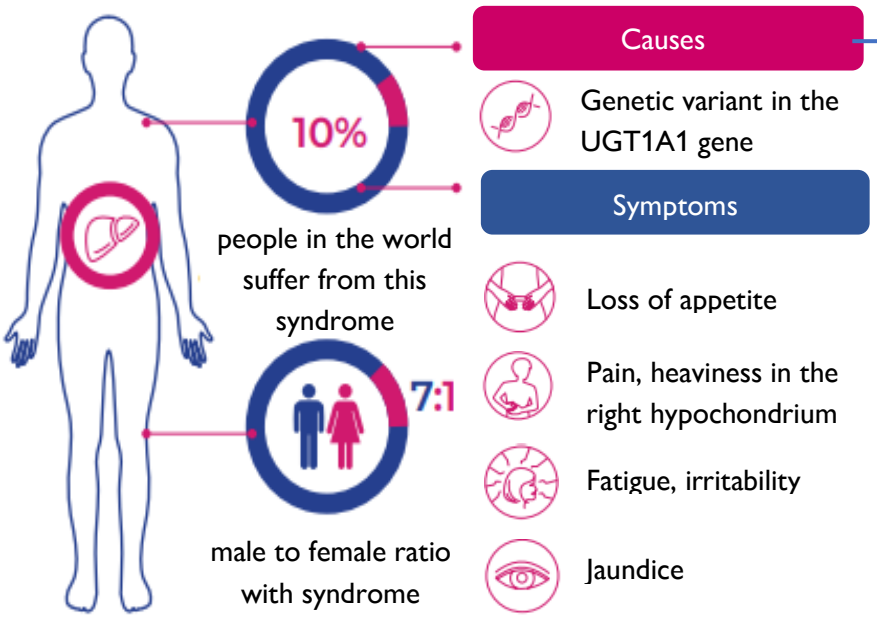
DNK s.r.o.
diagnostické centrum

Laboratory diagnosis of Gilbert's syndrome

Diagnostic kits for the detection of TA dinucleotides repeats in the UGT1A1 gene

Gilbert's syndrome

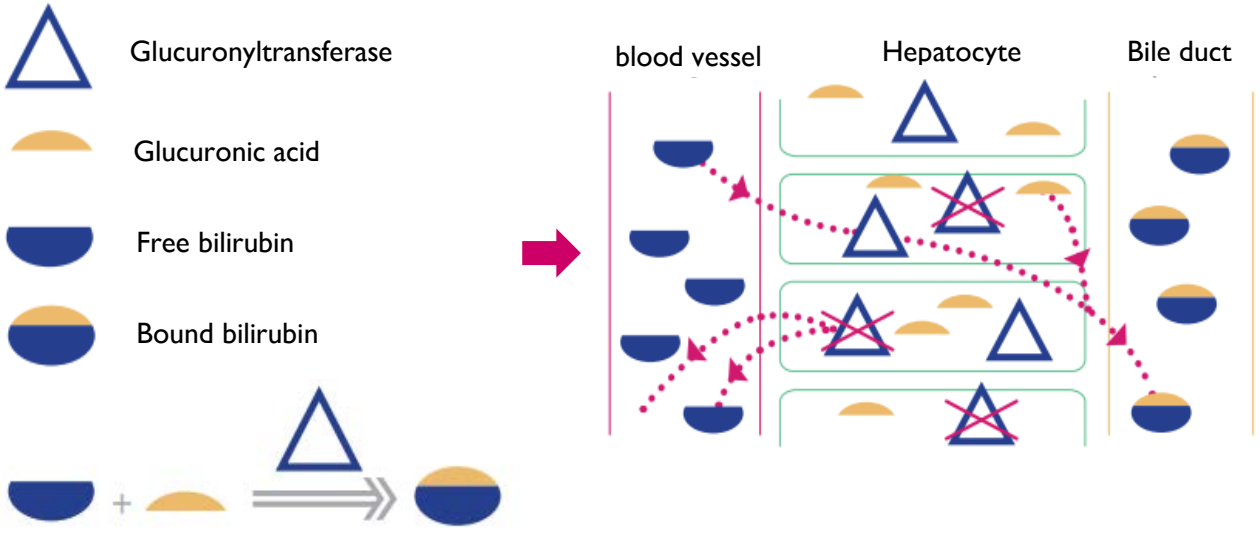
Gilbert's syndrome refers to chronic pathologies of the liver and is characterized by alternating periods of remission and episodes of jaundice. The manifestations of the syndrome can occur at any age, and it may be triggered by various factors: physical exertion, stressful situations, hunger strike, viral infections, alcohol intake and a number of medications.



