Medical EmExome: Clinical Exome Sequencing, Additional Family Member

Test Code: EXOMA  
Turnaround time: 6 weeks  
CPT Codes: 81416 x1

Condition Description

Traditionally there are two options when ordering exome sequencing. One can analyze only the affected individual (proband only) or a family trio (typically the affected person and his/her parents). Analyzing trios has shown to increase the diagnostic yield of exome sequencing by provide deeper insight into proband's findings. EGL Genetics (EGL) now offers an additional family member option to further increase the clinical efficacy of its exome sequencing. This test allows clinicians to send in an additional child or family member (affected or unaffected) to help sort out findings of unclear significance.

The sequencing design of the Medical EmExome at EGL provides >97% coverage of 22,000 genes, with a mean read depth of 100X. Of the ~4600 disease-associated genes analyzed, 3000 have 100% coverage (>20X) of all exons; twice the number of genes with complete coverage offered by competitors, making it the most comprehensive exome sequencing test available. This is also the highest coverage offered by any clinical exome sequencing performed in a CLIA-/CAP-certified laboratory.

Please note this test should be ordered at the same time as the “Medical EmExome: Clinical Exome Sequencing, Family Trios” (test code: EXOMT).

The Exome Coverage Tool can be used to view typical depth of sequence coverage obtained by exome sequencing performed by EGL. To access this too, please click here.

Indications

The Medical EmExome is indicated when there is a suspicion of a genetic etiology contributing to the proband's manifestations.

Methodology

Medical EmExome is performed on genomic DNA, using the Agilent V5 Plus designed to target the exome with greater coverage of known disease-associated genes. These targeted regions are then sequenced using the Illumina HiSeq 2500 sequencing system, with 100 basepair (bp) paired-end reads (similar to bidirectional Sanger sequencing) and an average coverage of 100X in the target region. (The target region includes the exon and 10 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.

Detection

Unknown.

Specimen Requirements

Submit only 1 of the following specimen types

**Type: Saliva**

Specimen Requirements:  
Oragene™ Saliva Collection Kit  
Orangene™ Saliva Collection Kit used according to manufacturer instructions. Please contact EGL for a Saliva Collection Kit for patients that cannot provide a blood sample.

Specimen Collection and Shipping:  
Please do not refrigerate or freeze saliva sample. Please store and ship at room temperature.

**Type: Liquid Buccal Swab**

Specimen Requirements:  
ORAcollect-DX (OCD-100) | Assisted saliva collection kit  
DNA Genotek ORAcollect-DX kit used according to manufacturer instructions. Please contact EGL for a saliva collection kit for patients that cannot provide a blood sample.

Specimen Collection and Shipping:  
Please do not refrigerate or freeze saliva sample. Please store and ship at room temperature.

**Type: DNA, Isolated**

Specimen Requirements:  
Microtainer  
8µg

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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.

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<th>Special Instructions</th>
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<td>Please submit medical records or clinic summary notes, and a signed consent form when ordering exome testing. Testing will not be initiated until these documents are received.</td>
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