

# Stickler syndrome...

affects connective tissues throughout the body, most notably the eyes, ears, face and joints.

is inherited in an autosomal dominant manner, with a 50 % recurrence in offspring of affected individuals. Family history is key in aiding diagnosis.

has a prevalence of 1 in 7500, but it is currently underdiagnosed.

is caused by mutations in the collagen genes. Molecular genetic testing is available in CLIA approved labs, although new clinical diagnostic criteria have been 98% reliable.

was originally published under the name Hereditary Progressive- Arthro-Ophthalmopathy.

is the most common cause of retinal detachments in children.

is believed to be the most common connective tissue disorder.

Information for this brochure has been gathered from a wide variety of published articles, including:

Stickler Syndrome Clinical Characteristics and Diagnostic Criteria, American Journal of Medical Genetics, 138A: 199-207, Wiley-Liss, Inc. 2005. (1)

Stickler Syndrome, Clair Francomano, Douglas J. Wilkin, Ruth M. Liberfarb, Management of Genetic Syndromes, Second Edition, Edited by Suzanne B. Cassidy and Judith E. Allanson, Wiley-Liss, Inc., 2005.

A full bibliography of Stickler-related medical research and publications is available at:  
[www.sticklers.org](http://www.sticklers.org)

Also available for order on the web site is a 30 minute video developed for health-care professionals.



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.

Stickler Involved People  
[www.sticklers.org](http://www.sticklers.org)  
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# Stickler Syndrome



## Recognition Diagnosis Treatment



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## Recognition

- myopia beginning in first decade of life and likely retinal detachment
- vitreous anomalies
- cataracts and glaucoma
  
- progressive sensorineural and/or conductive hearing loss
- chronic otitis media
  
- flattened facial structure
- palate abnormalities
- Pierre-Robin sequence
  
- premature degenerative joint changes
- excessive joint flexibility
- long bone and spinal abnormalities

Expression and severity of symptoms will vary among individuals in the same family



## Diagnosis

### Stickler Syndrome

#### Diagnostic Criteria <sup>(1)</sup>

A total of five points is required to determine the presence of Stickler syndrome and must include cleft palate, ocular abnormalities or a high frequency sensorineural hearing loss.

**Ocular** - characteristic vitreous degeneration or retinal detachment (2 points)

**Skeletal** (2 point maximum) - history of femoral head failure (Leggs-Perthes like disease or slipped epiphysis) (1 point); radiographically demonstrated osteoarthritis before age 40 (1 point); scoliosis, spondylolisthesis, or Scheuermann-like kyphotic deformity (1 point)

**Auditory** (2 point maximum) - high frequency sensori-neural hearing loss (threshold > 20, 30, 40 dB at 4-8 kHz at ages under 20, 20-40 and over 40 respectively) (2 points); hypermobile tympanic membranes (1 point)

**Oral-Facial** (2 points maximum) - cleft palate (open or submucous cleft or bifid uvula) (2 points); characteristic facies (malar hypoplasia, broad nasal bridge and micrognathia) (1 point)

**Family History/Molecular Data** - independently affected 1st degree relative in a pattern consistent with autosomal dominant inheritance or presence of COL2A1, COL11A1 or COL11A2 mutation associated with Stickler Syndrome (1 point)

(1) Developed by the National Institutes of Health

## Treatment and Evaluation

- ophthalmic assessment by an ophthalmologist or retinal specialist
- corrective lenses
- prophylactic laser or cryo retinal treatment
- hearing and otolaryngology evaluation
- hearing aids
- speech and feeding evaluation
- skeletal survey
- rheumatology consultation
- anti-inflammatory and pain medications
- surgery
- physical therapy and appropriate exercise
- genetic consultation

## Support

[www.sticklers.org](http://www.sticklers.org)

Stickler Involved People offers:

- web site and list serve
- annual conference
- newsletter, brochures and DVD