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 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) _____		

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:
○ Saliva Swab
○ Whole Blood
○ Dried Blood Spots
Other_____

Collection Date: MM/DD/YY
Was this sample collected in NY State: ○ Yes ○ No
Age of Manifestation:_____
ICD10 CODES: _____

BIOCHEMICAL SCREENING TESTS*

PANELS

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

SPECIALTY TESTING

○ 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.

SINGLE SITE ANALYSIS (Please include a copy of relative's report, if available.)

Gene | Variant Name (c.)

Proband's Name

Relationship to Proband | Original Accession#

Positive Control Sample: ○ To be sent later ○ Already at PKIG ○ Not available

PHYSICIAN STATEMENT

Confirmation of informed consent 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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General Test Requisition Form - Biochemical and Molecular

BILLING INFORMATION - INSTITUTION

Institution/Organization

Contact Name

aaa
Institution Phone

bbb
Institution Billing Address

Institution Fax

City / Town | State | Zip Code | Institution Email

Special Handling Notes (Internal Use Only):

BILLING INFORMATION - SELF PAY

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

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

Credit Card Number | CVV

Credit Card Billing Street Address | MM/YY Card Exp. Date | Cardholder Phone

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

Cardholder Signature

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General Test Requisition Form - Biochemical and Molecular

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

Clinical diagnosis: _____

Age of manifestation: _____ ICD-10 Codes: _____

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

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

Complete Test Menu

Associated Condition(s)	Test Type	Test Name	Test Code	Sample Type*
Multiple Carboxylase Deficiency	Gene Sequencing	*HLCS* Gene Sequencing	D5038	DBS, WB, SV
Multiple Sulfatase Deficiency	Gene Sequencing	*SUMF1* Gene Sequencing	D5058	DBS, WB, SV
Phenylketonuria (PKU)	Biochemical	PKU Monitoring - Phenylalanine	B0018	DBS, WB
Phenylketonuria (PKU)	Gene Sequencing	*PAH* Gene Sequencing	D5050	DBS, WB, SV
Propionic Acidemia	Common Variant Testing	Propionic Acidemia Mutation Panel	D0412	DBS, WB, SV
Propionic Acidemia	Gene Sequencing	*PCCA* and *PCCB* Gene Sequencing	D3014	DBS, WB, SV
Short Chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency (M/SCHADD)	Gene Sequencing	*HADH* Gene Sequencing	D5036	DBS, WB, SV
Short-chain Acyl-CoA Dehydrogenase Deficiency (SCADD)	Gene Sequencing	*ACADS* Gene Sequencing	D5003	DBS, WB, SV
Tyrosinemia	Biochemical	Tyrosinemia Monitoring - Succinylacetone and Tyrosine	B0022	DBS, WB
Tyrosinemia Type I	Gene Sequencing	*FAH* Gene Sequencing	D5023	DBS, WB, SV
Tyrosinemia Type I	Biochemical	Succinylacetone (SUAC)	B0021	DBS, WB
Tyrosinemia Type II	Gene Sequencing	*TAT* Gene Sequencing	D5059	DBS, WB, SV
Tyrosinemia Type III	Gene Sequencing	*HPD* Gene Sequencing	D5040	DBS, WB, SV
Very Long-chain Acyl-CoA Dehydrogenase Deficiency (VLCADD)	Gene Sequencing	*ACADVL* Gene Sequencing	D5005	DBS, WB, SV
		BIOTINIDASE DEFICIENCY		
Biotinidase Deficiency	Biochemical	Biotinidase Deficiency (Complete/Partial) - Biotinidase Deficiency Enzyme Analysis	B0001	DBS, WB
Biotinidase Deficiency	Common Variant Testing	Biotinidase Deficiency Mutation Panel	D0402	DBS, WB, SV
Biotinidase Deficiency	Gene Sequencing	*BTD* Gene Sequencing	D5014	DBS, WB, SV
		CYSTIC FIBROSIS		
Cystic Fibrosis	Biochemical	IRT Analysis (Not valid after 90 days of age)	B0005	DBS, WB
Cystic Fibrosis	Common Variant Testing	Cystic Fibrosis Mutation Panel	D0403	DBS, WB, SV
Cystic Fibrosis	Gene Sequencing	*CFTR* Gene Sequencing	D5016	DBS, WB, SV
		DUCHENNE MUSCULAR DYSTROPHY		
Duchenne Muscular Dystrophy (DMD)	Biochemical	Duchenne Muscular Dystrophy Creatine Kinase Activity	B0006	DBS, WB
Duchenne Muscular Dystrophy (DMD)	Deletion/ Duplication Testing	*DMD* Del/Dup Testing	D5125	DBS, WB, SV
Duchenne Muscular Dystrophy (DMD)	Gene Sequencing	*DMD* Gene Sequencing and Del/Dup Testing	D4045	DBS, WB, SV
		GALACTOSEMIA		
Galactosemia	Biochemical	Galactosemia Monitoring - Galactose-1-phosphate uridyltransferase Enzyme Analysis and Total Galactose	B0009	DBS, WB
Galactosemia	Common Variant Testing	Galactosemia Mutation Panel	D0405	DBS, WB, SV
Galactosemia	Gene Sequencing	*GALT* Gene Sequencing	D5029	DBS, WB, SV
Galactoepimerase Deficiency	Gene Sequencing	*GALE* Gene Sequencing	D5026	DBS, WB, SV
Galactokinase Deficiency	Gene Sequencing	*GALK* Gene Sequencing	D5027	DBS, WB, SV
		GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY		
Glucose-6-phosphate Dehyrogenase Deficiency	Biochemical	Glucose-6-phosphate Dehyrogenase Deficiency (screening only)	B0011	DBS, WB
Glucose-6-phosphate Dehyrogenase Deficiency	Common Variant Testing	Glucose-6-phosphate Dehyrogenase Deficiency Mutation Panel	D0404	DBS, WB, SV
Glucose-6-phosphate Dehyrogenase Deficiency	Gene Sequencing	*G6PD* Gene Sequencing	D5024	DBS, WB, SV
		LYSOSOMAL STORAGE DISORDERS - TESTING OPTIONS		
Lysosomal Storage Disorders	Biochemical	Lysosomal Storage Disease Enzyme Panel	B2040	DBS, WB
Lysosomal Storage Disorders	Gene Sequencing	Lysosomal Storage Disorder Gene Sequencing Panel (12 Genes)	D3001	DBS, WB, SV
Fabry Disease	Biochemical	Alpha-Galactosidase A Enzyme Analysis	B0007	DBS, WB
Fabry Disease	Biochemical	Globotriaosylsphingosine (lyso-Gb3) Monitoring	B0029	DBS, WB

