



General Test Requisition Form - Biochemical and Molecular

Please complete every field and tick box clearly.

PATIENT INFORMATION

<input type="text"/>	<input type="text"/>	<input type="text" value="MM/DD/YYYY"/>
Patient's First Name	Middle Initial	Patient's Date of Birth

<input type="text"/>	<input type="text"/>
Patient's Last Name	Patient ID/MR Number

Biological Sex: Male Female Unknown
 Gender Identity (if different from above):

Patient's Street Address

<input type="text"/>	<input type="text"/>	<input type="text"/>
City / Town	State	Zip Code

<input type="text"/>	<input type="text"/>
Country	Patient's Preferred Phone

Patient's Email

Ethnicity (check all that apply):

<input type="radio"/> African-American	<input type="radio"/> Asian (China, Japan, Korea)
<input type="radio"/> Caucasian/N. European/S. European	<input type="radio"/> Finnish
<input type="radio"/> French Canadian	<input type="radio"/> Hispanic
<input type="radio"/> Jewish - Ashkenazi	<input type="radio"/> Jewish - Sephardic
<input type="radio"/> Mediterranean	<input type="radio"/> Middle Eastern (Saudi Arabia, Qatar, Iraq, Turkey)
<input type="radio"/> Native American	<input type="radio"/> E. Indian
<input type="radio"/> Southeast Asian (Vietnam, Cambodia, Thailand)	<input type="radio"/> South Asian (India, Pakistan)
<input type="radio"/> Other (specify) <input type="text"/>	

ORDERING PROVIDER

Provider's First and Last Name

<input type="text"/>	<input type="text"/>
PKIG Ordering Provider Account Number	NPI

Clinic/Hospital/Institution Name

<input type="text"/>	<input type="text"/>
Provider's Email	Provider's Phone

Provider's Street Address

<input type="text"/>	<input type="text"/>	<input type="text"/>
City / Town	State	Zip Code

<input type="text"/>	<input type="text"/>
Country	Provider's Fax

SEND ADDITIONAL COPY OF RESULTS TO (If applicable)

Name

<input type="text"/>	<input type="text"/>
PKIG Ordering Provider Account Number	Phone Number

<input type="text"/>	<input type="text"/>
Email Address	Fax Number

PHYSICIAN CONFIRMATION OF INFORMED CONSENT AND MEDICAL NECESSITY

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. I understand and agree that, if the patient's insurance requires genetic counseling prior to performance of the ordered test, PerkinElmer will provide the patient information to a third party service so the patient can obtain genetic counseling. I understand and agree that a genetic counselor will be permitted to review the test(s) I have ordered and make changes based on clinical or payor related specifications, and that the genetic counselor will submit to the payor the required documentation in support of the test as ordered or with any recommended changes. I attest that all information on this TRF is true to the best of my knowledge. My signature applies to the entirety of the statement above and/or attached letter of medical necessity.

Signature _____ Date _____

PATIENT SAMPLE INFORMATION

SAMPLE TYPE: Saliva Swab Whole Blood Dried Blood Spots Urine Other _____

Collection Date:

Was this sample collected in NY State: Yes No

INDICATION FOR TESTING (Required)

ICD10 Code(s): _____
 Clinical Diagnosis: _____
 Age at Initial Presentation: _____

BIOCHEMICAL TESTS

SCREENING PANELS

- B0200 StepOne® Comprehensive Biochemical Profile
- B0210 Acylcarnitine Profile
- B2020 Amino Acid Profile
- B2040 Lysosomal Storage Disease Enzyme Panel
- B0024 Post-Mortem Screening Panel

DIAGNOSTIC AND MONITORING PANELS

- B0009 Galactosemia Monitoring
- B0018 PKU Clinical Monitoring
- B0022 Tyrosinemia Monitoring

COMPREHENSIVE GENETIC TESTING PANELS

- D3005 NeoSeq Newborn and Pediatric Gene Panel (1722 Genes)
- D3004 Expanded Newborns Screening (NBS) Gene Sequencing Panel (275 Genes)
- D3003 Newborn Screening and Lysosomal Storage Disease Panel (94 genes)

ADDITIONAL TESTING*

Test Code: _____
 Test Name: _____

*Additional testing options including DNA Mutation Screens and Gene Sequencing for individual conditions (or sets of conditions) can be found on pages 4 - 7.

