

Laboratory Service Report

1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #		
SAMPLEREPORT, GALTP	SA00047618	13 M	F	SA00047618		
Ordering Phys		•	•	DOB		
				06/10/2011		
Client Order #	Account Information			Report Notes		
SA00047618						
Collected	C7028846-DLMP ROC	HESTER				
07/11/2012 14:10		3050 SUPERIOR DRIVE				
Printed	ROCHESTER,MN 5590	01				
08/14/2012 11:22						

Reference Perform Test Flag Results Unit Value Site*

> REPORTED 07/12/2012 15:20 >=18.5

7.0 U/g Hb

Gal-1-P Uridyltransferase, RBC Comment.

> The galactose-1-phosphate uridyltransferase (GALT) activity in this sample is reduced, however, not consistent with classic galactosemia (galactose-1-phosphate uridyltransferase deficiency). Partial enzyme deficiency can be explained by a variety of genotypes. If clinically indicated or if carrier testing is desired, consider molecular genetic testing (MML #84366). If not already ordered, this could be done on the existing sample by calling MML within one month. Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions. Ultraviolet, Kinetic

> > REPORTED 07/12/2012 15:22

Not Applicable

MCR

MCR

MCR MCR

Gal-1-Phos Urdyltrns Phenotype, RBC

Galactose-1-Phosphate uridyltransferase activity and isoelectric focusing are not consistent with galactosemia. Biochemical phenotyping suggests DD genotype (DD Genotype: Duarte homozygote). Since presence of the Los Angeles variant may exhibit results similar to those of Duarte homozygotes, molecular testing is recommended to clarify the carrier status of this patient.

Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any

questions. Reviewed By

Isoelectric Focusing

Marie Quade

Performing Site:

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

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT, GALTP	07/11/2012 14:10	Final
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^{*} Report times for Mayo performed tests are CST/CDT