Please complete every field and tick box clearly.

Whole Exome Sequencing Requisition Form

PerkinElmer
For the Better

PATIENT INFORMATION

Patient's First Name | Patient's Last Name

Patient's Street Address | Patient's Date of Birth (MM/DD/YY) | Patient's Email

City / Town | State | Zip Code | Country | Patient's Preferred Phone

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)_____		

Biological Sex:		
○ Male	○ Female	○ Unknown

Gender Identity:_____
(If different from above)

PROVIDER INFORMATION

PROVIDER

Provider's First Name | Provider's Last Name

Clinic / Hospital / Institution Name | Provider's Phone

Provider's Street Address | Provider's Email

City / Town | State | Zip Code | Country | Provider's Fax

GENETIC COUNSELOR (IF APPLICABLE)

Genetic Counselor's Name | Genetic Counselor's Email

Genetic Counselor's Phone | Genetic Counselor's Fax

ADDITIONAL PROVIDER CONTACT

Additional Provider's Name | Additional Provider's Email

Additional Provider's Phone | Additional Provider's Fax

FAMILY INFORMATION

MOTHER

Mother's First Name | Mother's Last Name

Mother's Date of Birth (MM/DD/YY) | Date of Sample Collection (MM/DD/YY) | Time of Sample Collection (:) ○ AM ○ PM | Symptomatic / Asymptomatic (or clinically affected / clinically unaffected)

FATHER

Father's First Name | Father's Last Name

Father's Date of Birth (MM/DD/YY) | Date of Sample Collection (MM/DD/YY) | Time of Sample Collection (:) ○ AM ○ PM | Symptomatic / Asymptomatic (or clinically affected / clinically unaffected)

ADDITIONAL FAMILY MEMBER

Additional Family Member's Full Name | Relationship to Patient

Date of Birth (MM/DD/YY) | Date of Sample Collection (MM/DD/YY) | Time of Sample Collection (:) ○ AM ○ PM | Symptomatic / Asymptomatic (or clinically affected / clinically unaffected)

SPECIMEN INFORMATION

Date of Sample Collection (MM/DD/YY) | Time of Sample Collection (:) ○ AM ○ PM

SAMPLE TYPE: ○ Whole Blood ○ Dried Blood Spots ○ Saliva ○ Other_____

Age of Manifestation

NUMBER OF SAMPLES: ○ Solo ○ Trio (patient and parent(s)) ○ Trio Plus (additional family member beyond Trio)

TEST MENU

SAMPLES TO BE TESTED: ○ Patient only ○ Patient and parent(s) ○ Patient and other (sibling)

BIOINFORMATICS: ○ Data delivery FastQ, bam file format ○ Variant Call File (VCF) ○ Formatted Variant report

TEST CODE	TEST NAME
☐ EXOME	Whole Exome Sequencing, with interpretation (one sample)
☐ EXOMETECH	Whole Exome Sequencing, no interpretation (one sample)
☐ EXOMETRIO	Whole Exome Sequencing, with interpretation (three samples)
☐ EXOMETRIOTECH	Whole Exome Sequencing, no interpretation (three samples)
☐ EXOMESTAT	Whole Exome Sequencing, with interpretation (one sample) STAT
☐ EXOMESTATPLUS	Whole Exome Sequencing, with interpretation plus StepOne® Biochemical Profile STAT
☐ EXOMETECHSTAT	Whole Exome Sequencing, no interpretation (one sample) STAT
☐ EXOMETRIOSTAT	Whole Exome Sequencing, with interpretation (three samples) STAT
☐ EXOMETRIOSTATPLUS	Whole Exome Sequencing, with interpretation (three samples) plus StepOne® Biochemical Profile STAT
☐ EXOMETRIOTECHSTAT	Whole Exome Sequencing, no interpretation (three samples) STAT

