

# Biochemical Genetics Test Request

## **Client Information (required)**

Client Name		
Client Account No.		
Client Phone	Client Order	No.
Street Address		
City	State	ZIP Code

### Submitting Healthcare Professional (required)

Submitting Healthcare Professional Name (Last, First)		
Title/Credentials		
Phone (with area code)	Fax* (with area code)	
National Provider Identification (NPI)		
Email**		
** Any communication sent via email will comp * Fax number given must be from a fax machine HIPAA regulation.		

Note: It is the client's responsibility to maintain documentation of the order. New York State Patients: Informed Consent for Genetic Testing

"I hereby confirm that informed consent has been signed by an individual legally authorized to do so and is on file with this office or the individual's provider's office."

Signature

Note: Test requests without a signature will not be performed.

### Patient Information (required)

Patient ID (Medical Record No.)		
Patient Name (Last, First Middle)		
Sex Male Female	Birth Date (mm-dd-yyyy)	
Collection Date (mm-dd-yyyy)	Time	□ am □ pm

# Reason for Testing (required)

Has molecular/I	NIA testing already been performed?	
	DNA testing already been performed? If Yes, results:	
🗆 Yes 🗆 No		
□ Yes □ No Gene	If Yes, results:	
□ Yes □ No Gene Gene	If Yes, results: Variant	

#### Ship specimens to:

Mayo Clinic Laboratories 3050 Superior Drive NW Rochester, MN 55905

#### Customer Service: 800-533-1710

Visit www.MayoClinicLabs.com for the most up-to-date test and shipping information.

**Billing Information** 

· Payment terms are net 30 days.

Call the Business Office with billing-related questions: 800-447-6424 (US and Canada) 507-266-5490 (outside the US)

