# Thrombophilia Detection Kit

Version 1.0
November 2022

## THROMBOPHILIA - Etiology

**Prothrombin**, also known as **Factor II** (FII), is a protein involved in the blood clotting mechanism. The autosomal recessive G20210A mutation present in the 3'-untranslated region of FII leads to increased translation of the protein and thus increased risk of **hypercoagulability disorders or thrombophilia**[1-4].

**Factor V Leiden** (FVL) is the most common hereditary factor V (FV) variant of coagulation, which prevents FV inactivation by protein C, thus increasing the risk of **blood thrombus formation**[1-4].

**Methylenetetrahydrofolate reductase** (MTHFR) is involved in amino acid metabolism. Mutations (C677T and A1298C) leading to reduced activity of this enzyme are associated with increased levels of homocysteine in the blood. It is believed that this may be associated with the onset of several diseases, including **vascular occlusive disease** and homocystinuria[1-4].

### Epidemiology

The **G20210A mutation of FII is the second most frequent** variant implicated in hypercoagulability disorders in the Caucasian population with a **prevalence of 1% to 4%**, while **FV Leiden is the most common variant** with a frequency of **3-5%** and **20-40%** in patients with **venous thromboembolic disease**. Heterozygosity and homozygous mutation of FV **increase the probability of thrombosis up to 10- and 100-fold**, respectively[1-4].

## HYRIS INNOVATION

HYRIS MARKETPLACE™

Hyris offers an innovative approach of validating and integrating on its platform the best CE-IVD kits from reagent manufacturers on the market.

For the Thrombophilia kit, Hyris selected **Bosphore® Thrombophilia Detection Combo kit v4** from **Anatolia Geneworks**, with the following features:

- CE-IVD marked
- Validated on whole blood and oral swab (dry)
- Validated in Real-Time qPCR on DNA samples

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## TEST DESCRIPTION

The **Bosphore® Thrombophilia Detection Combo kit v4** detects the target genes in whole blood samples. Blood samples are incubated with Ex-Tract DNA solution and incubated at 80° C for 20 min to allow DNA extraction **without specific automated extractors**. At the end of incubation, dilution is performed to perform qPCR analysis.

The Bosphore® Thrombophilia Detection Combo kit v4 employs **multiplex qPCR technology**. DNA from FII, FV and MTHFR both *wild type* and mutant is amplified in parallel in four reactions using primers specific for the indicated allelic sequences. In this way, homozygous ***wild type*, heterozygous, and homozygous mutated individuals** can be identified for the mutations described above in the FII, FV, and MTHFR genes.

The **Bosphore® Thrombophilia Detection Combo kit v4** is a panel composed by four master mixes that identify the mutations in the genes FII, FV, MTHFR C677T and MTHFR A1298C.

The **Bosphore® Thrombophilia Detection Combo kit v4** contains the EX-Tract DNA that allows to work in direct amplification

## SUPPORTED BIOLOGICAL MATRICES:

- The **Bosphore® Thrombophilia Detection Combo kit v4** is compatible with **whole blood samples**.

## BIBLIOGRAPHY


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