BILLING INFORMATION

○ **INSTITUTIONAL BILLING**

Institution / Organization

Contact Name | **Institution Phone**

Institutional Billing Street Address | **Institution Email**

City / Town | **State** | **Zip Code** | **Institution Fax**

○ **SELF PAY** **Please make checks payable to: PerkinElmer Genetics, Inc.**

○ **Cashier Check** $_____ Amount Enclosed

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

CVV | **Credit Card Number**

Cardholder Billing Street Address | **Card Exp. Date** (MM/YY) | **Cardholder Phone**

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

Cardholder Signature

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

A. NEUROLOGY

1. Behavioral abnormality

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

2. Brain imaging

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

3. Developmental delay

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

4. Movement abnormality

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

5. Neuromuscular abnormality

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

6. Seizures

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

7. Others

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

B. METABOLISM

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

C. EYE

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

D. MOUTH, THROAT AND EAR

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

E. SKIN, INTEGUMENT AND SKELETAL

1. Skeletal

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

2. Skin and integument

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

F. CARDIOVASCULAR

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

G. GASTROINTESTINAL, GENITOURINARY, ENDOCRINE

1. Gastrointestinal

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

2. Genitourinary

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

3. Endocrine

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

H. Reproduction

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

I. Oncology

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

J. HEMATOLOGY AND IMMUNOLOGY

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

K. PRENATAL AND DEVELOPMENT

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

WHOLE EXOME SEQUENCING INFORMED CONSENT FORM

Informed consent is a process that provides education about genetics, and the options, benefits, limitations, and consequences of genetic testing. Genetic counseling provides the patient with informed consent prior to the decision to undergo testing and with the opportunity to review the results of the test in detail. Given the complexity of the Testing, genetic counseling and informed consent by a trained medical geneticist or genetic counselor is strongly recommended prior to and after undergoing this testing. By signing this form, you acknowledge that you have undergone genetic counseling or have been informed about the availability and importance of genetic counseling and that you have been provided with written information identifying a genetic counselor or medical geneticist. You can also visit www.nsgc.org to locate a genetic counselor in your area.

What is the Testing?

- The Testing is PKIG's Whole Exome Sequencing (WES) test. The Testing targets the region of the genome that contains the genes, called the exome. The exome is estimated to comprise approximately 1% of the genome, yet contains approximately 85% of disease-causing pathogenic variants ("mutations").
- The Testing is different from other genetic tests you may have had in the past because it sequences thousands of genes at the same time rather than sequencing only one or a few genes.
- Any WES test, including the Testing, may detect variants in known disease-associated genes or may detect variants in genes that have not yet been associated with disease. In the latter case, we may not be able to know with certainty that the variant is actually causing the disease in the patient.
- Based on WES studies, the Testing is expected to provide a diagnosis in nearly 28% of the cases for rare and ultra-rare disorders.

Why are parental samples needed?

- In order to interpret your results, other family members may also need to have the Testing or to have targeted testing depending on who else in the family is affected with the disorder and is available for testing. PKIG, in consultation with your ordering physician, will decide if other family members need to be tested.
- Parents are often the most informative family members to test to further interpret results; therefore, parental samples are often sent along with the patient's sample for testing.

How is the Testing performed and what are the risks of collecting the sample?

- The Testing requires 5-10 cc of blood, which has risks associated with obtaining the sample, such as bruising and bleeding from a blood draw. Alternate sample types such as saliva and dried blood spot cards may be used on request. DNA will be extracted from the blood sample and sequencing will be performed on the exome using next generation sequencing (NGS) technology. A list of sequence variants that potentially could be important to the patient's disease will be generated. Since NGS has a small false positive rate, each variant that is reported may need to be confirmed by a second detection method, such as Sanger sequencing.

