

Patient Information (required)

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AMINO ACID 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

Cystinuria

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

Maple Syrup Urine Disease

- ALLOI Allo-isoleucine, Blood Spot
- AAMSD Amino Acids, Maple Syrup Urine Disease Panel, Plasma
- MSUSC Branched-Chain Amino Acids, Self-Collect, Blood Spot

Homocystinuria

- CMMPP Cobalamin, Methionine, and Methylmalonic Acid Pathways, Plasma
- CMMPS Cobalamin, Methionine, and Methylmalonic Acid Pathways, Serum
- HCYSY Homocysteine, Total, Plasma
- HCYSY Homocysteine, Total, Serum

Phenylketonuria

- PKU Phenylalanine and Tyrosine, Plasma
- PKUBS Phenylalanine and Tyrosine, Blood Spot
- PKUSC Phenylalanine and Tyrosine, Self-Collect, Blood Spot

Tyrosinemia

- TYRBS Tyrosinemia Follow up Panel, Blood Spot
- TYRSC Tyrosinemia Follow up panel, Self-Collect, Blood Spot
- SUAC Succinylacetone, Blood Spot

CARBOHYDRATE METABOLISM

Congenital Disorders of Glycosylation

- CDG Carbohydrate Deficient Transferrin for Congenital Disorders of Glycosylation, Serum
- CDGN Congenital Disorders of N-Glycosylation, Serum
- OLIGU Oligosaccharide Screen, Random, Urine
- PMMIL Phosphomannomutase and Phosphomannose Isomerase, Leukocytes
- SORBU Sorbitol and Mannitol, Quantitative, Random, Urine

Galactosemia

- 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
- GCT Galactosemia Reflex, Blood
- GALE Uridine Diphosphate-Galactose 4' Epimerase, Blood

Transaldolase and Ribose-5-phosphate (RPI) Deficiencies

- TALDO Polyols, Quantitative, Urine

CHOLESTEROL BIOSYNTHESIS AND TRANSPORT

- CTXWB Cerebrotendinous Xanthomatosis, Blood
- CTXBS Cerebrotendinous Xanthomatosis, Blood Spot
- CTXP Cerebrotendinous Xanthomatosis, Plasma
- HSMBS Hepatosplenomegaly Panel, Blood Spot
- HSMWB Hepatosplenomegaly Panel, Blood
- HSMP Hepatosplenomegaly Panel, Plasma
- OXYWB Oxysterols, Blood
- OXYBS Oxysterols, Blood Spots
- OXNP Oxysterols, Plasma
- SLO Smith-Lemli-Opitz Screen, Plasma
- STER Sterols, Plasma

CONGENITAL ADRENAL HYPERPLASIA

- CAH2T Congenital Adrenal Hyperplasia Newborn Screen, Blood Spot
- CAH2I Congenital Adrenal Hyperplasia Profile for 21-Hydroxylase Deficiency, Serum

CREATINE DISORDERS

- CRDPP Creatine Disorders Panel, Plasma
- CRDPU Creatine Disorders Panel, Random, Urine

FAMILIAL AMYLOIDOSIS

- TTRX Amyloidosis, Transthyretin-Associated Familial, Reflex, Blood

FATTY ACID METABOLISM (BETA-OXIDATION)

- ACRN Acylcarnitines, Quantitative, Plasma
- ACRNS Acylcarnitines, Quantitative, Serum
- AGU20 Acylglycines, Quantitative, Random, Urine
- C4U C4 Acylcarnitine, Quantitative, Random, Urine
- CARN Carnitine, Plasma
- CARNS Carnitine, Serum
- CARNU Carnitine, Random, Urine
- FAO Fatty Acid Oxidation Probe Assay, Fibroblast Culture
- FAPCP Fatty Acid Profile, Comprehensive (C8-C26), Serum
- FAPM Fatty Acid Profile, Mitochondrial (C8-C18), Serum
- OAU Organic Acids Screen, Random, Urine

ORGANIC ACID METABOLISM

- C5OHU C5-OH Acylcarnitine, Quantitative, Random, Urine
- OAU Organic Acids Screen, Random, Urine
- O AUS Organic Acid Screen, Urine Spot

2-Hydroxyglutaric Aciduria

- 2HGA 2-Hydroxyglutaric Acid Chiral Analysis, Quantitative, Random, Urine

Biotinidase Deficiency

- BIOTS Biotinidase, Serum

Glutaric Acidemia

- C5DCU C5-DC Acylcarnitine, Quantitative, Random, 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
- 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
- MMAP Methylmalonic Acid, Quantitative, Plasma
- MMAS Methylmalonic Acid, Quantitative, Serum
- MMAU Methylmalonic Acid, Quantitative, Urine