Patient ID (Medical Record No.)	Client Account No.
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A	<b>MINO AC</b>	ID METABOLISM
	AAQP	Amino Acids, Quantitative, Plasma
	AAPD	Amino Acids, Quantitative, Random, Urine
	AACSF	Amino Acids, Quantitative, Spinal Fluid
	TRYPP	Tryptophan, Plasma
	TRYPU	Tryptophan, Random, Urine
Су	stinuria	
	CYSGP	Cystinuria Gene Panel
	CYSQN	Cystinuria Profile, Quantitative, 24 Hour, Urine
	CYSR	Cystinuria Profile, Quantitative, Random, Urine
Ho	mocystin	iuria
	CMMPP	Cobalamin, Methionine, and Methylmalonic Acid Pathways, Plasma
	CMMPS	Cobalamin, Methionine, and Methylmalonic Acid Pathways, Serum
	HCYSP	Homocysteine, Total, Plasma
	HCYSS	Homocysteine, Total, Serum
Maple Syrup Urine Disease		
Ma	aple Syrup	o Urine Disease
	a <b>ple Syru</b> Alloi	<b>o Urine Disease</b> Allo-isoleucine, Blood Spot
	ALLOI	Allo-isoleucine, Blood Spot Amino Acids, Maple Syrup Urine Disease
	ALLOI AAMSD	Allo-isoleucine, Blood Spot Amino Acids, Maple Syrup Urine Disease Panel, Plasma Branched-Chain Amino Acids, Self-Collect,
	ALLOI AAMSD MSUSC	Allo-isoleucine, Blood Spot Amino Acids, Maple Syrup Urine Disease Panel, Plasma Branched-Chain Amino Acids, Self-Collect, Blood Spot Maple Syrup Urine Disease Gene Panel
□ □ □ Ph	ALLOI AAMSD MSUSC MSUDP	Allo-isoleucine, Blood Spot Amino Acids, Maple Syrup Urine Disease Panel, Plasma Branched-Chain Amino Acids, Self-Collect, Blood Spot Maple Syrup Urine Disease Gene Panel
C C Ph	ALLOI AAMSD MSUSC MSUDP enylketor	Allo-isoleucine, Blood Spot Amino Acids, Maple Syrup Urine Disease Panel, Plasma Branched-Chain Amino Acids, Self-Collect, Blood Spot Maple Syrup Urine Disease Gene Panel
	ALLOI AAMSD MSUSC MSUDP enylketor PKUBS	Allo-isoleucine, Blood Spot Amino Acids, Maple Syrup Urine Disease Panel, Plasma Branched-Chain Amino Acids, Self-Collect, Blood Spot Maple Syrup Urine Disease Gene Panel nuria Phenylalanine and Tyrosine, Blood Spot
	ALLOI AAMSD MSUSC MSUDP enylketor PKUBS PHEGP	Allo-isoleucine, Blood Spot Amino Acids, Maple Syrup Urine Disease Panel, Plasma Branched-Chain Amino Acids, Self-Collect, Blood Spot Maple Syrup Urine Disease Gene Panel <b>nuria</b> Phenylalanine and Tyrosine, Blood Spot Phenylalanine Disorders Gene Panel
	ALLOI AAMSD MSUSC MSUDP enylketor PKUBS PHEGP PKU	Allo-isoleucine, Blood Spot Amino Acids, Maple Syrup Urine Disease Panel, Plasma Branched-Chain Amino Acids, Self-Collect, Blood Spot Maple Syrup Urine Disease Gene Panel <b>nuria</b> Phenylalanine and Tyrosine, Blood Spot Phenylalanine Disorders Gene Panel Phenylalanine and Tyrosine, Plasma Phenylalanine and Tyrosine, Self-Collect, Blood Spot
	ALLOI AAMSD MSUSC MSUDP enylketor PKUBS PHEGP PKU PKUSC	Allo-isoleucine, Blood Spot Amino Acids, Maple Syrup Urine Disease Panel, Plasma Branched-Chain Amino Acids, Self-Collect, Blood Spot Maple Syrup Urine Disease Gene Panel <b>nuria</b> Phenylalanine and Tyrosine, Blood Spot Phenylalanine Disorders Gene Panel Phenylalanine and Tyrosine, Plasma Phenylalanine and Tyrosine, Self-Collect, Blood Spot
	ALLOI AAMSD MSUSC MSUDP enylketor PKUBS PHEGP PKU PKUSC	Allo-isoleucine, Blood Spot Amino Acids, Maple Syrup Urine Disease Panel, Plasma Branched-Chain Amino Acids, Self-Collect, Blood Spot Maple Syrup Urine Disease Gene Panel <b>nuria</b> Phenylalanine and Tyrosine, Blood Spot Phenylalanine Disorders Gene Panel Phenylalanine and Tyrosine, Plasma Phenylalanine and Tyrosine, Self-Collect, Blood Spot
	ALLOI AAMSD MSUSC MSUDP enylketor PKUBS PHEGP PKU PKUSC rosinemia TYRGP	Allo-isoleucine, Blood Spot Amino Acids, Maple Syrup Urine Disease Panel, Plasma Branched-Chain Amino Acids, Self-Collect, Blood Spot Maple Syrup Urine Disease Gene Panel <b>nuria</b> Phenylalanine and Tyrosine, Blood Spot Phenylalanine Disorders Gene Panel Phenylalanine and Tyrosine, Self-Collect, Blood Spot

