



Patient ID <b>SA00056675</b>	Patient Name <b>TESTINGRNV, CBGTNORMAL</b>	Birth Date <b>1953-09-26</b>	Gender <b>F</b>	Age <b>59</b>
Order Number <b>SA00056675</b>	Client Order Number <b>SA00056675</b>	Ordering Physician <b>Client, Client</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>18 Apr 2013 00:00</b>		

## Galactocerebrosidase, Fibro

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1.23 nmol/h/mg Prot

MCR  
Reference Value  
≥1.20

### Reviewed By

JANICE HELGESON

MCR

Received: 19 Apr 2013 10:53

Reported: 19 Apr 2013 11:13

### Interpretation (CBGT)

MCR

In this specimen, the activity of galactocerebrosidase is not consistent with a possible diagnosis of Krabbe disease (OMIM 245200). Individuals with Saposin A deficiency (OMIM 611722) may have normal galactocerebrosidase activity in fibroblasts (Spiegel et al. Mol Genet Metab. 2005; 84: 160–166). If clinical presentation is suggestive of Krabbe disease, consider galactocerebrosidase testing in leukocytes.

#### ADDITIONAL INFORMATION

Radioisotopic Enzyme Assay

### Cryopreserve for Biochem Studies

MCR

Fibroblasts successfully cryopreserved.

Received: 19 Apr 2013 10:53

Reported: 19 Apr 2013 11:47

### Fibroblast Culture

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Fibroblasts successfully cultured.

Received: 19 Apr 2013 10:53

Reported: 19 Apr 2013 11:45

#### Performing Site Legend

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