

Laboratory Service Report

1-800-533-1710

MCR

Patient Name	Patient ID	Age		Order #
TESTINGRNV,CBGTABNORMAL Ordering Phys	SA00056676	12	M	SA00056676 DOB
CLIENT, CLIENT				11/01/2000
Client Order # SA00056676	Account Information			Report Notes
Collected 04/18/2013	3050 Superior Drive			
Printed 04/30/2013 09:39	Rochester, MN 55901			

Test	Flag Results	Unit	Reference Value	Perform Site*
llactocerebrosidase,Fibro		REPORTED 04/	19/2013 11:13	
Galactocerebrosidase, Fibro	L 0.00	nmol/h/mg Pro	t >=1.20	MCR
Interpretation (CBGT)				MCR
In this specimen, the activi reduced and consistent with (OMIM 245200). Please note the seen in patients with Saposis condition clinically similar clinically indicated, conside molecular genetic analysis of and/or measuring activity of fibroblasts (MML: 8297). A grecommended. In addition, the (http://www.huntershope.org) resources about Krabbe diseat families. Information about natural history and treatment found at http://www.clinical Biochemical Genetics consult call (1-800-533-1710) if you Radioisotopic Enzyme Assay	a diagnosis of Krabbe dischat low activity can also hat low activity can also hat low activity can also had deficiency (OMIM 6117: to Krabbe disease. If er further confirmation by the GALC gene (MML: 606: galactocerebrosidase in enetics consultation is entered to the Hunter's Hope Foundation provides information and se for health professional congoing studies into the tof Krabbe disease can be trials.gov. Please contact ant or genetic counselor	ease be 22), a y 96) n ls and e t the		
Reviewed By	JANICE HELGESO	ON		MCR
yopreserve for Biochem Studies Fibroblasts successfully cry	opreserved.	REPORTED 04/	19/2013 11:47	MCR
		REPORTED 04/	19/2013 11:46	

Fibroblast Culture

Fibroblasts successfully cultured.

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

Patient Name		Report Status
TESTINGRNV,CBGTABNORMAL	04/18/2013	Final
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