The Lantern Project™ Requisition Form

Lighting the way to rare disease diagnosis

This test requisition form can be used to submit a specimen as part of The Lantern Project testing program.
This program is brought to you at no additional charge by Sanofi Genzyme. Please complete every field and tick box clearly.
The Lantern Project is for diagnostic testing only. The testing options below are not appropriate for carrier testing.

PATIENT SAMPLE INFORMATION

SAMPLE TYPE:

- ○ Saliva Swab
- ○ Whole Blood
- ○ Dried Blood Spots

Collection Date: MM/DD/YY

Was this sample collected in NY State: ○ Yes ○ No

Age of Onset: _____

INDICATIONS FOR TESTING (MORE THAN ONE SELECTION MAY APPLY)

- ○ Clinical Suspicion
- ○ Family History
- ○ Newborn Screening Confirmation (please include previous testing results)

INCLUSION OF MEDICAL RECORDS, CLINICAL SUMMARY, PICTURES AND FAMILY HISTORY IS RECOMMENDED. CLINICAL INFORMATION IS CRUCIAL FOR ACCURATE INTERPRETATION OF RESULTS.

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) _____			

For general questions on the collection and return of samples, please call:
PerkinElmer Genetics at +1(866)354-2910 or by emailing Genomics@perkinelmer.com

For samples that are collected not utilizing the PerkinElmer Genomics test packs, ship sample, test requisition form, and informed consent form by preferred shipping method to PerkinElmer Genomics at:
PerkinElmer Genetics, 250 Industry Dr. Suite 400, Pittsburgh, PA 15275

MOBILE PHLEBOTOMY SAMPLE COLLECTION REQUEST*:

* Only to be requested if patient cannot have sample collected at provider's office.

KIT TYPE REQUESTED:

- ○ DBS Pack
- ○ Whole Blood Pack

VISIT TYPE:

- ○ ExamOne Office
- ○ Home

Patient Name | Patient Primary Phone Number

Requested Date | Patient Secondary Phone Number

Special Instructions

! To request mobile phlebotomy services, submit completed requisition form to PerkinElmer Genomics via email at Genomics@perkinelmer.com

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

PROVIDER SPECIALTY

- ○ Biochemical Genetics
- ○ Cardiology
- ○ Gastroenterology
- ○ Genetics
- ○ Hematology
- ○ Nephrology
- ○ Neurology
- ○ Neuromuscular or Rehab Med
- ○ Ophthalmology
- ○ Orthopedics/Metabolic Bone
- ○ Primary Care
- ○ Pulmonology
- ○ Rheumatology
- ○ Other (Please list): _____

○ Please also check if Pediatrics in above Specialties

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

☒ INSTITUTIONAL BILLING

Sanofi Genzyme (Institution/Organization Name) | B0100 (Provider /Genetic Counselor's Account #)

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 _____

The Lantern Project is not intended to and should not interfere in any way with a healthcare professional's or patient's independent judgment and freedom of choice in the treatment options for these diseases. Healthcare professionals and patients should always consider the full range of treatment options and select those most appropriate for the individual patient.

FOR INTERNAL USE ONLY				
Date Rec'd _____ Rec'd _____				
TEMP	SPEC	COL	#TUBES	VOL
R/C/F				
R/C/F				
R/C/F				

PerkinElmer For the Better | PerkinElmer GENOMICS

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Please complete every field and tick box clearly.

the lantern project

TEST MENU (Review Specimen Requirements prior to Submitting Sample. If appropriate testing option is not listed, please call to discuss)

Acid Sphingomyelinase Deficiency (ASMD, Niemann-Pick Type A and B)
- ○ SAN012 Acid sphingomyelinase enzyme assay
- ○ SAN012 > SAN013 Acid sphingomyelinase enzyme assay with reflex to *SMPD1* sequencing
- ○ SAN013 *SMPD1* sequencing
- ○ SAN600 *SMPD1* known familial variant testing (fill out section below)

