PerkinElmer For the Better | PerkinElmer GENOMICS

# Clinical Genomics Requisition Form

Please complete every field and tick box clearly.

## PATIENT INFORMATION

Patient's First Name | Middle Initial | Patient's Date of Birth (MM/ DD /YYYY)

Patient's Last Name | Patient ID Number

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:**
○ Saliva Swab
○ Whole Blood
○ Dried Blood Spots
Other_____

Collection Date: MM/DD/YY
Was this sample collected in NY State: ○ Yes ○ No
Age of Manifestation:_____
ICD10 CODES: _____

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

○ CNGnome™

**Whole Exome Sequencing Testing Options**

○ Exome – Proband Only
○ Exome – TRIO (Proband Report Only)*
○ Exome – TRIO (Proband and Parental Reports)*

**Whole Genome Sequencing Testing Options**

○ Genome – Proband Only
○ Genome – TRIO (Proband Report Only)*
○ Genome – TRIO (Proband and Parental Reports)*

**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

! *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

## PHYSICIAN STATEMENT

**Confirmation of informed consent 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_____





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# Clinical Genomics Requisition Form

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

○ Targeted Single Site Analysis

Gene | Variant Name (c.)

Positive Control Sample: ○ To be sent later ○ Already at PKIG ○ Not available

Proband's Name | Relationship to Proband | Original Accession#

## ADDITIONAL TECHNOLOGY ONLY MENU OPTIONS: NO INTERPRETATION INCLUDED (Please do not choose an option below if a test was selected on page 1)

| | | |
|---|---|---|
| **Exome** | ○ D1500 | Whole Exome Sequencing - Data Only (per sample) |
| **Genome** | ○ D2500 | Whole Genome Sequencing - Data Only (per sample) |

## AVAILABLE RAW DATA OPTIONS BY TEST TYPE*

**Data Only Tests (No Clinical Report Issued)**

○ FASTQ file

*BAM and VCF files are not available for Data Only testing options.*

**Full Report Tests (Clinical Report Issued)**

○ FASTQ file
○ BAM files
○ Variant Call File (VCF)

**Type of Data Delivery Requested:**

○ Electronic Transfer
○ Hard Drive

* All tests come with a clinical test report as standard unless a DATA ONLY testing option is selected. Please use this section to indicate raw data files desired and preferred delivery method. Additional costs may apply.

## 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 | Card Exp. Date (MM/YY) | Cardholder Phone

City / Town | State | Zip Code | Cardholder Printed Name as Appears on Card

Cardholder Signature



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# Clinical Genomics Requisition Form

## 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.**

### A. NEUROLOGY

**1. Behavioral abnormality**

- O 1.1 Autism
- O 1.2 Attention deficit disorder
- O 1.3 Psychiatric diseases

**2. Brain imaging**

- O 2.1 Abnormal myelination
- O 2.2 Abnormal cortical gyration
- O 2.3 Agenesis of corpus callosum
- O 2.4 Brain atrophy
- O 2.5 Cerebellar hypoplasia
- O 2.6 Heterotopia
- O 2.7 Holoprosencephaly
- O 2.8 Hydrocephalus
- O 2.9 Leukodystrophy
- O 2.10 Lissencephaly

**3. Developmental delay**

- O 3.1 Delayed motor development
- O 3.2 Delayed language development
- O 3.3 Developmental regression
- O 3.4 Intellectual disability

**4. Movement abnormality**

- O 4.1 Ataxia
- O 4.2 Chorea
- O 4.3 Dystonia
- O 4.4 Parkinsonism

**5. Neuromuscular abnormality**

- O 5.1 Muscular hypotonia
- O 5.2 Muscular hypertonia
- O 5.3 Hyperreflexia
- O 5.4 Spasticity

**6. Seizures**

- O 6.1 Febrile seizures
- O 6.2 Focal seizures
- O 6.3 Generalized seizures

**7. Others**

- O 7.1 Craniosynostosis
- O 7.2 Dementia
- O 7.3 Encephalopathy
- O 7.4 Headache / Migraine
- O 7.5 Macrocephaly
- O 7.6 Microcephaly
- O 7.7 Neuropathy
- O 7.8 Stroke

