PATIENT NAME	DHANUSH	Barcode No	20034301
Age/Gender	16 Years/M	Reg. No	0012101290399
Referring by	SVS Hospital	SPP Code	SPL-STS-086
REF. DOCTOR		Collected On	29 Jan 2021
Primary Sample	Whole Blood	Received On	29 Jan 2021
Sample Tested In	Whole Blood EDTA	Reported On	03 Feb 2021

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

Test Description	Test Report
UGT1A1 gene analysis	Genotype is consistent with Gilbert Syndrome

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	CONDTION
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). $(TA)_8$ allele in the UGT1A1 gene promoter of a Caucasian with Gilbert's syndrome Haematologica 84:106-109.

END OF THE REPORT

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