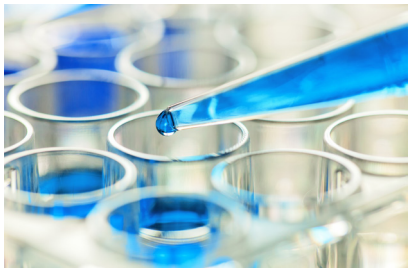
Greenwood Diagnostic Labs



LYSOSOMAL STORAGE DISORDERS

DIAGNOSTIC TESTING

COMPREHENSIVE SERVICES

The Greenwood Diagnostic Labs are CLIA certified and CAP accredited clinical laboratories with more than 30 years of experience with lysosomal storage disease testing. We offer a full range of testing for patients with a suspected lysosomal storage disorder including urine screening, enzyme analysis, and molecular confirmation by gene sequencing and dosage analysis.

Biochemical Testing
Diagnosis and monitoring

Molecular Testing
State of the art technology to confirm
disease-causing mutations

Sample Collection
Variety of sample types accepted
Sample collection kits (whole blood and DBS)
are available to simplify the sample collection process

User-Friendly Website
Printable requisition forms and detailed test information

Direct Access
Laboratory genetic counselors and laboratory directors are
available to answer any questions regarding tests and results

Online Results Portal
Provides convenient access to test reports

Institutional & Self-Pay Discounts
Please contact lab for more information

Mission Statement

The Greenwood Genetic Center is a nonprofit institute organized to provide clinical genetic services, diagnostic laboratory testing, educational programs and resources and research in the field of medical genetics.

FACULTY EXPERTISE

Greenwood Diagnostic Labs extensive experience with testing for lysosomal storage disorders is complemented by the expertise of the Greenwood Genetic Center's (GGC) clinical and research faculty. Projects such as The Glycoproteinoses Natural History Study facilitate ongoing collaboration among the Laboratory, Clinical, and Research divisions to increase understanding of these disorders and improve diagnostic testing for these patients.

Richard Steet, PhD

Heather Flanagan-Steet, PhD

Dr. Richard Steet, Director of the JC Self Research Institute, along with his colleague and spouse, Dr. Heather Flanagan-Steet, Director of Functional Studies, bring their long-time research on lysosomal storage disorders and congenital disorders of glycosylation to GGC. This work takes advantage of cell and animal-based models. A combination of molecular, developmental, and chemical approaches to unravel the complex pathogenesis of these disorders and explore new ways to treat them. Dr. Steet and Dr. Flanagan-Steet integrate with the Clinical and Diagnostic divisions of GGC to enhance our understanding of the genetic basis of birth defects and disabilities to better serve patients.

Dr. Pollard, Lead Director of the Biochemical Laboratory, has fourteen years of experience with clinical laboratory testing for lysosomal storage disorders, and is board certified in both biochemical and molecular genetics. Dr. Pollard has played a significant role in expanding the LSD test menu at Greenwood Diagnostic Labs; including diverse specimen types for enzyme testing, the addition of several individual gene sequencing tests, and improved biomarker analysis with tandem mass spectrometry assays. Dr. Pollard also has extensive experience with newborn screening follow-up, and leads several projects with industry partners to advance the treatment of biochemical disorders.

Laura Pollard, PhD

Sara Cathey, MD

Glycoproteinoses Natural History Study

Dr. Sara Cathey is a clinical geneticist at the Greenwood Genetic Center and Principal Investigator for the Longitudinal Studies of the Glycoproteinoses. The nine different glycoproteinoses are among the rarest lysosomal disorders. Dr. Cathey's project seeks to obtain clinical and laboratory information about individuals with these conditions in order to better characterize the natural course of the disease progression and to explore potential treatments.

GGC Senior Genetics Scholar

Dr. Jules Leroy is one of the Greenwood Genetic Center's Senior Scholars. Dr Leroy discovered Mucolipidosis II, or I cell disease. He works with Dr. Cathey in further delineating the clinical, biochemical, and molecular manifestations of this disorder and related conditions. He also works with the Diagnostic Laboratory and other clinicians to provide expertise on metabolic disorders and guidance in the statewide metabolic disease treatment program.

Jules Leroy, MD, PhD

BIOCHEMICAL TESTING

The demonstration of deficient enzyme activity is considered the gold-standard for diagnosing lysosomal storage disorders. Greenwood Diagnostic Labs offer individual enzyme analysis and multiple panels. In addition, urine and blood-based screening and monitoring is available to help further direct testing, and monitor treatment efficacy. To ease the sample collection process, the Biochemical Lab accepts a variety of sample types.

