

**Methylenetetrahydrofolate
reductase (MTHFR)
Mutation Analysis
Cenetron Test Code 7003**

Description:	The presence of a C-to-T transition at nucleotide 677 of the methylenetetrahydrofolate reductase (MTHFR) gene results in the production of a thermolabile enzyme variant. A link has been established between this mutation, hyperhomocysteinemia and venous thromboembolic events. Moreover, studies have suggested that the presence of the C677T allele increases the risk of thrombosis when found in a patient carrying at least one copy of the Factor V Leiden (R506Q) and/or prothrombin (Factor II) G20210A mutations, emphasizing the importance of assaying all three alleles. The Cenetron MTHFR Mutation Analysis assay distinguishes between the wild-type (C) and mutant (T) alleles by monitoring differentially labeled molecular beacons in a single, closed tube via thermal cycling and real-time fluorescence detection.
Clinical Utility:	<ul style="list-style-type: none">♦ Genotypic detection of the MTHFR gene at nucleotide 677♦ Prediction of possible risk for venous thromboembolism
Technical Information:	The assay uses molecular beacons, probes that fluoresce upon hybridization with their complimentary target. The two-allele specific beacons are labeled with different fluorophores. By using two different molecular beacons in each PCR reaction, three possible allelic combinations (genotypes) of two sequence variants can be distinguished simultaneously by the type of fluorescence detected. The alleles in a DNA sample are determined by using real-time fluorescence detection during amplification.
Assay Days:	Monday through Friday Turnaround: 72 hours after receipt of specimen
Specimen Requirements:	Required volume: 3mL of whole blood (minimum) <ul style="list-style-type: none">♦ Draw blood into vacutainer containing EDTA as anticoagulant (purple top)♦ Store and transport specimen at 2-8°C♦ Specimen must be received within one week of phlebotomy♦ Patient's name, I.D. or birth date, and date of sample acquisition must be marked on tube
References:	Heller C, Schobess R, et al. Abdominal venous thrombosis in neonates and infants: role of prothrombotic risk factors - a multicentre case-control study. <i>Br J Haematol</i> 111(2):534-539, Nov 2000. Conroy J.M., G. Trivedi, T. Sovd and M. Caggana. The Allele Frequency of Mutations in Four Genes that Confer Enhanced Susceptibility to Venous Thromboembolism in an Unselected Group of New York State Newborns. <i>Thrombosis Research</i> 99(4):317-324, Aug 2000. Salomon O., Steinberg D.M., et al. Single and Combined Prothrombotic Factors in Patients With Idiopathic Venous Thromboembolism: Prevalence and Risk Assessment. <i>Arterioscler Thromb Vasc Biol</i> 19(3):511-518, Mar 1999.