### B. METABOLISM

- O 1. Abnormal creatine kinase
- O 2. Decreased plasma carnitine
- O 3. Hyperalaninemia
- O 4. Hypoglycemia
- O 5. Increased CSF lactate
- O 6. Increased serum pyruvate
- O 7. Ketosis
- O 8. Lactic acidosis
- O 9. Organic aciduria

### C. EYE

- O 1. Blepharospasm
- O 2. Cataract
- O 3. Coloboma
- O 4. Glaucoma
- O 5. Microphthalmos
- O 6. Nystagmus
- O 7. Ophthalmoplegia
- O 8. Optic atrophy
- O 9. Ptosis
- O 10. Retinitis pigmentosa
- O 11. Retinoblastoma
- O 12. Strabismus
- O 13. Visual impairment

### D. MOUTH, THROAT AND EAR

- O 1. Abnormality of dental color
- O 2. Cleft lip / palate
- O 3. Conductive hearing impair.
- O 4. External ear malformation
- O 5. Hypodontia
- O 6. Sensoneural hearing impair.

### E. SKIN, INTEGUMENT AND SKELETAL

**1. Skeletal**

- O 1.1 Abnormal limb morphology
- O 1.2 Abnormal skeletal system
- O 1.3 Abnormal vertebral column
- O 1.4 Joint hypermobility
- O 1.5 Multiple joint contractures
- O 1.6 Polydactyly
- O 1.7 Scoliosis
- O 1.8 Syndactyly
- O 1.9 Talipes equinovarus

**2. Skin and integument**

- O 2.1 Abnormal skin pigmentation
- O 2.2 Abnormal hair
- O 2.3 Abnormal nail
- O 2.4 Hyperextensible skin
- O 2.5 Ichthyosis

### F. CARDIOVASCULAR

- O 1. Angioedema
- O 2. Aortic dilatation
- O 3. Arrhythmia
- O 4. Coarctation of aorta
- O 5. Defect of atrial septum
- O 6. Defect of ventricular septum
- O 7. Dilated Cardiomyopathy
- O 8. Hypertension
- O 9. Hypertrophic Cardiomyopathy
- O 10. Hypotension
- O 11. Lymphedema
- O 12. Malf. of heart and great vessels
- O 13. Myocardial infarction
- O 14. Stroke
- O 15. Tetralogy of Fallot
- O 16. Vasculitis

### G. GASTROINTESTINAL, GENITOURINARY, ENDOCRINE

**1. Gastrointestinal**

- O 1.1 Aganglionic megacolon
- O 1.2 Constipation
- O 1.3 Diarrhea
- O 1.4 High hepatic transaminases
- O 1.5 Gastroschisis
- O 1.6 Hepatic failure
- O 1.7 Hepatomegaly
- O 1.8 Obesity
- O 1.9 Pyloric stenosis
- O 1.10 Vomiting

**2. Genitourinary**

- O 2.1 Abnormal renal morphology
- O 2.2 Abnormal urinary system
- O 2.3 Hydronephrosis
- O 2.4 Renal agenesis
- O 2.5 Renal cyst
- O 2.6 Renal tubular dysfunction

**3. Endocrine**

- O 3.1 Diabetes mellitus
- O 3.2 Hypo / hyperparathyroidism
- O 3.3 Hypo / hyperthyroidism

**H. Reproduction**

- O 1. Abnormal external genitalia
- O 2. Abnormal internal genitalia
- O 3. Hypogonadism
- O 4. Hypospadias
- O 5. Infertility

**I. Oncology**

- O 1. Adenomatous polyposis
- O 2. Breast carcinoma
- O 3. Colorectal carcinoma
- O 4. Leukemia
- O 5. Myelofibrosis
- O 6. Neoplasm of the lung
- O 7. Neoplasm of the skin
- O 8. Paraganglioma
- O 9. Pheochromocytoma