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Associated Condition(s)	Test Type	Test Name	Test Code	Sample Type*
Fabry Disease	Gene Sequencing	*GLA* Gene Sequencing	D5033	DBS, WB, SV
Gaucher Disease	Biochemical	Glucocerebrosidase (Glucosylceramidase) Enzyme Analysis	B0010	DBS, WB
Gaucher Disease	Gene Sequencing	*GBA* Gene Sequencing	D5032	DBS, WB, SV
Krabbe Disease	Biochemical	Galactocerebrosidase Enzyme Analysis	B0012	DBS, WB
Krabbe Disease	Gene Sequencing	*GALC* Gene Sequencing	D5031	DBS, WB, SV
MPS I (Hurler Syndrome)	Biochemical	Alpha-L-Iduronidase Enzyme Analysis	B0013	DBS, WB
MPS I (Hurler Syndrome)	Gene Sequencing	*IDUA* Gene Sequencing	D5041	DBS, WB, SV
MPS II (Hunter Syndrome)	Biochemical	Iduronate 2-Sulfatase Enzyme Analysis	B0014	DBS, WB
MPS II (Hunter Syndrome)	Gene Sequencing	*IDS* Gene Sequencing	D5042	DBS, WB, SV
MPS IVA (Morquio A Syndrome)	Biochemical	Galactosamine-6-Sulfatase Enzyme Analysis	B0015	DBS, WB
MPS IVA (Morquio A Syndrome)	Gene Sequencing	*GALNS* Gene Sequencing	D5028	DBS, WB, SV
MPS IVB (GM1 Gangliosidosis)	Biochemical	β-galactosidase Enzyme Analysis	B0025	DBS, WB
MPS IVB (GM1 Gangliosidosis)	Gene Sequencing	*GLB1* Gene Sequencing	D5034	DBS, WB, SV
MPS VI (Maroteaux-Lamy Syndrome)	Biochemical	Arylsulfatase B Enzyme Analysis	B0016	DBS, WB
MPS VI (Maroteaux-Lamy Syndrome)	Gene Sequencing	*ARSB* Gene Sequencing	D5009	DBS, WB, SV
MPS VII (Sly Syndrome)	Biochemical	β-glucuronidase Enzyme Analysis	B0026	DBS, WB
Mucopolysaccharidosis VII	Gene Sequencing	*GUSB* Gene Sequencing	D5035	DBS, WB, SV
Multiple Sulfatase Deficiency	Gene Sequencing	*SUMF1* Gene Sequencing	D5058	DBS, WB, SV
Niemann Pick Disease Types A and B	Biochemical	ACID Sphingomyelinase Enzyme Analysis	B0017	DBS, WB
Niemann Pick Disease Types A and B	Gene Sequencing	*SMPD1* Gene Sequencing	D5057	DBS, WB, SV
Pompe Disease	Biochemical	ACID Alpha-Glucosidase Enzyme Analysis	B0019	DBS, WB
Pompe Disease	Gene Sequencing	*GAA* Gene Sequencing	D5025	DBS, WB, SV
Neuronal Ceroid Lipofuscinosis 2 (CLN2)	Biochemical	Tripeptidyl peptidase 1 Enzyme Analysis	B0027	DBS, WB
Neuronal Ceroid Lipofuscinosis 2 (CLN2)	Gene Sequencing	*TPP1* Gene Sequencing	D5090	DBS, WB, SV
SEVERE COMBINED IMMUNODEFICIENY				
Severe Combined Immunodeficiency (SCID)	Molecular DNA Screen	TREC Assay	D0416	DBS, WB
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*SCID* Gene Sequencing Panel (26 Genes)	D3006	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*ADA* Gene Sequencing	D5060	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*AK2* Gene Sequencing	D5061	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*ATM* Gene Sequencing	D5062	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*CD3D* Gene Sequencing	D5063	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*CD3E* Gene Sequencing	D5064	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*CD3Z* Gene Sequencing	D5065	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*CORO1A* Gene Sequencing	D5066	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*DCLRE1C* (Artemis) Gene Sequencing	D5067	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*DOCK8* Gene Sequencing	D5068	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*FOXN1* Gene Sequencing	D5069	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*IL2RG* SGene Sequencing	D5070	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*IL7R* Gene Sequencing	D5071	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*JAK3* Gene Sequencing	D5072	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*LIG4* Gene Sequencing	D5073	DBS, WB, SV

