gb GENETIC HFE

Clinical implications

Hereditary hemochromatosis (HHC) is a hereditary metabolic disorder caused by HFE gene mutation with an autosomal recessive type of inheritance and incomplete penetration (which means it does not have to always be shown in a phenotype). It is one of the most common hereditary disorders with an indicated prevalence of the majority C282Y mutation in a homozygous condition in 1:300–400 in the Caucasian population. HFE protein is bound to beta-2-microglobulin and this complex blocks the transferin receptor in the cells of the duodenum. There is no receptor blockade in the complex consisting of the mutated form of the protein which results in permanent increased iron absorption from the gastrointestinal tract into the blood stream and its accumulation in parenchymatous tissues and organs, especially in liver, pancreas, heart, gonads and in the skin. These organs can then be irreversibly damaged.

Principle of detection

The kit is intended for detection of mutations C282Y, H63D and S65C in the gene HFE 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
3208-025	gb GENETIC HFE	25
3208-050	gb GENETIC HFE	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 HFE C282Y	1.25×	Detection assay
	Standard WT HFE C282Y	10^4 cop/µl	Positive Control
	Standard MUT HFE C282Y	10^4 cop/µl	Positive Control
	Standard HET HFE C282Y	10^4 cop/µl	Positive Control
	Assay qPCR HFE H63D	1.25×	Detection assay
	Standard WT HFE H63D	10^4 cop/µl	Positive Control
	Standard MUT HFE H63D	10^4 cop/µl	Positive Control
	Standard HET HFE H63D	10^4 cop/µl	Positive Control
	Assay qPCR HFE S65C	1.25×	Detection assay
	Standard WT HFE S65C	10^4 cop/µl	Positive Control
	Standard MUT HFE S65C	10^4 cop/µl	Positive Control
	Standard HET HFE S65C	10^4 cop/µl	Positive Control
	Deionized Water		Negative Control

** Lid colour*



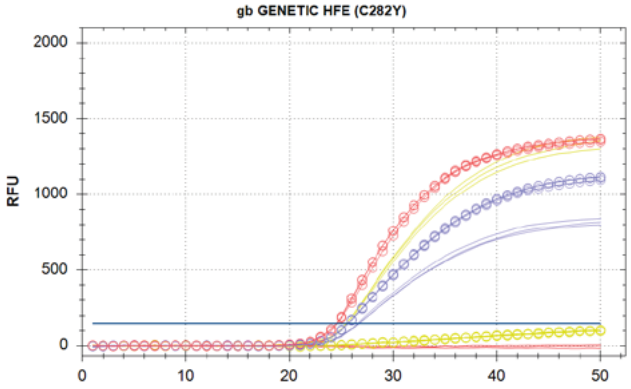


Fig. 1 – Detection of HFE (C282Y) standards on CFX96 device; yellow line – wild type; red line – mutant; violet line – heterozygote; smooth line – FAM channel; dotted line – HEX channel

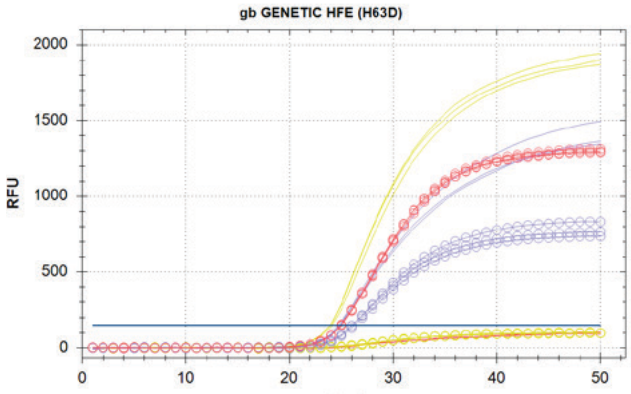


Fig. 2 – Detection of HFE (H63D) standards on CFX96 device; yellow line – wild type; red line – mutant; violet line – heterozygote; smooth line – FAM channel; dotted line – HEX channel

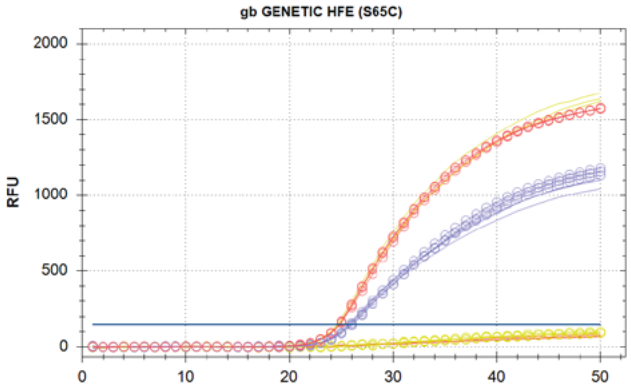


Fig. 3 – Detection of HFE (S65C) standards on CFX96 device; yellow line – wild type; red line – mutant; violet line – heterozygote; smooth line – FAM channel; dotted line – HEX channel

Validated for cyclers

- Rotor-Gene 3000/6000/Q (Corbett Research, Qiagen)
- iCycler iQ5/CFX96/CFX96 Touch (Bio-Rad)
- ABI 7500/7500 Fast/7900HT (Applied Biosystems)
- SmartCycler (Cepheid)
- MIC (Bio Molecular Systems)
- AriaMx (Agilent Technologies)
- Light Cycler 480/Cobas z480 (Roche Diagnostics)
- QuantStudio 5 (Applied Biosystems)

CE IVD