Tuberous Sclerosis

Precision Panel

Overview

Tuberous Sclerosis Complex (TSC) is an inherited neurocutaneous disorder that is characterized by pleomorphic features involving many organ systems.

One of the main features of this disorder is the presence of multiple benign hamartomas of the brain, eyes, heart, lung, liver, kidney and skin. It is an autosomal dominant genetic disorder caused by mutations in genes TSC1, TSC2 or IFNG that in turn affect cellular differentiation, proliferation and migration early in development. The most characteristic feature of TSC is adenoma sebaceum, which appears in late childhood or early adolescence. There is an increased risk of malignancy and susceptibility to epilepsy.

The Igenomix Tuberous Sclerosis Precision Panel can be used to make an accurate and directed diagnosis 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 Tuberous Sclerosis Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations:

- 1. Hamartomas and benign neoplasms in brain and various tissues (cerebral cortex, retina, heart, kidney)
- 2. Adenoma sebaceum (angiofibromas)
- 3. Hypopigmented macules
- 4. Shagreen patches
- 5. Epilepsy
- 6. Behavioral problems
- 7. Psychosocial difficulties

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 multidisciplinary treatment in the form of medical care with immunosuppressant medication as well as antiepileptic medications to prevent seizures. Early and routine surveillance is needed for malignancy screening. Surgical care for seizures may be necessary.
- 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)	CLINVAR**	HGMD**
IFNG	Tuberous Sclerosis	AD,AR	99.77	-	-
TSC1	Tuberous Sclerosis	AD	99.86	228 of 238	390 of 406
TSC2	Tuberous Sclerosis	AD	100	495 of 497	1157 of 1159

*Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial. D: Digenic; G: Gonosomal
**Number of clinically relevant pathogenic and likely pathogenic variants, according to ClinVar and HGMD.

Methodology

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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