CARBOHYDRATE METABOLISM		
Congenital	Disorders of Glycosylation	
□ CDG	Carbohydrate Deficient Transferrin for Congenital Disorders of Glycosylation, Serum	
CDGGP	Congenital Disorders of Glycosylation Gene Panel	
CDGN	Congenital Disorders of N-Glycosylation, Serum	
	Oligosaccharide Screen, Random, Urine	
D PMMIL	Phosphomannomutase and Phosphomannose Isomerase, Leukocytes	
	Sorbitol and Mannitol, Quantitative, Random, Urine	
Galactosem	lia	
□ GATOL	Galactitol, Quantitative, Urine	
🗆 GALK	Galactokinase, Blood	
GAL1P	Galactose-1-Phosphate, Erythrocytes	
GALT	Galactose-1-Phosphate Uridyltransferase, Blood	
□ GALTP	Galactose-1-Phosphate Uridyltransferase Biochemical Phenotyping, Erythrocytes	
🗆 GALP	Galactose, Quantitative, Plasma	
🗆 GALZ	Galactosemia, GALT Gene, Full Gene Analysis	
□ GCT	Galactosemia Reflex, Blood	
□ GALE	Uridine Diphosphate-Galactose 4' Epimerase, Blood	
Transaldola Deficiencies	se and Ribose-5-phosphate (RPI) s	
□ TALDO	Polyols, Quantitative, Urine	
CHOLESTA	ITIC LIVER DISEASE	
CHLGP	Cholestasis Gene Panel	
	EROL BIOSYNTHESIS AND	
TRANSPOR		
CTXWB	Cerebrotendinous Xanthomatosis, Blood	
□ CTXBS	Cerebrotendinous Xanthomatosis, Blood Spot	
🗆 СТХР	Cerebrotendinous Xanthomatosis, Plasma	
□ HSMBS	Hepatosplenomegaly Panel, Blood Spot	
□ HSMWB	Hepatosplenomegaly Panel, Blood	
□ HSMP	Hepatosplenomegaly Panel, Plasma	
□ охуwв	Oxysterols, Blood	
□ OXYBS	Oxysterols, Blood Spots	
□ OXNP	Oxysterols, Plasma	
🗆 SLO	Smith-Lemli-Opitz Screen, Plasma	
	Smith Lemli Opitz Syndrome, DHCR7 Gene, Full Gene Analysis	
□ STER	Sterols, Plasma	

