Lab Dept: Anatomic Pathology

Test Name: COL11A1 GENE SEQUENCING

General Information

Lab Order Codes: 11A1

Synonyms: Marshall syndrome COL11A1; Stickler syndrome, type II (STL2) COL11A1

CPT Codes: 81479 – Molecular Pathology, Unlisted Procedure

Test Includes: Direct DNA sequencing of PCR products generated from genomic DNA, sequencing of exons and exon-intron boundaries, point mutations, splice site mutations and small exonic deletions, insertions and indels.

Logistics

Test Indications: For confirmation of symptoms and the clinical diagnosis related to Marshall syndrome COL11A1 or Stickler syndrome, type II (STL2) COL11A1.

Stickler syndrome and related Marshall syndrome are autosomal dominantly inherited disorders caused by defects in three genes. Stickler syndrome, type II and Marshall syndrome are due to mutations in COL11A1.

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 about 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: Connective Tissue Gene Tests (CTGT Test: 1333)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time:	2 - 4 weeks
Special Instructions:	Please include a completed CTGT Request form with the patient or specimen to the laboratory.

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 blood collection
Special Processing:	Lab Staff: **Do Not** centrifuge. Specimen should be sent in original collection container. Send via overnight shipping to reach CTGT Monday through Friday. If weekend or holiday when drawn, store at refrigerated temperatures. Please include a CTGT Shipment Packing Slip with the shipment.
Patient Preparation:	None
Sample Rejection:	Mislabeled or unlabeled specimen

Interpretive

Reference Range:	Interpretive report
Critical Values:	N/A
Limitations:	Published estimates of test sensitivity for genes linked to certain disorders can be very inaccurate, and that it is difficult to predict the probability of detecting a mutation in any single gene for one individual. The following factors contribute to this challenge: Many disorders have overlapping phenotypes; some disorders are linked to mutations in more than one gene; in some instances genes remain to be linked to specific disorders; for most disorders, proper diagnosis requires that clinical findings are considered along with genetic findings.
Methodology:	Next Generation sequencing
References:	Connective Tissue Gene Tests May 2018

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Updates:

1/31/2013: CPT 2013 update
7/10/2013: CPT update CTGT, previously listed as 81408
5/15/2018: Method update