PerkinElmer For the Better | PerkinElmer GENOMICS

General Test Requisition Form - Biochemical and Molecular

Please complete every field and tick box clearly.

PATIENT INFORMATION

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

Patient's Last Name | Patient ID/MR 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) _____		

ORDERING PROVIDER

Provider's First and Last Name

PKIG Ordering Provider Account Number | NPI

Clinic/Hospital/Institution Name

Provider's Email | Provider's Phone

Provider's Street Address

City / Town | State | Zip Code

Country | Provider's Fax

SEND ADDITIONAL COPY OF RESULTS TO (If applicable)

Name

PKIG Ordering Provider Account Number | Phone Number

Email Address | Fax Number

PATIENT SAMPLE INFORMATION

SAMPLE TYPE:

○ Saliva Swab
○ Whole Blood
○ Dried Blood Spots
○ Other _____

Collection Date: MM/DD/YY

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

INDICATION FOR TESTING (Required)

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

BIOCHEMICAL TESTS

SCREENING PANELS

○ B0200 StepOne® Comprehensive Biochemical Profile†
Birth Time: _____ Collection Time: _____
Weeks' Gestation: _____ Birth Weight: _____
Transfusion status: ○ Yes ○ No
If yes, transfusion type: ○ Platelet Date: _____ Time: _____
○ Plasma Date: _____ Time: _____
○ RBC Date: _____ Time: _____

† DBS Only

○ 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 NEWBORN TESTING

○ D3005 NeoSeq Newborn and Pediatric Gene Testing
○ D3004 Expanded Newborn Screening (NBS) Gene Sequencing Test

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 single gene testing, please order test code D3100 - AnyGene™ Test: Single Gene Sequencing and Del/Dup Test. Please submit requested gene for testing at apps.perkinelmergenomics.com/genelist and include custom gene ID with request.

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. 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.

Signature _____ Date _____

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

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☐ INSTITUTIONAL BILLING

Institution/Organization Name

PerkinElmer Genomics Billing Account ID

Contact Name

Contact Phone

☐ PATIENT (SELF) PAYMENT

By providing payment information, you are authorizing PerkinElmer to process payment at the associated charge for tests ordered. Test cost may be confirmed by calling 877-475-4436. Payment is required prior to test initiation. The patient's sample will be placed on hold (for up to 30 days) until payment is secured. If the patient does not provide payment to PerkinElmer within 30 days, the test order may be canceled. Please note that failure by the patient to respond in a timely fashion to PerkinElmer's attempts to obtain payment may cause a delay in the receipt of the results report.

○ **CHECK:** $_____ Amount Enclosed (Please make checks payable to: PerkinElmer Genetics, Inc.)

○ **CREDIT CARD** (Please fill out all information):

Credit Card Number | Card Exp. Date (MM/YY) | CVV | Cardholder Printed Name as Appears on Card | Amount

Credit Card Billing Street Address | City / Town | State | Zip Code

Cardholder Signature

Cardholder Phone

○ **CONTACT PATIENT FOR PAYMENT INFORMATION**

Mobile Phone

Home Phone

Email Address

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General Test Requisition Form - Biochemical and Molecular

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)

Clinical diagnosis: _____

Age of manifestation: _____ ICD-10 Codes: _____

A. NEUROLOGY

1. Behavioral abnormality
- O 1.1 Autism
- O 1.2 Attention deficit disorder
- O 1.3 Psychiatric diseases

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

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

4. Movement abnormality
- O 4.1 Ataxia
- O 4.2 Chorea
- O 4.3 Dystonia
- O 4.4 Parkinsonism

5. Neuromuscular abnormality
- O 5.1 Muscular hypotonia
- O 5.2 Muscular hypertonia
- O 5.3 Hyperreflexia
- O 5.4 Spasticity

6. Seizures
- O 6.1 Febrile seizures
- O 6.2 Focal seizures
- O 6.3 Generalized seizures

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

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

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

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

E. SKIN, INTEGUMENT AND SKELETAL

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

2. Skin and integument
- O 2.1 Abnormal skin pigmentation
- O 2.2 Abnormal hair
- O 2.3 Abnormal nail
- O 2.4 Hyperextensible skin
- O 2.5 Ichthyosis

