DISEASE:
Autosomal recessive Stickler syndrome

NAME:	Autosomal recessive Stickler syndrome
DESCRIPTION:	A rare type of Stickler syndrome characterized by moderate to severe sensorineural hearing loss, high myopia, retinal degeneration, vitreous anomalies, and epiphyseal dysplasia. Midface hypoplasia, cleft palate, as well as additional skeletal manifestations (such as platyspondyly, scoliosis, and tibial and femoral bowing at birth) have also been observed.
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XREF(S):	Orphanet OMIM ICD-10 OMIM OMIM
ANALYTE(S):	COL9A1 COL9A2 COL9A3
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RELATED CONTENT

Related Genetic Tests

- Stickler syndrome (gene panel)
- cleft lip with/whitout cleft palate (virtual gene panel)

Related Laboratories

- Centre de Génétique Médicale UCL
- Centrum Medische Genetica - UZ Antwerpen

Related Analytes

- collagen type IX alpha 1 chain
- collagen type IX alpha 2 chain
- collagen type IX alpha 3 chain

Related Gene Panels

- Cleft lip and palate / dysmorphic facial features / craniofacial anomalies (255 genes)) - UCL
- Stickler syndrome (4 genes) - UZA