Multiple Epiphyseal Dysplasia: Deletion/Duplication Panel

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

**Condition Description**

Multiple epiphyseal dysplasia (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 osteoarthrosis. 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 osteoarthrosis.
- 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.

Please note that a “backbone” of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

**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**

Specimen Requirements:

In EDTA (purple top) or ACD (yellow top) tube:
- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml.
Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

**Type: Isolated DNA**

Specimen Requirements:

In microtainer: 10 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

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

**Special Instructions**

Radiographic results can help interpretation.

**Related Tests**

- Multiple Epiphyseal Dysplasia: Sequencing Panel