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PATIENT REPORT

Patient Age/Sex: 23 years Female

Specimen Collected: 29-Oct-25 14:16

UGT1A1 Genotyping |**Received: 29-Oct-25 14:16** **Report/Verified: 30-Oct-25 11:13**

Procedure	Result	Units	Reference Interval
UGT1A1 Genotyping Specimen	Whole Blood		
UGT1A1 Genotyping Allele 1	**(TA)7 or *28** *		
UGT1A1 Genotyping Allele 2	**(TA)7 or *28** *		
UGT1A1 Genotyping Interpretation	See Note f1 i1		
EER UGT1A1	See Note f2		

Result Footnote

f1: UGT1A1 Genotyping Interpretation

Indications for ordering:
- Determine sensitivity to irinotecan or related compounds.
- Confirm a diagnosis of Gilbert Syndrome.

Homozygous UGT1A1 (TA)7: Two copies of the UGT1A1 *28 (TA)7 variant were detected predicting a poor metabolizer status. This is associated with decreased UGT1A1 enzyme and increased risk for irinotecan toxicity, namely, neutropenia and diarrhea. Dose reduction is recommended. This genotype has been reported to be associated with Gilberts syndrome (benign familial hyperbilirubinemia).

This result has been reviewed and approved by [redacted]

f2: EER UGT1A1
Authorized individuals can access the ARUP Enhanced Report with an ARUP Connect account using the following link.

Your local lab can assist you in obtaining the patient report if you don't have a Connect account.

[redacted]

Test Information

i1: UGT1A1 Genotyping Interpretation

BACKGROUND INFORMATION: UDP Glucuronosyltransferase 1A1 (UGT1A1) Genotyping

CHARACTERISTICS: UGT1A1 is responsible for the clearance of drugs (e.g., irinotecan) and endobiotic compounds (e.g., bilirubin). Irinotecan's major active and toxic metabolite (SN-38) is inactivated by the UGT1A1 enzyme and then eliminated via the bile. UGT1A1 gene mutations cause accumulation of SN-38, which may lead to irinotecan-related toxicities (neutropenia, diarrhea).

CAUSE: Variations in TA repeat number in the TATAAA element of the 5'UGT1A1-promoter affects transcription efficiency. The common number of repeats is six [(TA)6, *1 allele], while seven repeats [(TA)7, *28 allele] is associated with reduced transcription activity. Homozygosity for the (TA)7 allele is also associated with Gilbert syndrome (benign familial hyperbilirubinemia).

ALLELES TESTED: *36 allele, (TA)5; *1 allele, (TA)6; *28 allele, (TA)7 and *37 allele, (TA)8.

CLINICAL SENSITIVITY/SPECIFICITY: Risk of irinotecan toxicity by genotype (Br J Cancer. 2004; 91:678-82).

Test Information

i1: UGT1A1 Genotyping Interpretation

6/6 (*1/*1): diarrhea 17 percent; neutropenia 15 percent
6/7 (*1/*28): diarrhea 33 percent; neutropenia 27 percent
7/7 (*28/*28): diarrhea 70 percent; neutropenia 40 percent

ALLELIC FREQUENCY:
*1(TA)6: Whites 0.61, Asians 0.84, African Americans 0.47
*28(TA)7: Whites 0.39, Asians 0.16, African Americans 0.43

METHODOLOGY: Polymerase chain reaction followed by size analysis using capillary electrophoresis.
ANALYTICAL SENSITIVITY AND SPECIFICITY: Greater than 99 percent.
LIMITATIONS: Variations in the UGT1A1 gene, other than those targeted, will not be detected. Clinical significance of the rare *36, (TA)5 and *37, (TA)8 alleles in predicting irinotecan toxicities is not well established. Genetic and non-genetic factors other than UGT1A1, may contribute to irinotecan toxicity and efficacy. Diagnostic errors can occur due to rare sequence variations.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. Food and Drug Administration. This test was performed in a CLIA-certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.