Stickler Syndrome
Basic Information for Patients and Families

Stickler syndrome is a genetic disorder affecting collagen throughout the body. Collagen is a primary part of connective tissue like bone, skin and cartilage. Stickler syndrome is an autosomal dominant genetic condition. This means it can occur equally in males or females (autosomal) and the gene's traits will be exhibited in the child if the gene is passed on by the affected parent (dominant). Stickler syndrome is also a progressive disorder, so the symptoms are likely to become more severe with age.

It is a disorder primarily characterized by problems with vision, joints and hearing. A person with Stickler syndrome, however, can live a long, happy and healthy life – it is a disorder, not a disease!

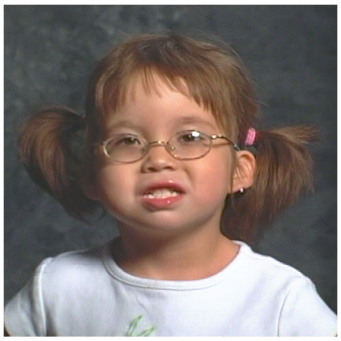
Diagnosis of Stickler syndrome can be made two ways. The first method is clinical diagnosis using the Diagnostic Criteria published by the National Institutes of Health in 2005. A diagnosis is made by looking at a patient's medical/family history and a thorough examination by a physician. The Diagnostic Criteria is a 12-point system which measures abnormalities in the following systems: eyes, ears, bones/joints, and face/palate, as well as family history or gene data. A score of 5 or more indicates Stickler syndrome. The criteria can be found at www.sticklers.org.

Stickler syndrome can also be diagnosed by using genetic testing. Stickler syndrome symptoms are caused by changes in certain genes. The known gene mutation locations are called COL2A1, COL11A1 and COL11A2. An inconclusive genetic test does not mean a person does not have Stickler syndrome. A person may have a gene mutation that has not yet been identified. There are at least three unknown gene mutation locations.

The prevalence of Stickler syndrome is estimated to be 1 in 7500, but it is thought to be frequently undiagnosed or misdiagnosed. When a mother or father has Stickler syndrome, there is a 50% chance that the Stickler-causing gene will be passed on to a child, with each pregnancy. Unlike some genetic syndromes, a person will not have a "carrier" gene for this disorder that can be passed on to the next generation without the parent having Stickler syndrome themselves. Occasionally, though, a person will have a spontaneous genetic mutation that will cause Stickler syndrome without either parent having the disorder.

Common Symptoms:

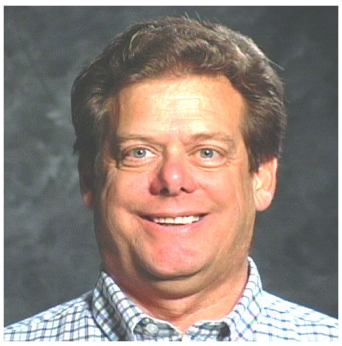
Eyes
- Myopia (near sightedness)
- Retinal detachment (retina separates from the back of the eye causing partial or total loss of sight)
- Cataracts (clouding of the eye lens)
- Astigmatism (cornea or lens not spherical)
- Vitreous degeneration (gel within eye liquefies and pulls away from the retina)
- 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

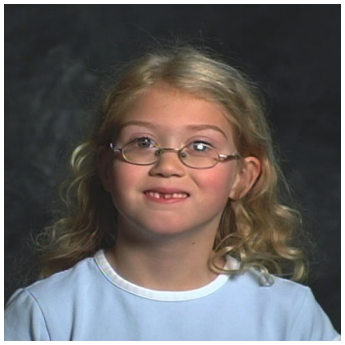
- Flat cheeks and nasal bridge (most noticeable in infants)
- Small jaw
- Palate abnormalities/obstructed airway
- Split uvula (the tissue that hangs down in the back of the throat is divided)
- Orthodontia issues
- Pierre-Robin sequence (small jaw, cleft palate, tongue placement abnormalities and breathing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

People with Stickler syndrome may have problems in many of the body systems listed or just a few. Affected family members may not exhibit the same symptoms. Many symptoms may be so mild that they go undiagnosed or do not need treatment.

Common treatments for people with Stickler syndrome can improve or correct many of the symptoms of this disorder. For example, glasses or contact lenses can correct vision and frequent retinal exams are important to track any changes or deterioration in the retina. Prophylactic (preventative) laser retinal surgery can be done to help the retina stay attached. Individuals with Stickler syndrome should have a base-line hearing evaluation and may need hearing aides. For bone and joint problems, a rheumatology evaluation including a full-body x-ray may be recommended. Skeletal issues may be improved by 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.

If you *think* you have Stickler syndrome ask your physician to refer you to a genetic counselor who can review your information and family history.

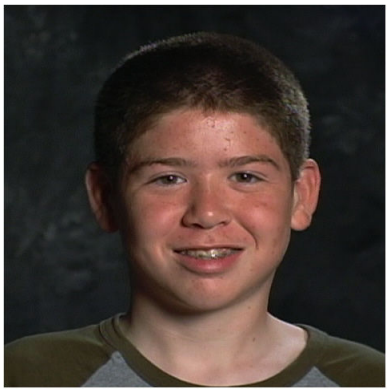
If you *know* you have Stickler syndrome, be sure to take educational materials to each of your physicians. Feel comfortable that your doctors are open to learning more about your medical condition. Recognize that there is a 50% chance that each of your children will be born with Stickler syndrome and be prepared through prenatal genetic counseling. Pursue an Individualized Education Program (IEP) or a 504 plan for any affected school-age child and be an advocate for your child. Consider modification of physical education class. Be smart about physical/sports involvement for the sake of your child's or your own joints and eyes. **Talk with your doctor.**



Stickler syndrome does not affect life expectancy or mental capacity. As noted above, it is a disorder, not a disease – affected individuals can live healthy, happy lives. This disorder does, however, have symptoms that should be monitored and managed. It is the most common cause of retinal detachment in children, so get frequent exams. Also, as with any major medical issue, it affects all family members, whether they have the disorder or not, so address spouse and sibling issues, as well.

Resources and References

- Stickler Syndrome Clinical Characteristics and Diagnostic Criteria, American Journal of Medical Genetics, 138A: 199-207, Wiley-Liss Inc., 2005.
- Stickler Syndrome, Francomano, Wilkin, Liberfarb, Management of Genetic Syndromes, 2nd edition, Wiley-Liss Inc., 2005.
- www.sticklers.org offers up-to-date information on the disorder, registration information for the annual conference, a physician-oriented brochure, information on a 30-minute DVD and a slide presentation.
- Dave Hawley's Stickler Syndrome page at http://members.aol.com/dhawley/stickler.html



This information has been prepared by Stickler Involved People. The mission of Stickler Involved People (SIP), a not-for-profit organization, is to educate and give support to all those affected by Stickler syndrome.

www.sticklers.org

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