

Test Definition: GBAMS

Gaucher Disease, Full Gene Analysis

Reporting Title: Gaucher Disease, Full Gene Analysis

Performing Location: Rochester

Specimen Requirements:

Forms:

1. New York Clients-Informed consent is required. Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions.

Molecular Genetics-Biochemical Disorders Patient Information Sheet (Supply T527) in Special Instructions

Specimen must arrive within 96 hours of collection.

Submit only 1 of the following specimens:

Specimen Type: Whole blood

Container/Tube:

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

Acceptable: Any anticoagulant Specimen Volume: 3 mL Collection Instructions:

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

Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Type: Cultured fibroblasts Container/Tube: T-75 or T-25 flask

Specimen Volume: 1 full T-75 or 2 full T-25 flasks

Specimen Stability Information: Ambient (preferred)/Refrigerated <24 hours

Specimen Type: Skin biopsy

Container/Tube: Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin. Tubes can be supplied upon request (Eagle's

minimum essential medium with 1% penicillin and streptomycin [Supply T115]).

Specimen Volume: 4-mm punch

Specimen Stability Information: Refrigerated (preferred)/Ambient

Acceptable:

Specimen Type: Blood spot

Container/Tube: Whatman Protein Saver 903 Paper

Specimen Volume: 5 blood spots

Collection Instructions:

- 1. Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours.
- 2. Do not expose specimen to heat or direct sunlight.
- 3. Do not stack wet specimens.
- 4. Keep specimen dry.

Specimen Stability Information: Ambient (preferred)/Refrigerated

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Specimen Type	Temperature	Time
Varies	Varies	

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Туре	Reportable
FBC	CG030	Source: • Autopsy • Chorionic Villi • Products of Conception or Stillbirth	Answer List	No
FBC	CG033	Reason for Referral	Plain Text	No

Result Codes:

Result ID	Reporting Name	Туре		LOINC®
33547	Specimen Alphanumeric			
33548	Specimen ID	Alphanumeric		
33549	Source	Alphanumeric		
33550	Order Date	Alphanumeric		
33551	Reason For Referral	Alphanumeric		
33552	Method	Alphanumeric		
33553	Result	Alphanumeric		
33554	Interpretation	Alphanumeric		
33555	Amendment	Alphanumeric		
33556	Reviewed By	Alphanumeric		
33557	Release Date	Alphanumeric		

Components:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
Billing only	NUCLEIC ACID HIGH RESOLUTE	22	83909		
Billing only	GENETIC EXAMINATION	1	83912		

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Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
Billing only	MOLECULE NUCLEIC AMPLI EACH	1	83898		
Billing only	MOLECULE GEL ELECTROPHOR	1	83894		
Billing only	MOLECULE ISOLATE NUCLEIC	1	83891		
Billing only	MOLECULAR DIAGNOSTICS	1	83892		
Billing only	GBA GENE	1	81251		

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
FBC	Fibroblast Culture for Genetic Test		Profile	No	Yes

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Туре	Unit	LOINC®
FBC	16167	Specimen	Alphanumeric		31208-2
FBC	16337	Specimen ID	Alphanumeric		
FBC	CG030	Source	Alphanumeric		
FBC	16168	Order Date	Alphanumeric		
FBC	CG033	Reason For Referral	Alphanumeric		42349-1
FBC	16172	Method	Alphanumeric		49549-9
FBC	16175	Interpretation	Alphanumeric		
FBC	16176	Amendment	Alphanumeric		
FBC	16177	Consultant	Alphanumeric		
FBC	16178	Report Date	Alphanumeric		



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Reference Values:

An interpretive report will be provided.