

Inborn Errors of Metabolism Test Request

Client Information (required)

Client Name		
Client Account No.		
Client Phone	Client Order No.	
Address		
City	State	Zip Code

Submitting Provider/Provider Name Information (required)

Submitting/Referring Provider <i>(Last, First)</i>
Fill in only if Call Back is required. Phone () _____ - _____ Fax * () _____ - _____
Provider's National I.D. (NPI)

**Fax number given must be from a fax machine that complies with applicable HIPAA regulation.*

Reason for Referral (required)

ICD-10 Diagnosis Code

Note: It is the client's responsibility to maintain documentation of the order.

New York State Patients: Informed Consent for Genetic Testing

<p>"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."</p> <p>Signature _____</p>
--

Note: It is the client's responsibility to maintain documentation of the order.

Ship specimens to:

Mayo Clinic Laboratories
3050 Superior Drive NW
Rochester, MN 55901

Customer Service: 855-516-8404

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

Patient Information (required)

Patient ID <i>(Medical Record No.)</i>		
Patient Name <i>(Last, First, Middle)</i>		
Gender <input type="checkbox"/> Male <input type="checkbox"/> Female	Birth Date <i>(Month DD, YYYY)</i>	
Collection Date <i>(Month DD, YYYY)</i>	Time <input type="checkbox"/> a.m. <input type="checkbox"/> p.m.	
Patient's Street Address		
Phone		
City	State	Zip Code

MCL Internal Use Only

Billing Information

- An itemized invoice will be sent each month.
- 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)

Provider Name

Provider Name

Patient Information (required)

Patient Name (Last, First, Middle)	
Birth Date (Month DD, YYYY)	ICD-10 Diagnosis Code

AMINO ACID METABOLISM

- AAMSD Amino Acids, Maple Syrup Urine Disease Panel, Plasma
- AAQP Amino Acids, Quantitative, Plasma
- AAPD Amino Acids, Quantitative, Random, Urine
- AACSF Amino Acids, Quantitative, Spinal Fluid
- HCMM Homocysteine (Total), Methylmalonic Acid, and Methylcitric Acid, Blood Spots
- HCYSS Homocysteine, Total, Serum
- HCYSU Homocysteine, Total, Urine
- 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
- OAU Organic Acids Screen, Urine
- SUAC Succinylacetone, Blood Spot
- TRYPP Tryptophan, Plasma
- TRYPU Tryptophan, Urine

Cystinuria

- CYSQN Cystinuria Profile, Quantitative, 24 Hour, Urine
- CYSR Cystinuria Profile, Quantitative, Random, Urine

Glutamate Formiminotransferase Deficiency

- GFDZ FTCD Gene, Full Gene Analysis

Maple Syrup Urine Disease

- ALLOI Allo-isoleucine, Blood Spot

Methylmalonic Acidemia & Homocystinuria

- MMAP Methylmalonic Acid (MMA), Quantitative, Plasma
- MMAS Methylmalonic Acid (MMA), Quantitative, Serum
- MMAU Methylmalonic Acid (MMA), Quantitative, Urine
- MHCZ Methylmalonic Aciduria and Homocystinuria, cbIC Type, Full Gene Analysis
- MHDZ Methylmalonic Aciduria and Homocystinuria, cbID Type, Full Gene Analysis

Phenylketonuria

- PKU Phenylalanine and Tyrosine, Plasma
- PKUBS Phenylalanine and Tyrosine, Blood Spot

CARBOHYDRATE METABOLISM

- CHOU Carbohydrate, Urine
 - GALP Galactose, Quantitative, Plasma
 - GALU Galactose, Quantitative, Urine
- #### Congenital Disorders of Glycosylation
- CDG Carbohydrate Deficient Transferrin for Congenital Disorders of Glycosylation, Serum
 - CDGP Congenital Disorders of Glycosylation Genetic Panels by Next-Generation Sequencing (NGS)
 - PMMIL Phosphomannomutase (PMM) and Phosphomannose Isomerase (PMI), Leukocytes

