gb GENETIC FRUCTO

Clinical implications

Hereditary fructose intolerance (HFI) is an autosomal recessive disorder caused by a deficiency of the enzyme aldolase B, which is essential for the metabolism of fructose. One person in 10,000 to 100,000 suffers from HFI. The most common mutations of the gene for aldolase B are A149P, A174D, N334K and deletion of del4E4. Hereditary fructose intolerance is characterized by clinical symptoms such as vomiting, nausea, diarrhea, growth restriction and metabolic disorders (hypoglykemia, hyperuricemia, hypomagnesemia, or lactic acidosis). HFI may also lead to liver and kidney failure. HFI first appears in infants who are not breastfed, but eat a diet containing sucrose and fructose.

Principle of detection

The kit is intended for detection of mutation A149P, A174D, N334K and deletion del4E4 in the ALDOB 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
3252-025	gb GENETIC FRUCTO	25
3252-050	gb GENETIC FRUCTO	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, IVDR certified**
- **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 ALDOB (A149P)	1.25×	Detection assay
	Assay qPCR ALDOB (A174D)	1.25×	Detection assay
	Assay qPCR ALDOB (N334K)	1.25×	Detection assay
	Assay qPCR ALDOB (del4E4)	10^4 cop/µll	Positive Control
	Standard WT ALDOB	10^4 cop/µl	Positive Control
	Standard MUT ALDOB	10^4 cop/µl	Positive Control
	Standard HET ALDOB	10^4 cop/µl	Positive Control
	Deionized Water		Negative Control

** Lid colour*



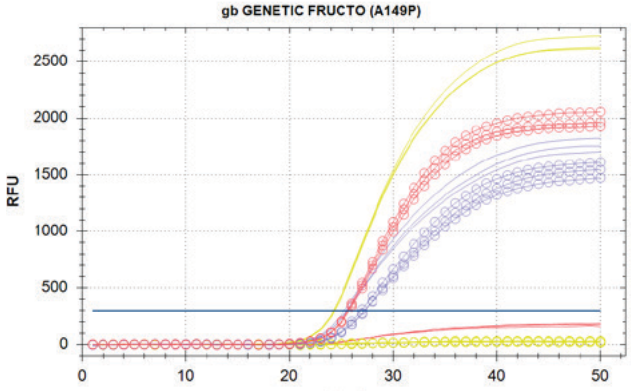


Fig. 1 – Detection of FRUCTO (A149P) standards on CFX96 device; yellow line – wild type; red line – mutant; violet line – heterozygote; smooth line – FAM channel; dotted line – HEX channel

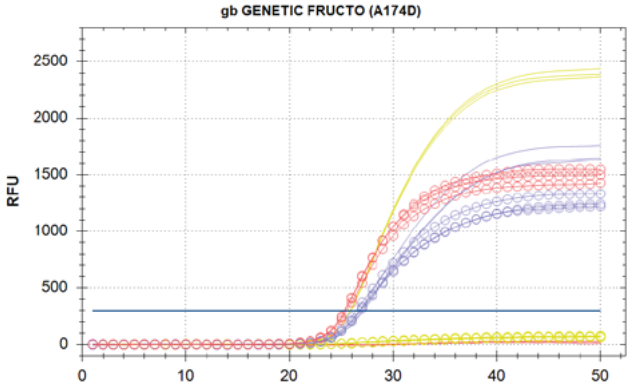


Fig. 2 – Detection of FRUCTO (A174D) standards on CFX96 device; yellow line – wild type; red line – mutant; violet line – heterozygote; smooth line – FAM channel; dotted line – HEX channel

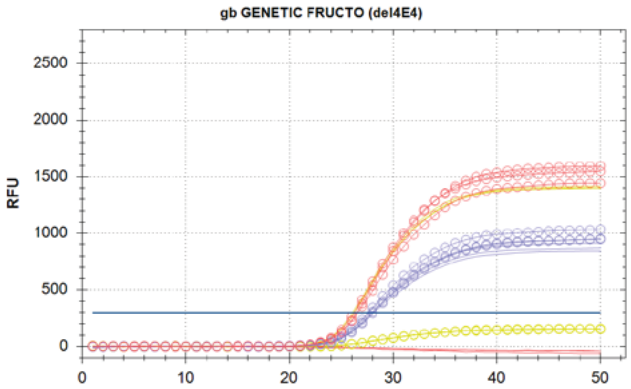


Fig. 3 – Detection of FRUCTO (del4E4) standards on CFX96 device; yellow line – wild type; red line – mutant; violet line – heterozygote; smooth line – FAM channel; dotted line – HEX channel

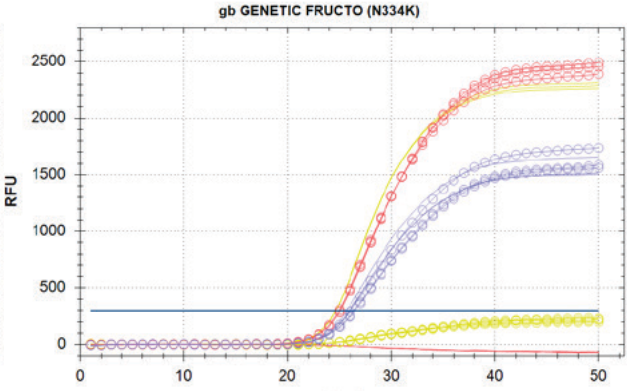


Fig. 4 – Detection of FRUCTO (N334K) standards on CFX96 device; yellow line – wild type; red line – mutant; violet line – heterozygote; smooth line – FAM channel; dotted line – HEX channel

Validated for cyclers

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