

## PATIENT INFORMATION

Patient's First Name Middle Initial

Patient's Last Name Patient's Date of Birth

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

## 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 informed consent for the testing ordered, including a discussion of the benefits and limitations. 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 informed consent and/or attached letter of medical necessity, if applicable (unless this box is checked).

Signature \_\_\_\_\_ Date \_\_\_\_\_

## PATIENT SAMPLE INFORMATION

**SAMPLE TYPE:**  Saliva Swab  Whole Blood  Dried Blood Spots  Other \_\_\_\_\_  
 Collection Date: MM/DD/YY  
 Age of Manifestation: \_\_\_\_\_  
 ICD10 CODES: \_\_\_\_\_

## TEST MENU (Please see page 2 for full menu and additional testing options.)

CNGnome™  
 Exome – Proband Only  Genome – Proband Only  
 Exome – TRIO\*  Genome - TRIO\*

### Available Optional Testing Enhancements for Exome/Genome (Additional charges apply)

Include CNGnome™ with Exome (already included with all Genome tests)  
 STAT – Results in 7-10 days for Exome OR 10-12 days for Genome  
 With StepOne™ – With comprehensive biochemical analysis  
 Receive Separate Parental Reports (only available with TRIO tests)

! \*Please fill out family member section below. Additional samples MUST be received within 3 weeks.

## FAMILIAL INFORMATION (Required with TRIO orders)

### BIOLOGICAL MOTHER:

Last name, First name  
 Date of Birth: MM/DD/YYYY  
 Symptomatic (clinically affected)?  Yes  No  
 Sample:  Included - Collection Date MM/DD/YY  To be sent later

### BIOLOGICAL FATHER:

Last name, First name  
 Date of Birth: MM/DD/YYYY  
 Symptomatic (clinically affected)?  Yes  No  
 Sample:  Included - Collection Date MM/DD/YY  To be sent later

### ADDITIONAL FAMILY MEMBER:

Last name, First name  
  
 Relationship to Patient  
 Date of Birth: MM/DD/YYYY  
 Symptomatic (clinically affected)?  Yes  No  
 Sample:  Included - Collection Date MM/DD/YY  To be sent later

## SINGLE SITE ANALYSIS (Please include a copy of relative's report, if available.)

Gene Variant Name (c.)  
  
 Proband's Name  
   
 Relationship to Proband Original Accession#  
 Positive Control Sample:  To be sent later  Already at PKIG  Not available

## BILLING INFORMATION - INSTITUTION

Institution/Organization

Contact Name

Institution Phone

Institution Billing Address

Institution Fax

City / Town

State

Zip Code

Institution Email

**Special Handling Notes** (Internal Use Only):

## BILLING INFORMATION - SELF PAY

**Check:** \$ \_\_\_\_\_ Amount Enclosed (Please make checks payable to: PerkinElmer Genetics, Inc.)

**Credit Card** (Please fill out all information):

Credit Card Number

CVV

Credit Card Billing Street Address

MM/YY

Card Exp. Date

Cardholder Phone

City / Town

State

Zip Code

Cardholder Printed Name as Appears on Card

Cardholder Signature

Please select additional testing option, if applicable.

## ADDITIONAL TECHNOLOGY ONLY MENU OPTIONS (NO INTERPRETATION INCLUDED)

<b>Exome</b>	<input type="radio"/> D1500	Whole Exome Sequencing, Proband Only - Data Only
	<input type="radio"/> D1510	Whole Exome Sequencing, TRIO - Data Only
	<input type="radio"/> D1520	STAT Whole Exome Sequencing, Proband Only - Data Only
	<input type="radio"/> D1530	STAT Whole Exome Sequencing, TRIO - Data Only
<b>Genome</b>	<input type="radio"/> D2500	Whole Genome Sequencing, Proband Only - Data Only
	<input type="radio"/> D2510	Whole Genome Sequencing, TRIO - Data Only
	<input type="radio"/> D2520	STAT Whole Genome Sequencing, Proband Only - Data Only
	<input type="radio"/> D2530	STAT Whole Genome Sequencing, TRIO - Data Only

## ADDITIONAL REQUESTS FOR SEQUENCING DATA\*

What type of data would you like to receive? <input type="radio"/> Raw Data in addition to Clinical Report <input type="radio"/> Raw Data Only (Must select a DATA ONLY test option)	Raw Data Option <input type="radio"/> FASTQ files <input type="radio"/> BAM files (Not available with DATA ONLY options) <input type="radio"/> Variant Call File (VCF) (Not available with DATA ONLY options)	Type of Data Delivery Requested: <input type="radio"/> Electronic Transfer <input type="radio"/> Hard Drive
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\* All tests come with a clinical test report as standard unless a DATA ONLY testing option is selected. Please use this section to select different reporting options involving raw data. Additional costs may apply.

