PerkinElmer
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Family Member Clinical Genomics Requisition Form

Please complete every field and tick box clearly.

PATIENT INFORMATION

Patient's First Name

Patient's Date of Birth MM/ DD /YYYY

Patient's Last Name

Biological Sex: ○ Male ○ Female ○ Unknown
Gender Identity (if different from above): _____

Patient's Street Address

City / Town | State | Zip Code

Country | Patient's Preferred Phone

Patient's Email

Ethnicity (check all that apply):	○ African-American	○ Asian (China, Japan, Korea)
○ Caucasian/N. European/S. European	○ Finnish	○ French Canadian
○ Hispanic	○ Jewish - Ashkenazi	○ Jewish - Sephardic	○ Mediterranean
○ Middle Eastern (Saudi Arabia, Qatar, Iraq, Turkey)	○ Native American	○ E. Indian
○ Southeast Asian (Vietnam, Cambodia, Thailand)	○ South Asian (India, Pakistan)	
○ Other (specify) _____		

PROVIDER

Provider's First and Last Name

Account # | Provider's Phone

Provider's Email

Clinic/Hospital/Institution Name

Provider's Street Address

City / Town | State | Zip Code

Country | Provider's Fax

ADDITIONAL PROVIDER/GENETIC COUNSELOR (IF APPLICABLE)

Provider/Genetic Counselor's Name

Provider /Genetic Counselor's Account # | Provider/Genetic Counselor's Phone

Provider/Genetic Counselor's Email | Provider/Genetic Counselor's Fax

PATIENT SAMPLE INFORMATION

SAMPLE TYPE: Collection Date: MM/DD/YY

○ Saliva Swab
○ Whole Blood
○ Dried Blood Spots
Other_____

TEST MENU (Familial Testing Only)

○ D1999 Family Member TRIO Testing

PROBAND INFORMATION

Last name, First name

Relation to Proband

PHYSICIAN STATEMENT

Confirmation of informed and medical necessity for genetic testing

The undersigned person (or representative thereof) ensures he/she is a licensed medical professional authorized to order genetic testing and confirms that the patient has given appropriate consent. I confirm that testing is medically necessary and that test results may impact medical management for the patient. Furthermore, all information on this TRF is true to the best of my knowledge. My signature applies to the attached letter of medical necessity (unless this box is checked). ☐

Signature_____ Date_____

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FAMILY MEMBER CONSENT FORM

PerkinElmer Genetics, Inc., and its affiliates, contractors and assigns, ("PerkinElmer") requires a completed Patient's Informed Consent Form (Consent). The Patient's Consent must be completed by you, the patient, or a legally authorized representative of the patient. For any patient below the age of majority, Consent must be completed by the patient's legally authorized representative.

Your health care provider (HCP) has recommended that you, or your child, receive biochemical or molecular genetics clinical testing (Test) as indicated on the PerkinElmer Test Requisition Form (Requisition). The purpose of the Consent is to provide you with a description of the Test ordered; known risks and benefits of the Test; retention of your sample and personal health (PH) information; research opportunities; and the reporting of secondary findings. Given the complexity of the type of Test ordered, it is recommended that you and/or your child receive genetic counseling by a trained genetic counselor or medical geneticist both prior to and after the Test. Your HCP, or their representative, will walk you through the Consent process and will provide you with this Consent form so that you can acknowledge that you have agreed to participate in the Test, retention of sample and reporting of secondary findings and that you or your child's participation is strictly voluntary. You also understand the importance of and availability of a genetic counselor or medical geneticist in you or your child's case.

TEST INFORMATION

A Test will be used to identify what, if any, DNA mutation(s) you or your child is carrying which is causing the specific disease or condition. Identifying the mutation may be useful for diagnostic and treatment purposes, and allows at-risk family members to be tested. Only the genes identified on the Requisition Form will be analyzed. It is recommended that you receive genetic counseling before and directly after having this genetic test. You can find a genetic counselor in your area at www.nsgc.org. Contact your HCP to find out if further testing or additional consultations are needed.

TEST METHOD

With your Consent to the HCP ordered Test, your HCP will take a sample of your (or your child's) blood, saliva, body fluid, tissue specimen, or other sample type. This sample will be prepared for isolation and purification of DNA. The Test will cover only the gene, disease or condition requested on the Requisition. Your specimen will be used for the purpose of attempting to determine if you or your child are carriers of a disease mutation or gene, or are affected with, or at increased risk of someday being affected with a genetic disease.

