
Lab Dept: Anatomic Pathology

Test Name: **STICKLER OR MARSHALL SYNDROME
(COL2A1/COL11A1) SEQUENCING**

General Information

Lab Order Codes: COL2

Synonyms: COL2A1 and COL11A1 Gene Sequencing; Stickler Syndrome I/II (STL1/STL2)

CPT Codes: 81479 x2 – Molecular Pathology, Unlisted Procedure

Test Includes: Sequencing of the COL2A1 and COL11A1 genes.

Logistics

Test Indications: Collagen II is a tough, fibrous protein that provides a major part of the strength of cartilage. It is also found in intervertebral disks and inner ear. Collagen XI is a quantitatively minor collagen associated with collagen II. Mutations in the COL2A1 gene can cause decreased synthesis of the protein or synthesis of defective protein. Defects in collagen II often result in structural or growth disorders frequently accompanied by eye abnormalities. Mutations in the COL11A1 gene can result in similar but milder phenotypes. Null allele mutations in the COL2A1 gene and more deleterious mutations (exon skippings and glycine substitutions) in the COL11A1 gene can cause phenotypes of Stickler, Marshall or Stickler-like syndromes.

Stickler Syndrome Types I and II are characterized by high myopia, retinal detachment, vitreoretinal degeneration, and cataracts. Some patients may have hearing loss, epiphyseal dysplasia and early-onset osteoarthritis. In addition, cleft palate and Robin sequence (cleft palate, small chin and glossoptosis) are seen in 30% of the patients.

Marshall Syndrome patients have hearing loss, myopia (low or high) vitreoretinal degeneration, retinal detachment, cataracts, midfacial hypoplasia, and cleft/palate/Robin sequence. These patients may also have epiphyseal dysplasia and early onset osteoarthritis.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: CTGT

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 2 to 3 months

Special Instructions: [See Patient Preparation](#). Request special forms for patient consent, physician signature and clinical history from Children's Laboratory or print [Request form](#) here. Select (Sequencing only) COL2A1 & COL11A1 (STL1/2) under Stickler Syndrome.

Note: These completed forms must accompany the specimen or come with the patient when they present at the lab to be drawn.

Specimen

Specimen Type: Whole blood

Container: Lavender top (EDTA) tube

Draw Volume: 6 mL (Minimum: 3 mL) blood

Processed Volume: Same as Draw Volume

Collection: Routine venipuncture

Special Processing: Lab Staff: **Do Not** process. Specimen should remain as whole blood in original collection container. Send Monday – Thursday via overnight shipping at refrigerated temperatures. Include patient consent and physician signature forms with the specimen. Forward promptly.

Note: For specimens collected Friday – Sunday (or on a holiday), they should be held in Children's Laboratories at refrigerated temperatures and shipped on Monday (or the next business day Monday – Thursday).

Patient Preparation: N/A

Sample Rejection: Frozen specimens; mislabeled or unlabeled specimens

Interpretive

Reference Range: No mutations detected

Critical Values: N/A

Limitations: The analysis may fail to detect a few mutations in patients with mutations in the COL2A1 and COL11A1 genes as a cause of the disease. Matrix DNA Diagnostics does not have an accurate estimate of how many mutations are missed, but they estimate that a mutation is detected in about 70-80% of the patients with Stickler/Marshall syndromes. Also, a few changes in the structure of the genes that are detected by the test are difficult to interpret in terms of whether they in fact cause the disease or are more neutral changes. Mutations in other genes are not detected.

Methodology: DNA Sequencing

References: [CTGT Web Page](#) (April 2010)

Updates: 4/19/2010: Moved from Tulane University to a Mayo forward to CTGT.
2/11/2013: CPT update
7/10/2013: CPT update CTGT, previously listed as 81408