

Patient ID <b>SA00047618</b>	Patient Name <b>SAMPLEREPOR, GALTP</b>	Birth Date <b>2011-06-10</b>	Gender <b>F</b>	Age <b>13 M</b>
Order Number <b>SA00047618</b>	Client Order Number <b>SA00047618</b>	Ordering Physician <b>Client, Client</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>11 Jul 2012 14:10</b>		

## Gal-1-P Uridyltransferase, RBC

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**7.0 U/g Hb**

Low

MCR

 Reference Value  
 ≥18.5

### Interpretation (GALT)

MCR

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.

#### ADDITIONAL INFORMATION

Ultraviolet, Kinetic

**Received:** 12 Jul 2012 15:06

**Reported:** 12 Jul 2012 15:20

## Gal-1-Phos Urdyltrns Phenotype,RBC

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MCR

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.

 Reference Value  
 Not Applicable

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

### Reviewed By

MCR

Marie Quade

#### ADDITIONAL INFORMATION

Isoelectric Focusing

**Received:** 12 Jul 2012 15:06

**Reported:** 12 Jul 2012 15:22

### Performing Site Legend

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