Gaucher Disease
- ○ SAN008, SAN012 > SAN009 + SAN004 or SAN013 Glucocerebrosidase enzyme assay (includes ASM enzyme assay in parallel) with reflex to GBA and Lyso-GL1 or SMPD1 sequencing as appropriate
- ○ SAN009 > SAN004 *GBA* sequencing with reflex to Lyso-*GL1*
- ○ SAN600 > SAN004 *GBA* known familial variant testing (fill out section below) with reflex to Lyso-*GL1*

Fabry Disease - Male Patient
- ○ SAN006 Alpha- galactosidase A enzyme assay
- ○ SAN006 > SAN007 + SAN005 Alpha-galactosidase A enzyme assay (males only) with reflex to *GLA* sequencing and Lyso-*GL3*
- ○ SAN007 > SAN005 *GLA* sequencing with reflex to Lyso-*GL3*
- ○ SAN600 > SAN005 *GLA* known familial variant testing (fill out section below) with reflex to Lyso-*GL3*

Fabry Disease - Female Patient
- ○ SAN007 > SAN005 *GLA* sequencing with reflex to Lyso-*GL3*
- ○ SAN600 > SAN005 *GLA* known familial variant testing (fill out section below) with reflex to Lyso-*GL3*

Mucopolysaccharidosis Type I (Hurler, Hurler/Sheie, Sheie Syndromes)
- ○ SAN010 Alpha- iduronidase enzyme assay
- ○ SAN010 > SAN011 Alpha-iduronidase enzyme assay with reflex to *IDUA* sequencing
- ○ SAN011 *IDUA* sequencing
- ○ SAN600 *IDUA* known familial variant testing (fill out section below)

Mucopolysaccharidosis – Unspecified
- ○ SAN001 > SAN011 MPS enzyme panel (MPS I, II, IIIB, IVA, IVB, VI, VII) (with *IDUA* sequencing reflex if MPS I enzyme deficient)

Pompe Disease
- ○ SAN014 Acid alpha- glucosidase enzyme assay
- ○ SAN014 > SAN015 Acid alpha-glucosidase enzyme assay with reflex to *GAA* sequencing
- ○ SAN015 GAA sequencing
- ○ SAN003 **STAT:** acid alpha-glucosidase enzyme with reflex to rapid GAA sequencing (for suspected infantile- onset disease and newborn screening confirmation only)
- ○ D0600 *GAA* known familial variant testing (fill out section below)

Focused Neuromuscular Disease Panel (Do NOT select this panel if you have also selected single gene GAA sequencing test)
- ○ SAN200 > SAN014 multigene panel (*GAA* positives will reflex to acid alpha-glucosidase enzyme assay (DBS or blood required for enzyme)

KNOWN FAMILIAL VARIANT TESTING* (*SMPD1, GBA, GLA, IDUA, GAA* ONLY)

Gene/Disease

Name of Family Member

Variant Name (c.)

Variant Name (c.)

Relationship of Family Member to Patient

Original Accession#

*Please provide copy of the family member's report, if available.

SPECIMEN REQUIREMENTS

Test	Testing	Acceptable Sample Type	DBS Card	EDTA	Heparin	Saliva
ASM (Niemann Pick A/B)	Enzyme	Dried Blood Spot / EDTA / Heparin*	Y	Y	Y*	N
ABG (Gaucher)	Enzyme	Dried Blood Spot / Heparin*	Y	N	Y	N
GAA (Pompe)	Enzyme	Dried Blood Spot / EDTA / Heparin*	Y	Y	Y*	N
GLA (Fabry)	Enzyme	Dried Blood Spot / EDTA / Heparin*	Y	Y	Y*	N
IDUA (MPS-I)	Enzyme	Dried Blood Spot / EDTA / Heparin*	Y	Y	Y*	N
ID2S (MPS-II)	Enzyme	Dried Blood Spot / EDTA / Heparin*	Y	Y	Y*	N
NAGLU (MPS-IIIB)	Enzyme	Dried Blood Spot / EDTA / Heparin*	Y	Y	Y*	N
GALNS (MPS-IVA)	Enzyme	Dried Blood Spot / EDTA / Heparin*	Y	Y	Y*	N
b-GAL (MPS-IVB)	Enzyme	Dried Blood Spot / EDTA / Heparin*	Y	Y	Y*	N
ARSB (MPS- VI)	Enzyme	Dried Blood Spot / EDTA / Heparin*	Y	Y	Y*	N
GUSB (MPS-VII)	Enzyme	Dried Blood Spot / EDTA / Heparin*	Y	Y	Y*	N
LysoGB-1	Biomarker	Dried Blood Spot / EDTA / Heparin*	Y	Y	Y*	N
LysoGB-3	Biomarker	Dried Blood Spot / EDTA	Y	Y	N	N
Single Gene/Multi-Gene	Sequencing	Dried Blood Spot / EDTA / Saliva+	Y	Y	N	Y

