

TEST DEFINITION

12/6/2011

Name/order code cross-reference

CODE NAME

UGT20 UGT1A1 SEQUENCE, HYPERBILI, SALIVA

TEST SETUP INFORMATION

ORDER RESULT

CODE CODE TITLE

CHECKING NORMALS

PRINT NORMALS

SITE *

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UGT2O(PROFILE) UGT1A1 SEQUENCE, HYPERBILI, SALIVA

UGTHO UGT1A1 Sequence, Hyperbilirubinemia MCR

TRANSPORT TEMP : AMBIENT

33028 UGT1A1 Hyperbilirubinemia Result

Units:

33029 REVIEWED BY

UNITS:

33030 UGT1A1 HYPERBILIRUBINEMIA INTERP

UNITS:

TEST CODE ALWAYS MESSAGE - [NP0384]

NP0384 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.

TEST SETUP INFORMATION

ORDER RESULT PERFORM CODE CODE TITLE CHECKING NORMALS PRINT NORMALS SITE *

____ UGT20 (CONTINUED...)

> 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.

TEST CODE ALWAYS MESSAGE - [TLDT] TLDT LABORATORY DEVELOPED TEST.

UGTSO UGT, FULL GENE SEQUENCING

TRANSPORT TEMP : AMBIENT\REFRIGERATED

UGTSQ UGT, FULL GENE SEQUENCING

UNITS:

*PERFORMING SITE LEGEND

MCR

MCR MAYO CLINIC DEPT. OF LAB MED AND PATHOLOGY

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RESULT CODES USED BY OTHER ORDER CODES

(This list shows other order codes that use these result codes. They may or may not be available to all clients.)

RESULT USED BY ORDER CODE CODE

UGTSQ UGT2, UGTI, UGTIO

END OF REPORT ***