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



General Test Requisition Form - Biochemical and Molecular

INSURANCE BILLING (Include a copy of both sides of insurance card)

<input type="text"/>	<input type="text"/>
Insurance Carrier	Insurance ID
<input type="text"/>	<input type="text" value="MM/DD/YYYY"/>
Policy Holder Name	Policy Holder DOB
Policy Holder Relationship to Patient: <input type="radio"/> Self <input type="radio"/> Parent <input type="radio"/> Spouse <input type="radio"/> Other: _____	

Benefit Investigation and Out-of-Pocket Cost Policy

PerkinElmer will contact the patient for any estimated out-of-pocket costs that are greater than \$100 USD before proceeding with testing. The patient's sample will be placed on hold (for up to 30 days) until authorization to proceed is received from the patient. If the patient does not respond to PerkinElmer within 30 days to discuss estimated out-of-pocket costs, the test order may be cancelled. Please note that failure by the patient to respond to PerkinElmer in a timely fashion regarding estimated out-of-pocket costs may cause a delay in the receipt of the results report.

Patient Billing Acknowledgement:

By signing this form, I certify that the insurance information that I have provided is accurate, complete and current and that no other coverage or insurance exists. I hereby authorize PerkinElmer Genetics, Inc. ("PerkinElmer") to bill my designated insurance carrier(s) and share health information as needed for the purposes of billing and reimbursement, and I request that payment of authorized benefits be made on my behalf to PerkinElmer for any services furnished the patient listed above by PerkinElmer. If any insurance benefits are remitted to me for services performed by PerkinElmer for the patient, I will forward said benefits to PerkinElmer. I authorize PerkinElmer to file an appeal on my behalf for any denial of payment and/or adverse benefit determination related to services and care provided. I agree to pay all charges for services provided by PerkinElmer to the patient which are not covered by my health insurance plan or which I am responsible for payment under my health insurance plan. Furthermore, I grant PerkinElmer permission to share health information with my insurance as needed for the purposes of billing and reimbursement.

Signature _____ Date _____

INSTITUTIONAL BILLING

<input type="text"/>	<input type="text"/>
Institution/Organization Name	PerkinElmer Genomics Billing Account ID
<input type="text"/>	<input type="text"/>
Contact Name	Contact Phone

PATIENT BILLING

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

Credit Card (Please fill out all information):

<input type="text"/>	<input type="text"/>
Credit Card Number	CVV
<input type="text"/>	<input type="text" value="MM/YY"/>
Credit Card Billing Street Address	Card Exp. Date Cardholder Phone
<input type="text"/>	<input type="text"/>
City / Town	State Zip Code
<input type="text"/>	Cardholder Printed Name as Appears on Card
<input type="text"/>	
Cardholder Signature	

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

ADDITIONAL OPTIONAL PHENOTYPE / PATIENT HISTORY SECTION (Check all that apply)