**PHENOTYPE(S) / PATIENT HISTORY (CHECK ALL THAT APPLIES)**

Clinical diagnosis: \_\_\_\_\_

Age of manifestation: \_\_\_\_\_ ICD-10 Codes: \_\_\_\_\_

DETAILED MEDICAL RECORDS, CLINICAL SUMMARY, PICTURES AND FAMILY HISTORY MUST BE ATTACHED. CLINICAL INFORMATION IS CRUCIAL FOR ACCURATE INTERPRETATION OF RESULTS.

<p><b>A. NEUROLOGY</b></p> <p><b>1. Behavioral abnormality</b></p> <p><input type="radio"/> 1.1 Autism</p> <p><input type="radio"/> 1.2 Attention deficit disorder</p> <p><input type="radio"/> 1.3 Psychiatric diseases</p> <p><b>2. Brain imaging</b></p> <p><input type="radio"/> 2.1 Abnormal myelination</p> <p><input type="radio"/> 2.2 Abnormal cortical gyration</p> <p><input type="radio"/> 2.3 Agenesis of corpus callosum</p> <p><input type="radio"/> 2.4 Brain atrophy</p> <p><input type="radio"/> 2.5 Cerebellar hypoplasia</p> <p><input type="radio"/> 2.6 Heterotopia</p> <p><input type="radio"/> 2.7 Holoprosencephaly</p> <p><input type="radio"/> 2.8 Hydrocephalus</p> <p><input type="radio"/> 2.9 Leukodystrophy</p> <p><input type="radio"/> 2.10 Lissencephaly</p> <p><b>3. Developmental delay</b></p> <p><input type="radio"/> 3.1 Delayed motor development</p> <p><input type="radio"/> 3.2 Delayed language development</p> <p><input type="radio"/> 3.3 Developmental regression</p> <p><input type="radio"/> 3.4 Intellectual disability</p> <p><b>4. Movement abnormality</b></p> <p><input type="radio"/> 4.1 Ataxia</p> <p><input type="radio"/> 4.2 Chorea</p> <p><input type="radio"/> 4.3 Dystonia</p> <p><input type="radio"/> 4.4 Parkinsonism</p> <p><b>5. Neuromuscular abnormality</b></p> <p><input type="radio"/> 5.1 Muscular hypotonia</p> <p><input type="radio"/> 5.2 Muscular hypertonia</p> <p><input type="radio"/> 5.3 Hyperreflexia</p> <p><input type="radio"/> 5.4 Spasticity</p> <p><b>6. Seizures</b></p> <p><input type="radio"/> 6.1 Febrile seizures</p> <p><input type="radio"/> 6.2 Focal seizures</p> <p><input type="radio"/> 6.3 Generalized seizures</p> <p><b>7. Others</b></p> <p><input type="radio"/> 7.1 Craniosynostosis</p> <p><input type="radio"/> 7.2 Dementia</p> <p><input type="radio"/> 7.3 Encephalopathy</p> <p><input type="radio"/> 7.4 Headache/ Migraine</p> <p><input type="radio"/> 7.5 Macrocephaly</p> <p><input type="radio"/> 7.6 Microcephaly</p> <p><input type="radio"/> 7.7 Neuropathy</p> <p><input type="radio"/> 7.8 Stroke</p>	<p><b>B. METABOLISM</b></p> <p><input type="radio"/> 1. Abnormal creatine kinase</p> <p><input type="radio"/> 2. Decreased plasma carnitine</p> <p><input type="radio"/> 3. Hyperalaninemia</p> <p><input type="radio"/> 4. Hypoglycemia</p> <p><input type="radio"/> 5. Increased CSF lactate</p> <p><input type="radio"/> 6. Increased serum pyruvate</p> <p><input type="radio"/> 7. Ketosis</p> <p><input type="radio"/> 8. Lactic acidosis</p> <p><input type="radio"/> 9. Organic aciduria</p> <p><b>C. EYE</b></p> <p><input type="radio"/> 1. Blepharospasm</p> <p><input type="radio"/> 2. Cataract</p> <p><input