**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
- OSTM1
- 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 72 hours of collection. Do not 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