nearsightedness) is one of the earliest classic signs of Stickler Syndrome. When the eye has a lot of myopia, the retina (inner lining of the back wall of the eye) is very thin and at risk for breaks, holes, retinal detachment and retinal scarring which can cause permanent blurry vision. Cataracts (clouding of the lens in the eye) can cause blurry vision as well, and can happen at a young age in people with Stickler Syndrome.

A group of face and jaw problems, called Pierre Robin sequence, is also common in people with Stickler Syndrome. Pierre Robin sequence includes cleft palate (problem with development of the roof of the mouth), 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 other gene mutations have also been linked with Stickler Syndrome. These mutations cause problems in how connective tissue (collagen) is made throughout the body. This collagen problem leads to the different signs in the body for Stickler Syndrome, as mentioned above.

The gene mutations that cause Stickler syndrome are often inherited (run in families) but some can happen sporadically (on their own without a family history).

HOW IS STICKLER SYNDROME DIAGNOSED?

Any child who gets large amounts of myopia at a young age should be checked to see if they have Stickler syndrome. Those with a family history of large amounts of myopia and retinal detachment early in life or other medical problems (like cleft palate, small face, hearing loss and very flexible joints) should also be checked for Stickler syndrome.

A full eye exam with an ophthalmologist can look for signs of Stickler syndrome. Many children can also be helped by an exam with a genetics doctor and gene testing. This can help diagnose most children who have Sticklers syndrome early so that they can get treatment as needed.

HOW IS STICKLER SYNDROME TREATED?

Early eye check-ups with regular, long-term follow-up is very important to help protect vision. The type of treatment depends on what problems with person with Stickler syndrome develops. Glasses and/or contact lenses are used to treat myopia (near-sightedness). Referral to a retina specialist who has experience treating children may be recommended, Laser or cryotherapy (freezing) treatments to thin spots in the retina may be recommended to help reduce the risk of detachment and vision loss. Additional retina surgery may be necessary to fix retinal detachments when or if they occur. Cataracts may need to be treated with glasses, contact lenses or surgery.

WHERE IS THERE MORE INFORMATION ABOUT STICKLER SYNDROME?

- Stickler Involved People
- https://aapos.org/glossary/stickler-syndrome

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