gb HEMO MTHFR (A1298C)

Clinical implications

Methylenetetrahydrofolate reductase (MTHFR) is the main regulatory enzyme involved in homocysteine and folic acid (folates) metabolisms. This mutation leads to enzyme activity reduction, increase of homocysteine level in blood and methionine deficiency. However, lower catalytic activity of MTHFR enzyme means higher utilisation of 5, 10-methylenetetrahydrofolate in DNA synthesis or repair. The risk of venous thrombosis increases in combination with other risk factors (e.g. mutation in MTHFR gene – C677T, mutation G20210A in factor II, Leiden mutation, long-time immobility, incorrect regimen, hormonal contraception) and grows with the number of mutated alleles in genotype.

Principle of detection

The kit is intended for detection of mutation A1298C in MTHFR 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
3203-025	gb HEMO MTHFR (A1298C)	25
3203-100	gb HEMO MTHFR (A1298C)	100

1 kit contains reagents to provide 25 or 100 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 MTHFR (A1298C)	1.25×	Detection assay
⚪	Deionized Water		Negative Control
🟡	Standard WT MTHFR (A1298C)	10^4 cop/µl	Positive Control
🔴	Standard MUT MTHFR (A1298C)	10^4 cop/µl	Positive Control
🟣	Standard HET MTHFR (A1298C)	10^4 cop/µl	Positive Control

** Lid colour*



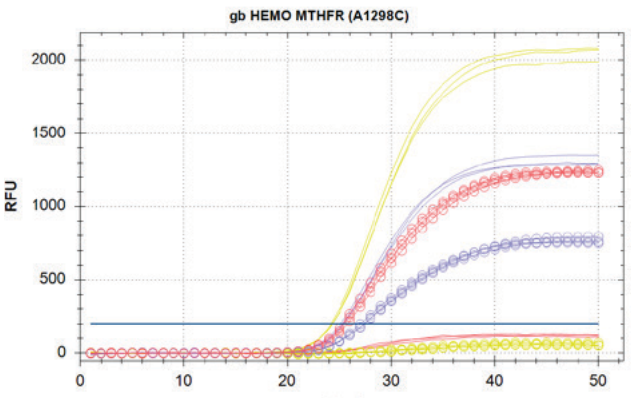


Fig. 1 – Detection of MTHFR (A1298C) standards on CFX96 device; yellow line – wild type; red line – mutant; violet line – heterozygote; smooth line – FAM channel; dotted line – HEX channel

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)**
- **Stratagene Mx3000P/Mx3005P (Agilent Technologies)**
- **Light Cycler 480/Cobas z480 (Roche Diagnostics)**
- **QuantStudio 5 (Applied Biosystems)**