Patient Name:	NA	**Booking ID:**	NA
Age:	NA	**Sample Type:**	NA
Gender:	NA	**Sample collection date:**	DD-MM-YYYY
Referring Clinician:	NA	**Sample receiving date:**	DD-MM-YYYY
Test Requested:	*UGT1A1 Genotyping for Gilbert Syndrome by PCR, Sanger Seq.*	**Reporting date:**	DD-MM-YYYY

UGT1A1 Genotyping for Gilbert Syndrome (Qualitative)

CLINICAL INFORMATION

N/A

RESULTS

GENOTYPE NM_000463	EFFECT OF POLYMORPHISM	UGT1A1 GENOTYPE STATUS
UGT1A1* 1 A(TA)6TAA	Normal enzyme activity	Not Detected
UGT1A1* 28 A(TA)7TAA	Minimal enzyme activity	Homozygous Variant Identified
UGT1A1* 37 A(TA)8TAA	Minimal enzyme activity	Not Detected
UGT1A1* 6 G71R	Reduced enzyme activity	Not Detected
UGT1A1* 7 Y486D	Reduced enzyme activity	Not Detected

DETAILED RESULT INFORMATION

UGT1A1*28/28 genotype was detected in the specimen provided. The patient is homozygous for (TA)7 genotype. The findings are suggestive of a diagnosis of Gilbert Syndrome.

CLINICAL INTERPRETATION

- ✓ Please correlate clinically.
- ✓ Results should be interpreted in context of clinical findings, relevant history, and other laboratory data.
- ✓ Gilbert Syndrome is an inherited from of unconjugated hyperbilirubinaemia resulting in mild jaundice occurring in the absence of haemolysis or underlying liver disease. Individuals with Gilbert Syndrome have a reduced level of UGT1A1, the enzyme required for the conjugation and clearance of bilirubin.
- ✓ Genetically, 90-95% of these individuals are homozygous for the presence of two extra bases (TA) in the promoter region of UGT1A1 and have a (TA) 7TAA sequence. This allele is called UGT1A1*28 & result in minimal enzyme activity. A missense change in the UGT1A1 gene, G71R, has also been identified in 30-40 % patients of neonatal hyper-bilirubinemia & implicated in Gilbert Syndrome. This allele, known as UGT1A1*6, also result in reduced enzyme activity.

TEST DESCRIPTION

This UGT1A1 gene mutation detection is a PCR and Sanger Sequencing technique based diagnostic test designed to detect mutations present at predefined nucleotide position in UGT1A1 gene in DNA extracted from whole blood. This test was developed and its analytical performance characteristics have been determined by Redcliffe labs. It has not been cleared or approved by FDA.

TEST LIMITATIONS

- ✓ This assay is based upon PCR and DNA Sequencing of UGT1A1 (RefSeq NM_000463).
- ✓ Test is DNA based, samples must be received at the laboratory under cool-pack conditions (4-8 ℃) within 72 hrs of collection to ensure preservation of intact DNA.
- ✓ Test results may vary if appropriate sample collection and transportation to lab not followed as per protocol.
- ✓ This test is laboratory developed and its performance were evaluated at Redcliffe Labs.
- ✓ The analytical sensitivity of the test allows detection of the genetic variant when the variant clone comprises at least 10-20% of the total genomic DNA. Mutations below the detection limits of the assay may not be detected. Typical detection limit for Sanger Sequencing assays is >10-20%.
- ✓ This report only includes variants that meets a level of evidence threshold for cause or contribute to disease.
- ✓ Gene transcript used for clinical reporting generally represents the canonical transcript, which is usually the longest coding transcript with strong/multiple supporting evidence.

DISCLAIMER

- ❖ Test has been performed assuming that the sample received belongs to the above-named individual(s) and that any stated relationships between individuals are accepted as true.
- ❖ This test is performed using a in house developed kit. The assay is designed to perform the reactions at the specified analytical sensitivity given that the template DNA is not heavily fragmented, and does not contain materials that could inhibit the amplification reaction.
- ❖ The results should be interpreted in the context of the patient's medical evaluation. Mutation identified in this gene does not guarantee activity of the drug in a given indication due to presence of contraindicated mutation in gene.
- ❖ The mutation information provided should only be utilized as a guide or aid and the decision to select any therapy option based on the information reported here resides solely with the discretion of the treating physician.
- ❖ This report should only be used as an aid and the treating physician should employ sound clinical judgment in arriving at any decision for patient care or treatment.

REFERENCE

- ❖ Beutler E, Gelbart T, Demina A. Racial variability in the UDP-glucuronosyltransferase 1 (UGT1A1) promoter: a balanced polymorphism for regulation of bilirubin metabolism? Proc Natl Acad Sci U S A . 1998;95(14):8170-8174.
- ❖ Liu X, Cheng D, Kuang Q, et al. Association of UGT1A1*28 polymorphisms with irinotecan-induced toxicities in colorectal cancer: a meta-analysis in Caucasians. Pharmacogenomics J . 2014;14(2):120-129.
- ❖ Gammal RS, Court MH, Haidar CE, et al. Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for UGT1A1 and atazanavir prescribing. Clin Pharmacol Ther . 2016;99(4):363-369.

.....End of Report.....

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Terms and Conditions of Reporting

1. The presented findings in the Reports are intended solely for informational and interpretational purposes by the referring physician or other qualified medical professionals possessing a comprehensive understanding of reporting units, reference ranges, and technological limitations. The laboratory shall not be held liable for any interpretation or misinterpretation of the results, nor for any consequential or incidental damages arising from such interpretation.

2. It is to be presumed that the tests performed pertain to the specimen/sample attributed to the Customer's name or identification. It is presumed that the verification particulars have been cleared out by the customer or his/her representation at the point of generation of said specimen / sample. It is hereby clarified that the reports furnished are restricted solely to the given specimen only.

3. It is to be noted that variations in results may occur between different laboratories and over time, even for the same parameter for the same Customer. The assays are performed and conducted in accordance with standard procedures, and the reported outcomes are contingent on the specific individual assay methods and equipment(s) used, as well as the quality of the received specimen.

4. This report shall not be deemed valid or admissible for any medico-legal purposes.

5. The Customers assume full responsibility for apprising the Company of any factors that may impact the test finding. These factors, among others, includes dietary intake, alcohol, or medication / drug(s) consumption, or fasting. This list of factors is only representative and not exhaustive.

Disclaimer: Method given in report are only indicative and can be changed depending upon type of machine and kit available at time of testing.

Not all tests at all locations are under NABL scope. Availability of tests under NABL scope varies from lab to lab.