Multiple Epiphyseal Dysplasia: Deletion/Duplication Panel

**Test Code:** MD100  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1

**Condition Description**

Multiple epiphyseal dysplasias (MED; also known as epiphyseal dysplasia, multiple, EDM) is a group of skeletal disorders with heterogeneous genetic causes. MED has seven subtypes with a continuum of clinical severity among these types. Clinical and radiographic features continue to be used reliably to assign patients to this general disease category. Identification of the precise genetic defect is important, however, to permit carrier testing and early prenatal diagnosis. Molecular analysis is likely to expand the clinical spectrum of MED and may also provide data relevant to prognosis and future therapeutic intervention. The overall incidence of MED is estimated to be 1 in 10,000 births. Although the phenotype range is broad, MED is mainly characterized with short stature and early-onset osteoarthritis. Radiographic findings for MED show a generalized abnormality of epiphyseal ossification without significant vertebral involvement. MED can be inherited in an autosomal dominant or autosomal recessive manner. The autosomal recessive form of MED includes features such as club foot and bilateral double-layered patellae.

**References:**
- GeneReviews

**Genes**
- COL2A1, COL9A1, COL9A2, COL9A3, COMP, MATN3, SLC26A2

**Indications**

This test is indicated for individuals with:

- Short stature and early-onset osteoarthritis.
- An abnormal radiographic findings show a generalized abnormality of epiphyseal ossification without significant vertebral involvement.

**Methodology**

**Deletion/Duplication Analysis:** DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that cover the entire genomic region.

**Detection**

**Deletion/Duplication Analysis:** Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient’s clinical and/or biochemical phenotype.

**Specimen Requirements**

*Submit only 1 of the following specimen types*

**Type: Whole Blood (EDTA)**

**Specimen Requirements:**
- EDTA (Purple Top)
- Infants and Young Children (2 years of age to 10 years old): 3-5 ml
- Older Children & Adults: 5-10 ml
- Autopsy: 2-3 ml unclotted cord or cardiac blood

**Specimen Collection and Shipping:**
Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

**Type: DNA, Isolated**

**Specimen Requirements:**
- Isolation using the Perkin Elmer™Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

**Specimen Collection and Shipping:**
Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

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Special Instructions

Radiographic results can help interpretation.

Related Tests

- Multiple Epiphyseal Dysplasia: Sequencing Panel