REALQUALITY RS-MTHFR C677T

code RQ-29

Kit for detection and genotyping of mutation C677T in the human gene coding for MTHFR by Real-Time PCR

CE IVD

INTRODUCTION

Phenomena of thrombophilia are commonly defined as blockage of blood circulation by clots, which originate in the veins or are stem from a thrombus in another area of the body. Thrombosis can be the result of events that activate the coagulation system, which include injury, surgery, immobilization, pregnancy and the use of estrogens and oral contraceptives. In addition, the genetic background of a person influences the individual risk of thrombosis.
Mutations in genes coding for factors of blood homeostasis and fibrinolysis may lead to a lifelong increased risk of thrombosis. Today, several of such genetic alterations are known.

The N^5,N^{10}-**methylenetetrahydrofolate reductase** (**MTHFR**) is an enzyme that plays an important role in the metabolization of the amino acid methionine. It catalyzes an essential step in the vitamin-B12-dependent remethylation of homocysteine to methionine. Some rare mutations can cause severe MTHFR deficiency, which may lead to homocysteinemia and severe clinical symptoms, like thrombosis and impairment of psychomotor development.

A genetic polymorphism commonly associated with severe MTHFR deficiency is defined by a C→T substitution (cytosine → thymine) at position 677 (C677T) of the *MTHFR* gene, which leads to the incorporation of amino acid alanine (A) instead of valine (V) at position 222 of the MTHFR protein. The altered MTHFR is known as "thermolabile MTHFR". Homozygous and heterozygous carriers of this mutation both show reduced MTHFR activity. In particular, homozygous carriers suffer from significantly increased blood levels of homocysteine.
In general, increased homocysteine levels are considered a risk factor of vascular diseases (e.g. arterial thrombosis).

Approx. 38% to 40% of the general population are heterozygous for the C677T mutation. Approx. 10% are homozygous.

Using Real-Time PCR this mutation can be detected quickly and with high specificity and sensitivity.

TECHNICAL CHARACTERISTICS

Number of tests: 48 or 96

Shelf life: 12 months

Sample material: DNA extract from whole peripheral blood

Positive controls: contain target (DNA) sequences corresponding to following genotypes regarding mutation *MTHFR C677T*: homozygous wild-type (WT), homozygous mutated (MUT) and heterozygous (HET)

Compatible platforms: Validated on

- Applied Biosystems 7500 Fast / Fast Dx, 7300 and StepOnePlus / StepOne Real-Time PCR System
- Dx Real-Time System and CFX96 Real-Time PCR Detection System (Bio-Rad)

The kit can be used on instruments that allow a reaction volume of 25 µL and read the FAM and CAL Fluor® Orange 560 fluorescence (detection channels FAM and JOE/HEX).

Analytical specificity: No non-specific pairing of primers and probes

Analytical sensitivity: 2 ng / reaction of DNA

Diagnostic sensitivity and specificity: 100 %

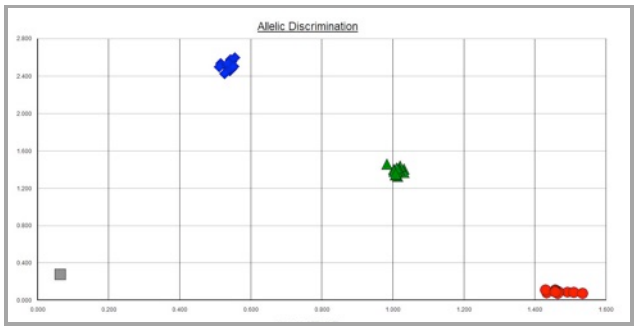

Fig. 1: Scatter plot on *Applied Biosystems 7300 Real-Time PCR System* using SDS software.

ORDERING INFORMATION

Code	Product	PKG
RQ-29-48/96	REALQUALITY RS-MTHFR C677T	48/96 tests

REFERENCES

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