

Test Definition: NIEM

Niemann-Pick Type C Detection, Fibroblasts

Reporting Title: Niemann-Pick Type C, Fibro

Performing Location: Rochester

Specimen Requirements:

This test is not recommended for prenatal testing.

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.

2. If not ordering electronically, submit a Biochemical Genetics Request Form (Supply T439) with the specimen.

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 (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

Specimen Type	Temperature	Time
Tissue	Varies	

Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
30173	Cholesterol Esterification	Alphanumeric		In Process
29746	Interpretation (NIEM)	Alphanumeric		59462-2
29748	Reviewed By	Numeric		N/A

CPT Code: 1 x 82658



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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	Туре	Unit	LOINC®
FIBR	8482	Fibroblast Culture	Alphanumeric		In Process
CRYOB	88832	Cryopreserve for Biochem Studies	Alphanumeric		In Process

Reference Values:

If the results indicate that the patient's cultured fibroblasts esterify cholesterol at a level which is <10% of normal cultured fibroblasts and when filipin staining shows excessive storage of free cholesterol, it will be stated that the patient is positive for Niemann-Pick type C disease. All samples will be stained by filipin to see if a milder biochemical phenotype is the likely cause of the Niemann-Pick disease-like clinical picture.