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

HYPEROXALURIA	
<input type="checkbox"/> HYOX	Hyperoxaluria Panel, Random, Urine

LYSOSOMAL METABOLISM AND STORAGE DISORDERS

Multi-Disorder Panels	
<input type="checkbox"/> ARSBB	Arylsulfatase B, Blood Spot
<input type="checkbox"/> ARSBW	Arylsulfatase B, Leukocytes
<input type="checkbox"/> GUSBB	Beta-Glucuronidase, Blood Spot
<input type="checkbox"/> GUSBW	Beta-Glucuronidase, Leukocytes
<input type="checkbox"/> CTSU	Ceramide Trihexosides and Sulfatides, Random, Urine
<input type="checkbox"/> I2SB	Iduronate-2-Sulfatase, Blood Spot
<input type="checkbox"/> I2SWB	Iduronate-2-Sulfatase, Leukocytes
<input type="checkbox"/> HSMWB	Hepatosplenomegaly Panel, Blood
<input type="checkbox"/> HSMP	Hepatosplenomegaly Panel, Plasma
<input type="checkbox"/> PLSD	Lysosomal and Peroxisomal Storage Disorders Screen, Blood Spot
<input type="checkbox"/> LSDS	Lysosomal Storage Disorders Screen, Random, Urine
<input type="checkbox"/> LSD6W	Lysosomal Storage Disorders, Six-Enzyme Panel, Leukocytes
<input type="checkbox"/> MPSBS	Mucopolysaccharidosis, Blood Spot
<input type="checkbox"/> MPSQU	Mucopolysaccharides Quantitative, Random, Urine
<input type="checkbox"/> MPS3B	Mucopolysaccharidosis III, Three-Enzyme Panel, Blood Spot
<input type="checkbox"/> MPS3W	Mucopolysaccharidosis III, Four-Enzyme Panel, Leukocytes
<input type="checkbox"/> MPS4B	Mucopolysaccharidosis IV Enzyme Panel, Blood Spot
<input type="checkbox"/> MPS4W	Mucopolysaccharidosis IV Enzyme Panel, Leukocytes
<input type="checkbox"/> MP8BS	Mucopolysaccharidoses, Eight-Enzyme Panel, Blood Spot
<input type="checkbox"/> MP9W	Mucopolysaccharidoses, Nine-Enzyme Panel, Leukocytes
<input type="checkbox"/> MSDBS	Multiple Sulfatase Deficiency, Blood Spot
<input type="checkbox"/> MSDW	Multiple Sulfatase Deficiency, Leukocytes
<input type="checkbox"/> OLIGU	Oligosaccharide Screen, Random, Urine
<input type="checkbox"/> OXNP	Oxysterols, Plasma

Fabry Disease	
<input type="checkbox"/> AGABS	Alpha-Galactosidase, Blood Spot
<input type="checkbox"/> AGAW	Alpha-Galactosidase, Leukocytes
<input type="checkbox"/> AGAS	Alpha-Galactosidase, Serum
<input type="checkbox"/> CTSU	Ceramide Trihexosides and Sulfatides, Random, Urine
<input type="checkbox"/> LGB3S	Globotriaosylsphingosine, Serum

Fucosidosis	
<input type="checkbox"/> FUCW	Alpha-Fucosidase, Leukocytes

Gaucher Disease	
<input type="checkbox"/> GBAW	Beta-Glucosidase, Leukocytes
<input type="checkbox"/> GPSYW	Glucopsychosine, Blood
<input type="checkbox"/> GPSY	Glucopsychosine, Blood Spot
<input type="checkbox"/> GPSYP	Glucopsychosine, Plasma

GM1 Gangliosidosis	
<input type="checkbox"/> BGA	Beta-Galactosidase, Leukocytes
<input type="checkbox"/> MPS4B	Mucopolysaccharidosis IV Enzyme Panel, Blood Spot
<input type="checkbox"/> MPS4W	Mucopolysaccharidosis IV Enzyme Panel, Leukocytes

Krabbe Disease	
<input type="checkbox"/> GALCW	Galactocerebrosidase, Leukocytes
<input type="checkbox"/> PSY	Psychosine, Blood Spot
<input type="checkbox"/> PSYCF	Psychosine, Spinal Fluid
<input type="checkbox"/> PSYR	Psychosine, Whole Blood

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"/> ARSU	Arylsulfatase A, 24 Hour, Urine
<input type="checkbox"/> ARSAW	Arylsulfatase A, Leukocytes
<input type="checkbox"/> ARSBW	Arylsulfatase B, Leukocytes
<input type="checkbox"/> ARSBB	Arylsulfatase B, Blood Spot
<input type="checkbox"/> CTSU	Ceramide Trihexosides and Sulfatides, Random, Urine

