



Patient ID <b>SA00056622</b>	Patient Name <b>SAMPLEREPOR, AGAS N</b>	Birth Date <b>1966-06-10</b>	Gender <b>F</b>	Age <b>46</b>
Order Number <b>SA00056622</b>	Client Order Number <b>SA00056622</b>	Ordering Physician <b>Client, Client</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>16 Apr 2013 00:00</b>		

## Alpha-Galactosidase, S

### Reason For Referral

Not provided

MCR

### Interpretation

MCR

In this specimen, the activity of alpha-galactosidase is normal, indicating this individual is most likely NOT a carrier for Fabry disease (OMIM #301500). However please note that enzyme analysis, in general, is 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 is the most reliable method of detecting carriers. Consider submitting a new blood specimen and ordering Fabry Disease Full Gene Analysis (MML 88264).

#### ADDITIONAL INFORMATION

Fluorometric Enzyme Assay

### Reviewed By

Marie Quade

MCR

**Received:** 17 Apr 2013 14:38

**Reported:** 17 Apr 2013 15:21

### Alpha-Galactosidase,S

0.098 U/L

MCR

**Reference Value**  
0.074-0.457

### Performing Site Legend

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