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:	Mislabled 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