type="radio"/> 3. Coloboma</p> <p><input type="radio"/> 4. Glaucoma</p> <p><input type="radio"/> 5. Microphthalmos</p> <p><input type="radio"/> 6. Nystagmus</p> <p><input type="radio"/> 7. Ophthalmoplegia</p> <p><input type="radio"/> 8. Optic atrophy</p> <p><input type="radio"/> 9. Ptosis</p> <p><input type="radio"/> 10. Retinitis pigmentosa</p> <p><input type="radio"/> 11. Retinoblastoma</p> <p><input type="radio"/> 12. Strabismus</p> <p><input type="radio"/> 13. Visual impairment</p> <p><b>D. MOUTH, THROAT AND EAR</b></p> <p><input type="radio"/> 1. Abnormality of dental color</p> <p><input type="radio"/> 2. Cleft lip / palate</p> <p><input type="radio"/> 3. Conductive hearing impair.</p> <p><input type="radio"/> 4. External ear malformation</p> <p><input type="radio"/> 5. Hypodontia</p> <p><input type="radio"/> 6. Sensoneural hearing impair.</p> <p><b>E. SKIN, INTEGUMENT AND SKELETAL</b></p> <p><b>1. Skeletal</b></p> <p><input type="radio"/> 1.1 Abnormal limb morphology</p> <p><input type="radio"/> 1.2 Abnormal skeletal system</p> <p><input type="radio"/> 1.3 Abnormal vertebral column</p> <p><input type="radio"/> 1.4 Joint hypermobility</p> <p><input type="radio"/> 1.5 Multiple joint contractures</p> <p><input type="radio"/> 1.6 Polydactyly</p> <p><input type="radio"/> 1.7 Scoliosis</p> <p><input type="radio"/> 1.8 Syndactyly</p> <p><input type="radio"/> 1.9 Talipes equinovarus</p> <p><b>OTHER:</b></p>	<p><b>2. Skin and integument</b></p> <p><input type="radio"/> 2.1 Abnormal skin pigmentation</p> <p><input type="radio"/> 2.2 Abnormal hair</p> <p><input type="radio"/> 2.3 Abnormal nail</p> <p><input type="radio"/> 2.4 Hyperextensible skin</p> <p><input type="radio"/> 2.5 Ichthyosis</p> <p><b>F. CARDIOVASCULAR</b></p> <p><input type="radio"/> 1. Angioedema</p> <p><input type="radio"/> 2. Aortic dilatation</p> <p><input type="radio"/> 3. Arrhythmia</p> <p><input type="radio"/> 4. Coarctation of aorta</p> <p><input type="radio"/> 5. Defect of atrial septum</p> <p><input type="radio"/> 6. Defect of ventricular septum</p> <p><input type="radio"/> 7. Dilated Cardiomyopathy</p> <p><input type="radio"/> 8. Hypertension</p> <p><input type="radio"/> 9. Hypertrophic Cardiomyopathy</p> <p><input type="radio"/> 10. Hypotension</p> <p><input type="radio"/> 11. Lymphedema</p> <p><input type="radio"/> 12. Malf. of heart and great vessels</p> <p><input type="radio"/> 13. Myocardial infarction</p> <p><input type="radio"/> 14. Stroke</p> <p><input type="radio"/> 15. Tetralogy of Fallot</p> <p><input type="radio"/> 16. Vasculitis</p> <p><b>G. GASTROINTESTINAL, GENITOURINARY, ENDOCRINE</b></p> <p><b>1. Gastrointestinal</b></p> <p><input type="radio"/> 1.1 Aganglionic megacolon</p> <p><input type="radio"/> 1.2 Constipation</p> <p><input type="radio"/> 1.3 Diarrhea</p> <p><input type="radio"/> 1.4 High hepatic transaminases</p> <p><input type="radio"/> 1.5 Gastroschisis</p> <p><input type="radio"/> 1.6 Hepatic failure</p> <p><input