

# gb GENETIC LACTO

## Clinical implications

Genetically determined lactose intolerance is a disorder of the metabolism of lactose (milk sugar) caused by decreased production of the lactase enzyme. Under usual conditions small intestinal cells produce lactase, which decomposes lactose into simpler sugars - glucose and galactose. These are then absorbed by the wall of the small intestine, getting into the bloodstream, and serves as an energy source. In case of lactose intolerance the undigested lactose is accumulated in the intestine and then decomposed by intestinal bacteria. Gastrointestinal symptoms include diarrhea, abdominal discomfort, stomach cramps or bloating as a result of lactose degradation. In Europe, lactose intolerance is associated with two mutations in the regulatory region of the gene LCT at positions -13910 and -22018. This gene is responsi-

ble for the level of the enzyme lactase. In the presence of alleles LCT -13910C and LCT -22018G the activity of lactase gene is much lower, which leads to lactase non-persistence. Haplotype LCT -13910T and -22018A are conversely associated with lactase persistence.

## Principle of detection

The kit is intended for detection of mutations LCT (C13910T) and LCT (G22018A) in the lactase gene in human genomic DNA. Detection is based on **real-time polymerase chain reaction (qPCR) using fluorescently labelled probes (allelic discrimination)**.

## Available products










Cat. No.	Product	rxn
3219-025	gb GENETIC LACTO	25
3219-050	gb GENETIC LACTO	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
- ready-to-use assay
- sample concentration 10-100 ng/µl
- positive and negative controls included
- FAM and HEX channels detection
- identical amplification profile as gb HEMO, gb GENETIC, gb PHARM kits

## Content of the diagnostic kit

* Component	Conc.	Purpose
 Assay qPCR LCT (C13910T)	1.25×	Detection assay
 Standard WT LCT (C13910T)	10 <sup>4</sup> cop/µl	Positive Control
 Standard MUT LCT (C13910T)	10 <sup>4</sup> cop/µl	Positive Control
 Standard HET LCT (C13910T)	10 <sup>4</sup> cop/µl	Positive Control
 Assay qPCR LCT (G22018A)	1.25×	Detection assay
 Standard WT LCT (G22018A)	10 <sup>4</sup> cop/µl	Positive Control
 Standard MUT LCT (G22018A)	10 <sup>4</sup> cop/µl	Positive Control
 Standard HET LCT (G22018A)	10 <sup>4</sup> cop/µl	Positive Control
 Deionized Water		Negative Control

\* Lid colour



## Validated for cyclers

- Rotor-Gene 3000/6000/Q (Corbett Research, Qiagen)
- iCycler iQ5/CFX96/CFX96 Touch (Bio-Rad)
- ABI 7300/7500/7500 Fast (Applied Biosystems)
- SmartCycler (Cepheid)
- MIC (Bio Molecular Systems)
- Light Cycler 480/Cobas z480 (Roche Diagnostics)
- QuantStudio 5 (Applied Biosystems)

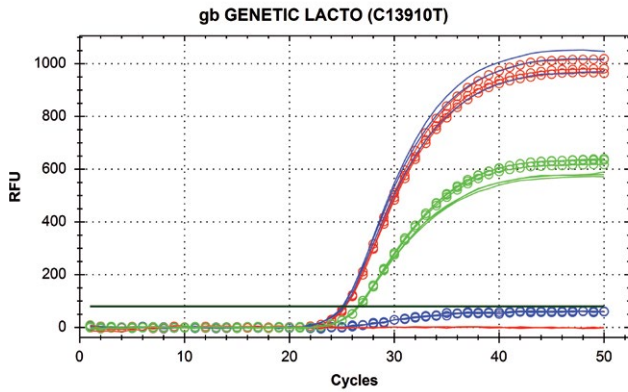


Fig. 1 – Detection of LACTO (C13910T) standards on CFX96 device; *blue line* – wild type; *red line* – mutant; *green line* – heterozygote; *smooth line* – FAM channel; *dotted line* – HEX channel

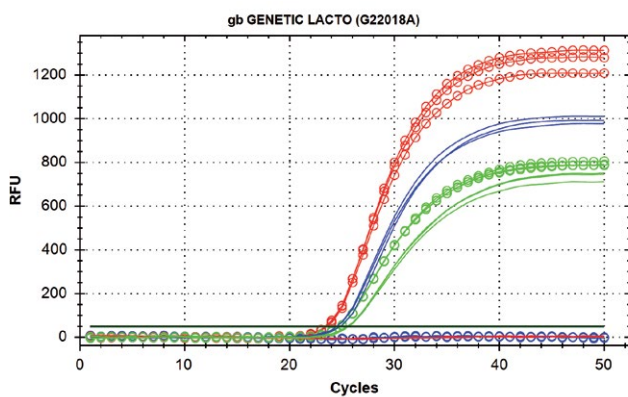


Fig. 2 – Detection of LACTO (G22018A) standards on CFX96 device; *blue line* – wild type; *red line* – mutant; *green line* – heterozygote; *smooth line* – FAM channel; *dotted line* – HEX channel