Medical EmExome: Clinical Exome Reanalysis

Test Code: EXOMR
Turnaround time: 3 weeks
CPT Codes: 81417 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%.

Sometimes a reanalysis of previous data may be desired if the patient has a new symptom or if new literature may affect the classification of a patient’s variants or implicate new genes in disease. This test uses the raw exome data from previous test and reanalyzes it to look for any new associations between a patient’s symptoms, genetic data, and disease. Please note that for this test to be ordered, the original exome testing must have been performed at EGL within the previous 3 years.

Will a particular gene be covered on the EmExome?

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. If a gene of specific interest does not have consistent (100%) coverage, please ask us about Sanger fill-in.

Will EGL release raw exome data?

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

Indications

This test should be ordered when a reanalysis of the Medical EmExome offered by EGL is desired. The original exome testing must have been performed at EGL Genetics within the previous 3 years.

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: 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.

Type: Saliva

Specimen Requirements:
Orangene™ 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.

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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.

Special Instructions
Please submit any updated medical records or clinic summary notes since the original Medical ExExome was ordered.

Related Tests
- EXOMT - Medical EmExome: Clinical Exome Sequencing, Trios
- EXOME - Medical EmExome: Clinical Exome Sequencing, Proband Only
- EXOMA - Medical EmExome: Clinical Exome Sequencing, Additional Family Member (EXOMT should be order first or at the same time)
- EXODD - Medical EmExome Array: Deletion/Duplication Analysis