Face2Gene ID (if applicable): _____

Clinical diagnosis: _____

Age of manifestation: _____ ICD-10 Codes: _____

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

OTHER: _____

Associated Condition(s)	Test Type	Test Name	Test Code	Sample Type*
AMINO ACID, ORGANIC ACID, FATTY ACID OXIDATION DISORDERS				
Multiple	Biochemical Assay	Acylcarnitine Profile	B0210	DBS, WB, gDNA
Multiple	Biochemical Assay	Amino Acid Profile	B2020	DBS, WB, gDNA
2,4 Dienoyl-CoA Reductase Deficiency (DE RED)	Full Gene Analysis	<i>NADK2</i> Gene Sequencing	D5049	DBS, WB, SV, gDNA
2-methylbutyryl Glycinuria	Full Gene Analysis	<i>ACADSB</i> Gene Sequencing	D5004	DBS, WB, SV, gDNA
3-methylcrotonyl-CoA Carboxylase Deficiency (3-MCC Deficiency)	Targeted Variant Testing	3-MCC Deficiency Mutation Panel	D0410	DBS
3-methylcrotonyl-CoA Carboxylase Deficiency (3-MCC Deficiency)	Full Gene Analysis	<i>MCCC1</i> and <i>MCCC2</i> Gene Sequencing	D3007	DBS, WB, SV, gDNA
3-methylglutaconic Aciduria, Type I	Full Gene Analysis	<i>AUH</i> Gene Sequencing	D5012	DBS, WB, SV, gDNA
Argininemia	Full Gene Analysis	<i>ARG1</i> Gene Sequencing	D5008	DBS, WB, SV, gDNA
Argininosuccinic Aciduria	Full Gene Analysis	<i>ASL</i> Gene Sequencing	D5010	DBS, WB, SV, gDNA
Beta-ketothiolase Deficiency	Full Gene Analysis	<i>ACAT1</i> Gene Sequencing	D5123	DBS, WB, SV, gDNA
Biopterin Defect in Cofactor Biosynthesis (BIOPT-BS)	Full Gene Analysis	<i>GCH1</i> and <i>PTS</i> Gene Sequencing	D3009	DBS, WB, SV, gDNA
Biopterin Defect in Cofactor Regeneration (BIOPT-REG)	Full Gene Analysis	<i>PCBD1</i> and <i>QDPR</i> Gene Sequencing	D3008	DBS, WB, SV, gDNA
Carnitine Palmitoyltransferase I Deficiency	Full Gene Analysis	<i>CPT1A</i> Gene Sequencing	D5017	DBS, WB, SV, gDNA
Carnitine Palmitoyltransferase II Deficiency	Full Gene Analysis	<i>CPT2</i> Gene Sequencing	D5018	DBS, WB, SV, gDNA
Carnitine Uptake Defect (CUD)	Full Gene Analysis	<i>SLC22A5</i> Gene Sequencing	D5054	DBS, WB, SV, gDNA
Carnitine-acylcarnitine Translocase (CACT) Deficiency	Full Gene Analysis	<i>SLC25A20</i> Gene Sequencing	D5056	DBS, WB, SV, gDNA
Citrullinemia Type I	Full Gene Analysis	<i>ASS1</i> Gene Sequencing	D5011	DBS, WB, SV, gDNA
Citrullinemia Type II	Full Gene Analysis	<i>SLC25A13</i> Gene Sequencing	D5055	DBS, WB, SV, gDNA
Cobalamin A/B Deficiency	Full Gene Analysis	<i>MMAA</i> and <i>MMAB</i> Gene Sequencing	D3010	DBS, WB, SV, gDNA
Cobalamin C Deficiency	Full Gene Analysis	<i>MMACHC</i> Gene Sequencing	D5046	DBS, WB, SV, gDNA
Cobalamin D Deficiency	Full Gene Analysis	<i>MMADHC</i> Gene Sequencing	D5047	DBS, WB, SV, gDNA
Glutaric Acidemia Type I	Targeted Variant Testing	Glutaric Acidemia Type I Mutation Panel	D0406	DBS
Glutaric Acidemia Type II	Full Gene Analysis	<i>ETFA</i> , <i>ETFB</i> , and <i>ETFDH</i> Gene Sequencing	D3011	DBS, WB, SV, gDNA
Glutaricaciduria, Type I	Full Gene Analysis	<i>GCDH</i> Gene Sequencing	D5030	DBS, WB, SV, gDNA
HMG-CoA Lyase Deficiency	Full Gene Analysis	<i>HMGCL</i> Gene Sequencing	D5039	DBS, WB, SV, gDNA
Homocystinuria	Full Gene Analysis	<i>CBS</i> Gene Sequencing	D5015	DBS, WB, SV, gDNA
Hypermethioninemia	Full Gene Analysis	<i>ADK</i> Gene Sequencing	D5007	DBS, WB, SV, gDNA
Isobutyryl-CoA Dehydrogenase Deficiency	Full Gene Analysis	<i>ACAD8</i> Gene Sequencing	D5001	DBS, WB, SV, gDNA
Isovaleric Acidemia	Targeted Variant Testing	Isovaleric Acidemia Mutation Panel	D0409	DBS
Isovaleric Acidemia	Full Gene Analysis	<i>IVD</i> Gene Sequencing	D5043	DBS, WB, SV, gDNA
Long-chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency (LCHADD)	Targeted Variant Testing	<i>LCHADD</i> Mutation Panel	D0407	DBS
Long-chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency (LCHADD)	Full Gene Analysis	<i>HADHA</i> and <i>HADHB</i> Gene Sequencing	D3012	DBS, WB, SV, gDNA
Maple Syrup Urine Disease	Targeted Variant Testing	Maple Syrup Urine Disease Mutation Panel	D0401	DBS
Maple Syrup Urine Disease	Full Gene Analysis	<i>BCKDHA</i> , <i>BCKDHB</i> , and <i>DBT</i> Gene Sequencing	D3013	DBS, WB, SV, gDNA
Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCADD)	Targeted Variant Testing	<i>MCADD</i> Mutation Panel	D0400	DBS
Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCADD)	Full Gene Analysis	<i>ACADM</i> Gene Sequencing	D5002	DBS, WB, SV, gDNA
Methylmalonic Acidemia	Targeted Variant Testing	Methylmalonic Acidemia Mutation Panel	D0411	DBS
Methylmalonic Acidemia	Full Gene Analysis	<i>MUT</i> Gene Sequencing	D5048	DBS, WB, SV, gDNA