С	DNGENIT	AL ADRENAL HYPERPLASIA
	CAH2T	Congenital Adrenal Hyperplasia Newborn Screen, Blood Spot
	CAH21	Congenital Adrenal Hyperplasia (CAH) Profile for 21-Hydroxylase Deficiency, Serum
	CYPZ	21-Hydroxylase Gene, CYP21A2, Full Gene Analysis
С	ONGENIT	AL LACTIC ACIDOSIS
	CLADP	Congenital Lactic Acidosis Gene Panel
0.5		
		DISORDERS
_	CRDPP	Creatine Disorders Panel, Plasma
	CRDPU	Creatine Disorders Panel, Random, Urine
CL	JSTOM G	ENE PANEL
	CGPH	Custom Gene Panel, Hereditary, Next-Generation Sequencing Gene List ID (if known) or Genes Requested for Testing:
FA	MILIALA	MYLOIDOSIS
	TTDV	
	TTRX	Amyloidosis, Transthyretin-Associated Familial, Reflex, Blood
	TTRX	
	TTRZ	Familial, Reflex, Blood
□ FA	TTRZ	Familial, Reflex, Blood <i>TTR</i> Gene, Full Gene Analysis
FA	TTRZ	Familial, Reflex, Blood <i>TTR</i> Gene, Full Gene Analysis <b>METABOLISM (BETA-OXIDATION)</b>
FA	TTRZ TTY ACID ACRN	Familial, Reflex, Blood <i>TTR</i> Gene, Full Gene Analysis <b>METABOLISM (BETA-OXIDATION)</b> Acylcarnitines, Quantitative, Plasma
<b>FA</b>	TTRZ TTY ACIE ACRN ACRNS	Familial, Reflex, Blood TTR Gene, Full Gene Analysis METABOLISM (BETA-OXIDATION) Acylcarnitines, Quantitative, Plasma Acylcarnitines, Quantitative, Serum
<b>FA</b>	TTRZ TTY ACIE ACRN ACRNS AGU20	Familial, Reflex, Blood TTR Gene, Full Gene Analysis <b>METABOLISM (BETA-OXIDATION)</b> Acylcarnitines, Quantitative, Plasma Acylcarnitines, Quantitative, Serum Acylglycines, Quantitative, Random, Urine C4 Acylcarnitine, Quantitative, Random,
<b>FA</b>	TTRZ TTY ACIE ACRN ACRNS AGU20 C4U CARN	Familial, Reflex, Blood TTR Gene, Full Gene Analysis <b>DMETABOLISM (BETA-OXIDATION)</b> Acylcarnitines, Quantitative, Plasma Acylcarnitines, Quantitative, Serum Acylglycines, Quantitative, Random, Urine C4 Acylcarnitine, Quantitative, Random, Urine Carnitine, Plasma
<b>FA</b>	TTRZ ACRN ACRNS AGU20 C4U	Familial, Reflex, Blood TTR Gene, Full Gene Analysis METABOLISM (BETA-OXIDATION) Acylcarnitines, Quantitative, Plasma Acylcarnitines, Quantitative, Serum Acylglycines, Quantitative, Random, Urine C4 Acylcarnitine, Quantitative, Random, Urine Carnitine, Plasma Carnitine, Serum
<b>FA</b>	TTRZ TTY ACID ACRN ACRNS AGU20 C4U CARN CARNS	Familial, Reflex, Blood TTR Gene, Full Gene Analysis METABOLISM (BETA-OXIDATION) Acylcarnitines, Quantitative, Plasma Acylglycines, Quantitative, Serum Acylglycines, Quantitative, Random, Urine C4 Acylcarnitine, Quantitative, Random, Urine Carnitine, Plasma Carnitine, Serum Carnitine, Random, Urine
<b>FA</b>	TTRZ ACRN ACRNS AGU20 C4U CARN CARNS CARNS	Familial, Reflex, Blood TTR Gene, Full Gene Analysis METABOLISM (BETA-OXIDATION) Acylcarnitines, Quantitative, Plasma Acylcarnitines, Quantitative, Serum Acylglycines, Quantitative, Random, Urine C4 Acylcarnitine, Quantitative, Random, Urine Carnitine, Plasma Carnitine, Serum
<b>FA</b>	TTRZ ACRN ACRNS AGU20 C4U CARN CARNS CARNU HFAOP	Familial, Reflex, Blood TTR Gene, Full Gene Analysis METABOLISM (BETA-OXIDATION) Acylcarnitines, Quantitative, Plasma Acylcarnitines, Quantitative, Serum Acylglycines, Quantitative, Random, Urine C4 Acylcarnitine, Quantitative, Random, Urine Carnitine, Plasma Carnitine, Serum Carnitine, Random, Urine Fatty Acid Oxidation Gene Panel Fatty Acid Oxidation Probe Assay,
<b>FA</b>	TTRZ TTY ACIE ACRN ACRNS AGU20 C4U CARN CARN CARNS CARNU HFAOP FAO	Familial, Reflex, Blood TTR Gene, Full Gene Analysis METABOLISM (BETA-OXIDATION) Acylcarnitines, Quantitative, Plasma Acylcarnitines, Quantitative, Serum Acylglycines, Quantitative, Random, Urine C4 Acylcarnitine, Quantitative, Random, Urine Carnitine, Plasma Carnitine, Serum Carnitine, Random, Urine Fatty Acid Oxidation Gene Panel Fatty Acid Oxidation Probe Assay, Fibroblast Culture Fatty Acid Profile, Comprehensive
<b>FA</b>	TTRZ ACRN ACRNS AGU20 C4U CARN CARNS CARNU HFAOP FAO FAPCP	Familial, Reflex, Blood TTR Gene, Full Gene Analysis DMETABOLISM (BETA-OXIDATION) Acylcarnitines, Quantitative, Plasma Acylcarnitines, Quantitative, Random, Urine C4 Acylcarnitine, Quantitative, Random, Urine C4 Acylcarnitine, Quantitative, Random, Urine Carnitine, Plasma Carnitine, Serum Carnitine, Random, Urine Fatty Acid Oxidation Gene Panel Fatty Acid Oxidation Probe Assay, Fibroblast Culture Fatty Acid Profile, Comprehensive (C8-C26), Serum Fatty Acid Profile, Mitochondrial (C8-C18),
<b>FA</b>	TTRZ ACRN ACRNS AGU20 C4U CARN CARNS CARNU HFAOP FAO FAPCP FAPCP	Familial, Reflex, Blood TTR Gene, Full Gene Analysis METABOLISM (BETA-OXIDATION) Acylcarnitines, Quantitative, Plasma Acylcarnitines, Quantitative, Serum Acylglycines, Quantitative, Random, Urine C4 Acylcarnitine, Quantitative, Random, Urine Carnitine, Plasma Carnitine, Plasma Carnitine, Serum Carnitine, Random, Urine Fatty Acid Oxidation Gene Panel Fatty Acid Oxidation Probe Assay, Fibroblast Culture Fatty Acid Profile, Comprehensive (C8-C26), Serum Fatty Acid Profile, Mitochondrial (C8-C18), Serum Medium-Chain Acyl-CoA Dehydrogenase
	TTRZ ACRN ACRNS AGU20 C4U CARN CARNS CARNU HFAOP FAO FAO FAPCP FAPM MCADZ	Familial, Reflex, Blood TTR Gene, Full Gene Analysis METABOLISM (BETA-OXIDATION) Acylcarnitines, Quantitative, Plasma Acylcarnitines, Quantitative, Serum Acylglycines, Quantitative, Random, Urine C4 Acylcarnitine, Quantitative, Random, Urine Carnitine, Plasma Carnitine, Plasma Carnitine, Serum Carnitine, Serum Carnitine, Random, Urine Fatty Acid Oxidation Gene Panel Fatty Acid Oxidation Probe Assay, Fibroblast Culture Fatty Acid Profile, Comprehensive (C8-C26), Serum Fatty Acid Profile, Mitochondrial (C8-C18), Serum Medium-Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency Full Gene Analysis

