Clinical Exome Sequencing

Medical EmExome: Clinical Exome Sequencing

What is Medical EmExome testing?
The Medical EmExome sequences the entire exome with enhanced coverage of ~5000 medically relevant and known disease-associated genes. This is the highest coverage offered by any clinical exome sequencing performed in a CLIA-/CAP-certified laboratory. The Medical EmExome also features the EmExome Boost Option (exome trios only), which allows clinicians to more deeply interrogate exons from the genes on any one next generation sequencing panel available at EGL (irrespective of number of genes) that is most relevant to the patient’s phenotype to guarantee 100% coverage of these highly suspected genes, at no additional cost. EGL is the first clinical laboratory to offer near complete coverage of all medically relevant genes with an exome boost option. A research protocol is also available for extended exome and genome testing for the discovery of novel disease genes.

What options are available for Medical EmExome testing?
EGL offers five distinct services for Medical EmExome testing:
- Medical EmExome, Family Trios: full-service exome sequencing for the proband and two family members when there is a suspicion of a genetic etiology contributing to the proband’s clinical manifestations. One EmExome Boost is included
- Medical EmExome, Proband Only: includes exome and mitochondrial genome sequencing, interpretation, and Sanger confirmation for reported variants
- Medical EmExome Array: the first exome array to detect intragenic copy number variation in the ~5000 medically relevant genes
- Sanger confirmation and interpretation of data from an outside laboratory
- Interpretation (exome or genome) of data from an outside laboratory

What is the clinical importance of Medical EmExome testing?
The identification of underlying disease-gene variants may have major implications for diagnostic and therapeutic approaches by guiding prediction of disease course and helping determine the need to test at-risk family members.

What are the advantages of Medical EmExome testing versus single-gene analysis?
Exome sequencing has the ability to: (1) identify variants in genes that were not tested due to an atypical clinical presentation; (2) identify clinical cases in which variants from different genes contribute to the different phenotypes in the same patient; and (3) cost-effectively provide a plethora of genetic data.

When is Medical EmExome testing appropriate for patients?
Medical EmExome testing is recommended when: (1) genetic testing for a suspected condition has yielded no positive results; (2) traditional diagnostic approaches have proven ineffective; or (3) a cost-effective alternative to whole genome testing is desirable.
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Variant Classification
As more information from human exome and genome sequencing projects becomes publicly available, and as more research is conducted on previously reported DNA variants, knowledge of variant classification increases. This knowledge will allow variants previously classified as variants of uncertain clinical significance to be reclassified as pathogenic variants or benign polymorphisms.

EGL is committed to ensuring clients and their patients are kept as up-to-date as possible concerning the classification of sequence variants. EGL is the first laboratory to develop a free, open-access variant catalog with no registration required. EmVClass provides web-based, current status of all sequence variants detected and analyzed by the laboratory. When new data emerges to support a variant classification change, EGL will issue amended reports for each patient with that variant, upon request.

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**CPT® is a registered trademark of the American Medical Association.

About Emory Genetics Laboratory (EGL)
EGL specializes in genetic diagnostic testing, with 45 years of clinical experience and board-certified laboratory directors and genetic counselors reporting out cases. EGL offers a combined 1100 molecular genetics, biochemical genetics, and cytogenetics tests under one roof and custom testing for all medically relevant genes, for domestic and international clients.

Equally important to improving patient care through quality genetic testing is the contribution EGL makes back to the scientific and medical communities. EGL is one of only a few clinical diagnostic laboratories to openly share data with the NCBI freely available public database ClinVar (>7000 variants on >500 genes) and is also the only laboratory with a free online database (EmVClass), featuring a variant classification search and report request interface, which facilitates rapid interactive curation and reporting of variants.

For more information about EGL and the nearly 1100 tests we offer:
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