Associated Condition(s)	Test Type	Test Name	Test Code	Sample Type*
Multiple Carboxylase Deficiency	Full Gene Analysis	<i>HLCS</i> Gene Sequencing	D5038	DBS, WB, SV, gDNA
Multiple Sulfatase Deficiency	Full Gene Analysis	<i>SUMF1</i> Gene Sequencing	D5058	DBS, WB, SV, gDNA
Phenylketonuria (PKU)	Biochemical Assay	PKU Monitoring - Phenylalanine	B0018	DBS, WB
Phenylketonuria (PKU)	Full Gene Analysis	<i>PAH</i> Gene Sequencing	D5050	DBS, WB, SV, gDNA
Propionic Acidemia	Targeted Variant Testing	Propionic Acidemia Mutation Panel	D0412	DBS
Propionic Acidemia	Full Gene Analysis	<i>PCCA</i> and <i>PCCB</i> Gene Sequencing	D3014	DBS, WB, SV, gDNA
Short Chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency (M/SCHADD)	Full Gene Analysis	<i>HADH</i> Gene Sequencing	D5036	DBS, WB, SV, gDNA
Short-chain Acyl-CoA Dehydrogenase Deficiency (SCADD)	Full Gene Analysis	<i>ACADS</i> Gene Sequencing	D5003	DBS, WB, SV, gDNA
Tyrosinemia	Biochemical Assay	Tyrosinemia Monitoring - Succinylacetone and Tyrosine	B0022	DBS, WB
Tyrosinemia Type I	Full Gene Analysis	<i>FAH</i> Gene Sequencing	D5023	DBS, WB, SV, gDNA
Tyrosinemia Type I	Biochemical	Succinylacetone (SUAC)	B0021	DBS, WB, gDNA
Tyrosinemia Type II	Full Gene Analysis	<i>TAT</i> Gene Sequencing	D5059	DBS, WB, SV, gDNA
Tyrosinemia Type III	Full Gene Analysis	<i>HPD</i> Gene Sequencing	D5040	DBS, WB, SV, gDNA
Very Long-chain Acyl-CoA Dehydrogenase Deficiency (VLCADD)	Full Gene Analysis	<i>ACADVL</i> Gene Sequencing	D5005	DBS, WB, SV, gDNA
BIOTINIDASE DEFICIENCY				
Biotinidase Deficiency	Biochemical Assay	Biotinidase Deficiency (Complete/Partial) - Biotinidase Deficiency Enzyme Analysis	B0001	DBS
Biotinidase Deficiency	Targeted Variant Testing	Biotinidase Deficiency Mutation Panel	D0402	DBS
Biotinidase Deficiency	Full Gene Analysis	<i>BTD</i> Gene Sequencing	D5014	DBS, WB, SV, gDNA
CYSTIC FIBROSIS				
Cystic Fibrosis	Biochemical Assay	IRT Analysis (Not valid after 90 days of age)	B0005	DBS
Cystic Fibrosis	Targeted Variant Testing	Cystic Fibrosis Mutation Panel	D0403	DBS
Cystic Fibrosis	Full Gene Analysis	<i>CFTR</i> Gene Sequencing	D5016	DBS, WB, SV, gDNA
DUCHENNE MUSCULAR DYSTROPHY				
Duchenne Muscular Dystrophy (DMD)	Biochemical Assay	Duchenne Muscular Dystrophy Creatine Kinase Activity	B0006	DBS
Duchenne Muscular Dystrophy (DMD)	Full Gene Analysis	<i>DMD</i> Gene Sequencing and Del/Dup Testing	D4045	DBS, WB, SV, gDNA
Duchenne Muscular Dystrophy (DMD)	Deletion/Duplication Analysis	<i>DMD</i> Del/Dup Testing	D5125	DBS, WB, SV, gDNA
FRIEDREICH'S ATAXIA				
Friedreich's Ataxia	Tandem Repeat Analysis	<i>FXN</i> Repeat Analysis	D5133	DBS, WB, gDNA
GALACTOSEMIA				
Galactosemia	Biochemical Assay	Galactosemia Monitoring - Galactose-1-phosphate uridylyltransferase Enzyme Analysis and Total Galactose	B0009	DBS
Galactosemia	Targeted Variant Testing	Galactosemia Mutation Panel	D0405	DBS
Galactosemia	Full Gene Analysis	<i>GALT</i> Gene Sequencing	D5029	DBS, WB, SV, gDNA
Galactosemia	Full Gene Analysis	<i>GALE</i> Gene Sequencing	D5026	DBS, WB, SV, gDNA
Galactokinase Deficiency	Full Gene Analysis	<i>GALK</i> Gene Sequencing	D5027	DBS, WB, SV, gDNA
GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY				
Glucose-6-phosphate Dehydrogenase Deficiency	Biochemical Assay	Glucose-6-phosphate Dehydrogenase Deficiency (screening only)	B0011	DBS
Glucose-6-phosphate Dehydrogenase Deficiency	Targeted Variant Testing	Glucose-6-phosphate Dehydrogenase Deficiency Mutation Panel	D0404	DBS
Glucose-6-phosphate Dehydrogenase Deficiency	Full Gene Analysis	<i>G6PD</i> Gene Sequencing	D5024	DBS, WB, SV, gDNA