Limitations of Whole Exome Sequencing by NGS

- A fraction of the exome cannot be sequenced to accurately determine if a pathogenic variant is present. Therefore, pathogenic variants in these regions will not be detected by this analysis.
- NGS cannot accurately sequence repetitive regions, such as trinucleotide repeats. This means that NGS cannot provide data on regions such as the fragile X syndrome repeat region, the Huntington disease repeat region, or the myotonic dystrophy repeat region.
- Results from the Testing may indicate that additional testing, such as full gene sequencing to complete exons with poor coverage or deletion/duplication analysis, is recommended.
- Large deletions and duplications are not evaluated in the Testing.
- Genetic changes identified may not necessarily predict the prognosis or severity of disease and it is possible that the genetic change may not affect management or treatment.

Potential risks associated with Whole Exome Sequencing:

- Discovery of variants indicating conditions not yet present - WES may show pathogenic variants in genes that lead to conditions for which the patient currently does not have symptoms (such as cancer, neuromuscular diseases, and adult onset disorders such as Alzheimer's disease). For some conditions, you may be given the option of knowing whether pathogenic variants are present.
- Uncertainty - We may not be able to tell you with certainty whether the variant(s) we find are directly related to the patient's disease. The interpretation of WES results will evolve over time as we learn more about normal and abnormal human genetic variation.
- Anxiety - Patients and family members may experience anxiety before, during, and/or after testing.
- Insurance access – The Testing may become part of your permanent medical record and, depending on the results, may have a material effect on your access to health insurance or life insurance coverage; for example, a life insurance company might ask you to provide genetic information indicating a disorder if this information is available to you. The federal Genetic Information and Non-Discrimination Act (GINA) prohibits the use of genetic information for discrimination in health insurance and employment. Individual states may also have laws concerning the use of genetic information.
- Testing multiple family members may reveal that familial relationships are not biologically what they were assumed to be. For example, the Testing may indicate nonpaternity (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.

A. MANDATORY DISCLOSURES (FOR BOTH MINORS AND ADULT PATIENTS):

1. Diagnostic findings related to disease - pathogenic variant(s), likely pathogenic variants(s), and variant(s) of uncertain significance in genes interpreted to be responsible for, or contributing to the patient's disease will be reported to your health care provider (HCP).
2. Diagnostic findings not related to disease in childhood onset conditions - 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.

B. OPTIONAL DISCLOSURES

1. **FOR BOTH MINORS AND ADULT PATIENTS (One election):**

1. **Pharmacogenetic variants:** Pharmacogenetic variants are 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.

◯ YES, report information regarding pharmacogenetic variants. Patient/Guardian Initials_____

◯ NO, DO NOT report information regarding pharmacogenetic variants. Patient/Guardian Initials_____

2. **FOR ADULT PATIENTS ONLY (18 years or older) (three elections)**

A. **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 Whole Exome Sequencing. 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.

◯ YES, report information regarding carrier status. Patient/Guardian Initials_____

◯ NO, DO NOT report information regarding carrier status. Patient/Guardian Initials_____

B. **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.

◯ YES, report information regarding adult-onset actionable conditions. Patient/Guardian Initials_____

◯ NO, DO NOT report information regarding adult-onset actionable conditions. Patient/Guardian Initials_____

C. **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.

○ YES, report information regarding adult-onset not currently actionable conditions. Patient/Guardian Initials _____
○ NO, DO NOT report information regarding adult-onset not currently actionable conditions. Patient/Guardian Initials _____

3. **FOR PARENTS OF EXOME TRIOS ONLY:**

Carrier status, pharmacogenetic variants, adult onset medically actionable disorders, and adult onset not currently medically-actionable disorders can be reported for parents undergoing the Testing as part of our Trios option. If these options are requested, a separate report will be provided per parent. Please initial next to the appropriate response.

Please note that we are unable to guarantee that all pathogenic variants in each option will be detected by the Testing. 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.

