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 in Special Instructions.

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)
Test Definition: FUCW
Alpha-Fucosidase, Leukocytes

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 in Special Instructions:
   - Informed Consent for Genetic Testing (T576)
   - Informed Consent for Genetic Testing-Spanish (T826)

2. **Biochemical Genetics Patient Information** (T602) in Special Instructions

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**

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<th>Gross hemolysis</th>
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Specimen Stability Information

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<th>Specimen Type</th>
<th>Temperature</th>
<th>Time</th>
<th>Special Container</th>
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<tr>
<td>Whole Blood ACD</td>
<td>Refrigerated (preferred)</td>
<td>6 days</td>
<td>YELLOW TOP/ACD</td>
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<tr>
<td></td>
<td>Ambient</td>
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**Clinical and 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. 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**


**Performance**

**Method Description**


**PDF Report**

No

**Day(s) Performed**

Preanalytical processing occurs 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 and 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 Regional Manager. 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. This test has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information
82657

LOINC® Information

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<td>Alpha-Fucosidase, Leukocytes</td>
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