



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

Gal-1-P Uridyltransferase, RBC

Gal-1-P Uridyltransferase, RBC



Low

7.0 U/g Hb

MCR

Reference Value
≥18.5

Comment

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

Gal-1-Phos Urdyltrns Phenotype,RBC

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.

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

Reference Value
Not Applicable

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	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292