

Thrombophilia Risk Panel

Background: Thrombosis is one of the most common types of blood coagulation disorders, affecting 1 in 1000 individuals with a fatality rate of 1-2%. Thrombosis can be categorized as inherited or acquired. Inherited thrombosis is characterized by increased risk of deep vein thrombosis, ectopic pregnancy, pulmonary embolism, myocardial infarction, cardiovascular disease and other complications related to abnormal blood coagulation.

Testing method: The eSensor® Thrombophilia Risk Test (TRT) is an in vitro diagnostic for the detection and genotyping of Factor II (Prothrombin) G20210A, Factor V (Factor V Leiden) G1691A and MTHFR (human 5, 10 methylenetetrahydrofolate reductase gene) C677T and A1298C mutations in patients with suspected thrombophilia from isolated genomic DNA obtained from whole blood samples. The test is intended to be used on the eSensor® XT-8™ System.

Turnaround Time: 5-7 business days

Sample requirements: Multiple DNA tests can be performed from a single tube of blood.

- 3 mL peripheral blood in lavender top tube (EDTA).
- Bone marrow aspirate (anticoagulated with either heparin or EDTA and, if possible, placed into tissue culture medium)

Factor II (Prothrombin) 20210 G -->A

Background: Inherited Thrombosis is associated with congenital predisposing risk factors such as Factor II (Prothrombin, FII)1 and Factor V (Leiden, FV) proteins involved in the blood coagulation enzyme activity cascade. The FII and FV mutations are present in ~2% and 5% of individuals with N. European ancestry respectively, but at much lower levels in other populations. The prothrombin mutation is inherited in an autosomal dominant manner.

Indications for Testing: This test is used in the evaluation of patients with deep vein thrombosis or pulmonary embolism, women with premature myocardial infarction, a family history of deep vein thrombosis or the prothrombin 20210A mutation, or the presence of another known genetic hypercoagulability condition.

CPT Code(s): 81240

Factor V (Leiden)

Background: Inherited Thrombosis is associated with congenital predisposing risk factors such as Factor II (Prothrombin, FII)1 and Factor V (Leiden, FV) proteins involved in the blood coagulation enzyme activity cascade. The FII and FV mutations are present in ~2% and 5% of

individuals with N. European ancestry respectively, but at much lower levels in other populations. Factor V Leiden is inherited in an autosomal dominant manner.

Indications for Testing: In 2001, the American College of Medical Genetics (ACMG) recommended that FV DNA testing should be performed in patients with any type of venous thrombosis (hepatic, mesenteric, cerebral and recurrent), with a strong family history of thrombotic disease, pregnant women with venous thrombosis, women with history of pregnancy difficulties such as miscarriages, placental abruption, intrauterine fetal growth retardation or still birth, or womentaking oral contraceptives and female smokers (under age 50) with myocardial infarction.

CPT Code(s): 81241

Methylenetetrahydrofolate reductase (MTHFR)

Background: Methylenetetrahydrofolate reductase (MTHFR) that converts homocysteine to methionine as part of the pathway that converts 5,10-methylenetetrahydrofolate to 5-methyltetrahydrofolate. The MTHFR C677T and A1298C mutations are present in ~40-50% of N. European ancestry, but the frequency varies considerably in other ethnic groups

Indications for Testing: This test is used in the evaluation of patients with hyperhomocysteinemia. Testing includes molecular genotyping of the MTHFR 677 C > T variant and MTHFR 1298A>C variant. Having two copies of 677C>T or having one copy of 677C>T and one copy of 1298A>C is associated with increased plasma homocysteine concentration without adequate folic acid intake.

CPT Code(s): 81291

Ship Specimens to:

Henry Ford Center for Precision Diagnostics
Henry Ford Hospital
Clinic Building, K6, Core Lab E-655
2799 W. Grand Blvd.
Detroit, MI 48202