

|                  |                    |              |               |
|------------------|--------------------|--------------|---------------|
| PATIENT NAME     | Mrs. PYARI KHATOON | Barcode No   | 24921481      |
| Age/Gender       | 20 Years/F         | Reg. No      | 0482309220183 |
| Referring by     |                    | SPP Code     | SPL-BH-110    |
| REF. DOCTOR      |                    | Collected On | 22-09-2023    |
| Primary Sample   | Whole Blood        | Received On  | 23-09-2023    |
| Sample Tested In | Whole Blood EDTA   | Reported On  | 29-09-2023    |

**Genetic analysis of Human UGT1A1 gene (Gilbert Syndrome) by PCR + Sequencing**

| Test Description     | Test Report                                         |
|----------------------|-----------------------------------------------------|
| UGT1A1 gene analysis | <b>Genotype is consistent with Gilbert Syndrome</b> |

| Alleles              | Effect of Polymorphism | No. of alleles detected |
|----------------------|------------------------|-------------------------|
| UGT1A1* 1 A(TA)6TAA  | Normal enzyme activity | 0                       |
| UGT1A1* 28 A(TA)7TAA | Low enzyme activity    | 2                       |

**COMMENTS:** Allele group observed in sample is Group No. 2

**INTERPRETATION: Grouping of allele distribution:**

| Group | Alleles   | No. of alleles detected | CONDITION |
|-------|-----------|-------------------------|-----------|
| 1     | UGT1A1*1  | 2                       | NORMAL    |
|       | UGT1A1*1  | 1                       | NORMAL    |
|       | UGT1A1*28 | 0                       | NORMAL    |
|       |           | 1                       | NORMAL    |
| 2     | UGT1A1*28 | 2                       | GILBERT'S |
|       | UGT1A1*1  | 0                       | GILBERT'S |

1. If alleles detected are that of group 1: **Finding is not consistent with Gilbert Syndrome.**
2. If alleles detected are that of group 2: Finding is consistent with Gilbert syndrome.

**UGT1A1 EXON MUTATION ANALYSIS :**

| MUTATION | EFFECT OF POLYMORPHISM | GENOTYPE DETECTED |
|----------|------------------------|-------------------|
| G71R     | Low enzyme activity    | MUTANT            |
| Y486D    | Low enzyme activity    | MUTANT            |

**COMMENTS:** Genotype is consistent with Gilbert Syndrome

**Gilbert's syndrome**, a chronic non-hemolytic unconjugated hyperbilirubinemia, is caused by a reduction in the activity of hepatic bilirubin UDP-glucuronosyltransferase (UGT1A1). This reduction has been shown to be due to a polymorphism in the promoter region of the UGT1A1 gene. The presence of seven thymine adenine (TA) repeats reduces the efficiency of transcription of the UGT1A1 gene. Other two mutations at Codon 71 and 486 also screened in this assay. These two mutations reduced UGT1A1 enzyme activity.

**METHOD:** Nucleic acid from clinical sample was extracted using an automated nucleic acid extraction platform (Qiasymphony, Germany); PCR was set as described by Iolascon *et.al.*, (1999) using an automated robotic liquid handling system (Qiagility, Germany) and the target DNA amplified using a Veriti thermal cycler (Life Technologies). PCR amplicon was purified using a Qiagen PCR product purification kit and sequenced on an ABI 3500 Genetic Analyzer using BigDye® Terminator v3.1 Cycle Sequencing Kit from Life Technologies. Nucleotide sequence was checked for number of TA repeats and mutations at Codon 71 and 486 by two independent persons skilled in the art of reading electropherograms and consensus data recorded for reporting.

**REFERENCE:**

IOLASCON A, MARIA FELICIA FAIENZA, MARTA CENTRA, SONIA STORELLI, LEOPOLDO ZELANTE, ANNA SAVOIA (1999). (TA)<sub>8</sub> allele in the UGT1A1 gene promoter of a Caucasian with Gilbert's syndrome Haematologica 84:106-109.

\*END OF THE REPORT\*



Dr. RUTURAJ

MD, MICROBIOLOGIST