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General Biochemical and Molecular Requisition Form

Testing can also be ordered via online portal – please scan or click on QR code.
Please complete every field and tick box clearly.

STEP 1: PATIENT INFORMATION

Patient's First Name | Middle Initial | Patient's Last Name

MM/ DD /YYYY
Patient's Date of Birth | Patient ID/MR Number/External Sample 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)_____

PATIENT SAMPLE INFORMATION

SAMPLE TYPE: ○ Whole Blood ○ Saliva Swab ○ Urine ○ Dried Blood Spots ○ C-Dried Blood Spots ○ DNA, Source: _____ ○ DHeparin - Plasma ○ EDTA - Plasma ○ Other: _____

Collection Date: MM/DD/YY
Was this sample collected in the State of NV, NY or OR?: ○ Yes ○ No
(If yes, separate consent is required. See forms section of website.)

INDICATION FOR TESTING

Clinical Diagnosis:_____ (medical records/clinical notes are required.) Age at Initial Presentation:_____

STEP 2: ORDERING PROVIDER AND REPORTING PREFERENCES

Provider's First and Last Name | NPI

Clinic/Hospital/Institution Name | Provider's Email

Provider's Street Address | City / Town | State | Zip Code | Country

Provider's Phone | Provider's Fax

How would you like to receive the report?:
○ Fax ○ Email ○ Portal

SEND ADDITIONAL COPY OF RESULTS TO (If applicable)

Name | Role with patient/Job title | Clinic/Hospital/Institution Name

Phone Number | Fax Number | Email Address

How would you like to receive the report?:
○ Fax ○ Email ○ Portal

STEP 3: BILLING INFORMATION

☐ **INSTITUTIONAL BILLING**

Institution/Organization Name | Billing Account ID | P.O. Number (if applicable)

Contact Name | Contact Phone

☐ **PATIENT (SELF) PAYMENT**

By providing payment information, you are authorizing Revvity Omics to process payment at the associated charge for tests ordered. Test cost is available on our website, or 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 Revvity Omics within 30 days, the test order may be canceled. Please note that failure by the patient to respond in a timely fashion to Revvity Omics attempts to obtain payment may cause a delay in the receipt of the results report.

○ **CREDIT CARD** (Please fill out all information below) ○ **CHECK:** $_____ Amount Enclosed (Please make checks payable to: Revvity Omics, Inc.)

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

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

Cardholder Signature | Cardholder Phone

○ **CONTACT FOR PAYMENT INFORMATION**

Name | Phone | Email Address

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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STEP 4: TEST MENU

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:_____

○ B0210 Acylcarnitine Profile
○ B2020 Amino Acid Profile
○ B2040 Lysosomal Storage Disease Enzyme Panel
○ B0024 Post-Mortem Screening Panel
○ BG100 Nicotinamide Adenine Dinucleotide (NAD) - C-DBS required*
○ GED1D Mucopolysaccharidosis type I (MPS I) marker - DBS required
○ GED2D Mucopolysaccharidosis type II (MPS II) marker - DBS required

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†

○ D3100 AnyGene™ Test: Single Gene Sequencing and Del/Dup Test
Please submit requested gene for testing at apps-omics.revvity.com/gene-dashboard, and include custom gene ID below

*Provide gene or custom panel ID here:*_____

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.

* DBS Required. Test code BG100 requires collection on C-DBS cards.

STEP 5: PHYSICIAN CONFIRMATION OF INFORMED CONSENT AND MEDICAL NECESSITY

The undersigned person (or designated representative thereof) certifies that: (a) he/she is a licensed medical professional authorized to order the testing ordered herein; (b) he/she fully complies with all applicable federal, state, and local laws, regulations, and rules, including but not limited to those governing genetic testing, informed consent, and patient consent and authorization requirements for the test(s) ordered; (c) he/she will obtain informed consent of the patient in compliance with all applicable laws and regulations, which shall include, to the extent applicable: (i) a statement of the purpose of the test(s) ordered; (ii) a statement that prior to signing the consent form, the consenting person discussed with the medical practitioner ordering the test the reliability of positive or negative test results and the level of certainty that a positive test result for that disease or condition serves as a predictor of such disease; (iii) a statement that the consenting person was informed about the availability and importance of genetic counseling and provided with written information identifying a genetic counselor or medical geneticist from whom the consenting person might obtain such counseling; (iv) a general description of each disease or condition tested for; and (v) the person or persons to whom the test results may be disclosed; (d) he/she will maintain, as part of the patient's record, documentation of the patient's informed consent and authorization for the test(s) ordered that complies with applicable laws and regulations, and will make such documentation available to Revvity upon request; (e) tests ordered are medically necessary and results may impact medical management for the patient; and (f) the information provided on this Test Requisition Form is complete, true, and accurate to the best of his/her knowledge.

