



Patient ID SA00062731	Patient Name SAMPLEREP, ANAT A	Birth Date 1958-06-24	Gender M	Age 55
Order Number SA00062731	Client Order Number SA00062731	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 24 Sep 2013 12:05		

A-N-Acetylglucosaminidase, Fibro

A-N-Acetylglucosaminidase

MCR



0.01 nmol/min/mg Prot

Reference Value
≥ 0.05

Low

Reviewed By

MCR

Daniel Kraft

Received: 24 Sep 2013 12:12

Reported: 24 Sep 2013 12:49

Interpretation (ANAT)

MCR

In this specimen, the activity of N-acetylglucosaminidase is significantly reduced. This result indicates that this individual is affected with Sanfilippo type B disease (N-acetylglucosaminidase deficiency). If clinically indicated, consider further confirmation by molecular genetic analysis of the NAGLU gene. Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.

ADDITIONAL INFORMATION

Colorimetric Enzyme Assay

Cryopreserve for Biochem Studies

MCR

Cryopreserve for Biochem Studies was cancelled on 09/24/2013 at 12:13; Test not performed.

Received: 24 Sep 2013 12:12

Reported: 24 Sep 2013 12:13

Fibroblast Culture

MCR

Fibroblast Culture was cancelled on 09/24/2013 at 12:13; Test not performed.

Received: 24 Sep 2013 12:12

Reported: 24 Sep 2013 12:13

Performing Site Legend

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