MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,NIEM A	M0002188	40	F	M0002188
Ordering Phys				**DOB** 09/28/1971
Client Order # M0002188	**Account Information**			**Report Notes**
Collected 06/27/2012 00:06	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 02/27/2013 12:34				

Test	Flag	Results	Unit	Reference Value	Perform Site*
			REPORTED 02/27/2013 12:10		
Cryopreserve for Biochem Studies					MCR
Fibroblasts successfully cryopreserved.					
			REPORTED 02/27/2013 12:10		
Fibroblast Culture					MCR
Fibroblasts successfully cultured.					
Niemann-Pick Type C, Fibro			REPORTED 02/21/2013 09:49		
Specimen		Fibroblasts			MCR
Reason For Referral		Not provided			MCR
Cholesterol Esterification					MCR
Cholesterol esterification = 3% of normal control cells.					
Interpretation					MCR
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). Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.					
Radiolabeled Lipid Extraction Using Thin Layer Chromatography					
Reviewed By		Matthew Fatehi			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,NIEM A	06/27/2012 00:06	Final
Page 1 of 1		** End of Report **

* Report times for Mayo performed tests are CST/CDT