
Reporting Title: Fabry Disease Full Gene Analysis**Performing Location:** Rochester**Ordering Guidance:**

The recommended first-tier test for males with suspected Fabry disease is alpha-galactosidase A enzyme activity in blood or serum. Order either AGAW / Alpha-galactosidase, Leukocytes or AGAS / Alpha-galactosidase, Serum.

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

Specimen Requirements:

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Submit only 1 of the following specimens:**Preferred:**

Specimen Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA) tube or yellow top (ACD) tube

Acceptable: Any anticoagulant

Specimen Volume: 3 mL

Collection Instructions:

1. Invert several times to mix blood.
2. Send specimen in original tube.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Acceptable:

Specimen Type: Blood spot

Supplies: Card - Blood Spot Collection (Filter Paper) (T493)

Container/Tube:

Preferred: Collection card (Whatman Protein Saver 903 Paper)

Acceptable: Ahlstrom 226 filter paper, or Blood Spot Collection Card

Specimen Volume: 2 to 5 Blood spots on collection card

Collection Instructions:

1. An alternative blood collection option for a patient older than 1 year of age is finger stick.
2. Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours.
3. Do not expose specimen to heat or direct sunlight.
4. Do not stack wet specimens.
5. Keep specimen dry.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Additional Information:

1. For collection instructions, see [Blood Spot Collection Instructions](#) in Special Instructions.
2. For collection instructions in Spanish, see [Blood Spot Collection Card-Spanish Instructions](#) (T777) in Special Instructions.
3. For collection instructions in Chinese, see [Blood Spot Collection Card-Chinese Instructions](#) (T800) in Special Instructions.

Instructions.

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. [Molecular Genetics: Biochemical Disorders Patient Information](#) (T527) in Special Instructions.

| Specimen Type | Temperature | Time | Special Container |
|---------------|-------------|------|-------------------|
| Varies | Varies | | |

Result Codes:

| Result ID | Reporting Name | Type | Unit | LOINC® |
|-----------|------------------------|--------------|------|---------|
| 53894 | Result Summary | Alphanumeric | | 50397-9 |
| 53895 | Result | Alphanumeric | | 76036-3 |
| 53896 | Interpretation | Alphanumeric | | 69047-9 |
| 53897 | Additional Information | Alphanumeric | | 48767-8 |
| 53898 | Specimen | Alphanumeric | | 31208-2 |
| 53899 | Source | Alphanumeric | | 31208-2 |
| 53900 | Released By | Alphanumeric | | 18771-6 |

LOINC® and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

81405-GLA (galactosidase, alpha) (eg, Fabry disease), full gene sequence

Reference Values:

An interpretive report will be provided.