illumina®

TruGenome™ Undiagnosed Disease Test

Patient Informed Consent

Some states/countries may have additional requirements for informed consent. Make sure that you comply with those requirements and provide a copy of any additional written informed consents, if applicable.

Introduction. This form describes the benefits, risks, and limitations of having your/your child's genome tested by sequencing. This is a voluntary test. Read this form carefully before making your decision about testing.

Purpose. The purpose of this test is to detect changes present in your/your child's DNA and to understand the potential consequences of these changes. Your/your child's medical history will be used to determine what genetic changes may be causing your/your child's clinical features/symptoms. This information may help physicians make more informed management decisions for your/your child's health. For more information on genetics, genetic disease, inheritance, or genetic testing, consult your/your child's physician or genetic counselor.

Test Procedure. A tube of your/your child's blood (or a portion of previously extracted genomic DNA) will be obtained and sent to Illumina, Inc. Illumina will analyze your/your child's genetic material, generate the DNA sequence for their genome, and identify changes in the sequence, called variants. The variants identified will be interpreted based on current available scientific information and according to standard guidelines. Clinical genome testing typically include samples from the biological parents and/or other biological relatives to help interpret the test results. This requires a tube of blood to be drawn from those relatives and submitted at the same time as your/your child's sample.

Types of Test Results

1. Positive: A change(s) in your/your child's DNA sequence was found which likely explains the cause of the clinical features/symptoms.
2. Negative: No reportable change in your/your child's DNA was found to explain the cause of the clinical features/symptoms.
3. Variant of Uncertain Significance: A change was found. However, we do not have enough information to know if the change explains your/your child's clinical features/symptoms or if the change is benign/normal.
4. Secondary Findings: For individuals undergoing clinical genome sequencing, the American College of Medical Genetics and Genomics (ACMG) recommends looking for changes in a predetermined set of genes for which medical management would be altered.
    a. A change in one of these genes could be identified in your/your child's DNA that may be unrelated to current clinical features/symptoms but is important for health.
    b. All individuals for whom a sample is received, will have the option to choose to receive these results. If individuals opt to not receive these results (opt out), no report will be issued.
5. Incidental Findings: During the course of testing your DNA, we could identify variants in genes that might be important for your/your child's health and medical management but are not part of the primary reason for testing and not included on the ACMG secondary findings gene list. This type of result is called an incidental finding. While these types of results are rare, you should discuss the possibility of incidental findings with your/your child's doctor. Currently, Illumina's policy is to report an incidental finding if detected; individuals do not have an option to decline this result.
6. Pharmacogenomics: This test also looks at changes in your/your child's DNA sequence for a predetermined set of genes that have a well-established drug-gene association, and thus could affect the type/effectiveness of medication used in certain situations.
    a. All individuals for whom a sample is received will have the option to choose to receive a pharmacogenomics report. If individuals opt to not receive these results (opt out), no report will be issued.

Your Family. These genetic test results may have implications for your relatives. Speak with your/your child's physician/genetic counselor about whether to share test results with others. If you decide to do this, consider the best way to communicate this information to them.

Benefits. Test results may help you and your/your child's physician make more informed choices about your/your child's health care. It is also possible that test results will not provide any immediate or perceived benefit.

Risks

1. Physical Risks: This test requires DNA most often provided from a sample of blood. Side effects of having blood drawn are uncommon, but may include dizziness, fainting, soreness, bleeding, bruising, and rarely, infection.
2. Discrimination Risks: Genetic information could potentially be used as a basis of discrimination. To address concerns regarding possible health insurance and employment discrimination, many US states and the US government have enacted laws to prohibit genetic discrimination in these circumstances. The laws may not protect against genetic discrimination in other circumstances such as when applying for life insurance or long-term disability insurance. Talk to your/your child's physician or genetic counselor if you have concerns about genetic discrimination before testing.
3. Other Risks: Test results may reveal information about yourself/your child, or relatives, that you would rather not know. Such information may pertain to ancestry, genetic risks, and predisposition to disease, including diseases that might not be curable. In a trio or parent/child analysis, it may be discovered that a family member is unrelated to the patient, such as in the case of adoption or nonpaternity. It may not be possible to prevent learning such information through this test. Talk to your/your child's physician or genetic counselor about the type of information that you do and do not want to know.

Implications and Limitations of Genetic Testing—While test results may aid in determining the cause of your/your child's clinical features/symptoms, it is also possible that these results may not provide any benefit. Much about genetics is not yet fully understood and your/your child's results may include variants that are of unknown clinical significance based on available information today. In addition, this test can only detect specific kinds of changes in DNA; other kinds of changes could also cause disease or lead to symptoms. Testing technology for clinical genome sequencing has limits, including the inability to sequence all parts of a person's genome and a known error rate (although it is low). Further testing of you and/or your family may be needed to confirm your/your child's test results, which could result in an expense to you.

Delivery of Test Results/Privacy—Illumina keeps test results confidential. Your/your child's test results will be sent to the physician that ordered the test. Speak with your/your child's physician if you would like a copy of the test results. You may request a copy of these test results from the laboratory directly. Illumina will release results to other health care providers involved in your/your child's medical care, or to another health care provider as directed by you (or a person legally authorized to act on your behalf) in writing, or otherwise as required or authorized by applicable law.

Use of Information—We may use your leftover specimen for additional laboratory purposes (including quality control, laboratory operations, and laboratory improvement). We may also use your de-identified health information (including genetic data) resulting from your sample for scientific research purposes. Such uses may result in the development of commercial products and services. You will not receive notice of any specific uses and you will not receive any compensation for these uses. All such uses will be de-identified, in compliance with applicable law, and only performed under the approval and oversight of an Institutional Review Board. You may opt out of having your leftover specimen used for additional laboratory purposes by checking the opt-out box below and returning this form to the laboratory or by contacting the laboratory and requesting to opt out. This does not apply to leftover specimens collected from New York State if you chose to opt-out below. New York state law requires Illumina to destroy samples after 60 days.

Reanalysis—Our knowledge of genetics continues to grow. As more information becomes available, an additional look at your/your child's sample may be beneficial to see if newly discovered genetic changes can explain your/your child's clinical features/symptoms.

Future Correspondence—Understanding of genetic variation is rapidly advancing, meaning that some of the changes we find in your/your child's genome might be better understood in the future. Illumina recommends that you keep in contact with your/your child's health care provider on an annual basis to learn of any new developments in genetics and to provide any updates to your/your child's personal or family history.

Cross-Border Data Transfer—If you are from outside the United States, your/your child's specimen and associated health information will be sent to the United States for the testing to be completed. As part of the testing, additional health information about you/your child will be created and maintained. Your country may consider the legal privacy protections in the United States to be inadequate.

Learn more

TruSight Undiagnosed Disease Test, illumina.com/clinical/illumina_clinical_laboratory/trugenome-clinical-sequencing-services.html

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I have read, or have had read to me, all the above statements and understand information regarding clinical genome sequencing. By signing this form, I authorize Illumina to perform the genetic testing as ordered by my physician.

☐ By selecting this box, I confirm that I do not give my permission to the use of my leftover de-identified specimen and health information resulting from my sample for additional laboratory purposes (which includes quality control, laboratory operations, and laboratory improvement).

Name of patient being tested (please print)	**Date of Birth (DD/MON/YYYY eg, 28/Feb/2024)**
Signature of Patient/Legal Guardian	**Date (DD/MON/YYYY eg, 28/Feb/2024)**
Provider signature	**Date (DD/MON/YYYY eg, 28/Feb/2024)**

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