
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.

Specimen Minimum Volume:

Blood: 1 mL
Blood Spots: 5 punches-3 mm diameter

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.