*Heparin is not preferred and will not allow for bundle testing if ordered. +Saliva will not allow for additional enzyme or biomarker testing if ordered.

SAMPLE TYPES	CODE	REQUIREMENTS
DRIED BLOOD SPOTS *PREFERRED SAMPLE TYPE*	**DBS**	***Collection and Processing Instructions:*** Follow kit instructions to fill all spots. Briefly, allow blood to saturate card until indicated areas are filled and blood has soaked through card. Air dry card at ambient temperature for at least 3 hours.
WHOLE BLOOD **EDTA (purple top)** **Heparin (green top)** *HEPARIN CANNOT BE USED FOR ANY DNA TESTING OR LYSO-GB3*	**WB**	***Collection and Processing Instructions:*** Infants (< 2-years): 2 to 3 mL; Children (>2-years): 3 to 5 mL; Older children and adults: 5 to 10 mL. ***Comments:*** Clotted or hemolyzed samples are not accepted.
SALIVA *CANNOT BE USED FOR ENZYME ASSAY OR BIOMARKER TESTING*	**SV**	***Collection and Processing Instructions:*** Collect saliva on an Oragene™ Saliva Collection Kit according to the manufacturer's instructions. Please contact PKIG to request the saliva collection kit for patients that cannot provide a blood sample.

For samples that are collected not utilizing the PerkinElmer Genomics test packs, ship sample, test requisition form, and informed consent form by preferred shipping method to PerkinElmer Genomics at:

PerkinElmer Genetics
250 Industry Dr. Suite 400
Pittsburgh, PA 15275

For general questions on the collection and return of sample results, please call: PerkinElmer Genetics at +1 (866) 354-2910 (Monday-Friday, 8:00AM - 5:00PM EST) or by emailing Genomics@perkinelmer.com

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

Clinical diagnosis: _____

Age of manifestation: _____ ICD-10 Codes: _____

INCLUSION OF MEDICAL RECORDS, CLINICAL SUMMARY, PICTURES AND FAMILY HISTORY IS RECOMMENDED. CLINICAL INFORMATION IS CRUCIAL FOR ACCURATE INTERPRETATION OF RESULTS.

A. NEUROLOGY

1. Brain Imaging

- ○ 1.1 Abnormal myelination
- ○ 1.2 Brain atrophy
- ○ 1.3 Cerebellar hypoplasia
- ○ 1.4 Hydrocephalus
- ○ 1.5 White matter lesions/hyperintensities
- ○ 1.6 Leukodystrophy
- ○ 1.7 Cerebrovascular abnormalities

2. Cognitive Dysfunction

- ○ 2.1 Delayed motor development
- ○ 2.2 Delayed language development
- ○ 2.3 Developmental regression
- ○ 2.4 Intellectual disability
- ○ 2.5 Autism
- ○ 2.6 ADHD
- ○ 2.7 Psychiatric Disorder
- ○ 2.8 Executive function issues
- ○ 2.9 Lewy Body dementia
- ○ 2.10 Learning disabilities