Mucopolysaccharidoses	
<input type="checkbox"/> MPSQU	Mucopolysaccharides Quantitative, Random, Urine
<input type="checkbox"/> MPSEB	Mucopolysaccharides Quantitative, Serum
<input type="checkbox"/> MPSWB	Mucopolysaccharidosis, Blood
<input type="checkbox"/> MPSBS	Mucopolysaccharidosis, Blood Spot

MPS Type I (Hurler/Scheie)	
<input type="checkbox"/> IDUAW	Alpha-L-Iduronidase, Leukocytes
<input type="checkbox"/> MPSEB	Mucopolysaccharides Quantitative, Serum

MPS Type II (Hunter)	
<input type="checkbox"/> I2SB	Iduronate-2-Sulfatase, Blood Spot
<input type="checkbox"/> I2SWB	Iduronate-2-Sulfatase, Leukocytes

MPS Type III	
<input type="checkbox"/> MPS3B	Mucopolysaccharidosis III, Three-Enzyme Panel, Blood Spot
<input type="checkbox"/> MPS3W	Mucopolysaccharidosis III, Four-Enzyme Panel, Leukocytes

MPS Type IV	
<input type="checkbox"/> MPS4B	Mucopolysaccharidosis IV Enzyme Panel, Blood Spot
<input type="checkbox"/> MPS4W	Mucopolysaccharidosis IV Enzyme Panel, Leukocytes

MPS VI	
<input type="checkbox"/> ARSBB	Arylsulfatase B, Blood Spot
<input type="checkbox"/> ARSBW	Arylsulfatase B, Leukocytes

MPS VII	
<input type="checkbox"/> GUSBW	Beta-Glucuronidase, Leukocytes
<input type="checkbox"/> GUSBB	Beta-Glucuronidase, Blood Spot

Multiple Sulfatase Deficiency	
<input type="checkbox"/> MSDBS	Multiple Sulfatase Deficiency, Blood Spot
<input type="checkbox"/> MSDW	Multiple Sulfatase Deficiency, Leukocytes

MPS Type IVB (Morquio B)	
<input type="checkbox"/> BGA	Beta-Galactosidase, Leukocytes

Niemann-Pick Types A and B	
<input type="checkbox"/> ASMW	Acid Sphingomyelinase, Leukocytes
<input type="checkbox"/> OXNP	Oxysterols, Plasma

Niemann-Pick Type C	
<input type="checkbox"/> NIEM	Niemann-Pick Type C Detection, Fibroblasts
<input type="checkbox"/> OXNP	Oxysterols, Plasma

Neuronal Ceroid Lipofuscinoses	
<input type="checkbox"/> NCLBS	Neuronal Ceroid Lipofuscinosis, Two-Enzyme Panel, Blood Spot
<input type="checkbox"/> NCLW	Neuronal Ceroid Lipofuscinosis, Two-Enzyme Panel, Leukocyte

Pompe Disease	
<input type="checkbox"/> GAAW	Acid Alpha-Glucosidase, Leukocytes
<input type="checkbox"/> HEX4	Glucotetrasaccharides, Random, Urine
<input type="checkbox"/> PDBS	Pompe Disease, Blood Spot
<input type="checkbox"/> PDCRF	Pompe Disease Cross-Reactive Immunological Material Status, Fibroblasts
<input type="checkbox"/> PDCRW	Pompe Disease Cross-Reactive Immunological Material Status, Leukocytes

Tay-Sachs and Sandhoff Diseases	
<input type="checkbox"/> NAGW	Hexosaminidase A and Total Hexosaminidase, Leukocytes
<input type="checkbox"/> NAGS	Hexosaminidase A and Total Hexosaminidase, Serum
<input type="checkbox"/> NAGR	Hexosaminidase A and Total, Leukocytes/Molecular Reflex, Whole Blood
<input type="checkbox"/> MUGS	Hexosaminidase A, Serum

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MITOCHONDRIAL ENERGY METABOLISM	
<input type="checkbox"/>	Q10 Coenzyme Q10, Reduced and Total, Plasma
<input type="checkbox"/>	TQ10 Coenzyme Q10, Total, Plasma
<input type="checkbox"/>	FAPM Fatty Acid Profile, Mitochondrial (C8-C18), Serum
<input type="checkbox"/>	GDF15 Growth Differentiation Factor 15, Plasma
<input type="checkbox"/>	LAPYP Lactate Pyruvate Panel, Plasma
<input type="checkbox"/>	MMPP Mitochondrial Metabolites, Plasma
<input type="checkbox"/>	OAU Organic Acids Screen, Random, Urine
<input type="checkbox"/>	PYRC Pyruvate, Spinal Fluid
<input type="checkbox"/>	PYR Pyruvic Acid, Blood