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

G. GASTROINTESTINAL, GENITOURINARY, ENDOCRINE

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

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

3. Endocrine
- O 3.1 Diabetes mellitus
- O 3.2 Hypo / hyperparathyroidism
- O 3.3 Hypo / hyperthyroidism

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

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

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

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

OTHER:

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Complete Test Menu

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	*NADK2* Gene Sequencing	D3100	DBS, WB, SV, gDNA
2-methylbutyryl Glycinuria	Full Gene Analysis	*ACADSB* Gene Sequencing	D3100	DBS, WB, SV, gDNA
3-methylcrotonyl-CoA Carboxylase Deficiency (3-MCC Deficiency)	Targeted Variant Testing	3-MCC Deficiency Mutation Panel	D0410	DBS
3-methylglutaconic Aciduria, Type I	Full Gene Analysis	*AUH* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Argininemia	Full Gene Analysis	*ARG1* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Argininosuccinic Aciduria	Full Gene Analysis	*ASL* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Beta-ketothiolase Deficiency	Full Gene Analysis	*ACAT1* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Carnitine Palmitoyltransferase I Deficiency	Full Gene Analysis	*CPT1A* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Carnitine Palmitoyltransferase II Deficiency	Full Gene Analysis	*CPT2* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Carnitine Uptake Defect (CUD)	Full Gene Analysis	*SLC22A5* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Carnitine-acylcarnitine Translocase (CACT) Deficiency	Full Gene Analysis	*SLC25A20* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Citrullinemia Type I	Full Gene Analysis	*ASS1* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Citrullinemia Type II	Full Gene Analysis	*SLC25A13* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Cobalamin C Deficiency	Full Gene Analysis	*MMACHC* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Cobalamin D Deficiency	Full Gene Analysis	*MMADHC* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Glutaric Acidemia Type I	Targeted Variant Testing	Glutaric Acidemia Type I Mutation Panel	D0406	DBS
Glutaricaciduria, Type I	Full Gene Analysis	*GCDH* Gene Sequencing	D3100	DBS, WB, SV, gDNA
HMG-CoA Lyase Deficiency	Full Gene Analysis	*HMGCL* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Homocystinuria	Full Gene Analysis	*CBS* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Hypermethioninemia	Full Gene Analysis	*ADK* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Isobutyryl-CoA Dehygrogenase Deficiency	Full Gene Analysis	*ACAD8* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Isovaleric Acidemia	Targeted Variant Testing	Isovaleric Acidemia Mutation Panel	D0409	DBS
Isovaleric Acidemia	Full Gene Analysis	*IVD* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Long-chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency (LCHADD)	Targeted Variant Testing	*LCHADD* Mutation Panel	D0407	DBS
Maple Syrup Urine Disease	Targeted Variant Testing	Maple Syrup Urine Disease Mutation Panel	D0401	DBS
Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCADD)	Targeted Variant Testing	*MCADD* Mutation Panel	D0400	DBS
Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCADD)	Full Gene Analysis	*ACADM* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Methylmalonic Acidemia	Targeted Variant Testing	Methylmalonic Acidemia Mutation Panel	D0411	DBS
Methylmalonic Acidemia	Full Gene Analysis	*MUT* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Multiple Carboxylase Deficiency	Full Gene Analysis	*HLCS* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Multiple Sulfatase Deficiency	Full Gene Analysis	*SUMF1* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Phenylketonuria (PKU)	Biochemical Assay	PKU Monitoring - Phenylalanine	B0018	DBS, WB
Phenylketonuria (PKU)	Full Gene Analysis	*PAH* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Propionic Acidemia	Targeted Variant Testing	Propionic Acidemia Mutation Panel	D0412	DBS
Short Chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency (M/SCHADD)	Full Gene Analysis	*HADH* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Short-chain Acyl-CoA Dehydrogenase Deficiency (SCADD)	Full Gene Analysis	*ACADS* Gene Sequencing	D3100	DBS, WB, SV, gDNA

