

Patient Name TESTINGRNV,GCTABNORMAL	Patient ID SA00066659	Age 29	Gender F	Order # SA00066659
Ordering Phys CLIENT,CLIENT			DOB 02/11/1985	
Client Order # SA00066659	Account Information			Report Notes
Collected 04/01/2014 00:00	C7028846-DLMP Rochester SDSC 2 - Client Support Rochester, MN 55901			
Printed 04/17/2014 09:13				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Galactosemia Reflex, B					
RECEIVED: 04/02/2014 12:52 REPORTED: 04/02/2014 13:00					
Gal-1-P Uridyltransferase, RBC	L	0.4	nmol/h/mg Hb	>=24.5	MCR
Interpretation (GALT)					
The galactose-1-phosphate uridyltransferase (GALT) activity in this sample is reduced and most consistent with the Duarte variant galactosemia (partial GALT deficiency; DG). Consider a galactose-free diet until the diagnosis is verified by molecular genetic testing. Molecular genetic test results to follow. Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.					
Enzyme reaction followed by LC-MS/MS					
Reviewed By	JANICE HELGESON				MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:
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Patient Name TESTINGRNV,GCTABNORMAL	Collection Date and Time 04/01/2014 00:00	Report Status Final
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* Report times for Mayo performed tests are CST/CDT