

TruGenome™ Technical Sequence Data

Frequently Asked Questions

Who should order the TruGenome Technical Sequence Data Service?

The TruGenome Technical Sequence Data Service is intended for clinical laboratories or clinical researchers who prefer to perform clinical analyses and interpretation for their patients directly. The Illumina Clinical Services Laboratory performs whole-genome sequencing under our CLIA-certified protocol and provides the technical data on an encrypted hard drive. The data provided are solely for use in clinical research or downstream clinical analysis in a different CLIA-certified, CAP-accredited laboratory.

What do I receive with the TruGenome Technical Sequence Data Service?

You will receive a technical report outlining the methodology used and specifications of the sequencing run. You will also receive 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. Variant call format (VCF) and genome VCF (gVCF) files include the nucleotide calls made at each position throughout the genome that pass Illumina Clinical Services Laboratory internal quality metrics. An interpreted clinical report is not provided with this service. The hard drive and encryption key are returned only to the ordering physician unless the patient authorizes these results to be released to another individual.

How do I access my patient's technical data on the hard drive?

You will be provided with an encryption key that you can use to access the hard drive.

What is a BAM file?

A BAM file contains sequence alignment data saved in binary format. If you order a technical deliverable, all sequencing reads are returned to you in BAM format and stored on an encrypted hard drive. There are many third-party and open-source applications that enable visualization of BAM files (eg, Integrative Genomics Viewer, UCSC Genome Browser, JBrowse). You can also access a BAM file using SAMtools, a set of command-line utilities developed for the Unix platform.

What is a gVCF file?

A gVCF file is a per-base representation of the entire genome in variant call format. This file contains all assayable positions in which your patient's genotype matches the reference base, contains a variant (single nucleotide, insertion, or deletion) from the reference base, or is undetermined. VCF specifications are available at www.1000genomes.org. A gVCF file can be accessed and analyzed using Unix command-line utilities or open-source tools (eg, tabix, gvcftools).

What is a VCF file?

A VCF file is a text file format used to represent different types of variant data such as single nucleotide variations (SNV), insertions, deletions, and structural variants. Each file contains meta-information, a header, and variant data on one or more samples.

What data do I receive in the VCF file?

Copy number variation (CNV) calls are provided in a VCF file. Each data record in the file specifies the boundaries for the CNV ('POS' column and 'END' tag in 'INFO' column) and the actual copy number (eg, 2-copy deletion, 1-copy deletion).

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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.

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