

STICKLER SYNDROME

Stickler syndrome is a group of genetic disorders that affects connective tissue, specifically collagen. The condition is characterized by facial abnormalities, ocular problems, hearing loss, and joint problems.

Stickler syndrome affects the eyes, ears, bones, joints, face, and teeth, but most people do not experience all of its features.

What other names do people use for Stickler syndrome?

Stickler syndrome is also referred to as hereditary arthro-ophthalmo-dystrophy, hereditary arthro-ophthalmopathy, and Stickler dysplasia.

How prevalent is Stickler syndrome?

Stickler syndrome affects an estimated 1 in 7,500 to 9,000 newborns.

What are the features of Stickler syndrome?

Features associated with Stickler syndrome are:

Eyes

- Nearsightedness (myopia)
- Retinal detachment
- Cataracts (clouding of the eye lens)
- Astigmatism (cornea or lens not spherical)
- Vitreous degeneration (gel within eye liquefies and pulls away from the retina)
- Strabismus (eyes move independently of each other)
- Glaucoma (elevated eye pressure)

Ears/Hearing

- Inner ear hearing loss
- Otitis media (frequent ear infections)

Bones/Joints

- Joint pain/enlarged joints
- Osteoarthritis (degenerative joint disease)
- Hyper-mobile joints (loose-jointed)
- Genu valgum (knock-kneed)
- Scoliosis (curvature of the spine)
- Legg-Perthes (degenerative hip disease)

Oral/Facial

- Flat cheeks and nasal bridge (most noticeable in infants)
- Small jaw
- Palate (roof of the mouth) abnormalities/obstructed airway
- Split uvula (the tissue that hangs down in the back of the throat is divided)
- Pierre-Robin sequence (small jaw, cleft palate, tongue placement abnormalities and breathing problems)

People with Stickler syndrome do not usually have all features that are associated with the condition. Features even vary among affected members of the same family. In some cases, a person may have multiple features, but only one problem is severe enough to be diagnosed. Stickler syndrome is also a progressive disorder; therefore, the symptoms are likely to worsen with age.

What causes Stickler syndrome?

Types I, II, and III Stickler syndrome are caused by mutations in the COL2A1, COL11A1, and COL11A2 genes, respectively. They are inherited in an autosomal dominant pattern, which means one copy of the altered gene is sufficient to cause the disorder. In some cases, an affected person inherits a gene mutation from an affected parent. In other cases, a person can be the first in their family to have Stickler syndrome. This would be the result of a new mutation.

Types IV and V Stickler syndrome are caused by mutations in the COL9A1 or COL9A2 gene. They are inherited in an autosomal recessive pattern. Autosomal recessive inheritance means both copies of the gene in an affected individual have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

The condition occurs equally in males and females.

How is Stickler syndrome diagnosed?

The diagnosis of Stickler syndrome can be made through a clinical exam and a genetic test.

Diagnosis through a clinical exam is based on diagnostic criteria, using a 12-point system established by the National Institutes of Health. The physician evaluates changes in the eyes, ears, bones and joints, face, and palate (roof of the mouth). Family medical history is also considered.

Genetic testing is another way to diagnose Stickler system. It can be used to test for the known gene mutation locations (COL2A1, COL11A1, COL11A2, COL9A1, and COL9A2). An inconclusive genetic test does not mean a person does not have Stickler syndrome because a person may have a gene mutation that has not yet been identified. There are at least three unknown gene mutation locations.

How is Stickler syndrome managed?

Common treatments for people with Stickler syndrome can improve or correct many of its features.

- Glasses or contact lenses can correct vision.

- Frequent retinal exams can track any changes or deterioration in the retina; then, preventative laser retinal surgery can be done before the retina detaches.
- Hearing aides can be used to overcome hearing loss.
- Bone and joint problems, which may require a rheumatology evaluation including a full-body x-ray, can be addressed with splints, braces, or aids; pain management/anti-inflammatory medications; and/or rehabilitation or physical therapy.
- Children with Stickler syndrome and/or Pierre-Robin sequence (PRS) may need a feeding evaluation, palate or jaw surgery, orthodontics, or speech therapy.

What is the life expectancy of someone with Stickler syndrome?

Stickler syndrome is not thought to alter life expectancy.

Do you have questions? Would you like more information?

- Call our help center, 800-862-7326, ext. 126 to speak with a nurse who can answer your questions and send you additional information.
- Visit our website at marfan.org. You can print information that interests you and ask questions online.