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#### **ORGANIC ACID METABOLISM** 🗆 AGU20 Acylglycines, Quantitative, Random, Urine 3MGAP 3-Methylglutaconic Aciduria Gene Panel C50HU C5-OH Acylcarnitine, Quantitative, Random, Urine □ KETGP Ketone Disorders Gene Panel 🗆 OAU Organic Acids Screen, Random, Urine □ OAUS Organic Acid Screen, Urine Spot 2-Hydroxyglutaric Aciduria 20HGP 2-Hydroxyglutaric Aciduria Gene Panel 2HGA 2-Hydroxyglutaric Acid Chiral Analysis, Quantitative, Random, Urine **Biotinidase Deficiency** □ BIOTS Biotinidase, Serum □ BTDZ Biotinidase Defiency, BTD Full Gene Analysis **Glutaric Acidemia** C5DCU C5-DC Acylcarnitine, Quantitative, Random, Urine □ GA2P Glutaric Aciduria Type II Gene Panel □ HGEM Hydroxyglutaric Acids, Glutaric Acid, Ethylmalonic Acid, and Methylsuccinic Acid, Blood Spot HGEMP Hydroxyglutaric Acids, Glutaric Acid, Ethylmalonic Acid, and Methylsuccinic Acid, Plasma HGEMS Hydroxyglutaric Acids, Glutaric Acid, Ethylmalonic Acid, and Methylsuccinic Acid, Serum □ TRYPP Tryptophan, Plasma TRYPU Tryptophan, Random, Urine Methylmalonic Acidemia/Cobalamin/ **Propionic Acidemia** CMMPP Cobalamin, Methionine, and Methylmalonic Acid Pathways, Plasma CMMPS Cobalamin, Methionine, and Methylmalonic Acid Pathways, Serum MMAGP Methylmalonic Aciduria Gene Panel Methylmalonic Aciduria-Propionic Aciduria MPAGP Combined Gene Panel MMAP Methylmalonic Acid, Quantitative, Plasma □ MMAS Methylmalonic Acid, Quantitative, Serum 🗆 MMAU Methylmalonic Acid, Quantitative, Urine

### **GLYCOGEN STORAGE DISORDERS**

□ GSDGP	Glycogen Storage Disease Gene Panel
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### HYPEROXALURIA

🗆 нүох	Hyperoxaluria Panel, Random, Urine
□ RSCGP	Nephrocalcinosis, Nephrolithiasis, and Renal Electrolyte Imbalance Gene Panel

