Congenital Afibrinogenemia

Precision Panel

Overview

Congenital fibrinogen disorders are a group of most frequent rare coagulation disorders, characterized by deficiency or defects in fibrinogen molecules. Congenital Afibrinogenemia is quantitative defect in fibrinogen resulting from mutations that affect plasma fibrinogen concentration and are frequently associated with a bleeding diathesis. Fibrinogen is a glycoprotein that is synthesized in the liver and circulates in the plasma and its physiologic function is in hemostasis. At the end of the coagulation cascade, fibrinogen is converted into fibrin creating a fibrin clot. It is involved as well in platelet aggregation and fibrinolysis. These disorders are generally inherited in an autosomal recessive pattern.

The Igenomix Congenital Afibrinogenemia Precision Panel can be used to make an accurate and directed diagnosis as well as a differential diagnosis of recurrent bleeding 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 Congenital Afibrinogenemia Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations:

- Umbilical cord bleeding present at birth and postpartum hemorrhage
- Epistaxis (nose bleeding)
- Hemarthrosis (bleeding in joint space)
- Gastrointestinal bleeding
- Menorrhagia
- Traumatic and surgical bleeding
- Intracranial hemorrhage in rare cases
- Recurrent spontaneous abortions may occur in affected female patients.

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.

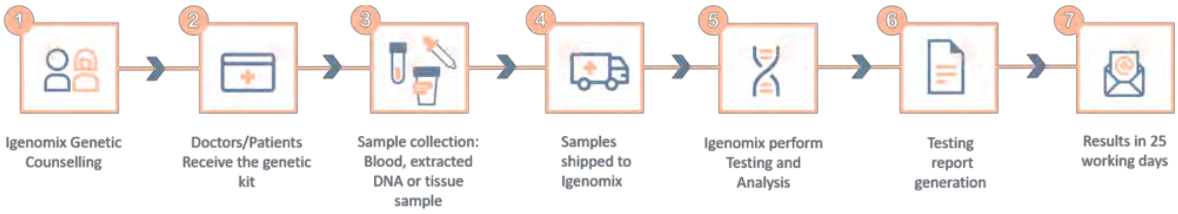
- Early initiation of treatment with a multidisciplinary team in the form of medical therapy with fibrinogen replacement therapy, and continuous monitoring for possible liver transplantation.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
FGA	Congenital Hypofibrinogenemia, Familial Afibrinogenemia , Familial Dysfibrinogenemia	AD,AR	100	153 of 154
FGB	Congenital Hypofibrinogenemia, Familial Afibrinogenemia , Familial Dysfibrinogenemia	AR	99.97	92 of 93
FGG	Congenital Hypofibrinogenemia, Familial Afibrinogenemia , Familial Dysfibrinogenemia	AR	99.95	136 of 138

*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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