Condition	Enzyme	Sample Type(s) Accepted	CPT Code	Price
Alpha-mannosidosis	α-mannosidase	DBS, Fibroblasts, Leukocytes	82657	$200
Aspartylglucosaminuria	Aspartyglucosaminidase	CSF, DBS, Leukocytes, Plasma	82657	$200
Beta-mannosidosis	β-mannosidase	DBS, Fibroblasts, Leukocytes	82657	$200
Fabry Disease	α-galactosidase	DBS, Fibroblasts, Leukocytes, Plasma	82657	$200
Fucosidosis	α-fucosidase	DBS, Fibroblasts, Leukocytes	82657	$200
Gaucher Disease	β-glucosidase	DBS, Fibroblasts, Leukocytes	82963	$200
GM1 Gangliosidosis	β-galactosidase	DBS, Fibroblasts, Leukocytes, Plasma	82657	$200
Hurler Syndrome (MPS I)	α-iduronidase	DBS, Fibroblasts, Leukocytes, Plasma	82657	$200
Hunter Syndrome (MPS II)	iduronate-2-sulfatase	DBS, Fibroblasts, Leukocytes, Plasma	82657	$200
Krabbe disease	Galactocerebrosidase	DBS	82657	$200
Maroteaux-Lamy Syndrome (MPS VI)	Arylsulfatase B	DBS, Fibroblasts, Leukocytes	82657	$200
Metachromatic Leukodystrophy	Arylsulfatase A	Fibroblasts, Leukocytes	82657	$200
Morquio Syndrome A (MPS IVA)	N-acetyl-galactosamine-6-sulfatase	DBS, Fibroblasts, Leukocytes	82657	$200
Morquio Syndrome B (MPS IVB)	β-galactosidase	DBS, Fibroblasts, Leukocytes, Plasma	82657	$200
Neuronal Ceroid Lipofuscinosis 1 (CLN1)	Palmitoyl-protein thioesterase 1	Leukocytes	82657	$200
Neuronal Ceroid Lipofuscinosis 2 (CLN2)	Tripeptidyl peptidase 1	DBS	82657	$200
Niemann-Pick disease A/B	Acid sphingomyelinase (DBS)	DBS	82657	$200
Pompe Disease, Glycogen Storage Disease Type II	α-glucosidase	DBS, Fibroblasts, Leukocytes	82657	$200
Sanfilippo syndrome A (MPS IIIA)	Heparan-N-sulfatase	Fibroblasts, Leukocytes	82657	$200
Sanfilippo syndrome B (MPS IIIB)	N-acetyl-alpha-D-glucosaminidase	DBS, Fibroblasts, Plasma	82657	$200
Sanfilippo syndrome C (MPS IIIC)	Acetyl CoA : glucosamine N acetyl transferase	Fibroblasts, Leukocytes	82657	$200
Sanfilippo syndrome D (MPS IIID)	N-acetyl glucosamine-6-sulfatase	Fibroblasts, Leukocytes	82657	$200
Schindler/Kanzaki Disease	N-acetyl-alpha galactosaminidase	DBS, Fibroblasts, Plasma	82657	$200
Sialidosis	α-neuraminidase-sialidase	Fibroblasts	82657	$200
Sly Syndrome (MPS VII)	β-glucuronidase	DBS, Fibroblasts, Leukocytes	82657	$200
Tay-Sachs/Sandhoff Disease	β-hexosaminidase	Leukocytes, Plasma	83080	$200

BIOCHEMICAL SAMPLE & SHIPPING REQUIREMENTS

Cerebral Spinal Fluid (CSF) : Send frozen via overnight shipping.

Dried Blood Spot (DBS) : Completely fill at least 3 circles with a single layer of blood for each circle. Dry spots 3-4 hours prior to sending.

Plasma (P) : Sodium heparin tube (green top) tube. Ship whole blood overnight at room temperature, OR spin down, remove plasma and send plasma frozen.

Leukocytes (L) : Ship whole blood in sodium heparin (green top) tube overnight. Samples should arrive within 24-48 hours of draw, OR isolated leukocyte pellet can be shipped frozen overnight.

Fibroblasts (F) : Fresh tissue should be placed in transport media (preffered) or sterile saline and shipped overnight. For cultured tissue, please send two T25 flasks overnight. If cultured tissue is being sent, a control flask is requested in addition to the patient sample.

