



Patient ID SA00062414	Patient Name SAMPLEREP, MANT A	Birth Date 1994-10-26	Gender F	Age 18
Order Number SA00062414	Client Order Number SA00062414	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 17 Sep 2013 10:36		

Cryopreserve for Biochem Studies

MCR

Cryopreserve for Biochem Studies was cancelled on 09/17/2013 at 11:19; Test not performed.

Received: 17 Sep 2013 11:19

Reported: 17 Sep 2013 11:19

Fibroblast Culture

MCR

Fibroblast Culture was cancelled on 09/17/2013 at 11:19; Test not performed.

Received: 17 Sep 2013 11:19

Reported: 17 Sep 2013 11:19

Alpha-Mannosidase, Fibroblasts

Alpha-Mannosidase, Fibroblasts

MCR



0.23 nmol/min/mg Prot

Reference Value
≥0.53

Reviewed By

MCR

Daniel Kraft

Received: 17 Sep 2013 11:19

Reported: 17 Sep 2013 13:47

Interpretation (MANT)

MCR

In this specimen, alpha-mannosidase activity is reduced. This result is consistent with a diagnosis of alpha-mannosidosis (OMIM 248500), a rare autosomal recessive lysosomal storage disorder. If clinically indicated, consider further confirmatory testing by molecular genetic analysis of the MAN2B1 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

Fluorometric Enzyme Assay

Performing Site Legend

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