**J. HEMATOLOGY AND IMMUNOLOGY**

- O 1. Abnormality of coagulation
- O 2. Anemia
- O 3. Immunodeficiency
- O 4. Neutropenia
- O 5. Pancytopenia
- O 6. Abnormal hemoglobin
- O 7. Splenomegaly
- O 8. Thrombocytopenia

**K. PRENATAL AND DEVELOPMENT**

- O 1. Dysmorphic facial features
- O 2. Failure to thrive
- O 3. Hemihypertrophy
- O 4. Hydrops fetalis
- O 5. IUGR
- O 6. Oligohydramnios
- O 7. Overgrowth
- O 8. Polyhydramnios
- O 9. Premature birth
- O 10. Short stature
- O 11. Tall stature

**OTHER:**

PerkinElmer Genetics, Inc., ("PerkinElmer") requires a completed Patient's Informed Consent Form (ICF) for testing to be performed. The ICF must be completed by the patient, or a legally authorized representative of the patient (or by the healthcare provider where permitted under applicable law or regulation). For any patient below the age of majority, the ICF must be completed by the patient's legally authorized representative.

The purpose of this ICF 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, if applicable. Given the complexity of the type of the Test, it is recommended that you and/or your child receive genetic counseling by a trained genetics professional before and after the testing is performed.

## TEST INFORMATION

Your healthcare provider ("HCP") has recommended that you or your child, receive enzymatic, biochemical or molecular genetics clinical testing ("Test") indicated on the submitted Test Requisition Form ("Requisition"). For more information on the reasons your HCP has ordered the Test, and the disorders your HCP is having you tested for, please consult with your HCP. You are free to decide if you want this Test performed or not. Providing a Sample and undergoing the Test is voluntary and you may withdraw your consent without penalty at any time.

*Enzyme/Biomarker Test:* This type of test measures the presence or absence of enzymes/biomarkers and/or their level of activity in an individual. Only the enzymes/biomarkers identified on the requisition will be tested. Results from this type of Test may indicate the presence of a specific condition or conditions, and follow-up confirmatory testing may be recommended.

*Genetic/Genomic Test:* This type of Test looks at the genes in your DNA. This Test is used to identify what, if any, DNA variant(s) you or your child is 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 determine with certainty which gene is actually causing the disease.

## TEST METHOD

If you consent to the Test, your HCP will take a sample of your and/or your child's blood, saliva, body fluid, tissue or other sample type. Your Sample will be sent to PerkinElmer's laboratories in the United States for the Test; the enzyme activity, biomarker tests, and select genetic testing assays will be conducted in Pennsylvania, USA, and all other genetic testing will be conducted in Connecticut, USA.

Under some circumstances, including inadequate or poor quality sample, an additional Sample may be required for Tests to be performed.

## TEST RESULTS

Your treating HCP has sole responsibility for all decisions concerning the possible management of your diagnosis and disease; PerkinElmer will not provide a diagnosis. PerkinElmer will report Test results only to your HCP via secure email, a secure internet portal, or fax. Your HCP is responsible for communicating with you regarding the results of the Test and may refer you or your child to a specialist for further clinical evaluation and confirmation of diagnosis, if applicable. Possible results include:

1. *Positive:* 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 positive genetic test may limit your access to health insurance or life assurance coverage; for example, a life insurance company might ask you to provide genetic information indicating a disorder if this information is available to you.
2. *Negative:* A negative result indicates that the enzyme/biomarker results were within normal ranges, or that no disease-causing variant was identified in the Test performed. No Test can rule out all genetic diseases or conditions. A negative result does not guarantee that you are free from genetic disorders or other medical conditions.
3. *Inconclusive/Variant of Uncertain Significance:* A variant of uncertain significance (VOUS) result indicates that a variant outside of the normal range was detected, but it is currently unknown if the variant is associated with a genetic disorder. A VOUS is not the same as a positive result and does not clarify whether there is an increased risk to develop a genetic disorder. The variant could be a benign change or it could be indicative of disease/disease-causing.
4. *Unexpected Results:* In rare instances, this Test may reveal an important genetic change that is not directly related to the reason for ordering this test. This information would be disclosed to your HCP if it potentially impacts medical care, and you have consented to receive this type of result

## TEST REPORT

Reported disease-causing variants are described as pathogenic variant(s), likely pathogenic variants(s), or variant(s) of uncertain significance in genes interpreted to be responsible for, or potentially contributing to, a disease or condition. In addition, variants in genes not known to be associated with disease but for which there is evidence to suggest an association with disease may also be reported.