Carrier Status

Mother
○ YES, report information regarding carrier status. Mother's Initials _____
○ NO, DO NOT report information regarding carrier status. Mother's Initials _____

Father
○ YES, report information regarding carrier status. Father's Initials _____
○ NO, DO NOT report information regarding carrier status. Father's Initials _____

Pharmacogenetic Variants

Mother
○ YES, report information regarding pharmacogenetic status. Mother's Initials _____
○ NO, DO NOT report information regarding pharmacogenetic status. Mother's Initials _____

Father
○ YES, report information regarding pharmacogenetic status. Father's Initials _____
○ NO, DO NOT report information regarding pharmacogenetic status. Father's Initials _____

Adult-Onset Medically-Actionable Disorders

Mother
○ YES, report information regarding adult-onset actionable conditions. Mother's Initials _____
○ NO, DO NOT report information regarding adult-onset actionable conditions. Mother's Initials _____

Father
○ YES, report information regarding carrier status. Father's Initials _____
○ NO, DO NOT report information regarding carrier status. Father's Initials _____

Adult-Onset Not Medically-Actionable Disorders

Mother
○ YES, report information regarding adult-onset not currently actionable conditions. Mother's Initials _____
○ NO, DO NOT report information regarding adult-onset not currently actionable conditions. Mother's Initials _____

Father
○ YES, report information regarding carrier status. Father's Initials _____
○ NO, DO NOT report information regarding carrier status. Father's Initials _____

USE, RETENTION, AND DISCLOSURE OF SAMPLE AND INFORMATION

The sample will be sent to PerkinElmer's laboratories in Pennsylvania or Connecticut for testing. You have the right to confidential treatment of the sample and your information. Unless required by law, PerkinElmer will not disclose your identifiable information to any person or entity except as you authorize in this form or with your further written consent. Your information will be kept confidential and accessible only to PerkinElmer's lab technicians and support personnel, including contractors, necessary for performing the Testing, analysis and reporting results.

Internal Use of Anonymized Data. You hereby consent to PerkinElmer retaining the results of the Testing and associated reports in an anonymized form untraceable to you ("Anonymized Data"), and using Anonymized Data for internal statistical and quality analysis, research, scientific and technical development, and market research purposes. Use of Anonymized Data for these purposes may improve identification of and therapies for existing and new diseases now or in the future.

You understand you are not required to give this consent to Anonymized Data retention and use, and whether or not you give this consent to Anonymized Data retention and use has no bearing on the Testing.

Patient: YES ○ | **NO** ○ Patient/Guardian Initials_____
Mother: YES ○ | **NO** ○ Mother's Initials_____
Father: YES ○ | **NO** ○ Father's Initials_____

External Use of Anonymized Data. You hereby consent to PerkinElmer sharing Anonymized Data with other biomedical and research institutions for statistical and quality analysis, research, scientific and technical development, and market research purposes. Sharing Anonymized Data with other biomedical and research institutions for these purposes may improve identification of and therapies for existing and new diseases now or in the future.

You understand you are not required to give this consent to sharing Anonymized Data with third-parties, and whether or not you give this consent has no bearing on the Testing.

Patient: YES ○ | **NO** ○ Patient/Guardian Initials_____
Mother: YES ○ | **NO** ○ Mother's Initials_____
Father: YES ○ | **NO** ○ Father's Initials_____

Internal Use of Anonymized Sample. You hereby consent to PerkinElmer retaining your sample indefinitely in anonymized form for internal quality control, assay development and improvement and other purposes relating to PerkinElmer's proper management and administration. Use of the Anonymized Sample for these purposes may improve identification of and therapies for existing and new diseases now or in the future.

You understand you are not required to give this consent to retain your sample in anonymized form, and whether or not you give this consent has no bearing on the Testing.

