Illumina, Inc.
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TruGenome™ Technical Sequence Data

Description

Indication

The TruGenome Technical Sequence Data test is intended for use for CLIA/CAP laboratories and institutions with approved IRB protocols for clinical research. Whole genome sequencing data is provided in two formats: gVCF and BAM. The gVCF represents all variant calls through the genome that pass the quality thresholds set within the Illumina Clinical Services Laboratory. The accuracy of the gVCF represents an accuracy consistent with other clinically actionable tests. The BAM format is appropriate solely for downstream CLIA/CAP laboratory use, or for use in clinical research under an approved IRB protocol. The BAM data files do not constitute an analytically validated result and are not appropriate for downstream clinical use unless processed by another clinical laboratory with an analytically validated pipeline.

Reasons for referral

- Medical institutions that would like to offer whole-genome sequencing as a laboratory-developed test, but do not have the infrastructure to support the sequencing. Clinical interpretation services will be performed at the medical institution.
- Research institutions with an approved IRB protocol that will incorporate whole genome sequencing into their research studies. Clinical interpretation services will be performed at the research institution.

Method

Whole-genome sequencing is performed for this test utilizing Illumina Sequencing-By-Synthesis (SBS) chemistry and paired-end read technology. Alignment and variant identification is performed with NCBI Human Genome Reference build 37.1.

Specifications

The TruGenome Technical Sequence test provides sequence for >90% of the reportable genome. We sequence to an average of ≥30 fold coverage. Our validations demonstrate that 30 fold coverage with quality scores of ≥Q30 results in the average call having greater than 99.99% accuracy in detecting SNVs in a diploid genome. Less than 3% of our total reported data are at 10 fold coverage, and these calls achieve 98% accuracy. Insertion and deletions events in the size range of 1-7 base pairs (+ /- 7 base pairs) have a sensitivity and specificity of approximately 80-85%, as determined through analysis of an extended, multigeneration family set that has been externally validated.

Deliverables

- A technical report describing the analytical performance of the sample
- A gVCF file with all variant calls through the genome
- Technical data in BAM file format (sequence information provided in a standard open source binary format. Li et al. 2009, http://samtools.sourceforge.net/). The data must be analyzed under an approved IRB protocol for research purposes or analyzed in a CLIA-accredited laboratory for clinical use.

Limitations

It is not technically possible to capture and sequence the entire human genome at present. It is anticipated that approximately 90-95% of the human reference genome will be assessed. Only single nucleotide substitutions and small insertion and deletion events are reported for this test. Other types of genetic variants that may also lead to genetic disease, but are not reported, include copy number variants, triplet repeat expansions, and other structural chromosomal rearrangements. If clinically indicated, additional testing and analyses, such as karyotyping, microarray or MLPA may be appropriate. The clinical sensitivity for the test varies depending on the gene and condition of interest. Clinical sensitivity is unknown.

It is important to note that due to the nature of whole-genome sequencing, the test results will have implications for the patient's family members. Genetic counseling is recommended.

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Lab Statement

The TruGenome Technical Sequence test is a Laboratory Developed Test. It was developed and its performance characteristics determined by the Illumina Clinical Services Laboratory (CLIA #05D1092911). It has not been cleared or approved by the U.S. Food and Drug Administration. Pursuant to the requirements of CLIA '88, this laboratory test has established and verified the test's accuracy and precision. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. We cannot accept orders from the state of New York at this time.

Required Information and Forms

- Completed Test Requisition Form with Ordering Physician signature, Clinical History, and Billing Information
- 100% pre-payment, unless other institutional arrangements have been made

Specimen Requirements

We require that all samples be collected and returned using our Sample Collection Kits. To request a collection kit, please fill out and return the kit request form

Blood: 4-8ml whole blood in a provided PAXgene DNA tube. Ship overnight at ambient temperature in the provided, pre-paid envelope. Specimens may be refrigerated (4˚C) for >5 days if needed to avoid weekend delivery.

Price and Turn-Around Time

TruGenome Technical Sequence Data: $5,000

Timeframe: 45 days

Contact

Please contact the lab with any questions you may have regarding test selection, ordering, sample submission and results interpretation and implications.

Phone: 858.736.8080

Fax: 858.255.5285

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