3. Movement Abnormality

- ○ 3.1 Ataxia
- ○ 3.2 Chorea
- ○ 3.3 Dystonia
- ○ 3.4 Parkinsonism

4. Neuromuscular

- ○ 4.1 Hypotonia
- ○ 4.2 Hypertonia
- ○ 4.3 Hyperreflexia
- ○ 4.4 Spasticity
- ○ 4.5 Exercise intolerance
- ○ 4.6 Muscle pain
- ○ 4.7 Muscle weakness - proximal
- ○ 4.8 Muscle weakness - distal

5. Others

- ○ 5.1 Encephalopathy
- ○ 5.2 Headache/migraine
- ○ 5.3 Macrocephaly
- ○ 5.4 Microcephaly
- ○ 5.5 Neuropathy
- ○ 5.6 Tia/stroke
- ○ 5.7 Abnormal EMG
- ○ 5.8 Abnormal NCV
- ○ 5.9 Abnormal muscle biopsy

B. METABOLISM/LABORATORY

- ○ 1 Elevated creatine kinase
- ○ 2 Elevated AST, ALT, GGT
- ○ 3 Elevated ferritin
- ○ 4 Elevated LDL
- ○ 5 Decreased HDL
- ○ 6 Elevated triglycerides
- ○ 7 Anemia
- ○ 8 Thrombocytopenia
- ○ 9 Elevated BUN
- ○ 10 Elevated creatinine
- ○ 11 Elevated urinary GAGs
- ○ Other _____

C. EYE

- ○ 1 Cataract
- ○ 2 Ophthalmoplegia
- ○ 3 Ptosis
- ○ 4 Strabismus
- ○ 5 Visual impairment
- ○ 6 Conjunctival vascular abn
- ○ 7 Corneal verticillata
- ○ 8 Retinal changes
- ○ 9 Retinal vessel abn
- ○ 10 Corneal clouding
- ○ 11 Retinal degeneration
- ○ 12 Amaurosis fugax
- ○ 13 Cherry red spot
- ○ Other _____

D. PULMONARY

- ○ 1 Reduced vital capacity
- ○ 2 Diaphragmatic weakness
- ○ 3 Sleep apnea
- ○ 4 Interstitial lung disease
- ○ 5 Reduced pulmonary function
- ○ Other _____

E. MOUTH,THROAT, EAR

- ○ 1 Conductive hearing loss
- ○ 2 Sensorineural hearing loss
- ○ 3 Enlarged tongue
- ○ 4 Tinnitus
- ○ 5 Recurrent otitis media
- ○ 6 Obstructive airway disease
- ○ 7 Chronic rhinitis
- ○ 8 Enlarged tonsils, adenoids
- ○ 9 Vertigo
- ○ Other _____

F. SKIN

- ○ 1 Angiokeratoma
- ○ Other _____

G. SKELETAL

- ○ 1 Short stature
- ○ 2 Joint contractures
- ○ 3 Scoliosis
- ○ 4 Kyphosis
- ○ 5 Dysostosis multiplex
- ○ 6 Osteopenia
- ○ 7 Osteonecrosis
- ○ 8 Bone marrow infiltration
- ○ 9 Erlenmeyer flask deformity
- ○ 10 Bone pain
- ○ 11 Joint stiffness
- ○ 12 Carpal tunnel syndrome
- ○ 13 Genu valgum
- ○ 14 Hip dysplasia
- ○ 15 Vertebral beaking
- ○ 16 Cervical stenosis
- ○ 17 Odontoid hypoplasia
- ○ 18 Phalangeal tapering
- ○ 19 Platyspondyly
- ○ 20 Epiphyseal flaring
- ○ Other _____

H. CARDIOVASCULAR

- ○ 1 Angioedema
- ○ 2 Aortic dilatation
- ○ 3 Arrythmia
- ○ 4 Coarctation of aorta
- ○ 5 Defect of atrial septum
- ○ 6 Defect of ventricular septum
- ○ 7 Dilated cardiomyopathy
- ○ 8 Hypertrophic cardiomyopathy
- ○ 9 Hypertension
- ○ 10 Hypotension
- ○ 11 Lymphedema
- ○ 12 Myocardial infarction
- ○ 13 Pulmonary hypertension
- ○ 14 Mitral valve regurg
- ○ 15 Atrial valve regurg
- ○ 16 Left ventricular hypertrophy
- ○ 17 Atrial fibrillation
- ○ 18 Exercise intolerance
- ○ Other _____