LYSOSOMAL METABOLISM		
AND STORAGE DISORDERS		
□ ctsu	Ceramide Trihexosides and Sulfatides, Random, Urine	
П немов	Hepatosplenomegaly Panel, Blood	
🗆 HSMP	Hepatosplenomegaly Panel, Plasma	
🗆 PLSD	Lysosomal and Peroxisomal Storage Disorders Screen, Blood Spot	
LSDGP	Lysosomal Storage Disease Gene Panel	
□ LSDS	Lysosomal Storage Disorders Screen, Random, Urine	
□ LSD6W	Lysosomal Storage Disorders, Six-Enzyme Panel, Leukocytes	
□ MPSBS	Mucopolysaccharidosis, Blood Spot	
□ mpsqu	Mucopolysaccharides Quantitative, Random, Urine	
□ MP8BS	Mucopolysaccharidoses, Eight-Enzyme Panel, Blood Spot	
□ MP9W	Mucopolysaccharidoses, Nine-Enzyme Panel, Leukocytes	
🗆 oligu	Oligosaccharide Screen, Random, Urine	
□ OXNP	Oxysterols, Plasma	
Fabry Disea	se	
🗆 FABRZ	Fabry Disease, Full Gene Analysis	
🗆 AGABS	Alpha-Galactosidase, Blood Spot	
🗆 AGAW	Alpha-Galactosidase, Leukocytes	
🗆 AGAS	Alpha-Galactosidase, Serum	
🗆 CTSU	Ceramide Trihexosides and Sulfatides, Random, Urine	
🗆 LGB3S	Globotriaosylsphingosine, Serum	
Fucosidosis	5	
□ FUCW	Alpha-Fucosidase, Leukocytes	
Gaucher Dis	sease	
🗆 GBAW	Beta-Glucosidase, Leukocytes	
🗆 GBAZ	Gaucher Disease, Full Gene Analysis	
□ GPSYW	Glucopsychosine, Blood	
GPSY	Glucopsychosine, Blood Spot	
□ GPSYP	Glucopsychosine, Plasma	
GM1 Gangliosidosis		
🗆 BGA	Beta-Galactosidase, Leukocytes	
□ MPS4B	Mucopolysaccharidosis IV Enzyme Panel, Blood Spot	
□ MPS4W	Mucopolysaccharidosis IV Enzyme Panel, Leukocytes	

Krabbe Disease	
□ GALCW	Galactocerebrosidase, Leukocytes
🗆 KRABZ	Krabbe Disease, Full Gene Analysis and Large (30 kb) Deletion
D PSY	Psychosine, Blood Spot
D PSYCF	Psychosine, Spinal Fluid
D PSYR	Psychosine, Whole Blood
Lysosomal Acid Lipase Deficiency	
🗆 LALB	Lysosomal Acid Lipase, Blood
□ LALBS	Lysosomal Acid Lipase, Blood Spot
Mannosidos	sis
MANN	Alpha-Mannosidase, Leukocytes
Metachrom	atic Leukodystrophy
🗆 ARSU	Arylsulfatase A, 24 Hour, Urine
□ ARSAW	Arylsulfatase A, Leukocytes
CTSU	Ceramide Trihexosides and Sulfatides, Random, Urine
Mucopolysa	accharidoses (MPS)
□ mpsqu	Mucopolysaccharides Quantitative, Random, Urine
MPSER	Mucopolysaccharides Quantitative, Serum
□ MPSWB	Mucopolysaccharidosis, Blood
□ MPSBS	Mucopolysaccharidosis, Blood Spot
MPS Type I	(Hurler/Scheie syndrome)
🗆 iduaw	Alpha-L-Iduronidase, Leukocytes
D MPS1Z	Hurler Syndrome, Full Gene Analysis
MPS Type II	(Hunter syndrome)
□ MPS2Z	Hunter Syndrome, Full Gene Analysis
🗆 I2SB	Iduronate-2-Sulfatase, Blood Spot
□ I2SWB Iduronate-2-Sulfatase, Leukocytes	
MPS Type II	I (Sanfilippo syndrome)
□ MPS3B	Mucopolysaccharidosis III, Three-Enzyme Panel, Blood Spot
□ MPS3W	Mucopolysaccharidosis III, Four-Enzyme Panel, Leukocytes
MPS Type I	/ (Morquio syndrome)
🗆 BGA	Beta-Galactosidase, Leukocytes
□ MPS4B	Mucopolysaccharidosis IV Enzyme Panel, Blood Spot
□ MPS4W	Mucopolysaccharidosis IV Enzyme Panel, Leukocytes
MPS VI (Maroteaux-Lamy syndrome)	
□ ARSBB	Arylsulfatase B, Blood Spot
□ ARSBW	Arylsulfatase B, Leukocytes
MPS VII (Sly syndrome)	
GUSBW	Beta-Glucuronidase, Leukocytes
□ GUSBB	Beta-Glucuronidase, Blood Spot

