Neurofibromatosis

Precision Panel

Overview

Neurofibromatosis type 1 (NF1) and type 2 (NF2) are neurocutaneous congenital disorders that affect organs of ectodermal origin including skin, central nervous system, and the eyes. All are inherited in an autosomal dominant pattern and are characterized by a high rate of mutational change occurring for the first time in an individual as well as variable expression. NF1 and NF2 differ with regards to their age of onset, clinical manifestations, gene loci, and gene protein products. However, in both conditions, the altered gene products have a crucial role in the dysregulation of tumor suppression. NF1, also known as von Recklinghausen syndrome, includes effects on multiple systems of the body and the major associated tumor is the neurofibroma. In addition, clinical manifestations include bone dysplasia, learning disabilities, and increased risk of malignancy. NF2 includes schwannomas of multiple cranial and spinal nerves, among others.

The Igenomix Neurofibromatosis Precision Panel can serve as a diagnostic tool 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, and their high or intermediate penetrance.

Indications

The Igenomix Neurofibromatosis Precision Panel is indicated in patients with a clinical suspicion or diagnosis of Neurofibromatosis type 1 or type 2 presenting with the following manifestations:

- Multiple neurofibromas
- Café au lait spots
- Lisch nodules (pigmented iris hamartomas)
- Seizures and/or focal neurologic signs
- Intellectual disability
- Bone involvement: scoliosis, short stature, fractures, cortical thinning etc
- Associated with other tumors: pheochromocytoma, Wilms tumor, meningiomas, ependymomas
- Bilateral vestibular schwannomas (acoustic neuroma)
- Early-onset cataracts

Clinical Utility

The clinical utility of this panel is:

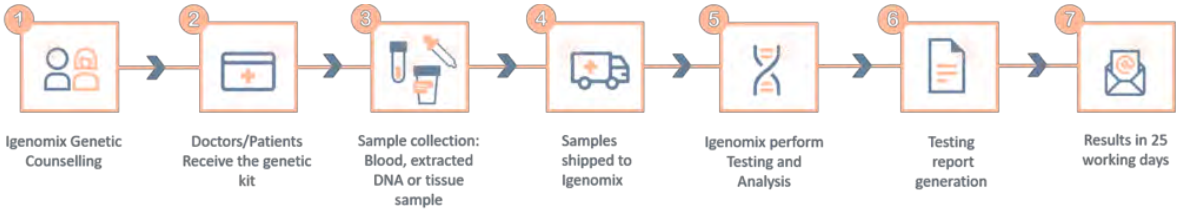
- The genetic and molecular diagnosis for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of treatment with a multidisciplinary team for appropriate surveillance for malignancy, appropriate laser treatment and/or chemotherapy, surgical resection of tumors as well as orthopaedic intervention.
- Risk assessment of asymptomatic family members according to the mode of inheritance
- Improved pathways from diagnosis to treatment in susceptible populations.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
LZTR1	Noonan Syndrome, Schwannomatosis	AD	99.99	136 of 136
MAP2K2	Cardiofaciocutaneous Syndrome, Neurofibromatosis-Noonan Syndrome	AD	100	37 of 37
NF1	Juvenile Myelomonocytic Leukemia, Neurofibromatosis-Noonan Syndrome, Familial Spinal Neurofibromatosis Type 1, Watson Syndrome, 17q11.2 Microduplication Syndrome, Hereditary Pheochromocytoma-Paraganglioma	AD	97.97	3082 of 3166
NF2	Familial Meningioma, Neurofibromatosis Type 2, Schwannomatosis	AD	100	359 of 362
SMARCB1	Coffin-Siris Syndrome, Rhabdoid Tumor Predisposition Syndrome, Schwannomatosis	AD	100	97 of 99
SPRED1	Legius Syndrome	AD	100	84 of 84

*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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