Galactosemia

- GATOL Galactitol, Quantitative, Urine
- GALK Galactokinase, Blood
- GAL1P Galactose-1-Phosphate (Gal-1-P), Erythrocytes
- GALT Galactose-1-Phosphate Uridyltransferase (GALT), Blood
- GALT1P Galactose-1-Phosphate Uridyltransferase Biochemical Phenotyping, Erythrocytes
- GAL14 Galactosemia Gene Analysis (14-Mutation Panel)
- GCT Galactosemia Reflex, Blood
- GALTZ GALT Gene, Full Gene Analysis
- GALE UDP-Galactose 4' Epimerase (GALE), Blood

Transaldolase and Ribose-5-phosphate (RPI) Deficiencies

- TALDO Polyols, Quantitative, Urine

CHOLESTEROL BIOSYNTHESIS & TRANSPORT

- CTXWB Cerebrotendinous Xanthomatosis, Blood
- CTXP Cerebrotendinous Xanthomatosis, Plasma
- GPSYW Glucopsychosine, Blood
- GPSYP Glucopsychosine, Plasma
- HSMBS Hepatosplenomegaly Panel, Blood Spot
- HSMWB Hepatosplenomegaly Panel, Blood
- HSMP Hepatosplenomegaly Panel, Plasma
- NIEM Niemann-Pick Type C Detection, Fibroblasts
- NPCZ Niemann-Pick Type C Disease, Full Gene Analysis
- OXYWB Oxysterols, Blood
- OXYBS Oxysterols, Blood Spots
- OXNP Oxysterols, Plasma
- SLO Smith-Lemli-Opitz Screen, Plasma
- STER Sterols, Plasma

CONGENITAL ADRENAL HYPERPLASIA

- CYPZ 21-Hydroxylase Gene (CYP21A2), Full Gene Analysis
- CAH21 Congenital Adrenal Hyperplasia Profile for 21-Hydroxylase Deficiency

CREATINE DISORDERS

- CRDPU Creatine Disorders Panel, Urine

FATTY ACID METABOLISM (BETA-OXIDATION) & ORGANIC ACID DISORDERS

- ACRN Acylcarnitines, Quantitative, Plasma
- ACRNS Acylcarnitines, Quantitative, Serum
- ACYLG Acylglycines, Quantitative, Urine
- AAQP Amino Acids, Quantitative, Plasma
- AAPD Amino Acids, Quantitative, Random, Urine
- AACSF Amino Acids, Quantitative, Spinal Fluid
- C4U C4 Acylcarnitine, Quantitative, Urine
- C5DCU C5-DC Acylcarnitine, Quantitative, Urine
- C5OHU C5-OH Acylcarnitine, Quantitative, Urine
- CARN Carnitine, Plasma
- CARNS Carnitine, Serum
- CARNU Carnitine, Urine
- FAO Fatty Acid Oxidation Probe Assay, Fibroblast Culture
- FAPCP Fatty Acid Profile, Comprehensive (C8-C26), Serum
- FAPEP Fatty Acid Profile, Essential, Serum
- FAPM Fatty Acid Profile, Mitochondrial (C8-C18), Serum
- HCYSU Homocysteine, Total, Urine
- OAU Organic Acids Screen, Urine
- PMSBB Postmortem Screening, Bile and Blood Spots

FATTY ACID METABOLISM (BETA-OXIDATION) & ORGANIC ACID DISORDERS

Biotinidase Deficiency

- BTDZ Biotinidase Deficiency, BTD Full Gene Analysis
- BIOTS Biotinidase, Serum

Carnitine-Acylcarnitine Translocase Deficiency

- CACTZ Carnitine-Acylcarnitine Translocase Deficiency, Full Gene Analysis

Carnitine Palmitoyltransferase II Deficiency

- CPT2Z Carnitine Palmitoyltransferase II Deficiency, Full Gene Analysis

Isovaleric Acidemia

- IVDA Isovaleryl-CoA Dehydrogenase (IVD) Gene Mutation Analysis (A282V)