When Whole Exome Sequencing (WES) or Whole Genome Sequencing (WGS) tests are ordered by your HCP, you have the option to receive some findings not directly related to the reason for ordering the Test. Please read the Secondary Findings section on page 3 of this consent form for more information, and reporting options.

## INFORMATION ABOUT PARENTAL AND FAMILIAL SAMPLES

In some circumstances, it may be helpful for additional family members to undergo testing 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 LIMITATIONS

Due to current limitations in technology and incomplete knowledge of diseases and genes, some variants may not be detected by the Test 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. In rare circumstances, Test results may be suggestive of a condition different from that which was originally considered for the purpose of consenting to this Test. The Test may also find variants or genes that lead to conditions for which you currently do not have symptoms or may not be related to your 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 Test 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.

Taking a blood or tissue sample from you and/or your child may lead to 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.

A positive test result may limit your access to health insurance or life assurance coverage; for example, a life insurance company might ask you to provide genetic information indicating a disorder if this information is available to you. Please refer to information on the Genetic Information Nondiscrimination Act (GINA) and applicable local laws for more information.



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## CONFIDENTIALITY

You have the right to confidential treatment of the Sample and your PHI. Your HCP will provide PerkinElmer with Personal Health Information ("PHI") such as your name, date of birth, gender and clinical symptoms to help track your sample and report results. To maintain confidentiality, the test results will only be released to the referring health care provider, to the ordering laboratory, to the patient/guardian, to other health care providers involved in your diagnosis and treatment, or as otherwise required by law or regulation. Unless required by law, PerkinElmer will not disclose your PHI to any person or entity except with your written consent.

You and your HCP can control how your Sample and PHI are processed. You have the right to request access to your PHI, request corrections of any errors in recorded PHI, or where PHI may be missing or incomplete ask that it be completed. You also have the right to ask that your PHI be erased, subject to law or regulation. You can contact your HCP for such requests and your HCP will contact PerkinElmer, or you can contact PerkinElmer directly by visiting www.perkinelmergenomics.com. If requests for access, correction, completion, or erasure cannot be fulfilled, you will be informed and provided with the reasons why your requests cannot be fulfilled.

## SAMPLE AND DATA RETENTION

Pursuant to laboratory best practices, your DNA sample will be retained by PerkinElmer for a minimum of two years and then destroyed. Additionally, your PHI, the data from the Tests (including those performed before any withdrawal of consent) and the related reports will be retained by PerkinElmer for a minimum of two years and then destroyed. In some instances, it may be beneficial to you for PerkinElmer to retain your sample for a longer period of time in order to conduct additional testing, and PerkinElmer will do so with appropriate documentation from you or your HCP.

PerkinElmer is requesting consent to keep you and/or your child's anonymized sample and data indefinitely. This consent is optional, and the Test will be performed whether or not you provide consent to the following:

- PerkinElmer will anonymize and retain your Sample indefinitely for internal quality control, test validation, assay development and improvement. By allowing PerkinElmer to retain your Sample, you understand and agree that you give up any property rights you may have in the Sample and are donating it to PerkinElmer Genetics, Inc. If you withdraw your consent, no additional tests or anonymization will be carried out on your Sample; no results will be reported and your sample, reports and data that have not been anonymized will be destroyed.
  - ❑ Check here if you would like to opt out of anonymized sample retention. Note, if not checked, this is interpreted as "consent given"
- PerkinElmer will anonymize your data and retain the anonymized data and related anonymized reports from your Tests indefinitely for internal statistical, quality analysis, research, scientific and technical development, and market research.
  - ❑ Check here if you would like to opt out of anonymized data retention. Note, if not checked, this is interpreted as "consent given"