Associated Condition(s)	Test Type	Test Name	Test Code	Sample Type*
LYSOSOMAL STORAGE DISORDERS - TESTING OPTIONS				
Lysosomal Storage Disorders	Biochemical Assay	Lysosomal Storage Disease Enzyme Panel	B2040	DBS, WB
Lysosomal Storage Disorders	Full Gene Analysis	Lysosomal Storage Disorder Gene Sequencing Panel (12 Genes)	D3001	DBS, WB, SV, gDNA
Fabry Disease	Biochemical Assay	Alpha-Galactosidase A Enzyme Analysis	B0007	DBS, WB
Fabry Disease	Biochemical Assay	Globotriaosylsphingosine (lyso-Gb3) Monitoring	B0029	DBS, WB
Fabry Disease	Full Gene Analysis	GLA Gene Sequencing	D5033	DBS, WB, SV, gDNA
Gaucher Disease	Biochemical Assay	Glucocerebrosidase (Glucosylceramidase) Enzyme Analysis	B0010	DBS, WB
Gaucher Disease	Biochemical Assay	Glucosylsphingosine (lyso-Gb1) Monitoring	B0030	DBS, WB
Gaucher Disease	Full Gene Analysis	GBA Gene Sequencing	D5032	DBS, WB, SV, gDNA
Krabbe Disease	Biochemical Assay	Galactocerebrosidase Enzyme Analysis	B0012	DBS, WB
Krabbe Disease	Biochemical Assay	Psychosine Biochemical Assay	B0028	DBS, WB
Krabbe Disease	Full Gene Analysis	GALC Gene Sequencing	D5031	DBS, WB, SV, gDNA
MPS I (Hurler Syndrome)	Biochemical Assay	Alpha-L-Iduronidase Enzyme Analysis	B0013	DBS, WB
MPS I (Hurler Syndrome)	Full Gene Analysis	IDUA Gene Sequencing	D5041	DBS, WB, SV, gDNA
MPS II (Hunter Syndrome)	Biochemical Assay	Iduronate 2-Sulfatase Enzyme Analysis	B0014	DBS, WB
MPS II (Hunter Syndrome)	Full Gene Analysis	IDS Gene Sequencing	D5042	DBS, WB, SV, gDNA
MPS IVA (Morquio A Syndrome)	Biochemical Assay	Galactosamine-6-Sulfatase Enzyme Analysis	B0015	DBS, WB
MPS IVA (Morquio A Syndrome)	Full Gene Analysis	GALNS Gene Sequencing	D5028	DBS, WB, SV, gDNA
MPS IVB (GM1 Gangliosidosis)	Biochemical Assay	β-galactosidase Enzyme Analysis	B0025	DBS, WB
MPS IVB (GM1 Gangliosidosis)	Full Gene Analysis	GLB1 Gene Sequencing	D5034	DBS, WB, SV, gDNA
MPS VI (Maroteaux-Lamy Syndrome)	Biochemical Assay	Arylsulfatase B Enzyme Analysis	B0016	DBS, WB
MPS VI (Maroteaux-Lamy Syndrome)	Full Gene Analysis	ARSB Gene Sequencing	D5009	DBS, WB, SV, gDNA
MPS VII (Sly Syndrome)	Biochemical Assay	β-glucuronidase Enzyme Analysis	B0026	DBS, WB
Mucopolysaccharidosis VII	Full Gene Analysis	GUSB Gene Sequencing	D5035	DBS, WB, SV, gDNA
Multiple Sulfatase Deficiency	Full Gene Analysis	SUMF1 Gene Sequencing	D5058	DBS, WB, SV, gDNA
Niemann Pick Disease Types A and B	Biochemical Assay	ACID Sphingomyelinase Enzyme Analysis	B0017	DBS, WB
Niemann Pick Disease Types A and B	Full Gene Analysis	SMPD1 Gene Sequencing	D5057	DBS, WB, SV, gDNA
Pompe Disease	Biochemical Assay	ACID Alpha-Glucosidase Enzyme Analysis	B0019	DBS, WB
Pompe Disease	Full Gene Analysis	GAA Gene Sequencing	D5025	DBS, WB, SV, gDNA
Neuronal Ceroid Lipofuscinosis 2 (CLN2)	Biochemical Assay	Tripeptidyl peptidase 1 Enzyme Analysis	B0027	DBS, WB
Neuronal Ceroid Lipofuscinosis 2 (CLN2)	Full Gene Analysis	TPP1 Gene Sequencing	D5090	DBS, WB, SV, gDNA
SEVERE COMBINED IMMUNODEFICIENCY				
Severe Combined Immunodeficiency (SCID)	Molecular DNA Screen	TREC Assay	D0416	DBS
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	SCID Gene Sequencing Panel (26 Genes)	D3006	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	ADA Gene Sequencing	D5060	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	AK2 Gene Sequencing	D5061	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	ATM Gene Sequencing	D5062	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	CD3D Gene Sequencing	D5063	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	CD3E Gene Sequencing	D5064	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	CD3Z Gene Sequencing	D5065	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	CORO1A Gene Sequencing	D5066	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	DCLRE1C (Artemis) Gene Sequencing	D5067	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	DOCK8 Gene Sequencing	D5068	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	FOXN1 Gene Sequencing	D5069	DBS, WB, SV, gDNA

