Lab Dept: Anatomic Pathology

Test Name: COL11A2 DELETION/DUPLICATION HDT ARRAY

General Information

Lab Order Codes: A2DD

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

CPT Codes: 81479 – Molecular Pathology, Unlisted Procedure

Test Includes: Deletion/Duplication HDT Array uses 60mer oligonucleotide sequences designed to selectively complement target areas with an extremely high degree of specificity. High resolution and sensitivity are achieved by assigning a number of probes to each exonic region to allow for a minimum of 3 probes within each 300-500 base section of the genomic DNA sequence. Non-coding intervening sequences are targeted at a minimum with approximately half of the density used for exonic regions. Many intervening sequences are more densely covered. The minimum CNV size detected by this high-density array is 300-500 nucleotides, which is the technical limit of the assay using these stringent parameters.

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: 4 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:	High-Density Targeted Array

References: Connective Tissue Gene Tests October 2010
(484) 224-2900 Fax (484) 244-2904

Updates: 1/31/2013: CPT 2013 update
7/10/2013: CPT update CTGT, previously listed as 81407