### For residents of NY State:

❑ By checking here I give PerkinElmer permission to store my sample for longer than 60 days. Note, if not checked, this is interpreted as "consent not given"

## 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.

## WITHDRAWAL OF CONSENT

I understand this consent is voluntary and is valid until I withdraw my consent. I understand I may withdraw my consent to sample and data retention, and to the Test at any time, that PerkinElmer will not perform the Test unless I provide consent to the Test. If I withdraw any consent, it will not affect actions taken before I withdrew my consent, including any anonymization of data or of my Sample. I understand that if I wish to withdraw my consent I should contact PerkinElmer via email at: Genomics@perkinelmer.com or toll-free by telephone +1-866-354-2910 to request withdrawal.

## PATIENT CONSENT TO TESTING

❑ By checking this box I attest:

I have read and understood the Informed Consent Form in its entirety, including the explanation of why my sample is being tested, how genetic testing is performed and the risks associated with genetic testing. I have had the opportunity to ask my HCP questions about the information contained herein, and understand that I am entitled to a copy of this ICF. My signature below acknowledges my free consent to the Test, and to any additional consents indicated above, and such testing in no way guarantees my health, the health of an unborn child, or the health of other family members.

Patient Signature (or Parent/Guardian if patient is minor) _____ Date _____

Patient Name _____ Name and Relationship (Parent/Guardian if patient is minor) _____

## FAMILY MEMBER CONSENT TO TESTING (if applicable)

❑ By checking this box I attest:

I have read and understood the Informed Consent Form in its entirety, including the explanation of why my sample is being tested, how genetic testing is performed and the risks associated with genetic testing. I have had the opportunity to ask my HCP questions about the information contained herein, and understand that I am entitled to a copy of this ICF. My signature below acknowledges my free consent to the Test, and to any additional consents indicated above, and such testing in no way guarantees my health, the health of an unborn child, or the health of other family members.

Family Member Signature _____ Date _____

Family Member Name _____ Relationship to Patient _____

## FAMILY MEMBER CONSENT TO TESTING (if applicable)

❑ By checking this box I attest:

I have read and understood the Informed Consent Form in its entirety, including the explanation of why my sample is being tested, how genetic testing is performed and the risks associated with genetic testing. I have had the opportunity to ask my HCP questions about the information contained herein, and understand that I am entitled to a copy of this ICF. My signature below acknowledges my free consent to the Test, and to any additional consents indicated above, and such testing in no way guarantees my health, the health of an unborn child, or the health of other family members.

Family Member Signature _____ Date _____

Family Member Name _____ Relationship to Patient _____

## SECONDARY FINDINGS: REQUIRED ONLY FOR WES/WGS

Since many different genes and conditions are being analyzed during the genetic Test, some findings not directly related to the reason for ordering the Test may be revealed. 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. Secondary findings are classified into four categories ("sections") 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 your 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 WES/WGS. You can choose whether or not you want us to report carrier status in genes that are not related to your disease. The Test is not designed to be a comprehensive carrier test. We are unable to guarantee that all conditions for which you are a carrier will be determined by the Test. You 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 genes defined as highly penetrant and medically actionable by the American College of Medical Genetics and Genomics:** As recommended by American College of Medical Genetics and Genomics (ACMG), secondary findings should be offered in a specific subset of highly penetrant and medically actionable genes 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. 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 Test will find all adult onset medically-actionable conditions for which you have a pathogenic variant. You may have a pathogenic variant for a condition in which there was little or no coverage in the Test and therefore will not be detected. Additional testing for health purposes should be discussed with your doctor or genetic counselor.
4. **Diagnostic findings in all other disease-causing genes not related to your clinical features:** Conditions that are medically-actionable but not included in section 3 (above), as well as conditions that are not medically-actionable (do not have recommended treatment or preventative measures), which may be childhood or adult onset. An example would be Alzheimer's disease. We are unable to guarantee that the Test will find all pathogenic variants in all disease-causing genes. You may have a pathogenic variant for a condition in which there was little or no coverage in the Test and therefore will not be detected. Additional testing for health purposes should be discussed with your doctor or genetic counselor.