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Complete Test Menu

Associated Condition(s)	Test Type	Test Name	Test Code	Sample Type*
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*NHEJ1* Gene Sequencing	D5074	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*ORAI1* Gene Sequencing	D5075	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*PNP* Gene Sequencing	D5076	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*PRKDC* Gene Sequencing	D5077	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*PTPRC* Gene Sequencing	D5078	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*RAC2* Gene Sequencing	D5079	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*RAG1* Gene Sequencing	D5080	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*RAG2* Gene Sequencing	D5081	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*RMRP* Gene Sequencing	D5082	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*STIM1* Gene Sequencing	D5083	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*TBX1* Gene Sequencing	D5084	DBS, WB, SV
Severe Combined Immunodeficiency (SCID)	Gene Sequencing	*ZAP70* Gene Sequencing	D5085	DBS, WB, SV
SICKLE CELL AND OTHER HEMOGLOBINOPATHIES				
Sickle Cell and Other Hemoglobinopathies	Biochemical	Isoelectric Focusing GEL Electrophoresis of Hemoglobiins	B0020	DBS, WB
Sickle Cell and Other Hemoglobinopathies	Common Variant Testing	Sickle Cell and Other Hemoglobinopathies Mutation Panel	D0408	DBS, WB, SV
SPINAL MUSCULAR ATROPHY (SMA)				
Spinal Muscular Atrophy (SMA)	Deletion/ Duplication Testing	*SMN1* Diagnostic Testing (by RT-PCR)	D5127	DBS, WB, SV
Spinal Muscular Atrophy (SMA)	Deletion/ Duplication Testing	*SMN1* Carrier Testing (by RT-PCR)	D5128	DBS, WB, SV
Spinal Muscular Atrophy (SMA)	Deletion/ Duplication Testing	*SMN2* Copy Number Testing (by RT-PCR)	D5129	DBS, WB, SV
OTHER				
Congenital Adrenal Hyperplasia (CAH)	Biochemical	Congenital adrenal hyperplasia - 17A Hydroxyprogesterone (17 *OHP*)	B0002	DBS, WB
Congenital Adrenal Hyperplasia (CAH)	Gene Sequencing and Deletion/ Duplication	*CYP21A2* Gene Sequencing and Del/Dup Testing (by MLPA)	D5019	DBS, WB, SV
Congenital Hypothyroidism	Biochemical	Thyroid-Stimulating Hormone (TSH)	B0003	DBS, WB
Congenital Hypothyroidism	Biochemical	Thyroxine (T4)	B0004	DBS, WB
Fragile X	Triplet Repeat Testing	*FMR1* Triplet Repeat (CGG) Testing	D4042	DBS, WB, SV
X-linked Adrenoleukodystrophy	Biochemical	X-Linked Adrenoleukodystrophy - C26:0 Lysophosphatidylcholine	B0023	DBS, WB
X-linked Adrenoleukodystrophy	Gene Sequencing	*ABCD1* Gene Sequencing	D5000	DBS, WB, SV
Multiple	Biochemical	Post Mortem - Includes: 17-Hydroxyprogesterone, Acylcarnitines, Galactose, and *TSH*	B0024	DBS, Vitreous Fluid, Bile, Liver Tissue Blot