Whole Blood (WB) : Ship whole blood in sodium heparin (green top) tube. Samples should arrive within 24-48 hours of draw. Ship overnight at room temperature.

Urine : Send frozen via overnight shipping.

ENZYME PANELS

DBS Lysosomal Storage Disease Enzyme Panel
Price: $800 CPT Code: 82657 (x4) Sample Type: D

Disorders	Enzymes
α-mannosidosis	α-mannosidase
β-mannosidosis	β-mannosidase
Aspartylglucosaminuria	Aspartylglucosaminidase
Fabry Disease	α-galactosidase
Fucosidosis	α-fucosidase
Gaucher Disease	β-glucosidase
Krabbe Disease	Galactocerebrosidase
MPS IVB/ GM1-Gangliosidosis	β-galactosidase
Neuronal Ceroid Lipofuscinosis	Tripeptidyl-peptidase 1
Niemannn-Pick Disease A/B	Acid Sphingomyelinase
Pompe Disease	α-glucosidase
Schindler/Kanzaki Disease	N-acetyl-alpha galactosaminidase

DBS Mucopolysaccharidosis (MPS) Enzyme Panel
Price: $800 CPT Code: 82657 (x4) Sample Type: F, WB

Disorders	Enzymes
Hunter syndrome (MPS II)	Iduronate-2-sulfatase
Hurler syndrome (MPS I)	α-iduronidase
Sanfilippo syndrome B (MPS IIIB)	N-acetyl-α-glucosaminidase
Maroteaux-Lamy syndrome (MPS VI)	Arylsulfatase B
Morquio syndrome A (MPS IVA)	N-acetyl galactosamine-6-sulfatase
Morquio syndrome B (MPS IVB)	β-galactosidase
Sly syndrome (MPS VII)	β-glucuronidase

Hydrops Enzyme Panel
Price: $800 CPT Code: 82657 (x4) Sample Type: F

Disorders	Enzymes
Gaucher Disease	β-glucosidase
GM1-Gangliosidosis	β-galactosidase
Sialidosis	α-neuraminidase (sialidase)
Sly syndrome (MPS VII)	β-glucronidase

Lysosomal Storage Disease Enzyme Panel
Price: $1,000 CPT Code: 82657 (x5) Sample Type: WB

Disorders	Enzymes
α-mannosidosis	α-mannosidase
β-mannosidosis	β-mannosidase
Aspartylglucosaminuria	Aspartylglucosaminidase
Fabry Disease	α-galactosidase
Fucosidosis	α-fucosidase
Gaucher Disease	β-glucosidase
Hurler syndrome (MPS I)	α-iduronidase
Krabbe Disease	Galactocerebrosidase
Metachromatic Luekodystrophy	Arylsulfatase A
MPS IVB/ GM1-Gangliosidosis	β-galactosidase
Niemann-Pick Disease A/B	Acid sphingomyelinase
Schindler/Kanzaki Disease	N-acetyl-alpha galactosaminidase
Tay-Sachs/Sandhoff Disease	β-hexosaminidase

Morquio syndrome (MPS IV) Panel, types A & B
Price: $400 CPT Code: 82657 (x2) Sample Type: F, L

Disorders	Enzymes
Morquio A syndrome (MPS IVA)	N-acetyl galactosamine-6-sulfatase
Morquio B syndrome (MPS IVB)	β-galactosidase

Mucopolysaccharidosis (MPS) Enzyme Panel
Price: $1,000 CPT Code: 82657 (x5) Sample Type: F, WB

Disorders	Enzymes
Hunter syndrome (MPS II)	Iduronate-2-sulfatase
Hurler syndrome (MPS I)	α-iduronidase
Maroteaux-Lamy syndrome (MPS VI)	Arylsulfatase B
Morquio syndrome A (MPS IVA)	N-acetyl galactosamine-6-sulfatase
Morquio syndrome B (MPS IVB)	β-galactosidase
Sanfilippo syndrome A (MPS IIIA)	Heparan-N-sulfatase
Sanfilippo syndrome B (MPS IIIB)	N-acetyl-alpha-D-glucosaminidase
Sanfilippo syndrome C (MPS IIIC)	Acetyl CoAglucosamine N acetyl trasnferase
Sanfilippo syndrome D (MPS IIID)	N-acetyl glucosamine-6-sulfatase
Sly syndrome (MPS VII)	β-glucuronidase