NEWBORN SCREENING	
Screening Panels	
<input type="checkbox"/>	LDALD Lysosomal and Peroxisomal Disorders Newborn Screen, Blood Spot
<input type="checkbox"/>	SNS Supplemental Newborn Screen, Blood Spot
Second Tier Tests	
<input type="checkbox"/>	ALLOI Allo-isoleucine, Blood Spot
<input type="checkbox"/>	CAH2T Congenital Adrenal Hyperplasia Newborn Screen, Blood Spot
<input type="checkbox"/>	GPSY Glucopsychosine, Blood Spot
<input type="checkbox"/>	HCMM Homocysteine (Total), Methylmalonic Acid, and Methylcitric Acid, Blood Spot
<input type="checkbox"/>	HGEM Hydroxyglutaric Acids, Glutaric Acid, Ethylmalonic Acid, and Methylsuccinic Acid, Blood Spot
<input type="checkbox"/>	KD2T Krabbe Disease Second-Tier Newborn Screen, Blood Spot
<input type="checkbox"/>	LPCBS Lysophosphatidylcholines, LC MS/MS, Blood Spot
<input type="checkbox"/>	MPSBS Mucopolysaccharidosis, Blood Spot
<input type="checkbox"/>	OXYBS Oxysterols, Blood Spot
<input type="checkbox"/>	PD2T Pompe Disease Second-Tier Newborn Screening, Blood Spot
<input type="checkbox"/>	PSY Psychosine, Blood Spot
<input type="checkbox"/>	SUAC Succinylacetone, Blood Spot

PEROXISOMAL BIOGENESIS & METABOLISM	
<input type="checkbox"/>	BAIPD Bile Acids for Peroxisomal Disorders, Serum
<input type="checkbox"/>	POXP Fatty Acid Profile, Peroxisomal (C22-C26), Plasma
<input type="checkbox"/>	POX Fatty Acid Profile, Peroxisomal (C22-C26), Serum
<input type="checkbox"/>	PIPA Pipecolic Acid, Serum
<input type="checkbox"/>	PIPU Pipecolic Acid, Random, Urine
<input type="checkbox"/>	PGRBC Plasmalogens, Blood
<input type="checkbox"/>	PGDBS Plasmalogens, Blood Spot

PORPHYRIAS	
Urine	
<input type="checkbox"/>	ALAU Aminolevulinic Acid, Urine
<input type="checkbox"/>	PBGU Porphobilinogen, Quantitative, Random, Urine
<input type="checkbox"/>	PQNU Porphyrins, Quantitative, 24 Hour, Urine
<input type="checkbox"/>	PQNRU Porphyrins, Quantitative, Random, Urine
Plasma	
<input type="checkbox"/>	PBALP Porphobilinogen and Aminolevulinic Acid, Plasma
<input type="checkbox"/>	PTP Porphyrins, Total, Plasma
Fecal	
<input type="checkbox"/>	FQPPS Porphyrins, Feces
Blood	
<input type="checkbox"/>	PEWE Porphyrins Evaluation, Washed Erythrocytes
<input type="checkbox"/>	PEE Porphyrins Evaluation, Whole Blood
<input type="checkbox"/>	PPFEW Protoporphyrins, Fractionation, Washed Erythrocytes
<input type="checkbox"/>	PPFE Protoporphyrins, Fractionation, Whole Blood
Enzymes	
<input type="checkbox"/>	PBGDW Porphobilinogen Deaminase, Washed Erythrocytes
<input type="checkbox"/>	PBGD_ Porphobilinogen Deaminase, Whole Blood
<input type="checkbox"/>	UPGC Uroporphyrinogen III Synthase (Co-Synthase), Erythrocytes
<input type="checkbox"/>	UPGDW Uroporphyrinogen Decarboxylase, Washed Erythrocytes
<input type="checkbox"/>	UPGD Uroporphyrinogen Decarboxylase, Whole Blood
POSTMORTEM BIOCHEMICAL TESTING	
<input type="checkbox"/>	PMSBB Postmortem Screening, Bile and Blood Spot
PURINE AND PYRIMIDINE METABOLISM	
<input type="checkbox"/>	PUPYP Purine and Pyrimidine Panel, Plasma
<input type="checkbox"/>	PUPYU Purine and Pyrimidine Panel, Random, Urine
<input type="checkbox"/>	SSCTU S-Sulfocysteine Panel, Urine

UREA CYCLE DISORDERS	
<input type="checkbox"/>	AAQP Amino Acids, Quantitative, Plasma
<input type="checkbox"/>	AAPD Amino Acids, Quantitative, Random, Urine
<input type="checkbox"/>	AAUCD Amino Acids, Urea Cycle Disorders Panel, Plasma
<input type="checkbox"/>	OAU Organic Acids Screen, Random, Urine
<input type="checkbox"/>	OROT Orotic Acid, Random, Urine

ADDITIONAL TESTS (INDICATE TEST NUMBER AND NAME)	