

Laboratory Service Report

1-800-533-1710

REPORTED 11/19/2011 14:03

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Patient Name REPORTVALIDATION, AUTOMATION DN	Patient ID RVDNPLB065	Age 40	Gender F	Order # RVDNPLB065
Ordering Phys				DOB 01/01/1971
Client Order # RVDNPLB065	Account Information			Report Notes
Collected 11/18/2011 11:32	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE			
Printed 11/21/2011 11:04	ROCHESTER,MN 55901			

Reference Perform
Test Flag Results Unit Value Site*

UGT1A1 Gene, Known Mutation, Saliva

UGT1A1 Gene, Known Mutation

The following familial mutation was NOT detected in the UGT1A1 gene: Heterozygous *28 (TA 6/7) (c.- $40_-39insTA$), heterozygous *12 (c.524T>A, p.L175Q).

Reviewed by Jamie Bruflat

UGT1A1 Gene, Known Mutation Interp

The above mutations within the UGT1A1 gene were previously identified in an affected member of this family. Since these mutations were not detected in this individual, this suggests that this patient is at no greater risk than someone in the general population for having unconjugated hyperbilirubinemia.

Bidirectional DNA sequence analysis was used to test for the presence of a specific mutation(s) in the UGT1A1 gene which was previously identified in an affected family member of this individual. The presence of UGT1A1 mutations does not necessarily confirm a diagnosis of unconjugated hyperbilirubinemia. Clinical correlation is recommended. A genetic consultation may be of benefit.

CAUTIONS: Rare polymorphisms exist that could lead to false negative or positive results. Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Large deletions or rearrangements are not detected by this assay, and these may affect UGT1A1 protein expression, and the ability to conjugate bilirubin.

Laboratory developed test.

UGT1A1 Known Mutation Sequencing Performed

* Performing Site:

MCR Mayo Clinic Dpt of Lab Med & Pathology
200 First St SW Rochester, MN 55905

Lab Director: Franklin R. Cockerill, III, M.D.

Patient Name REPORTVALIDATION, AUTOMATION DN	Collection Date and Time 11/18/2011 11:32	Report Status Final
Page 1 of 1		** End of Report **

^{*} Report times for Mayo performed tests are CST/CDT