Hereditary Hemorrhagic Telangiectasia

Precision Panel

Overview

Hereditary Hemorrhagic Telangiectasia (HHT), also known as Osler-Weber-Rendu disease (OWRD) is a rare autosomal dominant disease that affects blood vessels throughout the body, causing a vascular dysplasia that leads to an increase tendency of bleeding. The bleeding presents as arteriovenous malformations (AVMs) and telangiectasias in specific locations, a potential source of serios morbidity and mortality. It is characterized by nosebleeds, telangiectasias on the lips, hands and oral mucosa. Symptom on set may be delayed until the fourth decade of life or later, and prognosis is variable depending on the severity and location of the bleeding.

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

- Spontaneous, recurrent nosebleeds
- Telangiectasia (dilated veins)
- Family history of HHT (first-degree relative)
- Hepatic or pulmonary bleeding
- Gastrointestinal bleeding

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 and surgical treatment to decrease the amount of hemorrhage and minimizing sequelae of arteriovenous malformations.

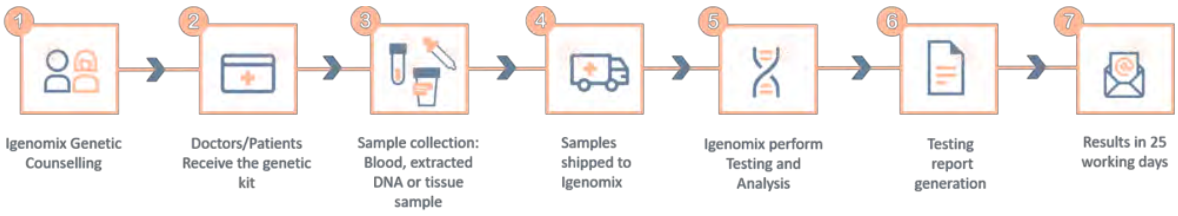
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.
- Facilitate gene discovery, identify genetic modifiers to explain clinical variability and potentially define and increased spectrum of hereditary telangiectasia disorders.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ACVRL1	Hereditary Hemorrhagic Telangiectasia	AD	100	457 of 462
ENG	Familial Cerebral Saccular Aneurysm, Generalized Juvenile Polyposis/Juvenile Polyposis Coli, Hereditary Hemorrhagic Telangiectasia	AD	100	467 of 471
EPHB4	Capillary Malformation-Arteriovenous Malformation, Nonimmune Hydrops Fetalis, And/Or Atrial Septal Defect, Vein Of Galen Aneurysmal Malformation	AD	100	65 of 65
GDF2	Hereditary Hemorrhagic Telangiectasia	AD	98.58	51 of 57
RASA1	Capillary Malformation-Arteriovenous Malformation, Parkes Weber Syndrome	AD	99.56	169 of 169
SMAD4	Juvenile Polyposis Syndrome, Juvenile Polyposis With Hereditary Hemorrhagic Telangiectasia, Generalized JuvenileWith Pulmonary Arteriovenous Malformation, Myhre Syndrome, Familial Thoracic Aortic Aneurysm And Aortic Dissection	AD	99.56	136 of 136

*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

1. Shovlin, C. L., Simeoni, I., Downes, K., Frazer, Z. C., Megy, K., Bernabeu-Herrero, M. E., Shurr, A., Brimley, J., Patel, D., Kell, L., Stephens, J., Turbin, I. G., Aldred, M. A., Penkett, C. J., Ouwehand, W. H., Jovine, L., & Turro, E. (2020). Mutational and phenotypic characterization of hereditary hemorrhagic telangiectasia. *Blood*, *136*(17), 1907–1918. https://doi.org/10.1182/blood.2019004560
2. Leng, H., Zhang, Q., & Shi, L. (2019). *Lin chuang er bi yan hou tou jing wai ke za zhi = Journal of clinical otorhinolaryngology, head, and neck surgery*, *33*(7), 591–592. https://doi.org/10.13201/j.issn.1001-1781.2019.07.004
3. McDonald, J., Wooderchak-Donahue, W., VanSant Webb, C., Whitehead, K., Stevenson, D. A., & Bayrak-Toydemir, P. (2015). Hereditary hemorrhagic telangiectasia: genetics and molecular diagnostics in a new era. *Frontiers in genetics*, *6*, 1. https://doi.org/10.3389/fgene.2015.00001
4. Major, T., Gindele, R., Szabó, Z., Jóni, N., Kis, Z., Bora, L., Bárdossy, P., Rácz, T., Karosi, T., & Bereczky, Z. (2019). A herediter haemorrhagiás teleangiectasia (Osler–Weber–Rendu-kór) genetikai diagnosztikája [Genetic diagnostics of hereditary hemorrhagic telangiectasia (Osler-Weber-Rendu disease)]. *Orvosi hetilap*, *160*(18), 710–719. https://doi.org/10.1556/650.2019.31380
5. Duffau, P., Lazarro, E., & Viallard, J. F. (2014). Maladie de Rendu-Osler [Hereditary hemorrhagic telangiectasia]. *La Revue de medecine interne*, *35*(1), 21–27. https://doi.org/10.1016/j.revmed.2013.02.022
6. McDonald, J., Bayrak-Toydemir, P., & Pyeritz, R. E. (2011). Hereditary hemorrhagic telangiectasia: an overview of diagnosis, management, and pathogenesis. *Genetics in medicine : official journal of the American College of Medical Genetics*, *13*(7), 607–616. https://doi.org/10.1097/GIM.0b013e3182136d32