Associated Condition(s)	Test Type	Test Name	Test Code	Sample Type*
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	<i>IL2RG</i> SGene Sequencing	D5070	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	<i>IL7R</i> Gene Sequencing	D5071	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	<i>JAK3</i> Gene Sequencing	D5072	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	<i>LIG4</i> Gene Sequencing	D5073	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	<i>NHEJ1</i> Gene Sequencing	D5074	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	<i>ORAI1</i> Gene Sequencing	D5075	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	<i>PNP</i> Gene Sequencing	D5076	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	<i>PRKDC</i> Gene Sequencing	D5077	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	<i>PTPRC</i> Gene Sequencing	D5078	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	<i>RAC2</i> Gene Sequencing	D5079	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	<i>RAG1</i> Gene Sequencing	D5080	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	<i>RAG2</i> Gene Sequencing	D5081	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	<i>RMRP</i> Gene Sequencing	D5082	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	<i>STIM1</i> Gene Sequencing	D5083	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	<i>TBX1</i> Gene Sequencing	D5084	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	<i>ZAP70</i> Gene Sequencing	D5085	DBS, WB, SV, gDNA
SICKLE CELL AND OTHER HEMOGLOBINOPATHIES				
Sickle Cell and Other Hemoglobinopathies	Biochemical Assay	Isoelectric Focusing GEL Electrophoresis of Hemoglobiins	B0020	DBS
Sickle Cell and Other Hemoglobinopathies	Targeted Variant Testing	Sickle Cell and Other Hemoglobinopathies Mutation Panel	D0408	DBS
SPINAL MUSCULAR ATROPHY (SMA)				
Spinal Muscular Atrophy (SMA)	Deletion/Duplication Analysis	SMA Diagnostic Test	D5134	DBS, WB, gDNA
Spinal Muscular Atrophy (SMA)	Deletion/Duplication Analysis	SMA Carrier Screen	D5135	DBS, WB, gDNA
Spinal Muscular Atrophy (SMA)	Deletion/Duplication Analysis	<i>SMN2</i> Copy Number Test	D5136	DBS, WB, SV, gDNA
OTHER				
Congenital Adrenal Hyperplasia (CAH)	Biochemical Assay	Congenital adrenal hyperplasia - 17A Hydroxyprogesterone (17 <i>OHP</i>)	B0002	DBS
Congenital Adrenal Hyperplasia (CAH)	Full Gene Analysis	<i>CYP21A2</i> Gene Sequencing and Del/Dup Testing (by MLPA)	D5019	DBS, WB, SV, gDNA
Congenital Hypothyroidism	Biochemical Assay	Thyroid-Stimulating Hormone (TSH)	B0003	DBS
Congenital Hypothyroidism	Biochemical Assay	Thyroxine (T4)	B0004	DBS
Fragile X	Triplet Repeat Testing	<i>FMR1</i> Triplet Repeat (CGG) Testing	D4042	DBS, WB, SV, gDNA
X-linked Adrenoleukodystrophy	Biochemical Assay	X-Linked Adrenoleukodystrophy - C26:0 Lysophosphatidylcholine	B0023	DBS, WB
X-linked Adrenoleukodystrophy	Full Gene Analysis	<i>ABCD1</i> Gene Sequencing	D5000	DBS, WB, SV, gDNA
Multiple	Biochemical Assay	Post Mortem - Includes: 17-Hydroxyprogesterone, Acylcarnitines, Galactose, and TSH	B0024	DBS

