



Patient ID M0001995	Patient Name SAMPLEREPORT, AGABS N	Birth Date 1971-09-28	Gender F	Age 40
Order Number M0001995	Client Order Number M0001995	Ordering Physician ,	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 26 Jun 2012 23:55		

Alpha-Galactosidase, BS

Reason For Referral

Not provided

MCR

Alpha-Galactosidase, BS

5.2 nmol/mL/h

MCR

Reference Value
≥2.8

Interpretation

MCR

In this specimen, the activity of alpha-galactosidase is normal, indicating this patient is most likely NOT a carrier for Fabry disease (OMIM #301500). However, please note that enzyme analysis is, in general, not sufficiently sensitive to detect all carrier females (see Hwu WL et al. Hum Mutat 2009; 30: 1-9; and Wilcox WR et al. Mol Genet Metab. 2008; 93: 112-128). Molecular genetic analysis of the GLA gene (MML 88264) is the most reliable method of detecting carriers. If not already ordered, this could be done on the existing sample by calling MML within one month.

ADDITIONAL INFORMATION

Fluorometric Enzyme Assay

Reviewed By

MCR

Marie Quade

Received: 26 Jun 2012 23:55

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