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Multiple Sulfatase Deficiency	
□ MSDBS	Multiple Sulfatase Deficiency, Blood Spot
MSDW	Multiple Sulfatase Deficiency, Leukocytes
Niemann-Pick Types A and B	
🗆 asmw	Acid Sphingomyelinase, Leukocytes
□ OXNP	Oxysterols, Plasma
Niemann-P	ick Type C
□ OXNP	Oxysterols, Plasma
Neuronal C	eroid Lipofuscinoses
□ NCLGP	Neuronal Ceroid Lipofuscinosis (Batten Disease) Gene Panel
□ NCLBS	Neuronal Ceroid Lipofuscinosis, Two-Enzyme Panel, Blood Spot
	Neuronal Ceroid Lipofuscinosis, Two-Enzyme Panel, Leukocytes
Pompe Dise	ease
🗆 GAAW	Acid Alpha-Glucosidase, Leukocytes
🗆 GAAZ	Pompe Disease Full Gene Analysis
□ HEX4	Glucotetrasaccharides, Random, Urine
D PDBS	Pompe Disease, Blood Spot
Tay-Sachs a	nd Sandhoff Diseases
□ NAGW	Hexosaminidase A and Total Hexosaminidase, Leukocytes
□ NAGS	Hexosaminidase A and Total Hexosaminidase, Serum
□ NAGR	Hexosaminidase A and Total, Leukocytes/Molecular Reflex, Whole Blood
□ MUGS	Hexosaminidase A, Serum
□ HEXBZ	Sandhoff Disease, HEXB Gene, Full Gene Analysis
□ HEXAZ	Tay-Sachs Disease, HEXA Gene, Full Gene Analysis
MITOCHONDRIAL DISEASES	
	Coenzyme O10 Reduced and Total Plasma

🗆 Q10	Coenzyme Q10, Reduced and Total, Plasma
□ TQ10	Coenzyme Q10, Total, Plasma
🗆 ғарм	Fatty Acid Profile, Mitochondrial (C8-C18), Serum
🗆 сміто	Combined Mitochondrial Full Genome and Nuclear Gene Panel
	Mitochondrial DNA Deletion Heteroplasmy, ddPCR
GDF15	Growth Differentiation Factor 15, Plasma
□ LAPYP	Lactate Pyruvate Panel, Plasma
	Mitochondrial Full Genome Analysis, Next-Generation Sequencing (NGS)
	Mitochondrial Metabolites, Plasma
	Nuclear Mitochondrial Gene Panel, Next- Generation Sequencing
D OAU	Organic Acids Screen, Random, Urine
D PYRC	Pyruvate, Spinal Fluid
D PYR	Pyruvic Acid, Blood

NEUROLO	GIC DISORDERS
□ FFRWB	Friedreich Ataxia, Frataxin, Quantitative, Blood
□ FFRBS	Friedreich Ataxia, Frataxin, Quantitative, Blood Spot
🗆 AFXN	Friedreich Ataxia, Repeat Expansion Analysis
□ SORD	Sorbitol and Xylitol, Quantitative, Random, Urine
NEWBORN	N SCREENING
Screening F	Panels
🗆 LDALD	Lysosomal and Peroxisomal Disorders Newborn Screen, Blood Spot
□ SNS	Supplemental Newborn Screen, Blood Spot
Second Tie	r Tests
🗆 ALLOI	Allo-isoleucine, Blood Spot
🗆 САН2Т	Congenital Adrenal Hyperplasia Newborn Screen, Blood Spot
GPSY	Glucopsychosine, Blood Spot
□ нсмм	Homocysteine (Total), Methylmalonic Acid, and Methylcitric Acid, Blood Spot
□ HGEM	Hydroxyglutaric Acids, Glutaric Acid, Ethylmalonic Acid, and Methylsuccinic Acid, Blood Spot
	Lysophosphatidylcholines, LC MS/MS, Blood Spot
	Mucopolysaccharidosis, Blood Spot
□ OXYBS	Oxysterols, Blood Spot
□ PD2T	Pompe Disease Second-Tier Newborn Screening, Blood Spot
D PSY	Psychosine, Blood Spot
	Succinylacetone, Blood Spot
PEROXISO	MAL BIOGENESIS & METABOLISM
	Bile Acids for Peroxisomal Disorders, Serum
	Fatty Acid Profile, Peroxisomal (C22-C26), Plasma
🗆 РОХ	Fatty Acid Profile, Peroxisomal (C22-C26),

