**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; Spondoeipiphyseal 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.