

Inborn Errors of Metabolism Test Request Form

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

Ship specimens to:

Mayo Medical Laboratories
3050 Superior Drive NW
Rochester, MN 55901

Customer Service: 855-516-8404

Visit www.MayoMedicalLaboratories.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

MML 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)

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Patient Name <i>(Last, First, Middle)</i>	
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GENERAL BIOCHEMICAL TESTS

- 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
- CRDPU Creatine Disorders Panel, Urine
- FAPCP Fatty Acid Profile, Comprehensive (C8-C26), Serum
- LYSDU Lysosomal Storage Disorders Screen, Urine
- OLIGU Oligosaccharide Screen, Urine
- OAU Organic Acids Screen, Urine
- PUPYU Purine and Pyrimidine Panel, Urine

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, cb1C Type, Full Gene Analysis
- MHDZ Methylmalonic Aciduria and Homocystinuria, cb1D 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
- 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

- NIEM Niemann-Pick Type C Detection, Fibroblasts
- NPCZ Niemann-Pick Type C Disease, Full Gene Analysis
- 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

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)

Malonyl-Coenzyme A Decarboxylase Deficiency

- MLYCZ MLYCD Gene, Full Gene Analysis

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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

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 Spots

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"/> PSY Psychosine, Blood Spots

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"/> 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"/> 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"/> KSQNU Keratan Sulfate Quantitative, Urine
<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"/> KSQNU Keratan Sulfate Quantitative, Urine
<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

Mucopolipidoses
<input type="checkbox"/> GNPTZ GNPTAB Gene, Full Gene Analysis
<input type="checkbox"/> MCIVP Mucopolipidosis 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 Spots
<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

Pompe Disease
<input type="checkbox"/> GAABS Acid Alpha-Glucosidase, Blood Spot
<input type="checkbox"/> GAAZ Pompe Disease, Full Gene Analysis

Sialidosis
<input type="checkbox"/> NEURF Neuraminidase, Fibroblasts

Tay-Sachs & Sandhoff Diseases
<input type="checkbox"/> MUGS Hexosaminidase A (MUGS), Serum
<input type="checkbox"/> NAGW Hexosaminidase A and Total Hexosaminidase, Leukocytes
<input type="checkbox"/> NAGS Hexosaminidase A and Total Hexosaminidase, Serum
<input type="checkbox"/> NAGAJ Hexosaminidase A and Total, Leukocytes/Ashkenazi Jewish
<input type="checkbox"/> NAGR Hexosaminidase A and Total, Leukocytes/Molecular Reflex
<input type="checkbox"/> HEXAZ Tay-Sachs Disease, HEXA Gene, Full Gene Analysis

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MITOCHONDRIAL ENERGY METABOLISM

- Q10 Coenzyme Q10, Reduced and Total, Plasma
- TQ10 Coenzyme Q10, Total, Plasma
- GDF15 Growth Differentiation Factor 15 (GDF15), Plasma
- LAA Lactate, Plasma
- LABF Lactate, Body Fluid
- MITON Mitochondrial Nuclear Gene Panel by Next-Generation Sequencing (NGS)
- MITOT Combined Mitochondrial Analysis, Mitochondrial Full Genome and Nuclear Gene Panel
- 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
- LSD6 Lysosomal Storage Disorders Newborn Screen, Blood Spot
- NBSE Newborn Screening Expanded Panel, Blood Spot
- NBSR Newborn Screen Recommended Panel, Blood Spot
- 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 Spots
- 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 Spots
- 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
- 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
- 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 Coproporphryria

- 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 Protoporphryria

- 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
- 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)