
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 x6 – Molecular Pathology, Unlisted Procedure

Test Includes: Analysis of different genes: COMP, COL9A1, COL9A2, COL9A3,
DTDST/SLC26A2 and MATN3.

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)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 3 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:	Sequence analysis
References:	Connective Tissue Gene Tests Lab Web Page May 2009 (484) 244-2900 Fax (484) 244-2904
Update:	2/7/2013: CPT update 7/10/2013: CPT update CTGT, previously listed as 81406