Hereditary Thrombophilias

Precision Panel

Overview

Hereditary Thrombophilias are states of hypercoagulability that increase the risk of patients to develop clots, venous thrombosis and arterial thrombosis. Out of these, venous thrombosis and pulmonary embolism carry the highest risk of morbidity and mortality. Some hereditary thrombophilias include Factor V Leiden, Prothrombin 20210A, Protein C deficiency, Protein S deficiency and Antithrombin deficiency. Out of these, Factor V Leiden is the most common form of inherited thrombophilia. Thrombophilias may have an autosomal dominant, autosomal recessive or X-linked inheritance. Association of inherited thrombophilias with pregnancy increases the risk of thromboembolic events and it may be related to other complications such as preeclampsia, recurrent miscarriages, early detachment of the placenta and prematurity.

The Igenomix Hereditary Thrombophilias Precision Panel can be used to make an accurate and directed diagnosis as well as a differential diagnosis of recurrent thrombosis ultimately leading to a better management and prognosis of the disease. It provides a comprehensive analysis of the genes involved in this disease using next-generation sequencing (NGS) to fully understand the spectrum of relevant genes involved.

Indications

The Igenomix Hereditary Thrombophilias Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations:

- History of recurrent thromboembolism
- Thrombosis at a young age
- Family history of thrombosis
- Thrombosis in unusual sites

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of treatment with a multidisciplinary team in the form of medical care with anticoagulants and management of bleeding.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.

- Improvement of delineation of genotype-phenotype correlation.
- Identification of causative genes of hereditary thrombophilias given the high degree of heterogeneity with various clinical presentations and prognoses.

Genes & Diseases

*Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial.
**Number of clinically relevant mutations according to HGMD

Methodology

Contact us

Call +34 963 905 310 or send an email to supportspain@igenomix.com for any of the following objectives:

- Get more information about the test.
- Request your kit.
- Request a pick up of the kit after collecting the sample.

References

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