Stickler syndrome

Overview

Stickler syndrome is a genetic disorder that can cause serious vision, hearing and joint problems. Also known as hereditary progressive arthro-ophthalmopathy, Stickler syndrome is usually diagnosed during infancy or childhood.

Children who have Stickler syndrome often have distinctive facial features — prominent eyes, a small nose with a scooped-out facial appearance and a receding chin. They are often born with an opening in the roof of the mouth (cleft palate).

While there is no cure for Stickler syndrome, treatments can help control symptoms and prevent complications. In some cases, surgery may be needed to correct some of the physical abnormalities associated with Stickler syndrome.

Symptoms

The signs and symptoms of Stickler syndrome — and the severity of those signs and symptoms — can vary widely from person to person, even within the same family.

- **Eye problems.** In addition to having severe nearsightedness, 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. It usually affects the ability to hear high frequencies.
- **Bone and joint abnormalities.** Children who have Stickler syndrome often have overly flexible joints and are more likely to develop abnormal curvatures of the spine, such as scoliosis. Osteoarthritis can begin in adolescence.

When to see a doctor

Regular follow-up visits, as well as yearly visits to doctors specializing in eye disorders, are crucial to monitor any progression of symptoms. Early treatment can help prevent life-altering complications. Hearing should be checked every six months in children through age 5 and then yearly thereafter.

Causes

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 jellylike material (vitreous) found within the eyes.

Risk factors

Your child is more likely to be born with Stickler syndrome if either you or your partner has the disorder.

Complications

Potential complications of Stickler syndrome include:

- **Difficulty breathing or feeding.** Breathing or feeding difficulties may occur in babies born with an opening in the roof of the mouth (cleft palate), a small lower jaw and a tendency for the tongue to drop back toward the throat.
- **Blindness.** Blindness can occur if retinal detachments aren't repaired promptly.
- **Ear infections.** Children with facial structure abnormalities associated with Stickler syndrome are more likely to develop ear infections than are children with normal facial structure.
- **Deafness.** Hearing loss associated with Sticker syndrome may worsen with time.
- **Heart problems.** Some people with Stickler syndrome may be at higher risk of heart valve problems.
- **Dental problems.** Most children who have Stickler syndrome have abnormally small jaws, so there often isn't enough room for the full complement of adult teeth. Braces or, in some cases, dental surgeries may be necessary.

By Mayo Clinic Staff