

Overview

Useful For

Screening for selected oligosaccharidosis

Genetics Test Information

Oligosaccharidoses are characterized by the abnormal accumulation of incompletely degraded oligosaccharides in cells and tissues and the corresponding increase of related free oligosaccharides in the urine.

Clinical features of the oligosaccharidoses often overlap; therefore, urine screening is an important tool in the initial workup for these disorders.

Enzyme or molecular analysis is required to make a definitive diagnosis.

Testing Algorithm

Oligosaccharide analysis may be considered in the workup of unexplained refractory epilepsy. For more information see:

[-Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm](#)

[-Congenital Disorders of Glycosylation: Screening Algorithm](#)

Special Instructions

- [Biochemical Genetics Patient Information](#)
- [Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm](#)
- [Congenital Disorders of Glycosylation: Screening Algorithm](#)
- [Congenital Disorders of Glycosylation Patient Information](#)

Method Name

Matrix-Assisted Laser Desorption/Ionization Time-of-Flight Mass Spectrometry (MALDI-TOF MS)

NY State Available

Yes

Specimen

Specimen Type

Urine

Ordering Guidance

This is the recommended test when clinical features are suggestive of, or when molecular testing results suggest, an oligosaccharidosis disorder that can be identified by this test.

The recommended screening test for the initial workup of a suspected lysosomal storage disorder, particularly when

clinical features are nonspecific, is LSDS / Lysosomal Storage Disorders Screen, Random, Urine.

Necessary Information

1. Patient's age is required.
2. [Biochemical Genetics Patient Information](#) (T602) is recommended. This information aids in providing a more thorough interpretation of results. Send information with specimen.

Specimen Required

Supplies: Urine Tubes, 10 mL (T068)
Container/Tube: Plastic, 10-mL urine tube
Specimen Volume: 8 mL
Pediatric Volume: 2 mL

Collection Instructions:

1. Collect a random urine specimen.
2. No added preservative.
3. Immediately freeze specimen.

Forms

1. [Biochemical Genetics Patient Information](#) (T602)
2. If not ordering electronically, complete, print, and send a [Biochemical Genetics Test Request](#) (T798) with the specimen.

Specimen Minimum Volume

2.5 mL

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Urine	Frozen (preferred)	365 days	
	Ambient	7 days	
	Refrigerated	15 days	

Clinical & Interpretive

Clinical Information

The oligosaccharidoses (glycoproteinoses) are a subset of lysosomal disorders (LD) caused by the deficiency of any one of the lysosomal enzymes involved in the degradation of complex oligosaccharide chains. They are characterized by the abnormal accumulation of incompletely degraded oligosaccharides in cells and tissues and the corresponding increase of related free oligosaccharides in the urine. Clinical diagnosis can be difficult due to the similarity of clinical features across disorders and their variable severity. Clinical features can include bone abnormalities, coarse facial features, corneal cloudiness, organomegaly, muscle weakness, hypotonia, developmental delay, and ataxia. Age of onset ranges from

early infancy to adulthood and can also present prenatally.

The oligosaccharidoses and other storage disorders detected by this assay include alpha-mannosidosis, beta-mannosidosis, aspartylglucosaminuria, fucosidosis, Schindler disease, GM1 gangliosidosis, Sandhoff disease, sialidosis, galactosialidosis, mucopolidoses types II and III, mucopolysaccharidosis IVA (Morquio A), mucopolysaccharidosis IVB (Morquio B), and Pompe disease (see Table). Additional conditions that may be picked up by this test include other mucopolysaccharidoses, Gaucher disease, and some congenital disorders of glycosylation (PMM2, NGLY1, MOGS, ALG1).

