Test Definition: FUCW

Alpha-Fucosidase, Leukocytes

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

For information see Lysosomal Disorders Diagnostic Algorithm, Part 1.

Special Instructions

- Informed Consent for Genetic Testing
- Biochemical Genetics Patient Information
- Informed Consent for Genetic Testing (Spanish)
- Lysosomal 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, 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 refrigerated within 6 days of collection to

be stabilized. Pre-analytical processing is performed Monday through Friday and Sunday. This test may be canceled if specimens are outside of stability when processing occurs. Collect and package specimens for arrival on days when processing is performed.

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

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 disease-causing variants in the *FUCA1* gene. Although the disorder is panethnic, the majority of reported patients with fucosidosis have been from Italy and the 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 June 9, 2025. 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;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;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 Friday, Sunday
Assay performed: Friday

Report Available

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
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