
Lab Dept: **Anatomic Pathology**

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

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.

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: 2-3 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 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

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](#) June 2018
(484) 224-2900 Fax (484) 244-2904

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