TruGenome Technical Sequence Data

Immediate access to trusted, clinical whole-genome sequencing.

Next-generation sequencing technologies are enabling unprecedented access to the human genome. As our knowledge of the genome increases, the idea that we can use this information to help uncover the underlying genetic basis of diseases is gaining popularity. In fact, there are several publications demonstrating the potential impact of whole-genome sequencing in the clinic[1,2]. This powerful technology offers you an opportunity to explore your patient's genome, possibly revealing information that may prove helpful when making decisions regarding his/her healthcare.

Today, some clinical laboratories have the expertise to analyze and interpret whole-genome sequencing data, but are not set up to perform the actual sequencing. With the Technical Service, you and your institution's clinical lab can take advantage of whole-genome sequencing in the CLIA-certified, CAP-accredited Illumina Clinical Services Laboratory. Now you can offer your patients genetic tests using clinical whole-genome sequencing done by Illumina with results based on your own interpretations.

What is the TruGenome Technical Data Service?
The TruGenome Technical Service uses the industry-leading Illumina next-generation sequencing (NGS) technology to provide accurate clinical whole-genome sequencing. Filters for calling bases have been established based on extensive validation experiments, and are set to ensure analytical sensitivity of > 98% and analytical specificity of > 99%. The test provides broad coverage of exomic, promoter, and regulatory regions. Variants are annotated to facilitate interpretation, although complete interpretation is not included. Turnaround time is 45 days, with a 14-day rapid turnaround option*.

What information do I receive from this service?
The TruGenome Technical Service delivers a report outlining the methodology used and specifications of the sequencing run and an encrypted hard drive containing sequencing data with all reads, quality scores, and variants for further analysis. BAM files contain aligned and unaligned reads of the genome. gVCF files include the nucleotide calls and the quality of each call made at each position that passed filter throughout the genome. If needed, support from the Illumina team of genetic specialists is available. A complete description of the TruGenome Technical Service is available at www.illumina.com/test_descriptions.

What is involved in using the Technical Service?
The first step is to determine that you have the need for whole-genome sequencing and access to the bioinformatics capabilities and clinical expertise necessary for analyzing and interpreting the data. Next, contact Illumina to arrange for sequencing in our CLIA-certified, CAP-accredited lab. You will receive a collection kit that includes consent forms and sample collection tubes that are bar-coded for sample tracking. Using the tubes provided, obtain a blood sample from your patient and return to the Illumina Clinical Services Laboratory. Clinical laboratory scientists with expertise in next-generation sequencing technologies will perform the whole-genome sequencing. Technical data will be provided to you on a hard drive for subsequent analysis and interpretation.

* The rapid turnaround test must be arranged in advance to make sure that a system is reserved to sequence the samples upon arrival.

Using the TruGenome Technical Sequence Data Service*

Contract with Illumina to perform whole-genome sequencing.

Collect patient's blood sample and send to Illumina.

Sample received and sequencing performed in the Illumina Clinical Service Laboratory.

Hard drive containing technical data in the form of BAM and gVCF files sent to customer.

About the Illumina Clinical Services Laboratory

The CLIA-certified, CAP-accredited Illumina Clinical Services Laboratory was established in 2009 for the purposes of offering human whole-genome sequencing services to physicians. It is staffed with certified Clinical Laboratory Scientists who have extensive training and expertise with Illumina NGS technology. Our team of Ph.D. geneticists and bioinformaticians, genetic counselors, and certified medical geneticists has over 50 years of combined experience analyzing human genetic data.

About Illumina NGS technology

Illumina sequencing by synthesis (SBS) technology is the most published NGS technology. Using a proprietary method, DNA sequence is detected in a base-by-base manner for highly accurate, reproducible sequencing results. Learn more about sequencing at www.illumina.com/sequencing.

Learn more

If you are considering whole-genome sequencing for your patients, we strongly encourage you to contact us before ordering so that we can help you choose the appropriate test and aid in diagnostic evaluation. We are available to discuss coverage of the targeted genes/regions, additional analysis and/or support that may be needed, and any other testing needs. To learn more about the Illumina Clinical Services Laboratory and the Technical Service, visit www.illumina.com/clinicallab.

* This graphic provides a high-level overview of the process for using the Technical Service from Illumina. For complete details, visit www.illumina.com/clinicallab.

References:
1. Bras J, Guerreiro R, Hardy J (2012) Use of next-generation sequencing and other whole-genome strategies to dissect neurological disease. Nat Rev Neurosci. 13:453–64.
2. Green ED, Guyer MS; National Human Genome Research Institute (2011) Charting a course for genomic medicine from base pairs to bedside. Nature. 470: 204–213.

Disclaimer:
This laboratory test was developed and its performance characteristics were determined by the Illumina Clinical Services Laboratory (CLIA-certified, CAP-accredited). Consistent with laboratory-developed tests, it has not been cleared or approved by the U.S. Food and Drug Administration. Patients who have any questions or concerns about what they might learn through their genome sequence information should be directed to contact their physician or a genetic counselor. Please note that Illumina does not accept orders for TruGenome Clinical Sequencing Services from New York.