Diagnostic criteria for Stickler syndrome

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


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.