PerkinElmer
For the Better

General Test Requisition Form - Biochemical and Molecular

Please complete every field and tick box clearly.

PerkinElmer GENOMICS

PATIENT INFORMATION

Patient's First Name | Middle Initial

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

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

Patient's Street Address

City / Town | State | Zip Code

Country | Patient's Preferred Phone

Patient's Email

Ethnicity (check all that apply):	○ African-American	○ Asian (China, Japan, Korea)
○ Caucasian/N. European/S. European	○ Finnish	○ French Canadian
○ Hispanic	○ Jewish - Ashkenazi	○ Jewish - Sephardic	○ Mediterranean
○ Middle Eastern (Saudi Arabia, Qatar, Iraq, Turkey)	○ Native American	○ E. Indian
○ Southeast Asian (Vietnam, Cambodia, Thailand)	○ South Asian (India, Pakistan)	
○ Other (specify) _____		

PROVIDER

Provider's First and Last Name

Account # | Provider's Phone

Provider's Email

Clinic/Hospital/Institution Name

Provider's Street Address

City / Town | State | Zip Code

Country | Provider's Fax

ADDITIONAL PROVIDER/GENETIC COUNSELOR (IF APPLICABLE)

Provider/Genetic Counselor's Name

Provider /Genetic Counselor's Account # | Provider/Genetic Counselor's Phone

Provider/Genetic Counselor's Email | Provider/Genetic Counselor's Fax

PATIENT SAMPLE INFORMATION

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

Collection Date: MM/DD/YY
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 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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BILLING INFORMATION - INSTITUTION

Institution/Organization

Contact Name

Institution Phone

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 | Card Exp. Date (MM/YY) | Cardholder Phone

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

Cardholder Signature

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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. Immunodefciency
- 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:

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

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	D5020	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

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

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 MLPA)	D0413	DBS, WB, SV
Spinal Muscular Atrophy (SMA)	Deletion/ Duplication Testing	*SMN1* Carrier Testing (by MLPA)	D0414	DBS, WB, SV
Spinal Muscular Atrophy (SMA)	Deletion/ Duplication Testing	*SMN2* Copy Number Testing (by MLPA)	D0415	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

GENERAL INFORMED CONSENT FORM

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

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

TEST INFORMATION

The type of Test you are consenting to is a genetic test that looks at the genes in your DNA. This Test is used to identify what, if any, DNA mutation(s) you or your child is carrying which is causing the specific disease or condition you are being tested for. Identifying the mutation may be useful for diagnostic and treatment purposes, and allows at-risk family members to be tested. Only the genes identified on the Requisition will be analyzed. In some cases, we may not be able to determine with certainty which gene is actually causing the disease.

TEST METHOD

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

TEST RESULTS

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

TEST LIMITATIONS

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

TEST RISKS

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

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

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

ANONYMIZATION

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

RETENTION

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

RESEARCH OPTIONS

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

WHY ARE PARENTAL SAMPLES NEEDED?

In some circumstances, it may be helpful for additional family members to undergo testing as well in order to provide information that can aid in the interpretation of the test results. These Tests could be part of a TRIO Test or as stand-alone targeted testing. PerkinElmer, in consultation with the HCP, will decide if other family members need to be tested. If the HCP recommends testing for additional family members, only the Test performed will be reported.

TEST REPORT

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

PATIENT'S CONSENT TO TESTING

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

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

PATIENT/ Representative Signature: _____

Print Patient Name:_____ Date_____ Time _____

Mother of Patient for TRIO testing authorization:

I have read the Consent as provided by the HCP and understand and agree to the following:

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

Print MOTHER Name: _____ Date_____ Time _____

Signature (Mother of Patient): _____

Father of Patient for TRIO testing authorization:

I have read the Consent as provided by the HCP and understand and agree to the following:

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

Print FATHER Name: _____ Date_____ Time _____

Signature (Father of Patient): _____