MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,UGT2	SA00055147	46	F	SA00055147
Ordering Phys UNKNOWN,PROVIDER				**DOB** 06/10/1966
Client Order # SA00055147	**Account Information**			**Report Notes**
Collected 03/15/2013 01:20	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 05/23/2013 11:36				

Test	Flag	Results	Unit	Reference Value	Perform Site*
UGT1A1 Sequence, Hyperbilirubinemia			REPORTED 05/23/2013 10:45		
UGT1A1 Sequence, Hyperbilirubinemia					
UGT1A1 Hyperbilirubinemia Result					MCR
This individual was shown to have the following mutation(s) in the UGT1A1 gene: homozygous *28(TA 7/7)(c.-40_-39insTA)					
UGT1A1 Hyperbilirubinemia Interp					MCR
Homozygosity for the *28 TA7 promoter repeat polymorphism is consistent with a diagnosis of UGT1A1-dependent unconjugated hyperbilirubinemia and Gilbert syndrome. 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.					
Reviewed by		Yvonne Philo			MCR
UGT, Full Gene Sequencing		Performed			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,UGT2	03/15/2013 01:20	Final
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* Report times for Mayo performed tests are CST/CDT