TEST RESULTS

Your treating HCP has sole responsibility for all decisions concerning the management of your diagnosis; PerkinElmer will not provide a diagnosis. PerkinElmer will report Test results only to your HCP via a secure internet portal. Your HCP will tell you the results of the Tests and if your test is positive, may refer you or your child to a specialist for further clinical evaluation and confirmation of diagnosis. A positive genetic test result may indicate that you are a carrier of, predisposed to, or have the specific disease or condition being tested for. A negative result from the Test ordered cannot rule out all genetic diseases or conditions as each disease or condition requires a specific test however the results of this Test may still assist your HCP in further testing and making a diagnosis.

TEST LIMITATIONS

Due to current limitations in technology and the lack of knowledge of mutations and genes, some mutations may not be detected by the Tests. There is a possibility that the result findings will be uninterpretable or of unknown significance. In rare circumstances, results may be suggestive of a condition different than that which was originally considered for purpose of consenting to this testing. The Test may find a mutation(s) that lead to conditions for which the patient currently does not have symptoms or find a mutation(s) that may not be related to the patient's disease.

TEST RISKS

Patients and family members may experience anxiety before, during, and/or after testing. Testing multiple family members may reveal that familial relationships are not biologically what they were assumed to be. For example, the Testing may indicate non-paternity (the stated father of an individual is not the biological father) or consanguinity (the parents of an individual are closely related by blood). These biological relationships may need to be reported to the health care provider who ordered the test.

Depending upon what type of sample your HCP collects from you or your child may include risks of mild pain, bruising, swelling, redness, and a slight risk of infection. Light-headedness, fainting or nausea may occur if your HCP collects blood or tissue samples. These side-effects are typically brief and transient, but you should contact your HCP if you or your child requires treatment. Under some circumstances an additional sample may be required for Tests to be performed.

Federal laws prohibit health insurers/employers from discriminating based on your genetic information. There are currently no federal laws that prohibit life insurance, long term care, or disability insurance companies from discriminating based on genetic information. Unless required by Law, PerkinElmer will not disclose your identifiable information to any person except as you have authorized on the records release and authorization at your HCP office.

SAMPLE RETENTION

Pursuant to laboratory best practices, your sample, personal information (PI), the data from the Test (including those performed before any withdrawal of Consent), and the related reports will be retained by PerkinElmer for two years per CLIA regulations. In some instances, there may be a benefit to having your or your child's sample and PI retained for a longer period for additional testing. If properly instructed by your HCP, PerkinElmer will retain your identified sample for a longer period. *For New York State HCP practices, your identifiable sample will be destroyed 60 days after test completion.*

SAMPLE ANONYMIZATION

PerkinElmer anonymizes and retains data and related reports from your Test indefinitely for its internal statistical and quality analysis, research, scientific and technical development, and market research. PerkinElmer anonymizes and retains your sample indefinitely for internal quality control, test validation, and assay development and improvement. Future analyses of the anonymized data, reports, and the sample may be conducted by third parties. By consenting, you understand and agree with PerkinElmer's use of data and specimens and that you give up property rights to the Sample and are donating the data and specimen to PerkinElmer.

RESEARCH OPTIONS

PerkinElmer may collaborate with scientists, researchers and drug developers to advance knowledge of genetic diseases. If there are opportunities to participate in future research relevant to the disorder in you, or your child, PerkinElmer may contact you or your HCP about the development of new testing, drug development, or other treatments.

TESTING TYPE

The type of Test you are consenting to is a genetic test that sequences thousands of genes at the same time. These Tests are called Whole Exome (WES) or Whole Genome (WGS) tests. These tests may detect variants in known disease-associated genes or may detect variants in genes that have not yet been associated with disease. The WES testing targets the region of the genome that contains the genes, called the exome. The WGS Testing involves sequencing the entire genome, which is the entire DNA in your cells. In some cases, we may not be able to know with certainty that the variant is actually causing the disease.

WHY ARE PARENTAL SAMPLES NEEDED?

In some circumstances, it may be helpful for additional family members to undergo testing as well in order to provide information that can aid in the interpretation of the WES/WGS test results. These tests could be part of a TRIO test or as standalone targeted testing. PerkinElmer, in consultation with your ordering physician, will decide if other family members need to be tested.

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WHAT IS REPORTED

It is mandatory to report any diagnostic findings for both you and/or your child related to disease or a different condition not related to the current condition. Related disease is a known pathogenic variant(s), a likely pathogenic variants(s), or a known variant(s) of uncertain significance in genes interpreted to be responsible for, or potentially contributing to the patient's disease. This also includes variants in genes not yet associated with disease but may be associated in the future. Conditions not related to disease in childhood onset, is a single pathogenic or likely pathogenic variant in genes that are known to cause autosomal dominant or X-linked childhood onset conditions, as well as two pathogenic or likely pathogenic variants in genes that are known to cause autosomal recessive childhood onset conditions, even if they are unrelated to the patient's disease, will be reported to your HCP.

