

## **Laboratory Service Report**

1-800-533-1710

REPORTED 11/19/2011 13:45

| Patient Name REPORTVALIDATION, AUTOMATION DN | Patient ID<br>RVDNPLB064                       | Age<br>40 | Gender<br>F | Order #<br>RVDNPLB064 |
|--|--|-----------|-------------|-----------------------|
| Ordering Phys                                |  |           |             | <b>DOB</b> 01/01/1971 |
| Client Order #<br>RVDNPLB064                 | Account Information                            |           |             | Report Notes          |
| <b>Collected</b> 11/18/2011 11:32            | C7028846-DLMP ROCHESTER<br>3050 SUPERIOR DRIVE |           |             |                       |
| <b>Printed</b> 11/21/2011 11:03              | ROCHESTER,MN 55901                             |           |             |                       |

Reference Perform
Test Flag Results Unit Value Site\*

## UGT1A1 Sequence, Hyperbili, Saliva

UGT1A1 Sequence, Hyperbilirubinemia UGT1A1 Hyperbilirubinemia Result

A mutation was NOT detected in the UGT1A1 gene.

Reviewed by Jamie Bruflat

UGT1A1 Hyperbilirubinemia Interp

Both copies of the UGT1A1 gene have the normal TA6 (\*1) promoter repeat and no additional UGT1A1 mutations were identified, which is consistent with normal activity of the UGT1A1 enzyme and would not be expected to cause marked unconjugated hyperbilirubinemia.

Bidirectional DNA sequence analysis was used to test for the presence of mutations in the promoter, exons, exon-intron boundaries, and 3'-untranslated region of the UGT1A1 gene that are associated with the diagnosis of unconjugated hyperbilirubinemia. A small percentage of individuals who have a diagnosis of unconjugated hyperbilirubinemia may have a mutation that is not identified by the methods described above. The presence of UGT1A1 mutations does not necessarily confirm a diagnosis of unconjugated hyperbilirubinemia. Breast-fed neonates may experience a physiologic unconjugated hyperbilirubinemia and jaundice from deconjugation of maternal bilirubin-glucuronides present in breast milk. Clinical correlation is recommended.

A genetic consultation may be of benefit.

A list of common polymorphisms identified for this patient is available from the laboratory upon request.

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.

UGT, Full Gene Sequencing

Performed

MCR

MCR

MCR

MCR

## \* Performing Site:

| MCR | Mayo Clinic Dpt of Lab Med & Pathology<br>200 First St SW Rochester, MN 55905 | Lab Director: |  |
|-----|---|---------------|--|

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

<sup>\*</sup> Report times for Mayo performed tests are CST/CDT