

Limb Malformation: Deletion/Duplication Panel

Test Code: MD174

Turnaround time: 2 weeks

CPT Codes: 81228 x1

Condition Description

Skeletal dysplasias are a heterogeneous group of more than 450 disorders with complex mechanisms. Clinical and biochemical 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 skeletal dysplasia and may also provide data relevant to prognosis and future therapeutic intervention.

Collectively, the incidence of skeletal dysplasia is estimated to be 1 in 5,000 births. Skeletal dysplasia is referred to as generalized disorders of cartilage and bone. Limb malformation consists of hypoplastic or absent bones of the limbs. These abnormalities can be isolated or associated with variety of other anomalies such as septal defects of the heart, renal anomaly, etc.

References:

- Alanay and Lachman. (2011) *J Clin Res Pediatr Endocrinol.* 3(4):163-78.
- Krakow and Rimoin. (2010) *Genet Med.* 12(6):327-41.
- Orioli, Castilla, and Barbosa-Neto. (1986) *J Med Genet.* 23:328-332.
- Seaver and Irons. (2009) *Genet Med.* 11(6):465-70.
- Warman et al. (2011) *Am J Med Genet A.* 155A(5):943-68.

Genes

[ARHGAP31](#), [BMP2](#), [BMPR1B](#), [CC2D2A](#), [CDH3](#), [CEP290](#), [CHSY1](#), [ESCO2](#), [FBLN1](#), [FBXW4](#), [FGF10](#), [FGFR2](#), [FGFR3](#), [FMN1](#), [GDF5](#), [GLI3](#), [GNAS](#), [GREM1](#), [HDAC4](#), [HOXD13](#), [IHH](#), [KIF7](#), [LMBR1](#), [LRP4](#), [MGP](#), [MKS1](#), [MYCN](#), [NIPBL](#), [NOG](#), [PIGV](#), [PITX1](#), [PTHLH](#), [RECQL4](#), [ROR2](#), [RPGRI1L](#), [SALL1](#), [SALL4](#), [SHH](#), [SOX9](#), [TBX15](#), [TBX3](#), [TBX5](#), [THPO](#), [TP63](#), [WNT3](#), [WNT7A](#)

Indications

This test is indicated for:

- Patients for whom there is a suspicion of skeletal dysplasia with limb malformation.

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: DNA, Isolated

Specimen Requirements:

Microtainer

3µg

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.

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.

Special Instructions

Please include radiographic findings for expert review at EGL Genetics.

Related Tests

- Limb Malformation: Sequencing Panel
- Skeletal Dysplasia: Sequencing Panel