Method Evolved for Recognition of Thrombophilia (MERT): Clinical Predictive Genetic Test for Venous Thrombosis

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Technology description

Venous thrombosis (VT) is one of the leading causes of mortality and morbidity resulting in approximately 300,000 hospitalizations and 50,000 fatalities per year in the United States with an incidence of 141 per 100,000 African-Americans, 104 per 100,000 Caucasians and 21 per 100,000 Asian/ Pacific Islanders. However, it is an avoidable disease if effective preventive measures such as early thromboprophylaxis are instituted.
It is highly beneficial to estimate individual thrombotic risk to aid in development of individualized risk-adapted prophylaxis.

Venous thrombosis is a multifactorial disorder and occurs as an outcome of a combination of environmental and genetic risk factors. In addition to well-established venous thrombosis associated acquired or environmental factors such as surgery, use of oral contraceptives and/or hormone replacement therapy, trauma, bone fractures, prolonged immobilization, advanced age, previous thrombosis history, malignancy and pregnancy, genetic predisposition via a number of variably penetrant genetic mutations or polymorphisms impart an increased risk for venous thrombosis.

In pregnant women, inherited thrombophilia can greatly increase the risk of adverse pregnancy outcomes such as miscarriages, intrauterine growth restriction, preeclampsia, placental abruption, or stillbirth as well as thrombosis during the recovery period after childbirth.

In addition to the differences in the prevalence of venous thrombosis among ethnic groups, there are accumulating data revealing differences in genetic determinants among ethnic groups such as differences in susceptibility associated genes and even in sequence alterations of the same gene. Furthermore some of the mutations and polymorphisms are mainly restricted to the specific populations. Such examples are FV Leiden, prothrombin G20210A polymorphisms. Whereas FV Leiden and prothrombin G0210A polymorphisms are the most prevalent risk factors for venous thrombosis in Caucasians, the patients from ethic populations other than Caucasians exhibit no or very rare FV Leiden or prothrombin G20210A polymorphisms.

This invention describes a highly-predictive genetic test to identify individuals with increased risk for venous thrombosis. It comprises a rapid, accurate and affordable genetic screen, utilizing genomic DNA microarray technology consisting of a combination of venous thrombosis associated mutations and polymorphisms that is applicable to diverse ethnic populations. Eight genes (antithrombin III, PC, PS, fibrinogen, factor V, prothrombin (factor II), MTHFR and ACE) are screened for the 143 known venous thrombosis-associated recurrent mutations and polymorphisms. This multi-gene test increases the predictive power for detection of genetic susceptibility to thrombosis over 20-fold compared to single-gene analysis, in multiple ethnic populations.
Market:
Individuals before or during exposure to situations that increase the risk of venous thrombosis, such as surgery, use of oral contraceptives and/or hormone replacement therapy, trauma, bone fractures, prolonged immobilization, long air journeys, advanced age, malignancy, or combinations thereof.
Pregnant women, or women who plan to become pregnant, as inherited thrombophilia is a significant risk factor for adverse pregnancy outcomes such as miscarriage, intrauterine growth restriction, preeclampsia, placental abruption, stillbirth and postpartum thrombotic events.

Application area

Rapid, cost-effective predictiv

Advantages

Rapid, cost-effective predictive test kit to identify pregnant women at risk for thrombophilia-associated adverse pregnancy outcomes such as miscarriage, intrauterine growth restriction, preeclampsia, placental abruption, or stillbirth as well as postpartum thrombosis.
Provides reduction of the yearly incidence of venous thrombosis by early identification of individuals at inherited risk, allowing protection before they develop symptoms by instituting effective preventive measures, such as early thromboprophylaxis or even decisions such as avoiding the use of oral contraceptives or hormone replacement therapy.
Provides advantages over currently available plasma-based thrombophilia screening panel by avoiding underdetermination of anticoagulant protein deficient individuals or by avoiding high rates of false positivity.
Allows individualized management and anticoagulation treatment of patients according to inherited thrombophilia status.

Institution

NIH - National Institutes of Health

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