Table. Conditions Identifiable by Test

Disease	Gene	Enzyme deficiency
Alpha- mannosidosis	MAN2B1	Alpha-mannosidase
	Phenotype: Considerably variable. Three clinical types have been suggested in untreated individuals. Type I Clinically recognized after age ten years, with myopathy, slow progression, and absence of skeletal abnormalities. Type 2 - Clinically recognized before age ten years, with myopathy, slow progression, and presence of skeletal abnormalities. Type 3 – Severe progression leading to early death from primary central nervous system involvement or infection. Enzyme replacement therapy is available for all forms.	
Beta-mannosidosis	MANBA	Beta-mannosidase
	Phenotype: Disease severity and progression is highly variable with onset from infancy to adulthood. Clinical features may include intellectual disability, respiratory infections, hearing loss, hypotonia, peripheral neuropathy, seizures, and behavioral problems.	
Aspartylglucosaminuria	AGA	Aspartylglucosaminidase
	Phenotype: Clinical features include developmental delay, intellectual disability, behavioral problems, recurrent infections, musculoskeletal features, and characteristic facial features. Clinical features worsen with age, and adults have progressive psychomotor decline.	
Fucosidosis	FUCA1	Alpha-L-fucosidase
	Phenotype: Continuum within a wide spectrum of severity; clinical features include neurodegeneration, coarse facial features, growth delay, recurrent infections, dysostosis multiplex, angiokeratoma, and elevated sweat chloride.	
Schindler disease	NAGA	Alpha-N-acetyl-galactosaminidase
	Phenotype: There are three types of Schindler diseases that differ in disease severity and age of onset. Type I is characterized by rapidly progressive neurodegeneration, typically by age 2 years. Type II is typically diagnosed in adulthood and characterized by angiokeratomas, mild cognitive impairment, and hearing loss. Type III is an intermediate form that presents as a variety of symptoms that may include intellectual disability, seizures, and autism spectrum disorder.	
GM1 gangliosidosis	GLB1	Beta-galactosidase
	Phenotype: Continuum of clinical features ranging from severe and	

	rapidly progressive disease to a milder and more slowly progressive course; infantile onset (type I) is characterized by early developmental delay/arrest followed by progressive neurodegeneration, skeletal dysplasia, facial coarseness, hepatosplenomegaly, and macular cherry red spot. Later onset forms (types II and III) are milder and observed as progressive neurologic disease and vertebral dysplasia. Adult onset presents mainly with dystonia.	
Sandhoff disease GM2 gangliosidosis, type II	HEXB	Beta-hexosaminidase A and B
	Phenotype: Infantile onset is characterized by rapidly progressive neurodegeneration, exaggerated startle reflex, "cherry red spot". Juvenile and late-onset forms of the disease can present with developmental regression and/or neurological impairment, such as ataxia, dystonia, spinocerebellar degeneration, and behavior changes.	
Sialidosis (ML I)	NEU1	Alpha-neuraminidase
	Phenotype: Continuum of clinical features ranging from severe disease (type II) to a milder and more slowly progressive course (type I). Clinical features range from early developmental delay, coarse facial features, short stature, dysostosis multiplex, and hepatosplenomegaly to late onset cherry-red spot myoclonus syndrome. Seizures, hyperreflexia, and ataxia have been reported in more than 50% of later-onset patients. A congenital form of the disease has been reported in which patients present with fetal hydrops or neonatal ascites.	
Galactosialidosis	CTSA	Cathepsin A causing secondary deficiencies in Beta-galactosidase and Alpha-neuraminidase
	Phenotype: Continuum of clinical features ranging from severe and rapidly progressive disease to a milder and more slowly progressive course; clinical features of the early infantile type include fetal hydrops, edema, ascites, visceromegaly, dysostosis multiplex, coarse facies, and cherry red spot. Most patients have milder presentations, which include ataxia, myoclonus, angiokeratoma, cognitive and neurologic decline.	
Mucopolipidosis II/III alpha/beta (ML II/III a/β)	GNPTAB(alpha/beta) GNPTG (gamma)	N-acetylglucosaminyl-1-phosphotransferase deficiency causing secondary intracellular deficiency of multiple enzyme activities
Mucopolipidosis III gamma (ML III?)	Phenotype: ML II is slowly progressive with features evident at birth. Common symptoms include skeletal abnormalities such as clubfeet, kyphosis, thoracic deformity, and deformed long bones, coarse facial features, gingival hyperplasia, and cardiovascular disease. ML III a/β is slowly progressive with onset in early childhood presenting as slowed growth, short stature, and joint pain and stiffness. ML III? presents similarly to ML III a/β but milder.	
Mucopolysaccharidosis IVB (Morquio B)	GLB1	Beta-galactosidase
	Phenotype: Progressive skeletal dysplasia with findings such as dysostosis multiplex, short stature, kyphoscoliosis, and genu/coxa valga. Corneal clouding is present in some individuals. Central nervous system	

	dysfunction, course facial features, and organ enlargement are not typical.	
Pompe disease (glycogen storage disease type II)	GAA	Alpha-glucosidase
	Phenotype: Infantile onset is characterized by prominent cardiomegaly, hypotonia, respiratory distress, and weakness with onset before age 12 months. Later onset disease includes individuals with onset before age 12 months without cardiomyopathy and all individuals with onset after age 12 months and is characterized by proximal muscle weakness and respiratory insufficiency. Clinically significant cardiac involvement is uncommon with late onset.	

