PerkinElmer For the Better | PerkinElmer GENOMICS

The Lantern Project™ Requisition Form

Lighting the way to rare disease diagnosis

Please complete every field and tick box clearly.

the lantern project

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.

PATIENT INFORMATION

Patient's First Name | Middle Initial

Patient's Last Name | Patient's Date of Birth (MM/ DD /YYYY)

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:
- ○ Dried Blood Spots
- ○ Whole Blood
- ○ Saliva Sample

Collection Date: MM/DD/YY

Age of Onset:_____

ICD10 CODES: _____

_____

INDICATIONS FOR TESTING (MORE THAN ONE SELECTION MAY APPLY)

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

TEST MENU

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

Gaucher Disease
- ○ Glucocerebrosidase enzyme assay with reflex to *GBA* sequencing (If normal, enzyme assay will reflex to acid sphingomyelinase enzyme assay)
- ○ *GBA* sequencing
- ○ *GBA* known familial variant testing (fill out section below)

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

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

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

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

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

Limb-Girdle Muscle Weakness Panel
- ○ 105 gene panel (*GAA* positives will reflex to acid alpha-glucosidase enzyme assay (DBS or blood required for enzyme)

KNOWN FAMILIAL VARIANT TESTING*

Gene/Disease | Variant Name (c.) | Variant Name (c.)

Name of Family Member

Relationship of Family Member to Patient | Original Accession#

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

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_____

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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 _____

GENERAL INFORMED CONSENT FORM

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

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

TEST INFORMATION

The type of Test you are consenting to is a genetic test that looks at the genes in your DNA. This Test is used to identify what, if any, DNA mutation(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

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

TEST RESULTS

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

TEST LIMITATIONS

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

TEST RISKS

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

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

Federal laws prohibit health insurers/employers from discriminating based on your genetic information. There are currently no federal laws that prohibit life insurance, long-term care, or disability insurance companies from discriminating based on genetic information. Unless required by Law, PerkinElmer will not disclose your identifiable information to any person except as you have authorized in this consent.

ANONYMIZATION

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

RETENTION

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

RESEARCH OPTIONS

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

WHY ARE PARENTAL SAMPLES NEEDED?

In some circumstances, it may be helpful for additional family members to undergo testing as well in order to provide information that can aid in the interpretation of the 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.

TEST REPORT

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

PATIENT'S CONSENT TO TESTING

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

- I understand that participation in this genetic testing is voluntary.
- I understand that PHI is protected by law and will not be used or linked to the results of any study or publication.
- I understand that my personal information is protected by law and will not be used or linked to the results of any study or publication.
- I understand that if my HCP practices in New York State, I agree that PerkinElmer may retain my anonymized samples indefinitely.
- I agree to give up property rights to my sample and am voluntarily donating my and/or my child's sample and data to PerkinElmer or a qualified third party for statistical, quality control, research, scientific, technical and marketing research purposes.
- I give PerkinElmer permission to conduct further analyses of my or my child's sample and data.
- I give PerkinElmer permission to use my sample anonymously in studies a to improve testing and for publication.
- I give PerkinElmer permission to inform my HCP, or myself, of any research opportunities that may be associated with my and/or my child's Test results and any secondary findings.
- I understand that if I wish to withdraw from the Test, or if I have any questions about the Test, that I may contact PerkinElmer via email at: Genomics@perkinelmer.com or by toll-free by telephone +1-866-354-2910 to request withdrawal.
- I understand that I (or my legal representative) are entitled to a copy of this Consent.

PATIENT/ Representative Signature: _____

Print Patient Name:_____Date_____ Time _____