Mucolipidosis II/III Screen
Price: $400 CPT Code: 82657 (x2) Sample Type: DBS, P

Three or four enzymes will be measured in plasma or DBS to detect elevated activity

Multiple Sulfatase Deficiency Enzyme Panel
Price: $400 CPT Code: 82657 (x2) Sample Type: F, DBS, WB

This panel contains 3 sulfastase enzymes: Arysulfatase B, Iduronate-2-sulfatase, and N-actyl-galactosamine-6-sulfatase

Neurological (Sphingolipidoses) Panel
Price: $600 CPT Code: 82657 (x3) Sample Type: WB

Disorders	Enzyme
Fabry Disease	α-galactosidase
Gaucher Disease	β-galactosidase
Krabbe Disease	Galactocerebrosidase
Metachromatic Luekodystrophy	Arylsulfatase A
MPS IVB/ GM1-Gangliosidosis	β-galactosidase
Neuronal Ceroid Lipofuscinosis 1	Palmitoyl-protein thioesterase 1
Neuronal Ceroid Lipofuscinosis 2	Tripeptidyl peptidase 1
Niemann Pick Disease A/B	Acid Spingomyelinase
Tay-Sachs/Sandhoff Disease	β-hexosaminidase

Oligosaccharidoses Panel
Price: $600 CPT Code: 82657 (x3) Sample Type: D, F*, L

Disorders	Enzymes
α-mannosidosis	α-mannosidase
β-mannosidosise	β-mannosidase
Aspartylglucosaminuria	Aspartylglucosaminidase
Fucosidosis	α-fucosidase
MPS IVB/GM1-Gangliosidosis	β-galactosidase
Schindler Disease	α-N-acetyl galactosaminidase
Sialidosis*	α-neurominidase-sialidase

* Only fibroblasts are accepted for Sialidosis

Sanfilippo Syndrome (MPS III) Enzyme Panel
Price: $800 CPT Code: 82657 (x4) Sample Type: F, WB

Disorders	Enzymes
Sanfilippo syndrome A (MPS IIIA)	Heparan-N-sulfatase
Sanfilippo syndrome B (MPS IIIB)	N-acetyl-alpha-D-glucosaminidase
Sanfilippo syndrome C (MPS IIIC)	Acetyl CoAglucosamine N acetyl transferase
Sanfilippo syndrome D (MPS IIID)	N-acetyl glucosamine-6-sulfatase

BIOMARKERS

URINE SCREENING TESTS

MPS ANALYSIS
CPT Code: 83864x3 Price: $450

- Quantification of individual glycosaminoglycans (dermatan sulfate, heparan sulfate, keratan sulfate, & chondiotin sulfate) via tandem mass spectrometry as well as quantification of total GAGs via DMB binding assay

Oligosaccharide Analysis
CPT Code: 84377 Price: $250

- Semi quantitative analysis via tandem mass spectrometry with improved sensitivity and specificity compared to traditional thin layer chromatography

Quantitative Sialic Acid, Total and free
CPT Code: 84275 Price: $200

- Measurement of siliac acid content in urine via HPLC and fluorescence

URINE MONITORING TESTS

Glucose Tetrasaccharide (Hex4) Monitoring
CPT Code: 82570, 83789 Price: $202

- Used to monitor treatment of Pompe Disease

Quantitative Glycosaminoglycans
CPT Code: 83864x2 Price: $300

- Used to monitor patients with a known MPS disorder who are receiving treatment
  - MPS I/II (Total GAGs, DS, HS)
  - MPS III (Total GAGs, HS)
  - MPS IV (Total GAGs, KS, CS)
  - MPSVI (Total GAGs, DS)
  - MPS VII (Total GAGs, DS, CS)

BLOOD-BASED SCREENING/MONITORING

Quantification of Chitotriosidase (Plasma)
CPT Code: 82657 Price: $200

- Used to monitor treatment of Gaucher Disease

MOLECULAR TESTING

Molecular testing can be used to confirm the diagnosis and to identify the disease causing mutation(s) within a family. Carrier testing is available. Prenatal diagnosis can be requested once the familial mutations are identified.

