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Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	2/2/1995
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	

UDP Glucuronosyltransferase 1A1 (UGT1A1) Genotyping

ARUP test code 0051332

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	

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. 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 Gilbert's syndrome (benign familial hyperbilirubinemia).

This result has been reviewed and approved by Rong Mao, M.D.

H=High, L=Low, *=Abnormal, C=Critical

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 alelle, (TA)7 and *37 allele, (TA)8.
CLINICAL SENSTIVITY/SPECIFICITY: Risk of irinotecan toxicity by genotype (Br J Cancer (2004) 91:678-82).
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: Caucasians 0.61, Asians 0.84, African Americans 0.47
*28(TA)7: Caucasians 0.39, Asians 0.16, African Americans 0.43

METHODOLOGY: Polymerase chain reaction followed by size analysis using capillary electrophoresis.
ANALYTICAL SENSTIVITY: 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.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
UGT1A1 Genotyping Specimen	18-178-108910	6/27/2018 12:02:00 PM	6/28/2018 11:01:41 AM	7/1/2018 5:52:00 PM
UGT1A1 Genotyping Allele 1	18-178-108910	6/27/2018 12:02:00 PM	6/28/2018 11:01:41 AM	7/1/2018 5:52:00 PM
UGT1A1 Genotyping Allele 2	18-178-108910	6/27/2018 12:02:00 PM	6/28/2018 11:01:41 AM	7/1/2018 5:52:00 PM
UGT1A1 Genotyping Interpretation	18-178-108910	6/27/2018 12:02:00 PM	6/28/2018 11:01:41 AM	7/1/2018 5:52:00 PM

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