Stickler Syndrome

WHAT IS STICKLER SYNDROME?

Stickler syndrome is an inherited condition that affects the connective tissue in the body. Connective tissue helps support many parts of the body. Stickler syndrome is present at birth and can cause health problems throughout life.

The condition was first described by Dr. Gunnar B. Stickler in 1965. It was first called “Hereditary Progressive Arthro-ophthalmopathy” because it often affects the joints and eyes.

WHAT ARE THE FEATURES OF STICKLER SYNDROME?

Stickler syndrome can affect many parts of the body. Common problems include:

- **Eyes:** Severe nearsightedness (myopia), tears of the retina, and retinal detachment
- **Ears:** Hearing loss and frequent ear infections
- **Face and mouth:** A small nose or lower jaw and a cleft palate
- **Bones and joints:** Loose joints, arthritis, and neck or back problems

Very strong myopia **(nearsightedness)** is one of the most common early signs of Stickler syndrome. When the eye has severe (high) myopia, the retina can become very thin. The retina is the thin layer at the back of the eye that senses light. A thin retina is more likely to develop tears or holes. This can lead to **retinal detachment**, which can cause permanent loss of vision.

Cataracts can also occur at a young age in people with Stickler syndrome. A cataract is a clouding of the lens inside the eye. Cataracts can cause blurry vision.

Some children with Stickler syndrome have a group of face and jaw problems called **Pierre Robin sequence**. This can include:

- A cleft palate, which is an opening in the roof of the mouth
- A tongue more likely to flop back in the throat
- A small lower jaw

WHAT CAUSES STICKLER SYNDROME?

Stickler syndrome is caused by a change, or **mutation**, in a gene. The gene most often involved is called **COL2A1**. Other genes that help make collagen can also cause Stickler syndrome.

Collagen is a protein that helps give strength and support to tissues in the body. Changes in these genes can cause the body to make collagen in an abnormal way. This leads to the many problems that can occur with Stickler syndrome.

Stickler syndrome often runs in families. However, it can also happen in a child who has no family history of the condition.

HOW IS STICKLER SYNDROME DIAGNOSED?

Children who develop severe myopia at a young age should be checked for Stickler syndrome. Children may also need to be checked if someone in their family has severe myopia, retinal detachment at a young age, or other signs of Stickler syndrome.

Other signs may include:

- Cleft palate
- A small lower jaw or other facial differences
- Hearing loss
- Very flexible or loose joints

A complete eye exam by an **ophthalmologist (eye M.D.)** can help find signs of Stickler syndrome. A visit with a **genetics doctor** may also be recommended. Genetic testing can help confirm the diagnosis in many children.

Finding Stickler syndrome early is important. Early diagnosis allows doctors to watch for problems and treat them when needed.

HOW IS STICKLER SYNDROME TREATED?

Regular eye exams are very important for children with Stickler syndrome. These exams often use dilating eye drops so the doctor can carefully examine the retina. Regular follow-up visits help protect vision.

Treatment depends on the problems that develop. Treatment may include:

- **Glasses or contact lenses** to correct myopia (nearsightedness)
- **Care from a retina specialist**, especially when there are changes in the retina
- **Laser treatment or cryotherapy (freezing treatment)** to treat weak areas of the retina and lower the risk of retinal detachment
- **Retinal surgery** if a retinal detachment develops
- **Cataract surgery** if a cataract causes enough vision loss to interfere with daily activities

SUMMARY FOR PATIENTS AND CAREGIVERS

Stickler syndrome is an inherited condition that can affect the eyes, ears, face, bones, and joints. Severe nearsightedness and problems with the retina are common. Because retinal tears or detachment can cause permanent vision loss, regular eye exams are very important. Early diagnosis and treatment can help protect vision. Children with Stickler syndrome may also need care from other specialists, such as genetics, hearing, or orthopedic doctors. If your child has Stickler syndrome, keep all recommended follow-up visits and tell the eye doctor right away about any new changes in vision.

WHERE IS THERE MORE INFORMATION ABOUT STICKLER SYNDROME?

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

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