
Lab Dept: **Anatomic Pathology**

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

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.

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, cartilate oligometric metrix 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.

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

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