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, CTXK, Dlx3, FERMT3, GALNT3, HPGD, LEMD3, LRP4, LRP5, LRP6, LRP10, RASGRP2, SCST, TBXAS1, TCIRG1, TGFBI, TNFRSF11A, TNFRSF11B, TNFRSF11C, 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.

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.

Reference Range

Please include radiographic findings for expert review at Emory Genetics Laboratory.

Specimen Requirements

Submit only 1 of the following specimen types

Type: Whole Blood

Specimen Requirements:
In EDTA (purple 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.

**Related Tests**

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