type="radio"/> 1.7 Hepatomegaly</p> <p><input type="radio"/> 1.8 Obesity</p> <p><input type="radio"/> 1.9 Pyloric stenosis</p> <p><input type="radio"/> 1.10 Vomiting</p> <p><b>2. Genitourinary</b></p> <p><input type="radio"/> 2.1 Abnormal renal morphology</p> <p><input type="radio"/> 2.2 Abnormal urinary system</p> <p><input type="radio"/> 2.3 Hydronephrosis</p> <p><input type="radio"/> 2.4 Renal agenesis</p> <p><input type="radio"/> 2.5 Renal cyst</p> <p><input type="radio"/> 2.6 Renal tubular dysfunction</p>	<p><b>3. Endocrine</b></p> <p><input type="radio"/> 3.1 Diabetes mellitus</p> <p><input type="radio"/> 3.2 Hypo / hyperparathyroidism</p> <p><input type="radio"/> 3.3 Hypo / hyperthyroidism</p> <p><b>H. Reproduction</b></p> <p><input type="radio"/> 1. Abnormal external genitalia</p> <p><input type="radio"/> 2. Abnormal internal genitalia</p> <p><input type="radio"/> 3. Hypogonadism</p> <p><input type="radio"/> 4. Hypospadias</p> <p><input type="radio"/> 5. Infertility</p> <p><b>I. Oncology</b></p> <p><input type="radio"/> 1. Adenomatous polyposis</p> <p><input type="radio"/> 2. Breast carcinoma</p> <p><input type="radio"/> 3. Colorectal carcinoma</p> <p><input type="radio"/> 4. Leukemia</p> <p><input type="radio"/> 5. Myelofibrosis</p> <p><input type="radio"/> 6. Neoplasm of the lung</p> <p><input type="radio"/> 7. Neoplasm of the skin</p> <p><input type="radio"/> 8. Paraganglioma</p> <p><input type="radio"/> 9. Pheochromocytoma</p> <p><b>J. HEMATOLOGY AND IMMUNOLOGY</b></p> <p><input type="radio"/> 1. Abnormality of coagulation</p> <p><input type="radio"/> 2. Anemia</p> <p><input type="radio"/> 3. Immunodeficiency</p> <p><input type="radio"/> 4. Neutropenia</p> <p><input type="radio"/> 5. Pancytopenia</p> <p><input type="radio"/> 6. Abnormal hemoglobin</p> <p><input type="radio"/> 7. Splenomegaly</p> <p><input type="radio"/> 8. Thrombocytopenia</p> <p><b>K. PRENATAL AND DEVELOPMENT</b></p> <p><input type="radio"/> 1. Dysmorphic facial features</p> <p><input type="radio"/> 2. Failure to thrive</p> <p><input type="radio"/> 3. Hemihypertrophy</p> <p><input type="radio"/> 4. Hydrops fetalis</p> <p><input type="radio"/> 5. IUGR</p> <p><input type="radio"/> 6. Oligohydramnios</p> <p><input type="radio"/> 7. Overgrowth</p> <p><input type="radio"/> 8. Polyhydramnios</p> <p><input type="radio"/> 9. Premature birth</p> <p><input type="radio"/> 10. Short stature</p> <p><input type="radio"/> 11. Tall stature</p>
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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; anonymization of personal health information (PHI); sample and data retention; research opportunities; and the reporting of secondary findings. 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, data and reporting of secondary findings and that you or your child's participation is strictly voluntary. You also understand 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 before and after the Test.

