

## Clinical implication

The enzyme UDP-glucuronosyltransferase (UGT) is responsible for the conjugation phase of bilirubin biotransformation. The activity of the enzyme is conditioned by the presence of polymorphisms in the UGT1A1 gene. In particular, the number of TA repeats (5 to 8 TAs) is monitored, with an increasing number of TA repeats being associated with both a reduced transcription rate and a lower enzyme level. The examinations are used to confirm the diagnosis of Gilbert's syndrome, or before the administration of irinotecan drug and other drugs metabolised by UGT.

A wild-type variation of the gene, an UGT1A1\*1 allele, carries 6 TA-repetitions. Homozygous UGT1A1\*28 variant is, in Caucasian and Afro-American populations, the most frequent cause of Gilbert syndrome, a benign disorder associated with

a mild chronic hyperbilirubinemia. In that case, the glucuronidation activity of UGT1A1 enzyme is reduced to 20–30%. Alleles UGT1A1\*36 and UGT1A1\*37 are mainly presented in Africans ethnic group. Genetic variations within the UGT1A1 gene have also been associated with the development of certain drug toxicities.

## Principle of detection

The real-time PCR diagnostic kit allows detection of variants of 5TA (UGT1A1\*36), 6TA (UGT1A1\*1), 7TA (UGT1A1\*28) and 8TA (UGT1A1\*37) polymorphisms. The detection is based on the **Q-FRET probe** and uses the **analysis of melting curves** for multiplex genotyping.



## Available products

Cat. No.	Product	rxn
3253-025	gb PHARM UGT1A1	25
3253-050	gb PHARM UGT1A1	50

1 kit contains reagents to provide 25 or 50 PCR reactions (20 µl volume of each reaction).

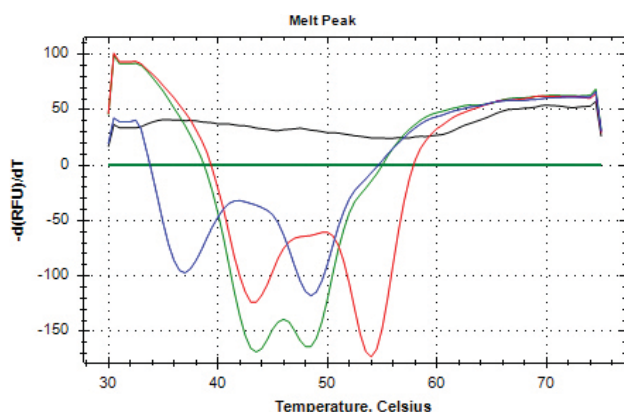
## Parameters of the diagnostic kit

- *in vitro* diagnostics
- CE IVD marked
- sample concentration 5-100 ng/µl
- positive and negative controls included
- melting curve analysis, FAM channel detection

## Content of the diagnostic kit

* Component	Conc.	Purpose
Assay UGT1A1	3.33×	Detection assay
Master Mix UGT1A1	2×	Master Mix
Standard HET1 5/7TA	10 <sup>4</sup> cop/µl	Positive Control
Standard HET2 6/8TA	10 <sup>4</sup> cop/µl	Positive Control
Deionized water		Negative Control

\*Lid colour



## Validated for cyclers

- CFX Opus 96 (Bio-Rad)
- CFX96/CFX96 Touch (Bio-Rad)
- Light Cycler 480/Cobas z480 (Roche Diagnostics)
- QuantStudio 5 (Applied Biosystems)
- Rotor-Gene 3000 (Corbett Research)

Fig. 1 – Example of melting curves analysis of Standard HET1 5/7 TA (blue), Standard HET2 6/8 TA (red) and NTC (black) in software CFX96 in FAM channel - identification of the number of TA repeats UGT1A1. In the figure is also shown the HET 6/7TA DNA sample (green) for illustration.