Patient: YES ○ | **NO** ○ Patient/Guardian Initials_____
Mother: YES ○ | **NO** ○ Mother's Initials_____
Father: YES ○ | **NO** ○ Father's Initials_____

CONSENT TO ADDITIONAL TESTING RESULTS DISCLOSURE

The results of the Testing are confidential and may not be released to anyone without your informed consent except as permitted or required by law. You agree that all identifiable reports from the Testing will be provided only to your HCP(s) to assist them with diagnosis and to provide you with the results. In addition to the ordering physician indicated above, you request that a copy of the results be provided to the following HCPs (physicians/counselors/clinicians):

_____

_____

PerkinElmer
For the Better

SIGNATURE AND ACKNOWLEDGEMENT

By signing below, you acknowledge that the risks, benefits, and limitation of the Testing have been explained to you and you have had a chance to have your questions answered in a satisfactory manner. You agree that you have read and will receive a copy of this consent form. You understand that this consent is voluntary and testing will not be performed unless you provide consent to the Testing. If you have given consent to the use of Anonymized Data or Samples, that consent is valid until you withdraw your consent. Your consent may be withdrawn at any time as to future activity, but if you withdraw your consent, it will not affect actions taken before it was withdrawn.

I, (name) _____, voluntarily request PerkinElmer Genetics, Inc., and its affiliates, contractors and assigns ("PKIG") to perform the testing indicated on this form (Testing) on myself and/or my child, (child's name)_____, and consent to the use and disclosure of the results of the testing, as well as the sample and related patient information, in accordance with this form.

Patient or Parent/Guardian Printed Name:_____

Patient or Parent/Guardian Signature:_____Date:_____

Patient Address (Street, City, State, Zip):_____

Mother Printed Name:_____

Mother Signature, Date:_____

Father Printed Name:_____

Father Signature, Date:_____

PHYSICIAN/COUNSELOR/CLINICIAN STATEMENT

I have provided genetic counseling and have explained the Testing to the patient/parent/guardian. The consent form and limitations of genetic testing were reviewed with the patient/parent/guardian. I accept responsibility for pre- and post-test genetic counseling.

Note to Ordering Clinician: PerkinElmer Genetics, Inc. encourages the discussion of the limitations and utility of a genetic test with the patient prior to specimen collection. This form is provided to address pertinent issues regarding the Testing. Specific information describing indications, methodology and detection can be found on the PerkinElmer, Inc. website at: www.perkinelmergenetics.com

Clinician Printed Name:_____

Clinician Signature:_____

Please mail requisition form, informed consent, and test sample to PerkinElmer Genetics 250 Industry Drive, Suite 400, Pittsburgh, PA 15275.

PerkinElmer
For the Better

Informed Consent Form - Oregon

Individuals residing in Oregon must also complete this page.

Section 1: Checklist

TO BE COMPLETED BY THE INDIVIDUAL ORDERING A GENETIC TEST.

The individual's DNA sample will be tested solely for the genetic characteristic below:

_____

(Name of genetic characteristic)

PROCESS TO FOLLOW PRIOR TO OBTAINING GENETIC INFORMATION:

After each of the points below have been clearly explained to the individual to be tested, or the individual's personal representative, please initial in the space provided to ensure that the informed consent procedure has been followed.

_____I have informed the individual that this genetic test is completely voluntary; that he/she has the option of withdrawing consent to the genetic test at any time.

_____I have explained to the individual the risks and benefits of having a genetic test, including:

- a description of the provisions of Oregon law pertaining to the confidentiality of genetic information;
- a statement of the potential consequences regarding insurability, employability, and social discrimination if the genetic test results become known to others;
- a statement explaining the implications of positive and negative test results, and the availability of support services, including genetic counseling.

_____I have informed the individual that it may be in his/her best interest to retain the DNA sample for future diagnostic testing, but also of his/her right to have the DNA sample promptly destroyed after the specific purpose for which it was tested (unless retention of the sample is otherwise authorized by law).

_____I have informed the individual about the meaning and purpose of the authorization form for disclosure of procedure to a third party payer, including:

- an explanation of the potential risks of disclosure to third-party payers that a genetic test has been performed;
- an explanation of the individual's option to pay out-of-pocket for the cost of the genetic testing procedure.

