

## Overview

### Useful For

Detection of fucosidosis

This test is **not useful for** establishing carrier status for fucosidosis.

### Genetics Test Information

Fucosidosis is an autosomal recessive lysosomal storage disorder caused by reduced or absent alpha-L-fucosidase enzyme activity.

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

### Testing Algorithm

See [Lysosomal Storage Disorders Diagnostic Algorithm, Part 1](#)

### Special Instructions

- [Informed Consent for Genetic Testing](#)
- [Biochemical Genetics Patient Information](#)
- [Informed Consent for Genetic Testing \(Spanish\)](#)
- [Lysosomal Storage Disorders Diagnostic Algorithm, Part 1](#)

### Method Name

Fluorometric

### NY State Available

Yes

## Specimen

### Specimen Type

Whole Blood ACD

### Ordering Guidance

If clinically suspicious of an oligosaccharidosis, a screening test is available. Order OLIGU / Oligosaccharide Screen, Random, Urine.

### 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
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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

Fucosidosis is an autosomal recessive lysosomal storage disorder caused by reduced or absent alpha-L-fucosidase enzyme activity. This enzyme is involved in degrading asparagine-linked, fucose-containing complex molecules (oligosaccharides and glycoasparagines) present in cells. Reduced or absent activity of this enzyme results in the abnormal accumulation of these molecules in the tissues and body fluids.

Severe and mild subgroups of fucosidosis, designated types I and II, have been described, although recent data suggests individual patients may represent a continuum within a wide spectrum of severity. The more severe type is characterized by infantile onset, rapid psychomotor regression, and severe neurologic deterioration. Additionally, dysostosis multiplex and elevated sweat sodium chloride are frequent findings. Death typically occurs within the first decade of life. Those with the milder phenotype express comparatively mild psychomotor and neurologic regression, radiologic signs of dysostosis multiplex, and skin lesions (angiokeratoma corporis diffusum). Normal sweat salinity, the presence of the skin lesions, and survival into adulthood most readily distinguish milder from more severe phenotypes.

Fucosidosis is an autosomal recessive condition resulting from two biallelic pathogenic variants in the *FUCA1* gene. Although the disorder is panethnic, the majority of reported patients with fucosidosis have been from Italy and southwestern United States. To date, about 100 cases have been reported worldwide.

An initial diagnostic workup includes a urine screening assay for several oligosaccharidosis (OLIGU / Oligosaccharide Screen, Random, Urine). If the screening assay is suggestive of fucosidosis, enzyme analysis of alpha-L-fucosidase can confirm the diagnosis.

**Reference Values**

> or =0.32 nmol/min/mg protein

**Interpretation**

Values below 0.32 nmol/min/mg protein are consistent with a diagnosis of fucosidosis.

**Cautions**

No significant cautionary statements

**Clinical Reference**

1. Enns GM, Steiner RD, Cowan TM: Lysosomal disorders. In: Sarafoglou K, Hoffmann GF, Roth KS, eds. Pediatric Endocrinology and Inborn Errors of Metabolism. McGraw-Hill Medical Division; 2009:747-748
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 February 17, 2022. Available at: <https://ommbid.mhmedical.com/content.aspx?sectionid=225545029>
3. Stepien KM, Ciara E, Jezela-Stanek A. Fucosidosis-clinical manifestation, long-term outcomes, and genetic profile-review and case series. Genes (Basel). 2020 Nov 22;11(11):1383. doi: 10.3390/genes11111383

**Performance****Method Description**

Incubation of 4-methylumbelliferyl-alpha-L-fucopyranoside with cell homogenates results in cleavage of the substrate by alpha-L-fucosidase yielding 4-methylumbelliferone (4-MU) and fucose. Free 4-MU can be quantitated by measurement of the fluorescence.(Beratis NG, Turner BM, Labadie G, Hirschhorn K: a-L-fucosidase in cultured skin fibroblasts from normal subjects and fucosidosis patients. Pediatr Res. 1977 Jul;11[7]:862-866; 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: Once per month

Report Available

30 to 45 days

Specimen Retention Time

WBC homogenate: 1 month

Performing Laboratory Location

Rochester

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
FUCW	Alpha-Fucosidase, Leukocytes	24047-3

Result ID	Test Result Name	Result LOINC® Value
8814	Alpha-Fucosidase, Leukocytes	24047-3
35635	Interpretation (FUCW)	59462-2
35634	Reviewed By	18771-6