PORPHYRI	PORPHYRIAS	
Urine		
	Aminolevulinic Acid, Urine	
D PBGU	Porphobilinogen, Quantitative, Random, Urine	
🗆 PQNU	Porphyrins, Quantitative, 24 Hour, Urine	
D PQNRU	Porphyrins, Quantitative, Random, Urine	
Plasma		
PBALP	Porphobilinogen and Aminolevulinic Acid, Plasma	
D PTP	Porphyrins, Total, Plasma	
Fecal		
□ FQPPS	Porphyrins, Feces	
Blood		
PEWE	Porphyrins Evaluation, Washed Erythrocytes	
D PEE	Porphyrins Evaluation, Whole Blood	
PPFWE	Protoporphyrins, Fractionation, Washed Erythrocytes	
PPFE	Protoporphyrins, Fractionation, Whole Blood	
Enzymes		
D PBGDW	Porphobilinogen Deaminase, Washed Erythrocytes	
D PBGD_	Porphobilinogen Deaminase, Whole Blood	
UPGC	Uroporphyrinogen III Synthase (Co-Synthase), Erythrocytes	
UPGDW	Uroporphyrinogen Decarboxylase, Washed Erythrocytes	
🗆 UPGD	Uroporphyrinogen Decarboxylase, Whole Blood	
Molecular		
□ APGP	Acute Porphyria Gene Panel	
D PCGP	Porphyria Comprehensive Gene Panel	
POSTMOR	TEM BIOCHEMICAL TESTING	
D PMSBB	Postmortem Screening, Bile and Blood Spot	
PURINE AN	ND PYRIMIDINE METABOLISM	
D PUPYP	Purine and Pyrimidine Panel, Plasma	
D PUPYU	Purine and Pyrimidine Panel, Random, Urine	
□ ssctu	S-Sulfocysteine Panel, Urine	
SIALIC AC	D DISORDERS	
SAU	Sialic Acid, Free and Total, Random, Urine	

Peroxisomal Disorder Gene Panel

Pipecolic Acid, Random, Urine

Pipecolic Acid, Serum

Plasmalogens, Blood

Plasmalogens, Blood Spot □ XALDZ X-Linked Adrenoleukodystrophy, Full Gene Analysis

Serum

D PDGP

🗆 PIPA

PIPU

D PGRBC

D PGDBS

N 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1	
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UREA CYC	LE DISORDERS
□ AAQP	Amino Acids, Quantitative, Plasma
🗆 AAPD	Amino Acids, Quantitative, Random, Urine
	Amino Acids, Urea Cycle Disorders Panel, Plasma
🗆 oau	Organic Acids Screen, Random, Urine
□ OROT	Orotic Acid, Random, Urine
UCDP	Urea Cycle Disorders Gene Panel
WILSON D	ISEASE
	Ceruloplasmin, Serum
🗆 CUU	Copper, 24 Hr, Urine
CUS1	Copper, Serum
□ WNDZ	Wilson Disease, ATP7B Full Gene Sequencing with Deletion/Duplication
WHOLE EX	KOME
U WESMT	Whole Exome and Mitochondrial Genome Sequencing
□ WESDX	Whole Exome Sequencing for Hereditary Disorders
□ WESR	Whole Exome Sequencing Reanalysis
WHOLE G	ENOME
	Whole Genome Sequencing for Hereditary Disorders

□ WGSR Whole Genome Sequencing Reanalysis

ADDITIONAL TESTS (INDICATE TEST ID AND NAME)