Done.
bp of flanking intronic region). The DNA sequence is mapped to, and analyzed in, comparison with the published human genome build UCSC hg19 reference sequence. The targeted coding exons and splice junctions of the known protein-coding RefSeq genes are assessed for the depth of coverage and data quality threshold values. The Medical EmExome bioinformatics analysis pipeline is used to compare sequence changes in the individual being tested to the reference sequence. All potential positive sequence variants in the proband are confirmed by conventional di-deoxy DNA sequence analysis (Sanger sequencing) using a separate DNA isolation.

The Medical EmExome also features the EmExome Boost Option, which allows clinicians to choose an EGL gene panel relevant to the patient’s phenotype to ensure coverage of ALL exons, at no additional cost.

Targeted sequencing of parental samples for the proband only option will be completed at no additional charge for exome sequencing only if needed (such as a variant of uncertain clinical significance).

**Specimen Requirements**

**Additional Specimen Collection/Handling Instructions Required for this Test**

Please send parental samples with the sample of the affected individual. If parental samples are not available, please notify the laboratory. No extra charge is applied for processing parental samples.

Please contact the lab before submitting any other specimen type.

**Type: Whole Blood**

**Specimen Requirements:**

In EDTA (purple top) or ACD (yellow top) tube: 5-10 ml for all ages

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.