Reference Values

An interpretive report will be provided.

Interpretation

This is a screening test; not all oligosaccharidoses are detected. The resulting excretion profile may be characteristic of a specific disorder; however, abnormal results require confirmation by enzyme assay or molecular genetic testing.

When abnormal results are detected with characteristic patterns, a detailed interpretation is given, including an overview of results and significance, a correlation to available clinical information, elements of differential diagnosis, recommendations for additional confirmatory studies (enzyme assay, molecular genetic analysis).

Cautions

This test may give false-negative results, especially in older patients with mild clinical presentations.

This test may give false-positive results for Pompe disease, especially in pediatric patients on infant formula.

Clinical Reference

1. Neufeld EF, Muenzer J. The mucopolysaccharidoses. In: Valle DL, Antonarakis S, Ballabio A, Beaudet AL, Mitchell GA. eds. The Online Metabolic and Molecular Bases of Inherited Disease. McGraw Hill; 2019. Accessed June 9, 2025. Available at <https://ommbid.mhmedical.com/content.aspx?bookId=2709§ionId=225544161>

2. Thomas GH. Disorders of glycoprotein degradation: Alpha-mannosidosis, beta-mannosidosis, fucosidosis, and sialidosis. In: Valle DL, Antonarakis S, Ballabio A, Beaudet AL, Mitchell GA. eds. The Online Metabolic and Molecular Bases of Inherited Disease. McGraw Hill; 2019. Accessed June 9, 2025. Available at <https://ommbid.mhmedical.com/content.aspx?bookid=2709§ionid=225545029>

3. Leslie N, Bailey L. Pompe Disease. In: Adam MP, Feldman J, Mirzaa GM, et al, eds. GeneReviews [Internet]. University of Washington, Seattle; 1993-2025. Updated November 21, 2019. Accessed June 9, 2025. Available at: <https://www.ncbi.nlm.nih.gov/books/NBK1261/>

4. Raas-Rothschild A, Spiegel R. Mucopolipidosis III Gamma. In: Adam MP, Feldman J, Mirzaa GM, et al, eds. GeneReviews [Internet]. University of Washington, Seattle; 1993-2025. Updated November 21,2019. Accessed June 9, 2025. Available at: www.ncbi.nlm.nih.gov/books/NBK24701/

5. Leroy JG, Cathey SS, Friez MJ. GNPTAB-Related Disorders.]. In: Adam MP, Feldman J, Mirzaa GM, et al, eds. GeneReviews [Internet]. University of Washington, Seattle; 1993-2025. Updated August 29, 2019.Accessed June 9, 2025. Available at: www.ncbi.nlm.nih.gov/books/NBK1828/

Performance

Method Description

Urine samples are extracted using Oasis HLB and carbograph columns and lyophilized overnight. Oligosaccharides are permethylated, replacing all hydroxy groups (-OH) with methoxy groups (-OCH3) and esterifies carboxyl groups (-COOH to -COOCH3). After permethylation, the tubes are centrifuged, and the supernatant removed from the sodium hydroxide pellet. The supernatant is quenched, neutralized, extracted onto an Oasis HLB column, eluted, and lyophilized again overnight. Specimens are resuspended, mixed with a matrix solution containing 2,5-dihydroxybenzoic acid, spotted onto a MALDI plate, and allowed to air dry. The plate is then analyzed using a matrix-assisted laser desorption/ionization tandem time-of-flight (MALDI TOF/TOF) 5800 Analyzer.(Xia B, Asif G, Arthur L, et al. Oligosaccharide analysis in urine by MALDI-TOF mass spectrometry for the diagnosis of lysosomal storage diseases. Clin Chem. 2013;59[9]:1357-1368, Hall PL, Lam C, Alexander JJ. Urine oligosaccharide screening by MALDI-TOF for the identification of NGLY1 deficiency. Mol Genet Metab. 2018;124[1]:82-86)

PDF Report

No

Day(s) Performed

Monday

Report Available

4 to 10 days

Specimen Retention Time

1 month

Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

Fees & Codes

Fees

- Authorized users can sign in to [Test Prices](#) for detailed fee information.
- Clients without access to Test Prices can contact [Customer Service](#) 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact [Customer Service](#).

Test Classification

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

84377

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
OLIGU	Oligosaccharide Screen, U	49284-3

Result ID	Test Result Name	Result LOINC® Value
64889	Oligosaccharide Screen, U	49284-3