Provider Name

Provider Name

Patient Information (required)

Patient Name (Last, First, Middle)	
Birth Date (Month DD, YYYY)	ICD-10 Diagnosis Code

FATTY ACID METABOLISM (BETA-OXIDATION) & ORGANIC ACID DISORDERS	
Malonyl-Coenzyme A Decarboxylase Deficiency	
<input type="checkbox"/> MLYCZ	MLYCD Gene, Full Gene Analysis
Medium-Chain Acyl-CoA Dehydrogenase Deficiency	
<input type="checkbox"/> MCADZ	Medium-Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency Full Gene Analysis
Short-Chain Acyl-CoA Dehydrogenase Deficiency	
<input type="checkbox"/> SCADZ	Short-Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency, Full Gene Analysis
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	
<input type="checkbox"/> VLCZ	Very Long Chain Acyl-CoA Dehydrogenase Deficiency, Full Gene Analysis

FREDREICH ATAXIA	
<input type="checkbox"/> FFRBS	Friedreich Ataxia, Frataxin, Quantitative, Blood Spot
<input type="checkbox"/> FFRWB	Friedreich Ataxia, Frataxin, Quantitative, Whole Blood

HYPEROXALURIA	
<input type="checkbox"/> AGXTG	Alanine:Glyoxylate Aminotransferase (AGXT) Mutation Analysis (G170R), Blood
<input type="checkbox"/> AGXTZ	AGXT Gene, Full Gene Analysis
<input type="checkbox"/> GRHPZ	GRHPR Gene, Full Gene Analysis
<input type="checkbox"/> HYOX	Hyperoxaluria Panel, Urine

LYSOSOMAL METABOLISM & STORAGE DISORDERS	
Multi-Disorder Panels	
<input type="checkbox"/> CTSA	Ceramide Trihexosides and Sulfatides, Urine
<input type="checkbox"/> GSDP	Glycogen Storage Disease Panel by Next-Generation Sequencing
<input type="checkbox"/> PLSD	Lysosomal and Peroxisomal Storage Disorders Screen, Blood Spot
<input type="checkbox"/> LSDP	Lysosomal Storage Disease Panel by Next-Generation Sequencing
<input type="checkbox"/> LYSDU	Lysosomal Storage Disorders Screen, Urine
<input type="checkbox"/> MPSSC	Mucopolysaccharides (MPS) Screen, Urine
<input type="checkbox"/> MPSQN	Mucopolysaccharides (MPS), Quantitative, Urine
<input type="checkbox"/> SFPAN	Mucopolysaccharidosis III, Multi-Gene Panel
<input type="checkbox"/> OLIGU	Oligosaccharide Screen, Urine
Fabry Disease	
<input type="checkbox"/> AGABS	Alpha-Galactosidase, Blood Spot
<input type="checkbox"/> AGA	Alpha-Galactosidase, Leukocytes
<input type="checkbox"/> AGAS	Alpha-Galactosidase, Serum
<input type="checkbox"/> CTSA	Ceramide Trihexosides and Sulfatides, Urine
<input type="checkbox"/> FABRZ	Fabry Disease, Full Gene Analysis

