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TruGenome™ Clinical Sequencing Services Methods and Technology

Frequently Asked Questions

What is clinical whole-genome testing?

Clinical whole-genome testing is the use of whole-genome sequencing and interpretation of the obtained genomic information in a clinical setting. The information is used for a comprehensive exploration of your patient's genetic data, which may be helpful in diagnosing or assessing your patient's risk for certain genetic diseases, carrier status, and response to medications. The information enables physicians and patients to make informed decisions that may lead to improved health care management.

What are the limitations of whole-genome sequencing?

There are several regions of the genome and types of genetic variants that are not covered well by any sequencing technology, including the technologies used in the Illumina Clinical Services Laboratory. Such excluded regions include areas containing large trinucleotide repeat expansions, gene duplications or pseudogenes, large insertions/deletions, or copy number variations. We are happy to review coverage of specific regions of interest with you. Depending upon the questions, additional complementary testing methods may be recommended.

What methods are used for genome sequencing and variant interpretation in the TruGenome™ Services and what is the expected accuracy?

Genome sequencing is performed via next-generation sequencing (NGS) using DNA extracted from whole blood. Variants are interpreted according to the American College of Medical Genetics (ACMG) guidelines, and include evaluation of all variants detected by using frequency, nature of variant (eg, amino acid change), and peer-reviewed literature reporting variant associations with disease.

Data are aligned and reported according to build 37.1 of the human reference genome. The analytical accuracy of these single nucleotide variant calls is > 97%, and calls are made for > 95% of the genome. Insertions/deletions are detected with > 80% accuracy. This test does not detect insertion/deletion events > 7 base pairs, translocations, trinucleotide repeats, or copy number variants. This test detects inherited variants, but not somatic variants, mosaicism, or heteroplasmy.

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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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