*DBS = Dried Blood Spots, WB = Whole Blood, SV = Saliva Swab

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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 looks at the genes in your DNA. 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 enzyme activity, biomarker tests, and select genetic testing assays will be conducted in Pennsylvania, USA, and all other genetic testing will be conducted in Connecticut, USA.

Under some circumstances, including inadequate or poor quality sample, an additional Sample may be required for Tests to be performed.

TEST RESULTS

Your treating HCP has sole responsibility for all decisions concerning the possible management of your diagnosis and disease; PerkinElmer will not provide a diagnosis. PerkinElmer will report Test results only to your HCP via secure email, a secure internet portal, or fax. Your HCP is responsible for communicating with you regarding the results of the Test and may refer you or your child to a specialist for further clinical evaluation and confirmation of diagnosis, if applicable. Possible results include:

1. *Positive:* A positive genetic test result may indicate that you are a carrier of, predisposed to, or have the specific disease or condition being tested for. A positive genetic test may limit your access to health insurance or life assurance coverage; for example, a life insurance company might ask you to provide genetic information indicating a disorder if this information is available to you.
2. *Negative:* A negative result indicates that the enzyme/biomarker results were within normal ranges, or that no disease-causing variant was identified in the Test performed. No Test can rule out all genetic diseases or conditions. A negative result does not guarantee that you are free from genetic disorders or other medical conditions.
3. *Inconclusive/Variant of Uncertain Significance:* A variant of uncertain significance (VOUS) result indicates that a variant outside of the normal range was detected, but it is currently unknown if the variant is associated with a genetic disorder. A VOUS is not the same as a positive result and does not clarify whether there is an increased risk to develop a genetic disorder. The variant could be a benign change or it could be indicative of disease/disease-causing.
4. *Unexpected Results:* In rare instances, this Test may reveal an important genetic change that is not directly related to the reason for ordering this test. This information would be disclosed to your HCP if it potentially impacts medical care, and you have consented to receive this type of result

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.

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. Please read the Secondary Findings section on page 3 of this consent form for more information, and reporting options.

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 test (WES or WGS), parents will have the option of receiving a full parental report for an additional charge. If selected, the respective parental consent section must be completed below.

TEST LIMITATIONS

Due to current limitations in technology and incomplete knowledge of diseases and genes, some variants may not be detected by the Test ordered. There is a possibility that the Test result that is uninterpretable or of unknown significance may require further testing when more information is gained. In rare circumstances, Test results may be suggestive of a condition different from that which was originally considered for the purpose of consenting to this Test. The Test may also find variants or genes that lead to conditions for which you currently do not have symptoms or may not be related to your current condition.

TEST RISKS

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

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.

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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 for a minimum of two years and then destroyed. In some instances, it may be beneficial to you for PerkinElmer to retain your sample for a longer period of time in order to conduct additional testing, and PerkinElmer will do so with appropriate documentation from you or your HCP.

PerkinElmer is requesting consent to keep you and/or your child's anonymized sample and data indefinitely. 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, no additional tests or anonymization will be carried out on your Sample; no results will be reported and your sample, reports and data that have not been anonymized will be destroyed.
  - ❑ Check here if you would like to opt out of anonymized sample retention. 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 internal statistical, quality analysis, research, scientific and technical development, and market research.
  - ❑ Check here if you would like to opt out of anonymized data retention. Note, if not checked, this is interpreted as "consent given"

For residents of NY State:

❑ By checking here I give PerkinElmer permission to store my sample for longer than 60 days. Note, if not checked, this is interpreted as "consent not given"

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.

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 _____