Complete, print and submit

Reset Form

MAYO CLINIC LABORATORIES

Inborn Errors of Metabolism Test Request

Client Information (required)

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

Patient Information (required)

Patient ID *(Medical Record No.)*		
Patient Name *(Last, First, Middle)*		
Gender ☐ Male ☐ Female	Birth Date *(Month DD, YYYY)*	
Collection Date *(Month DD, YYYY)*	Time ☐ a.m. ☐ p.m.	
Patient's Street Address		
Phone		
City	State	Zip Code

Submitting Provider/Provider Name Information (required)

Submitting/Referring Provider *(Last, First)*
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.*

MCL Internal Use Only

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

"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: 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

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)

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Provider Name

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Patient Information (required)

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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, cblC Type, Full Gene Analysis
- ☐ MHDZ Methylmalonic Aciduria and Homocystinuria, cblD 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
- ☐ GALTP 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)

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

Malonyl-Coenzyme A Decarboxylase Deficiency

- ☐ MLYCZ MLYCD Gene, Full Gene Analysis

Medium-Chain Acyl-CoA Dehydrogenase Deficiency

- ☐ MCADZ Medium-Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency Full Gene Analysis

Short-Chain Acyl-CoA Dehydrogenase Deficiency

- ☐ SCADZ Short-Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency, Full Gene Analysis

Very Long Chain Acyl-CoA Dehydrogenase Deficiency

- ☐ VLCZ Very Long Chain Acyl-CoA Dehydrogenase Deficiency, Full Gene Analysis

FREDREICH ATAXIA

- ☐ FFRBS Friedreich Ataxia, Frataxin, Quantitative, Blood Spot
- ☐ FFRWB Friedreich Ataxia, Frataxin, Quantitative, Whole Blood

HYPEROXALURIA

- ☐ AGXTG Alanine:Glyoxylate Aminotransferase (AGXT) Mutation Analysis (G170R), Blood
- ☐ AGXTZ AGXT Gene, Full Gene Analysis
- ☐ GRHPZ GRHPR Gene, Full Gene Analysis
- ☐ HYOX Hyperoxaluria Panel, Urine

LYSOSOMAL METABOLISM & STORAGE DISORDERS

Multi-Disorder Panels

- ☐ CTSA Ceramide Trihexosides and Sulfatides, Urine
- ☐ GSDP Glycogen Storage Disease Panel by Next-Generation Sequencing
- ☐ PLSD Lysosomal and Peroxisomal Storage Disorders Screen, Blood Spot
- ☐ LSDP Lysosomal Storage Disease Panel by Next-Generation Sequencing
- ☐ LYSDU Lysosomal Storage Disorders Screen, Urine
- ☐ MPSSC Mucopolysaccharides (MPS) Screen, Urine
- ☐ MPSQN Mucopolysaccharides (MPS), Quantitative, Urine
- ☐ SFPAN Mucopolysaccharidosis III, Multi-Gene Panel
- ☐ OLIGU Oligosaccharide Screen, Urine

Fabry Disease

- ☐ AGABS Alpha-Galactosidase, Blood Spot
- ☐ AGA Alpha-Galactosidase, Leukocytes
- ☐ AGAS Alpha-Galactosidase, Serum
- ☐ CTSA Ceramide Trihexosides and Sulfatides, Urine
- ☐ FABRZ Fabry Disease, Full Gene Analysis
- ☐ LGBWB Globotriaosylsphingosine, Blood
- ☐ LGBBS Globotriaosylsphingosine, Blood Spot
- ☐ LGB3S Globotriaosylsphingosine, Serum

Fucosidosis

- ☐ FUCW Alpha-Fucosidase, Leukocytes

Gaucher Disease

- ☐ BGL Beta-Glucosidase, Leukocytes
- ☐ GBAZ Gaucher Disease, Full Gene Analysis
- ☐ GAUP Gaucher Disease, Mutation Analysis, GBA
- ☐ GPSY Glucopsychosine, Blood Spot

GM1 Gangliosidosis

- ☐ BGAW Beta-Galactosidase, Blood
- ☐ BGABS Beta-Galactosidase, Blood Spot
- ☐ BGA Beta-Galactosidase, Leukocytes

