Lab Dept: Anatomic Pathology

Test Name: COL11A2 GENE SEQUENCING

General Information

Lab Order Codes: 11A2

Synonyms: Otospondylomegaepiphyseal dysplasia (OSMED) COL11A2; Stickler syndrome, type III (STL3) COL11A2; Weissenbacher-Zweymuller syndrome (WZS) COL11A2

CPT Codes: 81479 – Unlisted Molecular Pathology procuedure (COL11A2 Sequencing)

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 Otospondylomegaepiphyseal dysplasia (OSMED); Stickler syndrome, type III (STL3); or Weissenbacher-Zweymuller syndrome (WZS).

Otospondylomegaepiphyseal dysplasia (OSMED) is an autosomal recessive disorder characterized by sensorineural hearing loss, enlarged epiphyses, disproportionate shortness of the limbs, and vertebral body abnormalities. Cleft palate, small mandible, mid-facial hypoplasia and small upturned nose are also common findings. OSMED is typically caused by homozygous or compound heterozygous loss-of-function mutations in COL11A2.

Stickler syndrome, type III is the non-ocular form of the syndrome. Some patients may present predominantly with cleft palate/Robin sequence, hearing loss or early-onset osteoarthritis. Since COL11A2 is not expressed in the eye, these patients do not have eye findings.

Weissenbacher-Zweymuller syndrome (WZS) shares the same clinical features as OSMED, but it is milder.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: Connective Tissue Gene Tests (CTGT)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability:	Daily, 24 hours
Turnaround Time:	1 – 2 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 venipuncture
Special Processing:	Lab Staff: **Do Not** centrifuge. Specimen should be sent in original collection container. Send via overnight shipping with a cold pack 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:	Mislabled 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:	DNA sequencing
References:	Connective Tissue Gene Tests December 2014 (484) 224-2900 Fax (484) 244-2904

Updates: 1/31/2012: CPT 2013 udpate