

Gaucher Disease, Mutation Analysis, GBA

Reporting Title: Gaucher Disease, Mutation Analysis

Performing Location: Rochester

Specimen Requirements:

Forms:

1. Molecular Genetics-Congenital Inherited Diseases Patient Information Sheet (Supply T521) in Special Instructions

2. 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.

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/Frozen

Due to the complexity of prenatal testing, consultation with the laboratory is required for all prenatal testing. Prenatal specimens can be sent Monday through Thursday and must be received by 5 p.m. CST on Friday in order to be processed appropriately. All prenatal specimens must be accompanied by a maternal blood specimen. Order MCC/88636 Maternal Cell Contamination, Molecular Analysis on both the prenatal and the maternal specimens.

Specimen Type: Amniotic fluid

Container/Tube: Amniotic fluid container

Specimen Volume: 20 mL

Specimen Stability Information: Refrigerated (preferred)/Ambient

Specimen Type: Chorionic villi

Container/Tube: 15-mL tube containing 15 mL of transport media

Specimen Volume: 20 ma

Specimen Stability Information: Refrigerated

Acceptable:

Specimen Type: Confluent cultured cells

Container/Tube: T-25 flask Specimen Volume: 2 flasks

Collection Instructions: Submit confluent cultured cells from another laboratory.

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
AFC	CG132	Reason For Referral	Plain Text	No

Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
16672	Specimen	Alphanumeric		
16673	Specimen ID	Alphanumeric		
16674	Source	Alphanumeric		
16675	Order Date	Alphanumeric		
16676	Reason For Referral	Alphanumeric		
16677	Method	Alphanumeric		
16678	Result	Alphanumeric		
16679	Interpretation	Alphanumeric		
16680	Amendment	Alphanumeric		
16681	Reviewed By:	Alphanumeric		
16682	Release Date	Alphanumeric		

Components:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
Billing only	Molecl Diag Isol & Extra	1	83890		

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Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
Billing only	Nucleic Acid Probe, each	8	83896		
Billing only	Mut Ident Enzy Lig/Primer	8	83914		
Billing only	Enzyme Digest	1	83892		
Billing only	Separation and ID	1	83909		
Billing only	Amp of Pt Nuc Acid, 1st 2	1	83900		
Billing only	Interpretation and Report	1	83912		

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
FBC	Fibroblast Culture for Genetic Test		Profile	No	Yes
AFC	Amniotic Fluid Culture/Genetic Test		Profile	No	Yes
MCC	Maternal Cell Contamination, B		Profile	No	Yes

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Туре	Unit	LOINC®
FBC	16167	Specimen	Alphanumeric		
FBC	16337	Specimen ID	Alphanumeric		
FBC	CG030	Source	Alphanumeric		
FBC	16168	Order Date	Alphanumeric		
FBC	CG033	Reason For Referral	Alphanumeric		
FBC	16172	Method	Alphanumeric		
FBC	16175	Interpretation	Alphanumeric		
FBC	16176	Amendment	Alphanumeric		
FBC	16177	Consultant	Alphanumeric		
FBC	16178	Report Date	Alphanumeric		
AFC	16179	Specimen	Alphanumeric		
AFC	16338	Specimen ID	Alphanumeric		



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Test ID	Result ID	Reporting Name	Туре	Unit	LOINC®
AFC	16793	Source	Alphanumeric		
AFC	16180	Order Date	Alphanumeric		
AFC	CG132	Reason For Referral	Alphanumeric		
AFC	16184	Method	Alphanumeric		
AFC	16187	Interpretation	Alphanumeric		
AFC	16188	Amendment	Alphanumeric		
AFC	16189	Consultant	Alphanumeric		
AFC	16190	Report Date	Alphanumeric		
мсс	23332	Specimen	Alphanumeric		
мсс	23333	Specimen ID	Alphanumeric		
мсс	23334	Source	Alphanumeric		
мсс	23335	Order Date	Alphanumeric		
мсс	23338	Result	Alphanumeric		
мсс	23337	Amendment	Alphanumeric		
мсс	23341	Reviewed By:	Alphanumeric		
MCC	23339	Release Date	Alphanumeric		

Reference Values:

An interpretive report will be provided.