The University of Chicago Genetic Services Laboratories

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Exome Select

Exome Sequencing provides sequence data on the protein-coding regions of the human genome, referred to as the "exome". Our Exome Select testing option involves whole exome sequencing (proband only), with analysis limited to a customized gene list of interest. Exome Select may be an appropriate option for patients where the suspected or differential diagnosis has a limited number of associated candidate genes, but no panel exists that includes all candidate genes. It may also be useful for cases where clinical testing is not available for the gene(s) of interest. The gene list of interest may be provided to the laboratory by the ordering provider, or UCGS may assist in developing a gene list for providers interested in genes associated with a specific syndrome. The selected gene list must be approved in advance by a UCGS staff member.

If you are interested in ordering the Exome Select test, please contact one of our genetic counselors to discuss further, prior to sending a sample.

Test methods:

Exome sequencing is performed using the Agilent SureSelect Clinical Research Exome kit that is designed to target the exome with greater coverage of known disease-associated genes. Sequencing is performed using the Illumina technology and reads are aligned to the reference sequence. Approximately 90-95% of all exons are targeted at a minimum depth of 30X in the diagnostic Exome Sequencing test. Variants are identified and evaluated using a custom collection of bioinformatic tools and comprehensively interpreted by our team of directors and genetic counselors. All pathogenic and likely pathogenic variants are confirmed by Sanger sequencing.

Limitations

Not all the exons in the genome are targeted and captured due to certain inherent characteristics of the genome. In addition, there is limited or no coverage in regions outside of the exome. Certain types of mutations are not detectable by this test. This methodology will not detect low level mosaicism, copy number variations (i.e such as the deletion or duplication of an exon) and trinucleotide repeat expansions.

Reporting Results

UCGS will report on genetic variants that have been reported to be pathogenic, predicted to be pathogenic, possibly pathogenic as well as unclassified variants in the selected gene list. Genes that are not included in the Exome Select gene list will not be analyzed or reported.

Required Forms:

- Exome Sequencing Test Requisition Form
- Completed Clinical Checklist – in addition, please send detailed clinic notes, pedigree, results of previous genetic testing, and brain imaging reports if available.
- Completed Exome Sequencing Consent Form

Exome Select Panel

Sample specifications:	3 to10 cc of blood in a purple top (EDTA) tube
Cost:	$3000
CPT codes:	81415
Turn-around time:	8-10 weeks

***Note: We cannot bill insurance for the above test.**

References:

1. Bamshad MJ, Ng SB, Bigham AW et al. Exome sequencing as a tool for Mendelian disease gene discovery. Nat Rev Genet 2011: 12: 745-755.
2. Yang Y, Muzny DM, Reid JG et al. Clinical whole-exome sequencing for the diagnosis of mendelian disorders. N Engl J Med 2013: 369: 1502-1511.

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