SEQUENCING & DELETION/DUPLICATION	CPT CODE	PRICE
Alpha-mannosidosis : *MAN2B1* sequencing	81479	$1,500
Aspartylglucosaminuria : *AGA* sequencing	81479	$1,000
Batten Disease, NCL type 3 : CLN3 sequencing	81479	$1,000
Beta-mannosidosis : *MANBA* sequencing	81479	$1,000
Fabry Disease : *GLA* sequencing	81405	$1,000
Fucosidosis : *FUCA1* sequencing	81479	$1,000
Galactosialidosis : *CTSA* sequencing	81479	$1,200
Gaucher Disease : *GBA* sequencing	81479	$1,000
GM1 Gangliosidosis : *GLB1* sequencing	81479	$1,200
Hunter syndrome (MPS II) : *IDS* sequencing	81405	$1,000
Hunter syndrome (MPSII) : *IDS* deletion/duplication	81404	$500
Hurler syndrome (MPS I) : *IDUA* sequencing	81406	$1,000
Krabbe Disease : *GALC* sequencing	81406	$1,000
Maroteaux-Lamy syndrome (MPSVI) : *ARSB* sequencing	81479	$800
Metachromatic Leukodystrophy : *ARSA* sequencing	81405	$1,000
Morquio syndrome A (MPS IVA) : *GALNS* sequencing	81479	$1,000
Morquio syndrome B (MPS IVB) : *GLB1* sequencing	81479	$1,200
Mucolipidosis II & III, Alpha/Beta : *GNPTAB* sequencing	81479	$1,500
Mucolipidosis III Gamma : *GNPTG* sequencing	81479	$1,000
Neuronal Ceroid Lipofuscinosis, Type 1 : *PPT1* sequencing	81479	$800
Neuronal Ceroid Lipofuscinosis, Type 2 : *TPP1* sequencing	81479	$1,000
Pompe Disease, GSD, Type 2 : *GAA* sequencing	81406	$1,000
Sandhoff Disease : *HEXB* sequencing	81479	$900
Sanfilippo syndrome A (MPS IIIA) : SGSH sequencing	81479	$1,000
Sanfilippo syndrome B (MPS IIIB) : *NAGLU* sequencing	81479	$1,200
Sanfilippo syndrome C (MPS IIIC), : *HGSNAT* sequencing	81479	$1,500
Sanfilippo syndrome D (MPS IIID) : *GNS* sequencing	81479	$1,000
Sialidosis : *NEU1* sequencing	81479	$800
Sly syndrome (MPS VII) : *GUSB* sequencing	81479	$1,000
Tay-Sachs Disease : *HEXA* sequencing	81406	$1,000

ALL SINGLE GENES CAN BE SEQUENCED FROM A DRIED BLOOD SPOT

NGS Panels	CPT CODE	PRICE
Lysosomal Storage Disorder Panel (75 genes)	81479	$3,500
Neuronal Ceroid Lipofuscinosis Panel (9 genes)	81479	$2,500
Non-immune Hydrops Panel (87 genes)	81479	$3,500

Any of the above NGS panels may be useful for patients without a specific suspected storage disorder or where biochemical testing is not available. The entire coding region for all genes on the particular panel is included. Parental follow-up testing for variants of unknown significance are included at no charge.

NEWBORN SCREENING

FOLLOW-UP TESTING

Greenwood Diagnostic Labs understand the logistics and urgency needed to follow-up on an abnormal newborn screen, confirm the diagnosis, and promptly provide treatment.

Greenwood Diagnostic Labs has worked closely with the South Carolina NBS laboratory and the GGC metabolic treatment team to follow-up patients with an abnormal newborn screen for inborn errors of metabolism in the state of South Carolina for many years. We have recently expanded these services to other states for the follow-up of infants with an abnormal NBS for lysosomal storage disorders.

EXPEDITED TURNAROUND TIMES FOR MOLECULAR & ENZYME TESTING

Comprehensive Biomarker, Enzyme, & Molecular Analysis

Greenwood Diagnostic Labs have been performing follow-up testing for states that have already implemented NBS for LSD since this testing began more than three years ago. This includes the complex interpretation of of MPS I pseudo-deficiency.

- Enzyme analysis for 25 lysosomal enzymes, including those currently on the RUSP
- Sanger sequencing analysis for each of the 25 LSDs (can be performed from a Dried Blood Spot sample)
- Quantitative component glycosaminoglycan (GAG) analysis in urine via Mass Spectrometry
- Urine GLC4/HEX4 analysis for Pompe Disease
- Plasma chitotriosidase analysis for Gaucher Disease
- Molecular analysis from a DBS can be used as a second-tier NBS test, by using the DBS from the Newborn Screening Laboratory. This prevents the need for a second specimen collection and produces faster results.

Greenwood Diagnostic Labs

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