Lab Dept: Anatomic Pathology

Test Name: MULTIPLE EPIPHYSEAL DYSPLASIA SEQUENCING

General Information

Lab Order Codes:	MEDG (Blood)
Synonyms:	MED familial mutation, COMP gene; EDM1; MED; PSACH; Pseudoachondroplasia; Spondoepiphyseal Dysplasia
CPT Codes:	81479 x8 – Molecular Pathology, Unlisted Procedure
Test Includes:	Analysis of different genes: CANT, COL2A1, COMP, COL9A1, COL9A2, COL9A3, SLC26A2 and MATN3.

Logistics

Test Indications:	Multiple epiphyseal dysplasia (MED) is a clinically and genetically heterogeneous chondrodysplasia with either autosomal dominant or recessive inheritance. The phenotypic spectrum ranges from mild to severe. Patients usually present with joint pain and stiffness, waddling gait and/or mild short stature in childhood. Some patients, however, remain asymptomatic until adult age and present with early-onset osteoarthritis of the large weight-bearing joints. Typical radiographic findings include delayed and irregular ossification of the epiphyses in multiple joints. Spinal changes, if present, are mild.
Lab Testing Sections:	Anatomic Pathology - Sendouts
Referred to:	Connective Tissue Gene Tests Lab (CTGT Test: 5094)
Phone Numbers:	MIN Lab: 612-813-6280 STP Lab: 651-220-6550
Test Availability:	Daily, 24 hours
Turnaround Time:	2 - 4 weeks
Special Instructions:	A requisition form must be sent with any specimen. Forms can be obtained through the following link: CTGT Requisition Form.

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 for blood specimens, invert gently to mix
Special Processing:	Lab Staff: Send whole blood in original collection container labeled with patient name, date of birth and medical record number, including requisition. Store refrigerated, ship at room temperature. **Note:** Specimens stable approximately one week at refrigerated temp.
Patient Preparation:	None
Sample Rejection:	Mislabeled or unlabeled specimens

Interpretive

Reference Range:	Interpretive report
Critical Values:	N/A
Limitations:	N/A
Methodology:	Next Generation Sequence analysis
References:	Connective Tissue Gene Tests Lab Web Page May 2018 (484) 244-2900 Fax (484) 244-2904
Update:	2/7/2013: CPT update 7/10/2013: CPT update CTGT, previously listed as 81406 5/15/2018: Updated genes and CPT coding.