<input type="checkbox"/> LGBWB	Globotriaosylsphingosine, Blood
<input type="checkbox"/> LGBBS	Globotriaosylsphingosine, Blood Spot
<input type="checkbox"/> LGB3S	Globotriaosylsphingosine, Serum
Fucosidosis	
<input type="checkbox"/> FUCW	Alpha-Fucosidase, Leukocytes
Gaucher Disease	
<input type="checkbox"/> BGL	Beta-Glucosidase, Leukocytes
<input type="checkbox"/> GBAZ	Gaucher Disease, Full Gene Analysis
<input type="checkbox"/> GAUP	Gaucher Disease, Mutation Analysis, GBA
<input type="checkbox"/> GPSY	Glucopsychosine, Blood Spot
GM1 Gangliosidosis	
<input type="checkbox"/> BGAW	Beta-Galactosidase, Blood
<input type="checkbox"/> BGABS	Beta-Galactosidase, Blood Spot
<input type="checkbox"/> BGA	Beta-Galactosidase, Leukocytes
Krabbe Disease	
<input type="checkbox"/> CBGC	Galactocerebrosidase, Leukocytes
<input type="checkbox"/> KRABZ	Krabbe Disease, Full Gene Analysis and Large (30 kb) Deletion, PCR
<input type="checkbox"/> PSYWB	Psychosine, Blood
<input type="checkbox"/> PSY	Psychosine, Blood Spot
Lysosomal Acid Lipase Deficiency	
<input type="checkbox"/> LALB	Lysosomal Acid Lipase, Blood
<input type="checkbox"/> LALBS	Lysosomal Acid Lipase, Blood Spot
Mannosidosis	
<input type="checkbox"/> MANN	Alpha-Mannosidase, Leukocytes
Metachromatic Leukodystrophy	
<input type="checkbox"/> ARSAZ	ARSA Gene, Full Gene Analysis
<input type="checkbox"/> ARSU	Arylsulfatase A, 24 Hour, Urine
<input type="checkbox"/> ARSAW	Arylsulfatase A, Leukocytes
<input type="checkbox"/> CTSA	Ceramide Trihexosides and Sulfatides, Urine
MPS Type I (Hurler/Scheie)	
<input type="checkbox"/> IDSWB	Alpha-L-Iduronidase, Blood
<input type="checkbox"/> IDSBS	Alpha-L-Iduronidase, Blood Spot
<input type="checkbox"/> MPS1Z	Hurler Syndrome, Full Gene Analysis
<input type="checkbox"/> MPSWB	Mucopolysaccharidosis, Blood
<input type="checkbox"/> MPSBS	Mucopolysaccharidosis, Blood Spot
MPS Type II (Hunter)	
<input type="checkbox"/> MPS2Z	Hunter Syndrome, Full Gene Analysis
<input type="checkbox"/> I2SBS	Iduronate-2-Sulfatase, Blood Spot
<input type="checkbox"/> I2SW	Iduronate-2-Sulfatase, Whole Blood
<input type="checkbox"/> MPSWB	Mucopolysaccharidosis, Blood
<input type="checkbox"/> MPSBS	Mucopolysaccharidosis, Blood Spot
MPS Type IIIA (Sanfilippo Type A)	
<input type="checkbox"/> MP3AZ	Mucopolysaccharidosis IIIA, Full Gene Analysis

MPS Type IIIB (Sanfilippo Type B)	
<input type="checkbox"/> ANAS	Alpha-N-Acetylglucosaminidase, Serum
<input type="checkbox"/> MP3BZ	Mucopolysaccharidosis IIIB, Full Gene Analysis
MPS Type IIIC (Sanfilippo Type C)	
<input type="checkbox"/> MP3CZ	Mucopolysaccharidosis IIIC, Full Gene Analysis
MPS Type IIID (Sanfilippo Type D)	
<input type="checkbox"/> MP3DZ	Mucopolysaccharidosis IIID, Full Gene Analysis
MPS Type IVA (Morquio A)	
<input type="checkbox"/> G6SW	N-Acetylgalactosamine-6-Sulfatase, Leukocytes
<input type="checkbox"/> G6ST	N-Acetylgalactosamine-6-Sulfate Sulfatase, Fibroblasts
MPS Type IVB (Morquio B)	
<input type="checkbox"/> BGAW	Beta-Galactosidase, Blood
<input type="checkbox"/> BGABS	Beta-Galactosidase, Blood Spot
<input type="checkbox"/> BGA	Beta-Galactosidase, Leukocytes
MPS Type VI (Maroteaux-Lamy)	
<input type="checkbox"/> MPS6Z	Mucopolysaccharidosis VI, Full Gene Analysis
<input type="checkbox"/> ARSB	Arylsulfatase B, Fibroblasts
Mucopolidoses	
<input type="checkbox"/> GNPTZ	GNPTAB Gene, Full Gene Analysis
<input type="checkbox"/> MCIVP	Mucopolidosis IV, Mutation Analysis, IVS3(-2) A->G and del6.4kb
Multiple Sulfatase Deficiency	
<input type="checkbox"/> SUMFZ	Multiple Sulfatase Deficiency, Full Gene Analysis
Niemann-Pick Types A&B	
<input type="checkbox"/> NPABZ	Niemann-Pick Disease, Types A and B, Full Gene Analysis
<input type="checkbox"/> NPABP	Niemann-Pick Disease, Types A and B, Mutation Analysis
<input type="checkbox"/> OXYBS	Oxysterols, Blood Spot
<input type="checkbox"/> OXNP	Oxysterols, Plasma
Neuronal Ceroid Lipofuscinoses	
<input type="checkbox"/> NCLP	Neuronal Ceroid Lipofuscinosis (NCL, Batten Disease) Panel by Next-Generation Sequencing
<input type="checkbox"/> TPPTF	Tripeptidyl Peptidase 1 (TPP1) and Palmitoyl-Protein Thioesterase 1 (PPT1), Fibroblasts
<input type="checkbox"/> TPPTL	Tripeptidyl Peptidase 1 (TPP1) and Palmitoyl-Protein Thioesterase 1 (PPT1), Leukocytes