Signature_____ Date_____

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General Biochemical and Molecular Requisition Form

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

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

Clinical diagnosis: _____ Age of manifestation: _____

NEUROLOGY

1. Neurodevelopmental abnormality

- ○ 1.1 Autism
- ○ 1.2 Attention deficit disorder
- ○ 1.3 Global developmental delay
- ○ 1.4 Delayed motor development
- ○ 1.5 Delayed language development
- ○ 1.6 Developmental regression
- ○ 1.7 Intellectual disability

2. Brain imaging

- ○ 2.1 Abnormal myelination
- ○ 2.2 Agenesis of corpus callosum
- ○ 2.3 Brain atrophy
- ○ 2.4 Cerebellar hypoplasia
- ○ 2.5 Heterotopia
- ○ 2.6 Holoprosencephaly
- ○ 2.7 Hydrocephalus
- ○ 2.8 Leukodystrophy
- ○ 2.9 Lissencephaly

3. Movement abnormality

- ○ 3.1 Ataxia
- ○ 3.2 Chorea
- ○ 3.3 Dystonia
- ○ 3.4 Parkinsonism

4. Neuromuscular abnormality

- ○ 4.1 Muscular hypotonia
- ○ 4.2 Muscular hypertonia
- ○ 4.3 Hyperreflexia
- ○ 4.4 Spasticity

5. Seizures

- ○ 5.1 Febrile seizures
- ○ 5.2 Focal seizures
- ○ 5.3 Generalized seizures

6. Others

- ○ 6.1 Craniosynostosis
- ○ 6.2 Dementia
- ○ 6.3 Encephalopathy
- ○ 6.4 Headache / Migraine
- ○ 6.5 Macrocephaly
- ○ 6.6 Microcephaly
- ○ 6.7 Neuropathy
- ○ 6.8 Stroke

METABOLISM

- ○ 1. Creatine kinase
- ○ 2. Decreased plasma carnitine
- ○ 3. Hyperalaninemia
- ○ 4. Hypoglycemia
- ○ 5. Increased CSF lactate
- ○ 6. Increased serum pyruvate
- ○ 7. Ketosis
- ○ 8. Lactic acidosis
- ○ 9. Organic aciduria

EYE

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

MOUTH, THROAT AND EAR

- ○ 1. Abnormality of dental color
- ○ 2. Cleft lip / palate
- ○ 3. Conductive hearing impairment
- ○ 4. External ear malformation
- ○ 5. Hypodontia
- ○ 6. Sensoneural hearing impairment

SKIN, INTEGUMENT AND SKELETAL

1. Skeletal

- ○ 1.1 Abnormal limb morphology
- ○ 1.2 Abnormal vertebral column
- ○ 1.3 Joint hypermobility
- ○ 1.4 Multiple joint contractures
- ○ 1.5 Polydactyly
- ○ 1.6 Scoliosis
- ○ 1.7 Syndactyly
- ○ 1.8 Talipes equinovarus

2. Skin and integument

- ○ 2.1 Abnormal skin pigmentation
- ○ 2.2 Abnormal hair
- ○ 2.3 Abnormal nail
- ○ 2.4 Hyperextensible skin
- ○ 2.5 Ichthyosis

CARDIOVASCULAR

- ○ 1. Angioedema
- ○ 2. Aortic dilatation
- ○ 3. Arrhythmia
- ○ 4. Coarctation of aorta
- ○ 5. Defect of atrial septum
- ○ 6. Defect of ventricular septum
- ○ 7. Dilated cardiomyopathy
- ○ 8. Hypertrophic cardiomyopathy
- ○ 9. Lymphedema
- ○ 10. Malf. of heart and great vessels
- ○ 11. Myocardial infarction
- ○ 12. Tetralogy of Fallot

GASTROINTESTINAL, GENITOURINARY, ENDOCRINE

1. Gastrointestinal

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

2. Genitourinary

- ○ 2.1 Hydronephrosis
- ○ 2.2 Renal agenesis /hypoplasia
- ○ 2.3 Renal cyst
- ○ 2.4 Renal tubular dysfunction

3. Endocrine

- ○ 3.1 Diabetes mellitus
- ○ 3.2 Hypothyroidism
- ○ 3.3 Hyperparathyroidism
- ○ 3.4 Hypoparathyroidism
- ○ 3.5 Hyperthyroidism

REPRODUCTION

- ○ 1. Abnormal external genitalia
- ○ 2. Abnormal internal genitalia
- ○ 3. Hypogonadism
- ○ 4. Hypospadias
- ○ 5. Infertility

ONCOLOGY

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

HEMATOLOGY AND IMMUNOLOGY

- ○ 1. Abnormality of coagulation
- ○ 2. Anemia
- ○ 3. Immunodeficiency
- ○ 4. Neutropenia
- ○ 5. Pancytopenia
- ○ 6. Abnormal hemoglobin
- ○ 7. Splenomegaly
- ○ 8. Thrombocytopenia

PRENATAL AND DEVELOPMENT

- ○ 1. Failure to thrive
- ○ 2. Hemihypertrophy
- ○ 3. Hydrops fetalis
- ○ 4. IUGR
- ○ 5. Oligohydramnios
- ○ 6. Overgrowth
- ○ 7. Polyhydramnios
- ○ 8. Premature birth
- ○ 9. Disproportionate short stature
- ○ 10. Proportionate short stature
- ○ 11. Tall stature

OTHER (INCLUDING DYSMORPHIC FACIAL FEATURES AND OTHER DESCRIPTORS):

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

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

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

Complete Test Menu

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