

Patient ID SA00056673	Patient Name TESTINGRNV, CBGCABNORMAL	Birth Date 1987-08-10	Gender M	Age 25
Order Number SA00056673	Client Order Number SA00056673	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 18 Apr 2013 00:00		

Galactocerebrosidase, WBC

Galactocerebrosidase, WBC



Low

0.00 nmol/h/mg Prot

MCR

Reference Value
≥1.20

Interpretation (CBGC)

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

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 10:52

Reported: 19 Apr 2013 11:13

Performing Site Legend

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