

## **Thrombophilia Panel by Array Tape (Test #470)**

### **Factor V (Leiden) G1691A Mutation Detection**

### **Factor II Prothrombin G20210A Mutation Detection**

### **Methylenetetrahydrofolate Reductase (MTHFR) C677T Mutation Detection**

During normal homeostasis, activated protein C (APC) limits clot formation by proteolytic inactivation of factors Va and VIIIa with the help of protein S. The lack of proteolytic inactivation of factor Va (APC resistance) has been associated with the presence of a single point mutation in nucleotide 1,691 of the factor V gene. This G – A substitution, known as the factor V Leiden mutation, is an autosomal dominant mutation responsible for an increased lifetime risk of thrombosis in families and individuals carrying this mutation.

Circulating prothrombin is converted to the serine protease thrombin, an enzyme central to thrombosis and homeostasis. A guanine to adenine mutation in the prothrombin gene at nucleotide 20210 has been associated with increased plasma prothrombin concentrations. This G – A substitution mutation (G20210A) acts in an autosomal dominant fashion and is also responsible for an increased lifetime risk of thrombosis in families and individuals carrying this mutation.

The methylenetetrahydrofolate reductase (MTHFR) gene product catalyzes the reduction of 5, 10-methylenetetrahydrofolate to 5-methyltetrahydrofolate, which is the major circulating form of folate in the human body. Folate, in its 5-methyl form, participates in many complex biochemical pathways, including nucleotide synthesis, DNA methylation, methylation of various proteins, neurotransmitters, and phospholipids, and the re-methylation of homocysteine to methionine. Clinically, mild hyperhomocysteinemia has been identified as an independent risk factor for arterial disease, venous thrombophilia, complications of pregnancy, and neural tube defects. Mild homocysteinuria is attributed to the presence of a C677T mutation of the MTHFR gene in its homozygous form. Analysis of the MTHFR gene for the presence of the C677T mutation, therefore, can be used to explain persistent hyperhomocysteinemia, and assess risk for development of vascular disease, complications of pregnancy and neural tube defects.

### **SPECIMEN REQUIREMENTS**

Collect a minimum of 1 to 10 ml of whole blood in EDTA (purple top tube) or ACD (yellow top tube). Whole blood collected in Na Heparin is acceptable but not preferred. Ship whole blood specimens at ambient temperature. Do not freeze blood tube. During hot summer months, include a frozen ice pack in the shipping container. Do not allow the ice pack to come in direct contact with the specimen tubes. In winter, include an unfrozen ice pack to help moderate extremes in temperature. The DNA in whole blood is stable for at least 48 hours at 21°C, 5-7 days at 4°C.

<b>CPT Codes and Cost</b>	<b>Thrombophilia Panel by Array Tape</b>	<b>\$89.00</b>
Molec Diag, Ascertainment	83890	
Molec Diag, Isolation	83891	
Molecular Diag, Amplif x3	83898	
Molecular Diag, Mut. Scan x3	83903	
Interpretation and Report	83912	

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