MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,CBGCABNORMAL	SA00056673	25	M	SA00056673
Ordering Phys CLIENT,CLIENT				**DOB** 08/10/1987
Client Order # SA00056673	**Account Information**			**Report Notes**
Collected 04/18/2013	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 04/30/2013 09:32				

Test	Flag	Results	Unit	Reference Value	Perform Site*
			REPORTED 04/19/2013 11:13		
Galactocerebrosidase, WBC	L	0.00	nmol/h/mg Prot	>=1.20	MCR
Interpretation (CBGC)					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 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.
Radioisotopic Enzyme Assay

Reviewed By	JANICE HELGESON	MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:

Patient Name	Collection Date and Time	Report Status
TESTINGRNV,CBGCABNORMAL	04/18/2013	Final
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* Report times for Mayo performed tests are CST/CDT