



Patient ID <b>SA00057866</b>	Patient Name <b>SAMPLEREP, AGA N</b>	Birth Date <b>1966-06-10</b>	Gender <b>F</b>	Age <b>46</b>
Order Number <b>SA00057866</b>	Client Order Number <b>SA00057866</b>	Ordering Physician <b>Client, Client</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>29 May 2013 00:00</b>		

## Alpha-Galactosidase, Leukocytes

### Reason for Referral

Not provided

### ADDITIONAL INFORMATION

Fluorometric Enzyme Assay

MCR

### Alpha-Galactosidase, Leukocytes

MCR

**50.3 nmol/h/mg**

**Reference Value**  
≥23.1

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.

**Received:** 30 May 2013 11:12

**Reported:** 13 Jun 2013 08:33

### Performing Site Legend

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