Von Willebrand Disease

Precision Panel

Overview

Von Willebrand disease (VWD) is the most common inherited bleeding disorder with a heterogeneous clinical presentation and genetic background. The main feature relies on a deficiency or dysfunction of the protein named von Willebrand factor (vWF) resulting in an impaired primary homeostasis where platelets play a crucial role. Von Willebrand factor serves as a mediator for platelet adhesion during vascular injury and a reservoir and stabilizer for protein factor VIII, the absence of this protein causes a qualitative platelet disorder. Significant variability exists among family members that suffer from this disease depending on the amount on functioning circulating von Willebrand factor.

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

- Easy bruising
- Nosebleeds
- Gingival bleeding
- Prolonged bleeding
- Severe hemorrhage
- Menorrhagia
- Jaundice
- Splenomegaly
- Hematomas
- Petechiae
- Ecchymosis

Clinical Utility

The clinical utility of this panel is:

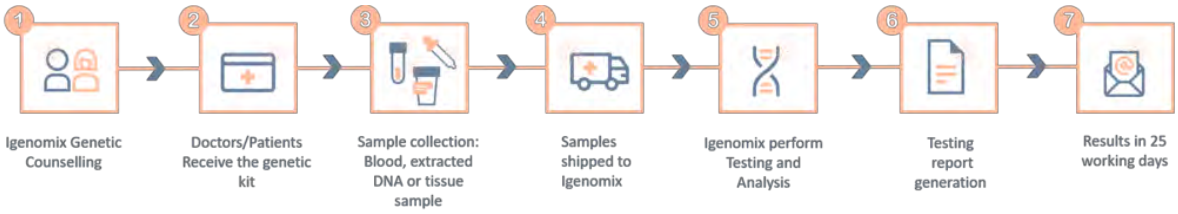
- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of treatment with a multidisciplinary team in the form of medical treatment with desmopressin, recombinant therapy and prevention of events that potentially increase risk of bleeding.
- 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**
F8	Hemophilia A	X,XR,G	99.89	-
GP1BA	Bernard-Soulier Syndrome, Nonarteritic Anterior Ischemic Optic Neuropathy, Von Willebrand Disease, Thrombocytopenia	AD,AR	99.98	73 of 73
GP1BB	Bernard-Soulier Syndrome, 22q11.2 Deletion Syndrome, Thrombocytopenia	AR	74.08	26 of 50
GP9	Bernard-Soulier Syndrome	AR	99.96	41 of 41
ITGA2B	Glanzmann Thrombasthenia, Thrombocytopenia	AD,AR	100	237 of 239
ITGB3	Glanzmann Thrombasthenia, Thrombocytopenia	AD,AR	99.44	178 of 179
NBEAL2	Gray Platelet Syndrome	AR	99.74	51 of 51
VWF	Von Willebrand Disease	AD,AR	98	933 of 1001

*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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