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|---|---|---|--------------------|------------------|
| Patient ID<br><b>SA00056623</b>                       | Patient Name<br><b>SAMPLEREPORT, AGAS A</b> | Birth Date<br><b>1966-06-10</b>             | Gender<br><b>F</b> | Age<br><b>46</b> |
| Order Number<br><b>SA00056623</b>                     | Client Order Number<br><b>SA00056623</b>    | Ordering Physician<br><b>Client, Client</b> | Report Notes       |                  |
| Account Information<br><b>C7028846 DLMP Rochester</b> |   | Collected<br><b>16 Apr 2013 00:00</b>       |                    |                  |

## Alpha-Galactosidase, S

### Reason For Referral

Not provided

MCR

### Alpha-Galactosidase,S



0.065 U/L

MCR

Reference Value  
0.074–0.457

### Interpretation

MCR

In this specimen, the activity of alpha-galactosidase is reduced, indicating this individual may be a carrier for Fabry disease (OMIM #301500). Many carriers are symptomatic. If clinically indicated, consider follow-up investigations to include molecular genetic analysis of the GLA gene (MML 88264). In addition, the Fabry Registry website (<http://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

MCR

Marie Quade

**Received:** 17 Apr 2013 14:52

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

### Performing Site Legend

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