Stickler Syndrome: Deletion/Duplication Panel

**Test Code:** MD236  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1

### Condition Description

Stickler syndrome is a genetically heterogeneous connective tissue disorder that typically results in abnormalities of the ocular, auditory, and skeletal systems. Individuals can have a characteristic flat facial appearance that results from underdeveloped bones in the midface. Pierre Robin sequence, hearing impairment/loss and joint hypermobility are common. Eye manifestations include high myopia, cataract, retinal detachment, and vitreous abnormalities. While the disorder is completely penetrant, much phenotypic variability exists. Stickler syndrome can be inherited in an autosomal dominant (COL2A1, COL11A1 and COL11A2 genes) or autosomal recessive (COL9A1 and COL9A2 genes) manner.

**References:**
- OMIM  
- GeneReviews

### Genes

COL2A1, COL11A2, COL11A1, COL9A1, COL9A2

### Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of Stickler syndrome.  
- Carrier testing in adults with a family history of Stickler syndrome.

### Methodology

**Deletion/Duplication Analysis:** DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that cover the entire genomic region. Please note that a "backbone" of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

**Detection**

**Deletion/Duplication:** Detection is limited to duplications and deletions. The CGH array will not detect point or intronic pathogenic variants. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

### Specimen Requirements

Submit only 1 of the following specimen types

**Type: Whole Blood**

Specimen Requirements:

In EDTA (purple top) tube:
- Infants (2 years): 3-5 ml  
- Older Children & Adults: 5-10 ml.

Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

**Type: Isolated DNA**

Specimen Requirements:

In microtainer: 10 ug  
Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.
Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.

### Special Instructions

Please include fundus photographs, electroretinogram (ERG) findings, visual field findings, and visual acuity, if available, for expert review and clinical correlation with test results.

### Related Tests

- Stickler Syndrome: Sequencing Panel
- Eye Disorders: Comprehensive Sequencing
- Eye Disorders: Deletion/Duplication Panel