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Complete Test Menu

Associated Condition(s)	Test Type	Test Name	Test Code	Sample Type
Tyrosinemia	Biochemical Assay	Tyrosinemia Monitoring - Succinylacetone and Tyrosine	B0022	DBS, WB
Tyrosinemia Type I	Full Gene Analysis	*FAH* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Tyrosinemia Type I	Biochemical	Succinylacetone (SUAC)	B0021	DBS, WB, gDNA
Tyrosinemia Type II	Full Gene Analysis	*TAT* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Tyrosinemia Type III	Full Gene Analysis	*HPD* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Very Long-chain Acyl-CoA Dehydrogenase Deficiency (VLCADD)	Full Gene Analysis	*ACADVL* Gene Sequencing	D3100	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	*BTD* Gene Sequencing	D3100	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	D3100	DBS
Cystic Fibrosis	Full Gene Analysis	*CFTR* Gene Sequencing	D3100	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	*DMD* Gene Sequencing and Del/Dup Testing	D4045	DBS, WB, SV, gDNA
Duchenne Muscular Dystrophy (DMD)	Deletion/ Duplication Analysis	*DMD* Del/Dup Testing	D5125	DBS, WB, SV, gDNA
		FRIEDREICH'S ATAXIA		
Friedreich's Ataxia	Tandem Repeat Analysis	*FXN* Repeat Analysis	D5133	DBS, WB, gDNA
		GALACTOSEMIA		
Galactosemia	Biochemical Assay	Galactosemia Monitoring - Galactose-1-phosphate uridyltransferase Enzyme Analysis and Total Galactose	B0009	DBS
Galactosemia	Targeted Variant Testing	Galactosemia Mutation Panel	D0405	DBS
Galactosemia	Full Gene Analysis	*GALT* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Galactoepimerase Deficiency	Full Gene Analysis	*GALE* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Galactokinase Deficiency	Full Gene Analysis	*GALK* Gene Sequencing	D3100	DBS, WB, SV, gDNA
		GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY		
Glucose-6-phosphate Dehyrogenase Deficiency	Biochemical Assay	Glucose-6-phosphate Dehyrogenase Deficiency (screening only)	B0011	DBS
Glucose-6-phosphate Dehyrogenase Deficiency	Targeted Variant Testing	Glucose-6-phosphate Dehyrogenase Deficiency Mutation Panel	D0404	DBS
Glucose-6-phosphate Dehyrogenase Deficiency	Full Gene Analysis	*G6PD* Gene Sequencing	D3100	DBS, WB, SV, gDNA
		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

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Associated Condition(s)	Test Type	Test Name	Test Code	Sample Type
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	D3100	DBS, WB, SV, gDNA
SEVERE COMBINED IMMUNODEFICIENY				
Severe Combined Immunodeficiency (SCID)	Molecular DNA Screen	TREC Assay	D0416	DBS
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*ADA* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*AK2* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*ATM* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*CD3D* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*CD3E* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*CD3Z* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*CORO1A* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*DCLRE1C* (Artemis) Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*DOCK8* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*FOXN1* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*IL2RG* SGene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*IL7R* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*JAK3* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*LIG4* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*NHEJ1* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*ORAI1* Gene Sequencing	D3100	DBS, WB, SV, gDNA

Associated Condition(s)	Test Type	Test Name	Test Code	Sample Type
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*PNP* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*PRKDC* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*PTPRC* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*RAC2* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*RAG1* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*RAG2* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*RMRP* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*STIM1* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*TBX1* Gene Sequencing	D3100	DBS, WB, SV, gDNA
Severe Combined Immunodeficiency (SCID)	Full Gene Analysis	*ZAP70* Gene Sequencing	D3100	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	*SMN2* Copy Number Test	D5136	DBS, WB, SV, gDNA
OTHER				
Congenital Adrenal Hyperplasia (CAH)	Biochemical Assay	Congenital adrenal hyperplasia - 17A Hydroxyprogesterone (17 *OHP*)	B0002	DBS
Congenital Adrenal Hyperplasia (CAH)	Full Gene Analysis	*CYP21A2* 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	*FMR1* 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	*ABCD1* Gene Sequencing	D3100	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

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U.S. CLINICAL INFORMED CONSENT FORM

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:

1. *Positive:* A positive genetic test result may indicate that you are a carrier of, predisposed to, or have the specific disease or condition being tested for. A positive genetic test may limit your access to health insurance or life assurance coverage; for example, a life insurance company might ask you to provide genetic information indicating a disorder if this information is available to you.
2. *Negative:* A negative result indicates that 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 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.
4. *Unexpected Results:* In rare instances, this Test may reveal an important genetic change that is not directly related to the reason for ordering this test. This information would be disclosed to your HCP if it potentially impacts medical care, and you have consented to receive this type of result

TEST REPORT

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