Provider Name

Provider Name

Patient Information (required)

Patient Name (Last, First, Middle)	
Birth Date (Month DD, YYYY)	ICD-10 Diagnosis Code

Pompe Disease

- HEX4 Glucotetrasaccharides, Urine
- PDBS Pompe Disease, Blood Spot
- GAAZ Pompe Disease, Full Gene Analysis
- PD2T Pompe Disease Second-Tier Newborn Screening, Blood Spot

Sialidosis

- NEURF Neuraminidase, Fibroblasts

Tay-Sachs & Sandhoff Diseases

- MUGS Hexosaminidase A (MUGS), Serum
- NAGW Hexosaminidase A and Total Hexosaminidase, Leukocytes
- NAGS Hexosaminidase A and Total Hexosaminidase, Serum
- NAGR Hexosaminidase A and Total, Leukocytes/Molecular Reflex
- HEXAZ Tay-Sachs Disease, HEXA Gene, Full Gene Analysis

MITOCHONDRIAL ENERGY METABOLISM

- Q10 Coenzyme Q10, Reduced and Total, Plasma
- TQ10 Coenzyme Q10, Total, Plasma
- MITOT Combined Mitochondrial Analysis, Mitochondrial Full Genome and Nuclear Gene Panel
- GDF15 Growth Differentiation Factor 15 (GDF15), Plasma
- LAA Lactate, Plasma
- LABF Lactate, Body Fluid
- MITON Mitochondrial Nuclear Gene Panel by Next-Generation Sequencing (NGS)
- MITOP Mitochondrial Full Genome Analysis by Next-Generation Sequencing (NGS)
- PDHC Pyruvate Dehydrogenase Complex (PDHC), Fibroblasts
- PYRC Pyruvate, Spinal Fluid
- PYR Pyruvic Acid, Blood

NEWBORN SCREENING

Screening Panels

- LDALD Lysosomal and Peroxisomal Disorders Newborn Screen, Blood Spot
- NBSE Newborn Screening Expanded Panel, Blood Spot
- NBSR Newborn Screen Recommended Panel, Blood Spot
- SMNDX Spinal Muscular Atrophy Diagnostic Assay by Deletion/Duplication Analysis
- SNS Supplemental Newborn Screen, Blood Spot