## TEST INFORMATION

The type of Test you are consenting to is a genetic test that looks at the thousands of genes in your DNA. These Tests are called Whole Genome (WGS) or Whole Exome (WES). These tests may identify known disease-associated genes or unknown disease-associated genes. The WES Test looks at the exome which is part of the genome that contains the genes. The WGS Test looks at the entire genome, which is the entire DNA in your cells. The Test you will be consenting to will be used to identify DNA mutation(s) or genes you and/or your child may be carrying which is causing the specific disease or condition you are being tested for. 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 will be analyzed. In some cases, we may not be able to know with certainty which gene is actually causing the disease.

## TEST METHOD

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

## 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. The HCP will tell you the results of the Tests and if the 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. The results of this Test may still assist your HCP with 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 ordered. There is a possibility that the Test result that is uninterpretable or of unknown significance, may require further testing when more information is gained about mutations and genes. In rare circumstances, Test results may be suggestive of a condition different than that which was originally considered for purpose of consenting to this Test. The Test may also find mutation(s) or genes that lead to conditions for which the patient currently does not have symptoms or may not be related to the current condition.

## 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 HCP who ordered the test.

The type of sample your HCP collects from you and/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 and/or your child require 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.

## ANONYMIZATION

PerkinElmer anonymizes the sample and data where all PHI is removed. PerkinElmer is requesting consent to keep you and/or your child's sample and data indefinitely in anonymized format. Anonymized samples are retained for internal quality control and test development, validation and improvements. Anonymized data and samples including Tests performed before any withdrawal of Consent are retained for internal statistics, quality, analysis, research, scientific, technical and market research purposes. 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 samples and that you give up property rights to the sample and are donating the data and sample to PerkinElmer.

## RETENTION

PerkinElmer is required to retain reports in traceable form including your PHI for two years according to CLIA regulation with the exception of reports for individuals with a New York State HCP which will be retained for seven years. *New York State samples will be destroyed 48-hours after test completion if consent is not obtained.* By PerkinElmer having access to your and/or your child's sample and data, it allows for prompt access in the event additional testing or analysis is requested by the HCP. PerkinElmer is requesting consent to keep you and/or your child's anonymized sample and data indefinitely.

## 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 disease in you and/or your child, PerkinElmer may contact you or your HCP about the development of new testing, drug development, or other treatments.

## 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 stand-alone targeted testing. PerkinElmer, in consultation with the HCP, will decide if other family members need to be tested. If the HCP recommends testing for additional family members, only the Test performed will be reported. If undergoing a TRIO test (WES or WGS) parents will have the option of receiving a full parental report for an additional charge. If selected, the respective parental consent section must be completed below.

## TEST REPORT

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 mutations is known by pathogenic variant(s), likely pathogenic variant(s), or variant(s) of uncertain significance in genes interpreted to be responsible for, or potentially contributing to disease or condition. This also includes variants in genes not yet associated with disease but which 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 the 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. Reportable secondary findings will be confirmed by an alternate test method. **SECONDARY FINDINGS WILL ONLY BE REPORTED IF CONSENT IS GIVEN BY PATIENT OR PARENT.** Secondary findings are classified into four categories 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 medically-actionable disorders not related to disease:** As recommended by American College of Medical Genetics and Genomics (ACMG), secondary findings should be reported in a specific subset of medically-actionable genes (ACMG 59) associated with various inherited disorders for all individuals undergoing WGS or WES. 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](http://www.acmg.net). 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.
- 4. 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.

## PATIENT’S CONSENT TO TESTING

On behalf of myself and/or my child, I have read the Consent as provided by the HCP and understand and agree to the following:

- I understand that 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 the HCP.
- I understand that my personal health information (PHI) is protected by law and will not be used or linked to the results of any study or publication.
- I understand that the HCP practices in New York State, I agree that PerkinElmer may retain the anonymized sample indefinitely.
- I agree to give up property rights to and am voluntarily donating my and/or my child’s sample and data to PerkinElmer or a qualified third party for internal statistical, quality control, research, scientific, technical and market research purposes.
- I give PerkinElmer permission to conduct future analyses of my and/or my child’s anonymized sample and data.
- I give PerkinElmer permission to inform the HCP, or myself, of any research opportunities that may be associated with my and/or my child’s Test results.
- 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](mailto:Genomics@perkinelmer.com) or by 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.

## PATIENT 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).

Print Patient Name: \_\_\_\_\_ Date \_\_\_\_\_ Time \_\_\_\_\_

Signature (Patient/Representative): \_\_\_\_\_

### Mother of Patient for TRIO testing authorization and optional disclosure consent : (Required if Parental Reports are desired and selected on the test requisition form):

- 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).

Print Mother Name: \_\_\_\_\_ Date \_\_\_\_\_ Time \_\_\_\_\_

Signature (Mother of Patient): \_\_\_\_\_

### Father of Patient for TRIO testing authorization and optional disclosure consent : (Required if Parental Reports are desired and selected on the test requisition form):

- 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).

Print Father Name: \_\_\_\_\_ Date \_\_\_\_\_ Time \_\_\_\_\_

Signature (Father of Patient): \_\_\_\_\_