### Lab Dept: Anatomic Pathology

### Test Name: COL2A1 GENE SEQUENCING

#### General Information

**Lab Order Codes:** C2A1

**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:** 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 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 Test: Varies by syndrome)

**Phone Numbers:**
- MIN Lab: 612-813-6280
- STP Lab: 651-220-6550

**Test Availability:** Daily, 24 hours

**Turnaround Time:** 2 -4 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 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: Next Generation Sequencing

References: Connective Tissue Gene Tests April 2018
(484) 224-2900  Fax (484) 244-2904

Updates: 1/13/2013: CPT 2013 updates
7/10/2013: CPT update CTGT, previously listed as 81408