SECONDARY FINDINGS

Since many different genes and conditions are being analyzed during genetic Test, the tests may reveal some findings not directly related to the reason for ordering the Test. These findings are called "secondary" and can provide information that was not anticipated when the Test was ordered. Secondary findings are variants found in genes that are unrelated to the individual's reported clinical features.

As recommended by American College of Medical Genetics and Genomics (ACMG), PerkinElmer will report secondary findings identified in a specific subset of medically actionable genes associated with various inherited disorders for all individuals undergoing WGS or WES. Reportable secondary findings will be confirmed by an alternate test method. Please refer to the latest version of the ACMG Recommendations for Reporting of Secondary Findings in Clinical Exome and Genome Sequencing for complete details at www.acmg.net. In addition to the 59 genes recommended by ACMG, PerkinElmer will report secondary findings listed below.

1. **Pharmacogenetic variants:** Changes in the DNA that do not cause a disease but may be related to how your body processes certain medications, such as chemotherapy drugs, antipyretics, antidepressants, anticoagulants, and others. These variants may not be important to you if you are not taking the medications involved, but may tell you how well the medications will work or if you will have side effects if you do take the medications now or in the future.

2. **Carrier Status for Autosomal Recessive Conditions (ex. cystic fibrosis):** A recessive condition is one in which two pathogenic variants in the same gene are required in order to show symptoms of the disease (one variant is inherited from each parent). Someone who has only one pathogenic variant does not show symptoms and is called a carrier. However, if we find a pathogenic variant in a recessive gene that is related to the patient's disease, we will report it as a diagnostic finding. Further testing may be necessary to look for a second pathogenic variant in that gene not identified by WGS. You can choose whether or not you want us to report carrier status in genes that are not related to the patient's disease. The Testing is not designed to be a comprehensive carrier test. We are unable to guarantee that all conditions for which the individual is a carrier will be determined by the Testing. An individual may be a carrier for a condition in which there was little or no coverage in the Testing and therefore will not be detected. Additional carrier testing for reproductive purposes should be discussed with your doctor or genetic counselor.

3. **Diagnostic findings in adult onset currently medically non-actionable disorders not related to disease:** Conditions that are not currently medically-actionable do not have recommended treatment or preventative measures. An example would be Alzheimer's disease. We are unable to guarantee that the Testing will find all adult onset medically non-actionable conditions for which the individual has a pathogenic variant. An individual may have a pathogenic variant for a condition in which there was little or no coverage in the Testing and therefore will not be detected. Additional testing for health purposes should be discussed with your doctor or genetic counselor.

4. **Diagnostic findings in adult onset medically-actionable disorders not related to disease:** Medically-actionable conditions are those for which there is currently recommended treatment or preventative actions that can be taken to reduce the risk of developing the disease. An example would be hereditary cancer syndromes such as Lynch syndrome. We are unable to guarantee that the Testing will find all adult onset medically-actionable conditions for which the individual has a pathogenic variant. An individual may have a pathogenic variant for a condition in which there was little or no coverage in the Testing and therefore will not be detected. Additional testing for health purposes should be discussed with your doctor or genetic counselor.

FAMILY MEMBER'S CONSENT TO TESTING

I have read the Consent as provided by my HCP and understand and give permission to the following:

- I understand that my, or my child's participation in this genetic testing is voluntary.
- I understand that by signing this Consent I am giving PerkinElmer permission to perform a Test as was ordered by my HCP.
- I understand that my personal information is protected by law and will not be used or linked to the results of any study or publication.
- I understand that if my HCP practices in New York State, I agree that PerkinElmer may retain my anonymized specimen for more than 60 days after Test completion.
- I give PerkinElmer permission to use my specimen anonymously in studies at PerkinElmer to improve testing and for publication.
- I give PerkinElmer permission to inform my HCP, or myself, of any research opportunities that may be associated with my or my child's Test results and any secondary findings.
- I understand that if I wish to withdraw from the Test, or if I have any questions about the Test, that I may contact PerkinElmer via email at: Genomics@perkinelmer.com or toll- free by telephone 1-866-354-2910 to request withdrawal.
- I understand that I (or my legal representative) are entitled to a copy of this Consent.

FAMILY MEMBER TESTING AUTHORIZATION AND OPTIONAL DISCLOSURE CONSENT

☐ Check this box if you wish to receive a report on pharmacogenetic variants (see Secondary Findings section above for details).

☐ Check this box if you wish to receive a report on carrier status –must be 18 years or older (see Secondary Findings section above for details).

☐ Check this box if you wish to receive a report on adult onset medically-actionable conditions–must be 18 years or older (see Secondary Findings section above for details).

☐ Check this box if you wish to receive a report on adult-onset not currently actionable conditions.–must be 18 years or older (see Secondary Findings section above for details).

Family Member Name: _____ Date_____ Time _____

Signature (Family Member): _____