**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