Diagnostic Criteria

- Based on findings of long-term NIH study
- Evaluates molecular data, family history, characteristic ocular, auditory and skeletal abnormalities
- 12 points possible on 9 criteria
- Diagnosis (for Type 1 SS) requires 5 points minimum AND presence of cleft palate, ocular abnormalities or high frequency sensorineural hearing loss

Stickler Syndrome Diagnostic Criteria

Oral-Facial Abnormalities (2 points maximum)

(2 points) Cleft palate (open cleft, submucous cleft, or bifid uvula)
(1 point) Characteristic facies (malar hypoplasia, broad nasal bridge, and micrognathia)

Ocular Abnormalities (2 points maximum)

(2 points) Characteristic vitreous degeneration or retinal detachment

Auditory Abnormalities (2 points maximum)

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

Skeletal Abnormalities (2 points maximum)

(1 point) history of 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

Family History / Molecular Data

(1 point) 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