DBS = Dried Blood Spots, WB = Whole Blood, SV = Saliva Swab, gDNA = Genomic DNA, UR = Urine

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 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 for Genetic/Genomic Tests include:

- 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.
- Negative:** A negative result indicates 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.
- Inconclusive/Variant of Uncertain Significance:** A variant of uncertain significance (VOUS) result indicates that a DNA change 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.
- 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 variant(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. For testing performed on prenatal samples or for screening of apparently healthy individuals, only variants classified as pathogenic or likely pathogenic will 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 called “Secondary Findings”. When Secondary Findings are requested, only Pathogenic or Likely Pathogenic findings will be reported, where applicable. Please read the Secondary Findings sections on page 3 and/or 4 of this consent form for more information, and available reporting options. For prenatal samples, secondary findings for the proband are not available.

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 WES or WGS test, family members will have the option to receive information about secondary findings either as a part of the proband report or as a standalone parental report. A full analysis of the parental samples for secondary findings will only be completed if standalone reports are selected (for an additional charge). If family members elect to receive information about secondary findings either as part of the proband report or as a standalone report, the family member must sign all applicable sections on page 3 and/or 4 of this form.

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.

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 indefinitely, unless otherwise noted. 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. PerkinElmer may also work with scientists or researchers from academic or commercial institutions who have received the necessary approvals to conduct a research study. In some instances, these scientists or researchers may like to contact you directly about your interest in participating in a specific research study.

By checking here I would like to opt out of PerkinElmer being able to provide my contact information to outside researchers to contact me directly about applicable research studies.

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