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**Lab Dept:**                    **Anatomic Pathology**

**Test Name:**                **COL11A1 DELETION/DUPLICATION HDT ARRAY**

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***General Information***

**Lab Order Codes:**        11DD

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

**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.

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***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)

**Phone Numbers:**         MIN Lab: 612-813-6280

STP Lab: 651-220-6550

<b>Test Availability:</b>	Daily, 24 hours
<b>Turnaround Time:</b>	1 – 2 weeks
<b>Special Instructions:</b>	Please include a completed CTGT <a href="#">Request form</a> with the patient or specimen to the laboratory.

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### ***Specimen***

<b>Specimen Type:</b>	Whole blood
<b>Container:</b>	Lavender top (EDTA) tube
<b>Draw Volume:</b>	6 mL (Minimum: 3 mL) blood
<b>Processed Volume:</b>	Same as Draw Volume
<b>Collection:</b>	Routine venipuncture
<b>Special Processing:</b>	Lab Staff: <b>Do Not</b> 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 <a href="#">CTGT Shipment Packing Slip</a> with the shipment.
<b>Patient Preparation:</b>	None
<b>Sample Rejection:</b>	Mislabeled or unlabeled specimen

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### ***Interpretive***

<b>Reference Range:</b>	Interpretive report
<b>Critical Values:</b>	N/A
<b>Limitations:</b>	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.
<b>Methodology:</b>	High-Density Targeted Array

**References:**

[Connective Tissue Gene Tests](#) December 2014  
(484) 224-2900 Fax (484) 244-2904

**Updates:**

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