



Patient ID SA00064503	Patient Name TESTINGRNV, OLITCABNORMAL	Birth Date 1985-02-11	Gender F	Age 28
Order Number SA00064503	Client Order Number SA00064503	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 18 Nov 2013 00:00		

Cryopreserve for Biochem Studies

MCR

Fibroblasts successfully cryopreserved.

Received: 19 Nov 2013 12:27

Reported: 19 Nov 2013 12:47

Fibroblast Culture

MCR

Fibroblasts successfully cultured.

Received: 19 Nov 2013 12:27

Reported: 19 Nov 2013 12:49

Oligosaccharidoses Screen, Fibro

Oligosaccharidoses Screen, Fibro

MCR

In this specimen, beta-mannosidase activity is reduced. This result is consistent with a diagnosis of beta-mannosidosis (OMIM 248510), a rare autosomal recessive lysosomal storage disorder. If clinically indicated, consider further confirmatory testing by molecular genetic analysis of the MANBA gene. A genetics consultation is recommended. Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.

ADDITIONAL INFORMATION

Enzyme reaction followed by FIA-MS/MS

Reviewed By

MCR

JANICE HELGESON

Received: 19 Nov 2013 12:27

Reported: 19 Nov 2013 12:39

Performing Site Legend

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