_____I have asked the individual whether he/she has any further questions; and if so, I have provided the individual with an opportunity to ask questions and receive answers from either a genetic counselor, or a person who is sufficiently knowledgeable to give accurate and understandable answers about genetic testing and its implications.

_____I have asked the individual to read, complete, sign and date this consent form; and provided the individual a copy of this completed form for his/her personal records.

The above referenced information was explained by me, to the individual being tested, and the individual being tested signed this consent form in my presence.

Name of individual ordering genetic test:_____

Signature of individual ordering genetic test:_____ **Date:**_____

Section 2: Informed Consent Of Individual Consenting to Testing

TO BE COMPLETED BY THE INDIVIDUAL CONSENTING TO A GENETIC TEST.

It has been explained to me that the procedure to be undertaken is a test of my DNA sample to obtain genetic information solely for the purpose(s) listed below. It has also been explained that consent to this procedure is completely voluntary. I have been told that there are risks and potential consequences regarding employability, insurability and social discrimination that may result from the collection of my genetic information.

Please **check one**:

☐ I have been asked if I want a more detailed explanation of the risks and benefits of genetic testing. I am satisfied with the explanation provided to me and do not need any more information.

☐ I have requested and received further explanation for the proposed genetic test and more information about the potential risks and consequences for the test for me and my family. I am satisfied with the additional information provided to me and do not need any more information.

☐ I have requested further explanation of the proposed genetic test and more information about the potential risks and consequences for the test for me and my family, and do not consent to the collection of my genetic information at this time.

☐ I consent to the collection of my genetic information for the purpose of_____and acknowledge that the results of this test or procedure will be recorded in my confidential medical record.

Name of individual consenting:_____

Signature of individual consenting:_____ **Date:**_____

Section 3: Notice of Your Right to Decline Participation in Future Anonymous or Coded Genetic Research

TO BE COMPLETED BY THE INDIVIDUAL BEING TESTED.

_____**(NAME OF HEALTH CARE PROVIDER)**

The State of Oregon has laws to protect the genetic privacy of individuals. These laws give you the right to decline to have your health information or biological samples used for research. A biological sample may include a blood sample, urine sample, or other materials collected from your body. You can decide whether to allow your health information or biological samples to be available for genetic research. Your decision will not affect the care you receive from your health care provider or your health insurance coverage.

Research is important because it gives us valuable information on how to improve health, such as ways to prevent or improve treatment for heart disease, diabetes, and cancer. Under Oregon law, a special team reviews all genetic research before it begins. This team makes sure that the benefits of the research are greater than any risks to participants.

In anonymous research, personal information that could be used to identify you, like your name or medical record number, cannot be linked to your health information or biological sample. In coded research, personal information that could be used to identify you is kept separate from your health information or biological sample so it would be very difficult for someone to link your personal information to your health information or biological sample. Your identity is protected in both types of research.

If you want to allow your health information and biological sample to be available for anonymous or coded genetic research, you don't have to do anything. If you make this choice, your health information or biological sample may be used for anonymous or coded genetic research without further notice to you.

If you want to decline to have your health information and biological sample available for anonymous or coded genetic research, **you must tell your health care provider** by:

- Completing this form and giving it to your health care provider
- Completing this form and mailing it to your health care provider the address provided

Your decision is effective on the date your health care provider receives this form.

If you have any questions or concerns about this notice, please contact your health care provider.

No matter what you decide now, you can always change your mind later. If you change your mind, tell your health care provider your decision in writing by a means indicated by your health care provider. If you change your mind, the new decision will apply only to health information or biological samples collected after your health care provider receives written notice of your new decision.

☐ I decline to have my health information and biological samples available for anonymous or coded genetic research.

Printed Name:_____ **Date of Birth:**_____

Signature:_____ **Date:**_____

PerkinElmer
For the Better

Informed Consent Form - Nevada

Individuals residing in Nevada must also complete this page.

