

Patient ID M0001996	Patient Name SAMPLEREPORT, AGABS A	Birth Date 1971-09-28	Gender F	Age 40	
Order Number M0001996	Client Order Number M0001996	Ordering Physician ,	Report Notes		
Account Information C7028846 DLMP Rochester		Collected 26 Jun 2012 23:56			

Alpha-Galactosidase, BS

Reason For Referral

Not provided

MCR

Alpha-Galactosidase, BS



0.5 nmol/mL/h

Reference Value
≥2.8

MCR

Interpretation

In this specimen, the activity of alpha-galactosidase enzyme is reduced indicating that this individual is a carrier for Fabry disease (OMIM #301500). Carriers may or may not be symptomatic. If clinically indicated, consider follow-up investigations to include molecular genetic analysis of the GLA

MCR

gene (MML 88264). If not already ordered, this could be done on the existing sample by calling MML within one month. In addition, the Fabry Registry website (<http://www.Fabryregistry.com>) provides information and resources for health professionals and families.

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

Reviewed By

Marie Quade

MCR

Received: 26 Jun 2012 23:56

Reported: 06 Feb 2013 16:01

Performing Site Legend

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