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**Lab Dept:**                   **Anatomic Pathology**

**Test Name:**               **MULTIPLE EPIPHYSEAL DYSPLASIA (MED)  
KNOWN MUTATION**

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***General Information***

**Lab Order Codes:**       MEDGK (Blood)

**Synonyms:**               MED Known familial mutation, COMP gene

**CPT Codes:**              81403 – Molecular Pathology procedure, Level 4

**Test Includes:**           Testing of a relative for a specific known mutation (carrier testing) where testing was previously done on family member(s). The previously tested proband DNA serves as a positive control.

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***Logistics***

**Test Indications:**       Multiple epiphyseal dysplasia 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. Mutation in five different genes, cartilage oligomeric matrix protein (COMP), collagen IX (COL9A1, COL9A2 and COL9A3) and matrilin-3 (MATN3) can cause the autosomal dominantly inherited forms for MED. Mutations in the diastrophic dysplasia sulfate transporter gene (DTDST or SLC26A2) can cause the recessively inherited form of the disorder.

**Lab Testing Sections:**   Anatomic Pathology - Sendouts

**Referred to:**             Connective Tissue Gene Tests Lab (CTGT test: 1387)

**Phone Numbers:**        MIN Lab: 612-813-6280

STP Lab: 651-220-6550

**Test Availability:**       Daily, 24 hours

**Turnaround Time:**       1 - 2 weeks

**Special Instructions:** A requisition form must be sent with any specimen. Forms can be obtained through the following link:  
[CTGT Requisition](#)  
Select Known Familial Mutation and indicate the mutation on the form.

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### ***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: Do Not centrifuge. Send whole blood in original collection. Store at refrigerated temperatures. Ship at room temp.  
  
Specimens drawn on weekends or holidays should be stored refrigerated until specimen can be shipped to arrive Monday – Friday.  
  
**Note:** Specimens stable approximately one week at refrigerated temp.

**Patient Preparation:** None

**Sample Rejection:** Mislabeled or unlabeled specimens

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### ***Interpretive***

**Reference Range:** Interpretive report

**Critical Values:** N/A

**Limitations:** N/A

**Methodology:** Sanger sequencing

**References:** [Connective Tissue Gene Tests Lab Web Page](#) April 2018  
phone: (484)244-2900 fax: (484)244-2904

**Update:** 2/7/2013: CPT update  
10/29/2014: Added minimum draw volume  
4/17/2018: Updated TAT, method