Custom Gene Sequencing

Customized, Sanger-based sequence analysis of rare disease genes

Description:

The human genome is comprised of about 20,000 genes. Of these, more than 6,500 genes have been associated with genetic conditions. Availability of genetic testing for a clinically relevant rare disease gene, in a CAP/CLIA certified diagnostic laboratory, allows many patients to receive a clinically-actionable diagnosis that may have important ramifications for their long-term medical management and treatment. Confirmation of a clinical diagnosis may also reduce or eliminate the need for further diagnostic testing and unnecessary health surveillance.

Indications:

Sequencing for clinically relevant genes associated with rare conditions may be available, barring patent protection. Customized genetic testing is especially important for rare conditions with a suspected genetic etiology for which genetic testing is limited or unavailable on a clinical basis.

Specific indications include:

- Confirmation of diagnosis in a symptomatic individual
- Confirmation of test results generated in research setting
- Presymptomatic testing/carrier testing of at-risk relatives, following identification of mutation(s) in the proband
- Prenatal diagnosis of an at-risk fetus, after confirmation of mutation(s) in the parent(s).

Testing Methodology:

PCR-based sequencing of the coding regions and the exon/intron boundaries of the specified gene.

Sensitivity:

Analytical Sensitivity: The sensitivity of PCR-based DNA sequencing is over 99% for the detection of nucleotide base changes, small deletions and insertions in the regions analyzed.

In general, mutations in regulatory regions or other untranslated regions are not detected by this test. Multiple exon deletions, large insertions, genetic recombinational events and rare, primer site mutations are not identified by this methodology. However, these types of mutations may be tested by special arrangement with the laboratory.

Clinical Sensitivity: Clinical sensitivity for each gene will be variable. An estimated clinical sensitivity can be provided upon request, following review of the medical literature.

Specimen:

At least 3mLs of whole blood in lavender top (EDTA) tube. Label tube with patient's name, birth date, and date of collection. Phlebotomist must initial tube to verify patient's identity.

Please call for information on alternate specimen types.

Turn-Around Time:

Up to 42 days, depending on complexity of gene.

For Additional Information:

Please call 513-636-4474 to discuss your patient with a genetic counselor.

Please call 1-866-450-4198 for current pricing, CPT codes, insurance preauthorization or with any billing questions.

Results:

All results will be reported to the referring health care provider as specified on the test requisition form. Abnormal results will be called to the referring health care provider.

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changing the outcome together

Laboratory of Genetics and Genomics
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Phone: (513) 636-4474
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Email: LabGeneticCounselors@cchmc.org
www.cincinnatichildrens.org/genetics

Shipping Instructions:

Please enclose **test requisition** with sample.
All information must be completed before sample can be processed.

Place samples in styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Saturday.

Ship to:

Cytogenetics and Molecular Genetics Laboratories
3333 Burnet Avenue NRB 1042
Cincinnati, OH 45229
513-636-4474

References:

Grody, W.W., Richards, C.S. (2008). New quality assurance standards for rare disease testing. Genet Med 10 (5): 320-4.

Ledbetter, D.H., Faucett, W.A. (2008). Issues in genetic testing for ultra-rare diseases: background and introduction. Genet Med 10 (5): 309-13.