Diagnostic criteria for Stickler syndrome

(Rose P.S. et al., Am J Med Genet. 138A:199-207, 2005)

Orofacial abnormalities (2 points maximum):
- Cleft palate (open cleft, submucous cleft, or bifid uvula): major, 2 points
- Characteristic facial features (malar hypoplasia, broad or flat nasal bridge, and micro/retrognathia): 1 point

Ocular abnormalities (2 points maximum):
- Characteristic vitreous changes or retinal abnormalities (lattice degeneration, retinal hole, retinal detachment or retinal tear): major, 2 points

Auditory abnormalities (2 points maximum):
- High-frequency sensorineural hearing loss: major, 2 points
  - Age < 20 years: threshold ≥ 20 dB at 4-8 kHz
  - Age 20-40 years: threshold ≥ 30 dB at 4-8 kHz
  - Age > 40 years: threshold ≥ 40 dB at 4-8 kHz
- Hypermobile tympanic membranes: 1 point

Skeletal abnormalities (2 points maximum):
- Femoral head failure (slipped epiphysis or Legg-Perthes-like disease): 1 point
- Radiographically demonstrated osteoarthritis before age 40: 1 point
- Scoliosis, spondylolisthesis, or Scheuermann-like kyphotic deformity: 1 point

Family history/molecular data*:
- Independently affected first-degree relative in a pattern consistent with autosomal dominant inheritance or presence of a *COL2A1*, *COL11A1*, or *COL11A2* mutation associated with Stickler syndrome: 1 point

**does not account for families with autosomal recessive Stickler syndrome*

Diagnosis of Stickler syndrome
≥5 points, with:
≥1 major 2-point manifestation
AND
absence of features suggestive of a more severe skeletal dysplasia or other syndrome

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From *Robin N.H. et al. Stickler Syndrome. 2000 Jun 9 [Updated 2014 Nov 26]. In: Pagon RA, Adam MP, Ardinger HH, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2015.* http://www.ncbi.nlm.nih.gov/books/NBK1302/:

The diagnosis of Stickler syndrome is confirmed in individuals with a heterozygous pathogenic variant in *COL2A1*, *COL11A1*, or *COL11A2* or biallelic pathogenic variants in *COL9A1*, *COL9A2*, or *COL9A3*.