**Secondary findings will only be reported if consent is given by the Patient or Parent/Guardian.** Each individual receiving secondary findings will need to fill out the appropriate section(s) below to indicate which secondary findings that they will receive. If a box is not checked, it is assumed that the applicable individual does not want to receive the corresponding secondary finding(s).

### PATIENT SECONDARY FINDINGS CONSENT

❑ Check this box if you wish to receive a report on pharmacogenetic variants (see Secondary Findings section #1 above for details).
❑ Check this box if you wish to receive a report on carrier status – (see Secondary Findings section #2 above for details).
❑ Check this box if you wish to receive a report on pathogenic or likely pathogenic findings in genes defined as highly penetrant and medically actionable by the American College of Medical Genetics and Genomics (see Secondary Findings section #3 above for details).
❑ Check this box if you wish to receive a report including pathogenic or likely pathogenic findings in all other disease-causing genes (see Secondary Findings section #4 above for details).

For categories 3 and 4 above, please select if you would like only pediatric findings, only adult findings, or both. The lack of selection will result in return of ALL results ("both"). Please note, it is recommended that the patient be 18 years or older for the return of adult findings.

❑ Pediatric Findings Only ❑ Adult Findings Only ❑ Both Pediatric and Adult Findings

Patient Signature (or Parent/Guardian if patient is minor) _____ Date _____

Patient Name _____ Name and Relationship (Parent/Guardian if patient is minor) _____

### FAMILY MEMBER(S) SECONDARY FINDINGS CONSENT

*If parental reports are desired, the appropriate selection needs to be made on the test requisition form **in addition** to completing this section. Filling out this section alone does not mean parental reports will be returned.*

❑ Check this box if you wish to receive a report on pharmacogenetic variants (see Secondary Findings section #1 above for details).
❑ Check this box if you wish to receive a report on carrier status – (see Secondary Findings section #2 above for details).
❑ Check this box if you wish to receive a report on pathogenic or likely pathogenic findings in genes defined as highly penetrant and medically actionable by the American College of Medical Genetics and Genomics (see Secondary Findings section #3 above for details).
❑ Check this box if you wish to receive a report including pathogenic or likely pathogenic findings in all other disease-causing genes (see Secondary Findings section #4 above for details).

For categories 3 and 4 above, please select if you would like only pediatric findings, only adult findings, or both. The lack of selection will result in return of ALL results ("both"). Please note, it is recommended that the patient be 18 years or older for the return of adult findings.

❑ Pediatric Findings Only ❑ Adult Findings Only ❑ Both Pediatric and Adult Findings

Family Member Signature _____ Date _____

Family Member Name _____ Relationship to Patient _____

### FAMILY MEMBER(S) SECONDARY FINDINGS CONSENT

*If parental reports are desired, the appropriate selection needs to be made on the test requisition form **in addition** to completing this section. Filling out this section alone does not mean parental reports will be returned.*

❑ Check this box if you wish to receive a report on pharmacogenetic variants (see Secondary Findings section #1 above for details).
❑ Check this box if you wish to receive a report on carrier status – (see Secondary Findings section #2 above for details).
❑ Check this box if you wish to receive a report on pathogenic or likely pathogenic findings in genes defined as highly penetrant and medically actionable by the American College of Medical Genetics and Genomics (see Secondary Findings section #3 above for details).
❑ Check this box if you wish to receive a report including pathogenic or likely pathogenic findings in all other disease-causing genes (see Secondary Findings section #4 above for details).

For categories 3 and 4 above, please select if you would like only pediatric findings, only adult findings, or both. The lack of selection will result in return of ALL results ("both"). Please note, it is recommended that the patient be 18 years or older for the return of adult findings.

❑ Pediatric Findings Only ❑ Adult Findings Only ❑ Both Pediatric and Adult Findings

Family Member Signature _____ Date _____

Family Member Name _____ Relationship to Patient _____



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