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

- Ocular Abnormalities (2 points maximum)
  - Characteristic vitreous degeneration or retinal detachment (2 points)

- Auditory Abnormalities (2 points maximum)
  - 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
  - Hypermobile tympanic membranes (1 point)

- Skeletal Abnormalities (2 points maximum)
  - History of femoral head failure (slipped epiphysis or Legg-Perthes like disease) (1 point)
  - Radiographically demonstrated osteoarthritis before age 40 (1 point)
  - Scoliosis, pectus defects, or Scheuermann-like kyphotic deformity (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 mutations associated with Stickler syndrome (1 point)