



Patient ID <b>SA00056676</b>	Patient Name <b>TESTINGRNV, CBGTABNORMAL</b>	Birth Date <b>2000-11-01</b>	Gender <b>M</b>	Age <b>12</b>
Order Number <b>SA00056676</b>	Client Order Number <b>SA00056676</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

### Galactocerebrosidase, Fibro



0.00 nmol/h/mg Prot

Low

MCR

Reference Value  
≥1.20

Foundation (<http://www.huntershope.org>) provides information and resources about Krabbe disease for health professionals and families. Information about ongoing studies into the natural history and treatment of Krabbe disease can be found at <http://www.clinicaltrials.gov>. Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.

#### ADDITIONAL INFORMATION

Radioisotopic Enzyme Assay

#### Reviewed By

JANICE HELGESON

MCR

Received: 19 Apr 2013 10:54

Reported: 19 Apr 2013 11:13

### Interpretation (CBGT)

MCR

In this specimen, the activity of galactocerebrosidase is reduced and consistent with a diagnosis of Krabbe disease (OMIM 245200). Please note that low activity can also be seen in patients with Saposin A deficiency (OMIM 611722), a condition clinically similar to Krabbe disease. If clinically indicated, consider further confirmation by molecular genetic analysis of the GALC gene (MML: 60696) and/or measuring activity of galactocerebrosidase in fibroblasts (MML: 8297). A genetics consultation is recommended. In addition, the Hunter's Hope

### Cryopreserve for Biochem Studies

MCR

Fibroblasts successfully cryopreserved.

Received: 19 Apr 2013 10:54

Reported: 19 Apr 2013 11:47

### Fibroblast Culture

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Fibroblasts successfully cultured.

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### Performing Site Legend

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