Skeletal Dysplasia With Increased Bone Density: Deletion/Duplication Panel

Test Code: MD172  
Turnaround time: 2 weeks  
CPT Codes: 81228 x1

Condition Description

Skeletal dysplasias are a heterogeneous group of more than 450 disorders with complex genetic etiologies. Clinical and radiographic features can be used reliably to assign patients to general disease categories, but reaching a specific diagnosis, especially at a young age, may be difficult. Identification of the precise genetic defect is important, however, to permit appropriate genetic counseling, anticipatory guidance, and early prenatal diagnosis.

Collectively, the incidence of skeletal dysplasias is estimated to be 1 in 5,000 births. Skeletal dysplasias with increased bone density are characterized by hypermineralization of some or all of the skeleton.

References:

Genes

ANKH, CA2, CLCN7, COL1A1, CTSK, DLX3, FERMT3, GALNT3, HPGD, LEMD3, LRP4, LRP5, QSTM1, RASGRP2, SOST, TBXAS1, TCIRG1, TGFB1, TNFRSF11A, TNFRSF11B, TNFSF11, TYROBP

Indications

This test is indicated for:
- Patients for whom there is a suspicion of skeletal dysplasia with abnormal radiographic findings indicating increased bone density.

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.

Reference Range

Please include radiographic findings for expert review at EGL Genetics.

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 24 hours of collection. Do not refrigerate or freeze.

Type: DNA, Isolated

Specimen Requirements:
Microtainer  
3µg
Isolation using the Perkin Elmer™ 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.

**Related Tests**

- Skeletal Dysplasia With Increased Bone Density: Sequencing Panel
- Skeletal Dysplasia: Sequencing Panel