Reporting Title: Sphingomyelinase, Fibroblasts
Performing Location: Rochester

Specimen Requirements:

This test is not recommended for prenatal testing.

Forms: 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.

Submit only 1 of the following specimens:

Specimen Type: Cultured fibroblasts
Container/Tube: T-75 or T-25 flask
Specimen Volume: 1 full T-75 flask or 2 full T-25 flasks
Specimen Stability Information: Ambient/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/Ambient

Specimen Type	Temperature	Time
Tissue	Varies	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
30035	Specimen	Alphanumeric		31208-2
30036	Specimen ID	Numeric		N/A
30037	Source	Alphanumeric		N/A
30038	Order Date	Alphanumeric		N/A
30039	Reason For Referral	Alphanumeric		42349-1
30043	Method	Alphanumeric		In Process
30044	Sphingomyelinase, Fibroblasts	Numeric	U/g Prot	In Process
30045	Interpretation	Alphanumeric		59462-2
30046	Amendment	Alphanumeric		In Process

Result ID	Reporting Name	Type	Unit	LOINC®
30047	Reviewed By	Numeric		N/A
30048	Release Date	Alphanumeric		N/A

CPT Code: 1 × 82657

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
FIBR	Fibroblast Culture	1	88233	Yes	Yes
CRYOB	Cryopreserve for Biochem Studies	1	88240	Yes	No

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
FIBR	8482	Fibroblast Culture	Alphanumeric		In Process
CRYOB	88832	Cryopreserve for Biochem Studies	Alphanumeric		N/A

Reference Values:

1.53-7.18 U/g of cellular protein