I. GASTROINTESTINAL

- ○ 1 Abdominal pain
- ○ 2 Diarrhea
- ○ 3 Constipation
- ○ 4 Nausea
- ○ 5 Vomiting
- ○ 6 Liver failure
- ○ 7 Hepatomegaly
- ○ 8 Splenomegaly
- ○ 9 Umbilical/inguinal hernia
- ○ Other _____

J. RENAL

- ○ 1 Renal cyst
- ○ 2 Renal tubular dysfunction
- ○ 3 Glomerulosclerosis
- ○ 4 Proteinuria
- ○ 5 Albuminuria
- ○ Other _____

K. HEMATOLOGY AND IMMUNOLOGY

- ○ 1 Anemia
- ○ 2 Pancytopenia
- ○ 3 Thrombocytopenia
- ○ 4 Hypercoagulation
- ○ 5 Hypocoagulation
- ○ 6 Splenomegaly
- ○ 7 Multiple myeloma
- ○ 8 MGUS
- ○ 9 Other malignancy
- ○ 10 Polyclonal gammopathy
- ○ Other _____

L. PRENATAL, DEVELOPMENT, MORPHOLOGY

- ○ 1 Dysmorphic features
- ○ 2 Hydrops fetalis
- ○ 3 IUGR
- ○ 4 Oligohydramnios
- ○ 5 Polyhydramnios
- ○ 6 Macrocephaly
- ○ 7 Coarse features
- ○ 8 Short stature
- ○ 9 Fine motor issues
- ○ 10 Gross motor issues
- ○ Other _____

INFORMED CONSENT FORM

PerkinElmer Genetics, Inc., ("PerkinElmer or PKIG") 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. There is no cost to you for the Test(s) in the Lantern Project and the test(s) are paid for you by Sanofi Genzyme. If you receive a diagnosis after using this testing program, you are in no way obligated to be treated with a medication Sanofi Genzyme manufactures.

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 analyzes one or more segments of your DNA depending on the assay requested. 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 majority of testing will be performed at our laboratory headquarters in Pittsburgh, PA.

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 result indicates the enzyme/biomarker results are below normal ranges. 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. The Focused Neuromuscular Disease Panel tests 99 genes, some of which lead to symptoms only in adulthood. Children undergoing this test may receive results that will not impact their health for many years.

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. Your/your child's symptoms can be an integral part of interpreting test results. Please ensure your HCP has filled out the Patient History section of the requisition.

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 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. 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. Genetic testing can also reveal unexpected differences in the genetic makeup of an individual (e.g., a male with two copies of an x-linked gene rather than the expected one).

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.

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. No identifying information will be disclosed to Sanofi Genzyme, the sponsor of this testing program.

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 for ongoing test development, scientific research, and/or other activities. 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 to use of your anonymized sample, no further anonymization will be performed.

  ❑ Check here if you would like to opt out of anonymized sample retention (NY State residents, please see section below). 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 statistical and quality analysis, research, scientific and technical development, and market research. PerkinElmer may also share your anonymized data and anonymized report with third parties.

  ❑ Check here if you would like to opt out of anonymized data retention. Note, if not checked, this is interpreted as "consent given"

REQUIRED FOR SAMPLES COLLECTED IN NEW YORK STATE ONLY

No tests other than those authorized shall be performed on the biological sample submitted for testing, and any material derived from the sample (i.e., DNA); this includes testing for internal research and/or quality control purposes. The sample shall be destroyed no more than 60 days after the sample was taken or at the end of the testing process, whichever occurs later, unless indicated below.

❑ By checking here and signing at right, I consent to PerkinElmer keeping my sample for longer than 60 days, and to using my de-identified sample for internal research and/or quality control purposes. Note, if not checked and signed, this is interpreted as "consent not given."

_____
Patient/Guardian Signature

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. No identifying information will be disclosed to Sanofi Genzyme, the sponsor of this testing program.

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