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TruGenome™ Predisposition Screen

Frequently Asked Questions

Who should order the TruGenome Predisposition Screen?

The TruGenome Predisposition Screen is intended for healthy adults over the age of 21 interested in learning about their risk for a specific set of monogenic adult-onset conditions, assessing their carrier status, and understanding their response to certain drugs. This information can help physicians make informed management and treatment decisions for their patient's health based on information provided from genome sequencing.

What is interpreted by the TruGenome Predisposition Screen?

The TruGenome Predisposition Screen includes analysis and interpretation of 1,691 genes that have well-established associations to a set of 1,232 conditions (as found in the NIH Genetic Testing Registry and Online Mendelian Inheritance in Man [www.omim.org], and 11 genes associated with response to 16 different drugs (as specified by the U.S. Food and Drug Administration (FDA) or the Clinical Pharmacogenomics Implementation Consortia [CPIC]). This test does not screen for variants associated with complex diseases known to have multiple genetic and environmental contributors. A complete list of genes and associated conditions and a list of alleles associated with specific drug responses are available.

What will I receive with the TruGenome Predisposition Screen?

You will receive a clinical report that includes an interpretation of genomic findings, details on clinically significant findings and their associations to conditions and drug response, references from which clinical associations were drawn, and an activation code that allows the patient to visualize and explore his/her genome information in the MyGenome Web App.

How do I view my genomic data?

Go to www.understandyourgenome.com to set up your login username and password. Select the Explore room. Enter your activation code (provided with your clinical report) to access and view your genomic data.

Why does the TruGenome Predisposition Screen test for only certain diseases/conditions?

These diseases/conditions are included because they are caused by variants (mutations) in well-characterized genes. Other diseases/conditions either do not have strong enough data supporting clinical association of specific genes or variants to the disease or, currently, the underlying genetics are not best evaluated using next-generation sequencing. Example: several genes contribute to Type 2 diabetes risk; however, none of these genes individually contribute enough to be able to use the information for medical intervention. Characteristics such as age and body mass index are far more predictive than genetics for this condition. Another example: Fragile X is caused by a trinucleotide repeat expansion that it is not detected well by current next-generation sequencing methods, so alternative methods must be used.

What is a "well-characterized" condition or disease?

A well-characterized monogenic disease is one that multiple groups have studied and for which the relationships between a single gene and the disease are understood with a high degree of confidence.

Will I receive updates to my clinical report as new variant-to-disease associations are discovered?

The Illumina Clinical Services Laboratory will provide updates to the original clinical report in the following situations:

1. If, through our standard process of interpreting variants within other patients' DNA, we change a classification of a variant such that it either now would be considered clinically reportable (pathogenic or likely pathogenic) or else was previously thought to be clinically reportable, and now is considered not clinically significant. Example: data from a new clinical study result in a variant classification change from variant of unknown significance to pathogenic. In this example, all previous reports generated with that variant will be updated and a new report will be sent to the original ordering physician.
2. The ordering physician requests to have a patient's genome reinterpreted. Requests for reinterpretation can be made at any time; however, they are limited to one time per year. An additional fee is required for this request.
3. A new version of the test is available containing new features or enhancements. An additional fee is required for this update.

Illumina • 1.800.809.4566 toll-free (U.S.) • +1.858.202.4566 tel • techsupport@illumina.com • www.illumina.com

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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