Krabbe Disease

- ☐ CBGC Galactocerebrosidase, Leukocytes
- ☐ KRABZ Krabbe Disease, Full Gene Analysis and Large (30 kb) Deletion, PCR
- ☐ PSYWB Psychosine, Blood
- ☐ PSY Psychosine, Blood Spot

Lysosomal Acid Lipase Deficiency

- ☐ LALB Lysosomal Acid Lipase, Blood
- ☐ LALBS Lysosomal Acid Lipase, Blood Spot

Mannosidosis

- ☐ MANN Alpha-Mannosidase, Leukocytes

Metachromatic Leukodystrophy

- ☐ ARSAZ ARSA Gene, Full Gene Analysis
- ☐ ARSU Arylsulfatase A, 24 Hour, Urine
- ☐ ARSAW Arylsulfatase A, Leukocytes
- ☐ CTSA Ceramide Trihexosides and Sulfatides, Urine

MPS Type I (Hurler/Scheie)

- ☐ IDSWB Alpha-L-Iduronidase, Blood
- ☐ IDSBS Alpha-L-Iduronidase, Blood Spot
- ☐ MPS1Z Hurler Syndrome, Full Gene Analysis
- ☐ MPSWB Mucopolysaccharidosis, Blood
- ☐ MPSBS Mucopolysaccharidosis, Blood Spot

MPS Type II (Hunter)

- ☐ MPS2Z Hunter Syndrome, Full Gene Analysis
- ☐ I2SBS Iduronate-2-Sulfatase, Blood Spot
- ☐ I2SW Iduronate-2-Sulfatase, Whole Blood
- ☐ MPSWB Mucopolysaccharidosis, Blood
- ☐ MPSBS Mucopolysaccharidosis, Blood Spot

MPS Type IIIA (Sanfilippo Type A)

- ☐ MP3AZ Mucopolysaccharidosis IIIA, Full Gene Analysis

MPS Type IIIB (Sanfilippo Type B)

- ☐ ANAS Alpha-N-Acetylglucosaminidase, Serum
- ☐ MP3BZ Mucopolysaccharidosis IIIB, Full Gene Analysis

MPS Type IIIC (Sanfilippo Type C)

- ☐ MP3CZ Mucopolysaccharidosis IIIC, Full Gene Analysis

MPS Type IIID (Sanfilippo Type D)

- ☐ MP3DZ Mucopolysaccharidosis IIID, Full Gene Analysis

MPS Type IVA (Morquio A)

- ☐ G6SW N-Acetylgalactosamine-6-Sulfatase, Leukocytes
- ☐ G6ST N-Acetylgalactosamine-6-Sulfate Sulfatase, Fibroblasts

MPS Type IVB (Morquio B)

- ☐ BGAW Beta-Galactosidase, Blood
- ☐ BGABS Beta-Galactosidase, Blood Spot
- ☐ BGA Beta-Galactosidase, Leukocytes

MPS Type VI (Maroteaux-Lamy)

- ☐ MPS6Z Mucopolysaccharidosis VI, Full Gene Analysis
- ☐ ARSB Arylsulfatase B, Fibroblasts

Mucolipidoses

- ☐ GNPTZ GNPTAB Gene, Full Gene Analysis
- ☐ MCIVP Mucolipidosis IV, Mutation Analysis, IVS3(-2) A->G and del6.4kb

Multiple Sulfatase Deficiency

- ☐ SUMFZ Multiple Sulfatase Deficiency, Full Gene Analysis

Niemann-Pick Types A&B

- ☐ NPABZ Niemann-Pick Disease, Types A and B, Full Gene Analysis
- ☐ NPABP Niemann-Pick Disease, Types A and B, Mutation Analysis
- ☐ OXYBS Oxysterols, Blood Spot
- ☐ OXNP Oxysterols, Plasma

Neuronal Ceroid Lipofuscinoses

- ☐ NCLP Neuronal Ceroid Lipofuscinosis (NCL, Batten Disease) Panel by Next-Generation Sequencing
- ☐ TPPTF Tripeptidyl Peptidase 1 (TPP1) and Palmitoyl-Protein Thioesterase 1 (PPT1), Fibroblasts
- ☐ TPPTL Tripeptidyl Peptidase 1 (TPP1) and Palmitoyl-Protein Thioesterase 1 (PPT1), Leukocytes

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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 Protoporphyria

- ☐ 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 Coproporphyria

- ☐ 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 Protoporphyria

- ☐ 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)

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