

## Medical EmExome: Clinical Exome Sequencing, Proband Only

**Test Code:** EXOME

**Turnaround time:** 6 weeks

**CPT Codes:** 81415 x1

### Condition Description

#### What is the Medical EmExome?

The human exome is the complete coding (exonic) region of the genome. It is estimated to encompass approximately 1-2% of the genome, yet contains approximately 85% of disease-causing pathogenic variants. The Medical EmExome is expertly curated to target genes known or suspected to cause disease. The design provides >96% coverage of 19,000 genes, with a mean read depth of 100X. For the ~5,400 disease-associated genes analyzed, we typically get coverage >98%.

#### What gene coverage levels can be expected?

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

#### Will EGL release raw exome data?

Yes, upon request for data less than 3 years old.

#### Will EGL re-analyze data?

Yes, upon request for data less than 3 years old.

### Indications

This test is indicated for individuals with a complex or ambiguous phenotype or for individuals with clinical features of a genetic disorder for whom previous testing has been non-diagnostic.

### Methodology

Medical EmExome is performed on genomic DNA using in solution hybridization to enrich for the exome. These targeted regions are then sequenced using next-generation sequencing technology at an average coverage of 100X in the target regions. This sequencing typically provides >96% coverage of the ~19,100 genes in the exome at >20X. Intronic variants within 10 nucleotides from the exon/intron boundaries are analyzed, unless prohibited by the complexity of the sequence. 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 genes associated with disease are assessed for the depth of coverage and data quality threshold values. EGL has developed an EmExome bioinformatics analysis pipeline to compare sequence changes in the individual being tested to the reference sequence. High-quality variants which pass EGL's quality filters are not confirmed by Sanger sequencing. Reportable variants that do not pass the quality filters are confirmed using bidirectional Sanger sequence analysis.

### Detection

Based on published studies, WES is expected to provide a diagnosis in 20-30% of the cases for rare and ultra-rare disorders.

### Specimen Requirements

**Submit only 1 of the following specimen types**

#### 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: 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: Saliva****Specimen Requirements:**

Oragene™ Saliva Collection Kit

Oragene™ 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: DNA, Isolated****Specimen Requirements:**

Microtainer

8µg

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.

**Special Instructions**

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.