Second Tier Tests

- ALLOI Allo-isoleucine, Blood Spot
- CAH2T Congenital Adrenal Hyperplasia (CAH) Newborn Screen, Blood Spot
- GPSY Glucopsychosine, Blood Spot
- HCMM Homocysteine (Total), Methylmalonic Acid, and Methylcitric Acid, Blood Spot
- HGEM Hydroxyglutaric Acids, Glutaric Acid, Ethylmalonic Acid, and Methylsuccinic Acid, Blood Spot
- KD2T Krabbe Disease Second-Tier Newborn Screen, Blood Spot
- LPCBS Lysophosphatidylcholines by LC MS/MS, Blood Spot
- MPSBS Mucopolysaccharidosis, Blood Spot
- OXYBS Oxysterols, Blood Spot
- PD2T Pompe Disease Second-Tier Newborn Screening, Blood Spot
- PSY Psychosine, Blood Spot
- SUAC Succinylacetone, Blood Spot

PEROXISOMAL BIOGENESIS & METABOLISM

- BAIPD Bile Acids for Peroxisomal Disorders, Serum
- POXP Fatty Acid Profile, Peroxisomal (C22-C26), Plasma
- POX Fatty Acid Profile, Peroxisomal (C22-C26), Serum
- PDP Peroxisomal Disorder Panel by Next-Generation Sequencing
- PIPA Pipecolic Acid, Serum
- PIPU Pipecolic Acid, Urine
- XALDZ X-Linked Adrenoleukodystrophy, Full Gene Analysis

PORPHYRIAS

- APPAN Acute Porphyria, Multi-Gene Panel
- PBGU Porphobilinogen, Quantitative, Random, Urine
- FQPPS Porphyrins, Feces
- PQNU Porphyrins, Quantitative, 24 Hour, Urine
- PQNRU Porphyrins, Quantitative, Random, Urine
- PTP Porphyrins, Total, Plasma

Aminolevulinic Acid Dehydratase Deficiency Porphyria

- ALAUR Aminolevulinic Acid (ALA), Urine
- ALADW Aminolevulinic Acid Dehydratase (ALA-D), Washed Erythrocytes
- ALAD Aminolevulinic Acid Dehydratase (ALAD), Whole Blood

Acute Intermittent Porphyria

- HMBSZ HMBS Gene, Full Gene Analysis
- PBGDW Porphobilinogen Deaminase (PBGD), Washed Erythrocytes
- PBGD_ Porphobilinogen Deaminase (PBGD), Whole Blood

Congenital Erythropoietic Porphyria

- UPGC Uroporphyrinogen III Synthase (Co-Synthase) (UPG III S), Erythrocytes

Erythropoietic Protoporphria

- FECHZ Ferrochelatase (FECH) Gene, Full Gene Analysis
- PEWE Porphyrins Evaluation, Washed Erythrocytes
- PEE Porphyrins Evaluation, Whole Blood
- PPFWE Protoporphyrins, Fractionation, Washed Erythrocytes
- PPFE Protoporphyrins, Fractionation, Whole Blood

Hereditary Coproporphria

- CPOXZ CPOX Gene, Full Gene Analysis

Porphyria Cutanea Tarda

- UPGDW Uroporphyrinogen Decarboxylase (UPG D), Washed Erythrocytes
- UPGD Uroporphyrinogen Decarboxylase (UPG D), Whole Blood

Variegate Porphyria

- PPOXZ PPOX Gene, Full Gene Analysis

X-linked Dominant Protoporphria

- PPFWE Protoporphyrins, Fractionation, Washed Erythrocytes
- PPFE Protoporphyrins, Fractionation, Whole Blood

PURINE & PYRIMIDINE METABOLISM & UREA CYCLE DISORDERS

- AAPD Amino Acids, Quantitative, Random, Urine
- AAUCD Amino Acids, Urea Cycle Disorders Panel, Plasma
- OAU Organic Acids Screen, Urine
- OROT Orotic Acid, Urine
- PUPYP Purine and Pyrimidine Panel, Plasma
- PUPYU Purine and Pyrimidine Panel, Urine

WILSON DISEASE

- CUU Copper, 24 Hour, Urine
- CUS Copper, Serum
- WDZ Wilson Disease, Full Gene Analysis

ADDITIONAL TESTS (INDICATE TEST NUMBER AND NAME)

--