Test Definition: MANN

Alpha-Mannosidase, Leukocytes

Overview

Useful For

Diagnosis of alpha-mannosidosis

This test is **not useful for** establishing carrier status for alpha-mannosidosis.

Genetics Test Information

Alpha-mannosidosis is an autosomal recessive lysosomal storage disorder caused by reduced or absent acid alpha-mannosidase enzyme activity.

Determining enzymatic activity is the next step of the diagnostic workup for an individual clinically suspicious for an oligosaccharidosis with a positive screening result suggestive of alpha-mannosidosis.

Special Instructions

- Informed Consent for Genetic Testing
- Biochemical Genetics Patient Information
- Informed Consent for Genetic Testing (Spanish)

Method Name

Fluorometric

NY State Available

Yes

Specimen

Specimen Type

Whole Blood ACD

Ordering Guidance

If clinically suspicious of an oligosaccharidosis, screening tests are available. Order either OLIGU / Oligosaccharide Screen, Random, Urine or LSDS / Lysosomal Disorders Screen, Random, Urine which includes a combined analysis of ceramide trihexosides, mucopolysaccharides, oligosaccharides, sulfatides, and total and free sialic acid.

Shipping Instructions

For optimal isolation of leukocytes, it is recommended the specimen arrive refrigerate within 6 days of collection to be stabilized.
Collect specimen Monday through Thursday only and not the day before a holiday. Specimen should be collected and packaged as close to shipping time as possible.

Specimen Required

Container/Tube:

Preferred: Yellow top (ACD solution B)
Acceptable: Yellow top (ACD solution A)
Specimen Volume: 6 mL
Collection Instructions: Send specimen in original tube. **Do not aliquot**.

Forms

1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available:
-Informed Consent for Genetic Testing (T576)
-Informed Consent for Genetic Testing-Spanish (T826)
2. Biochemical Genetics Patient Information (T602)
3. If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

Specimen Minimum Volume

5 mL

Reject Due To

Gross hemolysis	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole Blood ACD	Refrigerated (preferred)	6 days	YELLOW TOP/ACD
	Ambient	6 days	YELLOW TOP/ACD

Clinical & Interpretive

Clinical Information

Alpha-mannosidosis is an autosomal recessive lysosomal storage disorder caused by reduced or absent acid alpha-mannosidase enzyme activity. This enzyme is involved in glycoprotein catabolism, with absent or reduced activity resulting in the accumulation of undigested mannose-containing complex oligosaccharides in the lysosomes, disrupting the normal functioning of cells.

Clinical features and severity of symptoms are widely variable within alpha-mannosidosis but, in general, the disorder is characterized by skeletal abnormalities, immune deficiency, hearing impairment, and intellectual disability. Three clinical subtypes of the disorder have been described and vary with respect to age of onset and clinical presentation. Type 1 is generally classified by a mild presentation and slow progression with onset after 10 years of age and absence of skeletal abnormalities. Type 2 is generally a more moderate form with slow progression and onset prior to 10 years of age with skeletal abnormalities and myopathy. Type 3 is the most severe form with onset in early infancy, skeletal abnormalities such as dysostosis multiplex, and severe central nervous system involvement. Although treatment is mostly supportive and aimed at preventing complications, hematopoietic stem cell transplant has been reported to be a feasible therapeutic option. The incidence of alpha-mannosidosis is estimated at 1 in 500,000 live births.

An initial diagnostic workup may include a screening assay for several oligosaccharides in urine, OLIGU / Oligosaccharide Screen, Random, Urine. If the urine oligosaccharide screening assay is suggestive of alpha-mannosidosis, enzyme analysis of acid alpha-mannosidase can confirm the diagnosis. Molecular analysis of the *MAN2B1* gene allows for detection of a disease-causing variant in affected individuals and subsequent carrier detection in relatives (see CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing; specify *MAN2B1* gene list ID: IEMCP-MUMNLV).

Reference Values

> or =0.54 nmol/min/mg protein

Interpretation

Values below 0.54 nmol/min/mg protein are consistent with a diagnosis of alpha-mannosidosis.

Cautions

No significant cautionary statements

Clinical Reference

1. Malm D, Nilssen O. Alpha-mannosidosis. In: Adam MP, Ardinger HH, Pagon RA, et al, eds. GeneReviews [Internet]. University of Washington, Seattle. 2001. Updated July 18, 2019. Accessed June 10, 2025. Available at www.ncbi.nlm.nih.gov/books/NBK1396/
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 10, 2025. Available at https://ommbid.mhmedical.com/content.aspx?sectionid=225545029
3. Mynarek M, Tolar J, Albert MH, et al. Allogeneic hematopoietic SCT for alpha-mannosidosis: an analysis of 17 patients. Bone Marrow Transplant. 2012;47(3):352-359. doi:10.1038/bmt.2011.99
4. Guffon N, Tylki-Szymanska AT, Borgwardt L, et al. Recognition of alpha-mannosidosis in paediatric and adult patients: Presentation of a diagnostic algorithm from an international working group. Mol Genet Metab. 2019;126(4):470.474. doi:10.1016/j.ymgme.2019.01.024

Performance

Method Description

The deficiency of alpha-D-mannosidase is demonstrable using the artificial substrate 4-methylumberiferal alpha-D-mannopoyranoside.(Gehler J, Cantz M, Tolksdorf M, Spranger J, Gilbert E, Drube H. Mucopolysaccharidosis. VII. Beta-glucuronidase deficiency. Humangenetik. 1974;23[2]:149-158. doi:10.1007/BF00282212; Cowan T, Pasquali M. Laboratory investigations of inborn errors of metabolism. In: Sarafoglou K, Hoffman GF, Roth KS, eds. Pediatric Endocrinology and Inborn Errors of Metabolism. 2nd ed. McGraw-Hill; 2017:1139-1158)

PDF Report

No

Day(s) Performed

Preanalytical processing: Monday through Saturday
Assay performed: Friday

Report Available

2 to 8 days

Specimen Retention Time

WBC homogenate 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

82657

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
MANN	Alpha-Mannosidase, Leukocytes	24053-1

Result ID	Test Result Name	Result LOINC® Value
35639	Alpha-Mannosidase, Leukocytes	24053-1
35640	Interpretation (MANN)	59462-2
35641	Reviewed By	18771-6