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**Lab Dept:** Anatomic Pathology

**Test Name:** COL2A1 DELETION/DUPLICATION HDT ARRAY

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***General Information***

**Lab Order Codes:** C2DD

**Synonyms:** Achondrogenesis, Type II (ACG2)/ACG1B; Avascular Necrosis of Femoral Head, Primary (ANFH); Kniest Dysplasia; Osteoarthritis with mild chondrodysplasia; Platyspondylic Lethal Skeletal Dysplasia, Torrence Type (PLSDT); Stickler Type 1 (STL1); Spondyloepimetaphyseal Dysplasia, Studwick Type (SEMD); Spondyloperipheral Dysplasia; Spondyloepiphyseal Dysplasia Congenital (SEDC)

**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 Achondrogenesis, Type II (ACG2)/ACG1B; Avascular Necrosis of Femoral Head, Primary (ANFH); Kniest Dysplasia; Osteoarthritis with mild chondrodysplasia; Platyspondylic Lethal Skeletal Dysplasia, Torrence Type (PLSDT); Stickler Type 1 (STL1); Spondyloepimetaphyseal Dysplasia, Studwick Type (SEMD); Spondyloperipheral Dysplasia; or Spondyloepiphyseal Dysplasia Congenital (SEDC).

**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.

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### ***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:** Mislabeled or unlabeled specimen

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### ***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](#) December 2014  
(484) 224-2900 Fax (484) 244-2904

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