



Patient ID SA00062931	Patient Name SAMPLEREPOR, NIEM A	Birth Date 1999-11-25	Gender M	Age 13
Order Number SA00062931	Client Order Number SA00062931	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 04 Oct 2013 11:12		

Cryopreserve for Biochem Studies

MCR

Cryopreserve for Biochem Studies was cancelled on 10/04/2013 at 11:16; Test not performed.

Received: 04 Oct 2013 11:16

Reported: 04 Oct 2013 11:16

Fibroblast Culture

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Fibroblast Culture was cancelled on 10/04/2013 at 11:16; Test not performed.

Received: 04 Oct 2013 11:16

Reported: 04 Oct 2013 11:16

Niemann-Pick Type C, Fibro

Cholesterol Esterification

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Cholesterol esterification = 4% of normal control cells.

Interpretation (NIEM)

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The esterification of cholesterol in response to stimulation of LDL cholesterol uptake is significantly depressed relative to normal and similar to the Niemann-Pick type C positive control. Filipin staining of free cholesterol is also abnormal as seen in NPC. These observations are consistent with the diagnosis of NPC. To date there are two clearly defined allelic variants. Molecular genetic testing to evaluate for mutations within the NPC1 and NPC2 genes is currently offered by the Mayo Molecular Genetics Laboratory (Test code 89015, NPCMS).

ADDITIONAL INFORMATION

Radiolabeled Lipid Extraction Using Thin Layer Chromatography

Reviewed By

MCR

Daniel Kraft

Received: 04 Oct 2013 11:16

Reported: 04 Oct 2013 14:23

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905