As used in this document, "genetic information" means any information that is obtained from a genetic test.

1. I understand that no insurer or corporation that provides health insurance, carrier serving small employers or health maintenance organization may:

(a) Require me or any member of my family to take a genetic test;

(b) Require me to disclose whether I or any member of my family has taken a genetic test;

(c) Request my genetic information or the genetic information of a member of my family; or

(d) Determine the rates or any other aspect of the coverage or benefits for health care for me or my family based on whether I or any member of my family has taken a genetic test or based on my genetic information or the genetic information of any member of my family.

2. I also understand that:

(a) I have the right to receive the results of a genetic test, in writing, within 10 working days after the person conducting the test has received the results. The written results must indicate that, except as otherwise provided in chapter 629 of NRS, my genetic information may not be obtained, retained or disclosed without first obtaining my in formed consent.

(b) It is unlawful for a person or entity to obtain my genetic information without my informed consent, unless the information is obtained;

(1) By a federal, state, county or city law enforcement agency to establish the identity of a person or a dead human body;

(2) To determine the parentage or identity of a person in certain circumstances;

(3) To determine the paternity of a person in certain circumstances;

(4) For use in a study where the identities of the persons from whom the genetic information is obtained are not disclosed to the person conducting the study;

(5) To determine the presence of certain inheritable disorders in an infant in certain circumstances; or

(6) Pursuant to an order of a court of competent jurisdiction.

(c) It is unlawful for a person to retain genetic information that identifies me without first obtaining my informed consent, unless retention of the genetic information is:

(1) Necessary to conduct a criminal investigation, an investigation concerning the death of a person or a criminal or juvenile proceeding;

(2) Authorized pursuant to an order of a court of competent jurisdiction; or

(3) Necessary for certain medical facilities to maintain my medical records.

(d) If I have authorized a person to retain my genetic information, I may request that the person destroy the genetic information. Such a person shall destroy the information, unless retention of the information is:

(1) Necessary to conduct a criminal investigation, an investigation concerning the death of a person or a criminal or juvenile proceeding;

(2) Authorized by an order of a court of competent jurisdiction;

(3) Necessary for certain medical facilities to maintain my medical records; or

(4) Authorized or required by state or federal law.

(e) Except as otherwise provided by federal law or regulation, a person who obtains my genetic information for use in a study shall destroy the information upon completion of the study or my withdrawal from the study, whichever occurs first, unless I authorize the person conducting the study to retain my genetic information after the study is completed or upon my withdrawal from the study.

(f) It is unlawful for a person to disclose or to compel another person to disclose my identity if I was the subject of a genetic test or to disclose to another person genetic information that allows the other person to identify me without first obtaining my informed consent, unless the information is disclosed:

(1) To conduct a criminal investigation, an investigation concerning the death of a person or a criminal or juvenile proceeding;

(2) To determine the parentage or identity of a person in certain circumstances;

(3) To determine the paternity of a person in certain circumstances;

(4) Pursuant to an order of a court of competent jurisdiction;

(5) By a physician after I am deceased and my genetic information will assist in the medical diagnosis of persons related to me by blood;

(6) To a federal, state, county or city law enforcement agency to establish the identity of a person or dead human body;

(7) To determine the presence of certain inheritable preventable disorders in an infant in certain circumstances; or

(8) By an agency of criminal justice in certain circumstances.

I,_____(name of person giving consent), hereby give my consent to_____

(name of health care provider person obtaining genetic information) to obtain my genetic information;

I,_____(name of person giving consent), hereby give my consent to PerkinElmer Genetics, Inc. to disclose my genetic information to_____ (name of health care provider ordering test).

This consent document is valid until_____(date of expiration).

If the person tested is unable to sign, please indicate the reason here: _____

_____

Signature of consenting person or his or her legal representative: _____ **Date:** _____

Witness: _____ **Date:** _____