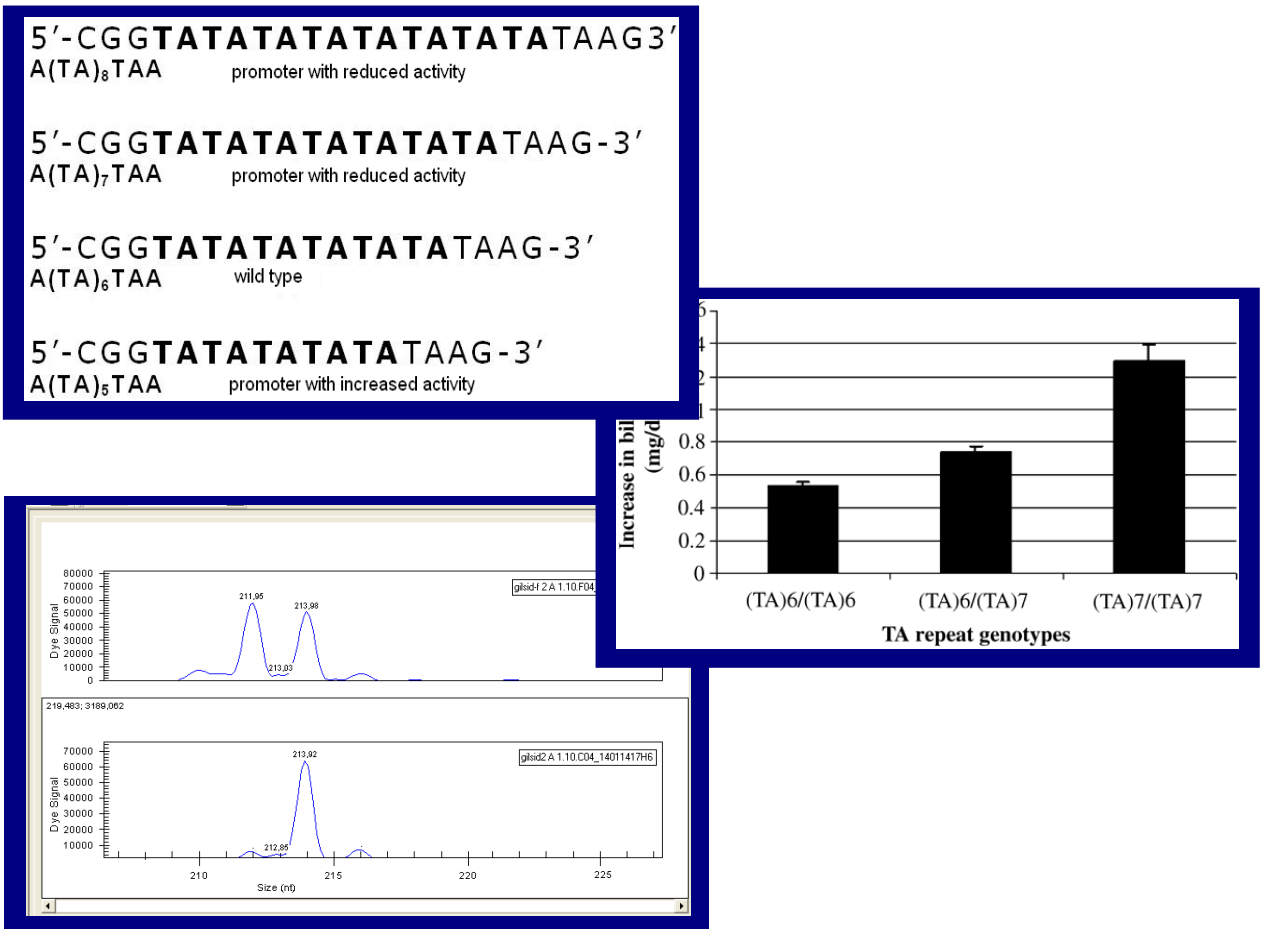
System for the molecular analysis of the Gilbert syndrome by automated fragment analysis

Gilbert Syndrome Kit-FL

cod. GS.01FL



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The **Gilbert's Syndrome** (GS) is a form of chronic and moderate iperbilirubinemia (not conjugated) caused by a lower hepatic glucuronidation of bilirubin by **UDP-glucuronosyltransferase 1** (UGT1A1) enzyme.
Most patients affected by this disease are homozygous for a TA insertion in TATA-box of UGT1A1 gene promoter. The transcription of **A(TA)$_7$TAA** allele is lower of almost 70% in respect to **A(TA)$_6$TAA** wild type allele, being UGT1A1 the only enzyme with a substantial bilirubin glucuronidation activity. The presence of TA in both alleles explains the decrease of conjugated bilirubin in patients with GS.
In different population was found a variable number of TA repetition, ranging from 5 to 8, with an inverse relation between promoter activity and number of TA repetition. This alleles are extremely rare in Caucasian population.
The *Gilbert sydrome kit- FL* amplifies the TATA-box contained in UGT1A1 (UDP-glucuronosyltransferase) gene promoter by a primers pair one of which is marked with a fluorescent dye. The amplicons are separated by capillary electrophoresis by an automatic sequencer. This technique allows, with an extremely accurate manner, to assess the length of amplicons and to distinguish mutated alleles from wild type alleles, also if they differs by only a base pair.

Kit contents

Label	Contents
D4-GS MASTER MIX	Mix for amplification of UGT1A1 gene promoter
ExperTaq polymerase	Taq DNA polimerase

How does the kit work?

The **Gilbert Sindrome kit- FL** can distinguish the wild-type alleles from the different mutations through a capillary electrophoresis of amplified products by capillary electrophoresis.
This technique can separate alleles with a difference of only a **single nucleotide**.

Why we should utilized the kit?

The kit is utilized because is an easy, fast & simple system for screening and **differential diagnosis** of Gilbert Syndrome in patients with hyperbilirubinemia. The presence of other pathology associated with increase of blood-bilirubin can be excluded.
The technique utilized is easy, highly sensible, easily to read and fast to achieve, only 4 hours to have the final result.

Starting samples: peripheral blood

DNA isolation method: QIAamp DNA blood mini kit, QIAcube, QIAsymphony (Qiagen), High Pure PCR template preparation kit (Roche).

DNA Sequencer: CEQ 8000/8800 Genetic Analysis System(Beckman Coulter); 310, 3100, 3130, 3730, 3500 Genetic Analyzers (Applied Biosystmes).

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