The ***UGT1A1*** gene encodes the enzyme **UDHT1**, which plays a key role in bilirubin metabolism. With an increase in the number of TA repetitions in the promoter region of the gene (polymorphism **5TA/6TA/7TA/8TA**), there is a decrease in the amount of enzyme. This leads to a violation of the conjugation of free bilirubin, then to an increase in its concentration in the blood and the development of Gilbert's syndrome.

A further increase in the level of free bilirubin can lead to the development of cholelithiasis, cholecystitis or duodenitis. In addition, other more serious pathologies of the gastrointestinal tract, similar in symptoms and requiring treatment, can be masked under undiagnosed Gilbert syndrome. With mild manifestations of the syndrome, it is possible to adjust the patient's lifestyle until the complete disappearance of discomfort caused by hyperbilirubinemia.

UDHT1 is also involved in the glucuronization of many drugs, therefore, with an initial lack of the enzyme, toxic reactions to the drug and the manifestation of Gilbert's syndrome can develop.



The most objective confirmation of the diagnosis of Gilbert's syndrome is genetic methods.

Diagnostic Kit RealBest-Genetics UGT1A1 (cat. No. 3813)

The kit is designed to determine the number of repetitions (5TA/6TA/7TA/8TA) in the promoter region of the uridine diphosphate glucuronosyltransferase 1 gene (UGT1A1) by real-time PCR method with melting curve detection.

Usage of the diagnostic kit:

- for differential diagnosis of Gilbert's syndrome with other diseases of the gastrointestinal tract, infectious pathologies, oncological processes
- when assessing the possibility of therapy with the use of hepatotoxic drugs
- to prevent adverse reactions to drug therapy (irinocecan, atazanavir, etc.) due to dose modification or replacement with another medicine

Interpretation of testing

The test results in genotyping of polymorphisms 5TA/6TA/7TA/8TA. Carriers of the 6TA allelic variant synthesize as much of the enzyme as the body needs.

An increase in the number of repetitions of the TA dinucleotide motif leads to a decrease in enzyme synthesis, and the degree of UDHT1 deficiency depends on the genotype of this polymorphism. To determine the metabolic state of the patient, it is important to identify all alleles, since their combination determines the overall activity of the enzyme UDHT1.

N-repeats	6TA	5TA	7TA	8TA	5TA	7TA	8TA
		Caucasian			African Americans		
Frequency, %	norm	< 1	35-40	< 1	3,5	-	7-8
UGT1A1 synthesis		↑	↓	↓↓	↑	↓	↓↓

Features and advantages of the diagnostic kit

- **Ready Master Mix for PCR**: Simplification of analysis procedures and high stability of test quality
- **Specimens**: whole blood or buccal epithelium
- **Number of tests**: kits are designed for the analysis of 48 samples including control samples
- **Multiplexes:** determination of the polymorphism genotype 5TA/6TA/7TA/8TA in two tubes
- **Compatible devices**: CFX96 (Bio-Rad, USA), DT-96 a DTprime (DNA-Technology, Russia), Gentier 96E/R (Xi'an TianLong, Science and Technology Co., Ltd., China)
- **High stability of the kit:** storage at a temperature of 2–8 ° C; transport up to 26 °C not more than 10 days

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