Cystic Fibrosis
Precision Panel

Overview

Cystic Fibrosis (CF) is the most common lethal inherited disease in white persons. It is a life-limiting autosomal recessive genetic disorder, with highest prevalence in Europe, North America and Australia. The disease is caused by mutation of a gene that encodes a chloride-conducting transmembrane channel that regulates anion transport and mucociliary clearance in the airways and other exocrine glands. This leads to an exocrine gland dysfunction that involves multiple organ systems resulting in chronic respiratory infections, pancreatic enzyme insufficiency, infertility and associated complications in untreated patients. End-stage lung disease is the principal cause of death. Most of the carriers of the CF gene are asymptomatic, therefore the importance of prenatal diagnosis as well as parent screening can identify those patients at risk of transmitting the mutation to their offspring.

The Igenomix Cystic Fibrosis Precision Panel can be used as a diagnostic and screening 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.

Indications

The Igenomix Cystic Fibrosis Precision Panel is indicated in those cases where there is a clinical suspicion of CF or family history of CF with or without the following manifestations:

- Meconium ileus
- Rectal prolapse
- Pancreatic insufficiency: fat-soluble vitamin deficiency, malabsorption of fats, proteins and carbohydrates
- Failure to thrive
- Foul-smelling flatus
- Recurrent abdominal pain and abdominal distention
- Chronic and recurrent cough
- Recurrent pneumonia
- Shortness of breath on exertion
- Upper respiratory tract infections
- Undescended testicles
- Infertility

Clinical Utility

The clinical utility of this panel is:

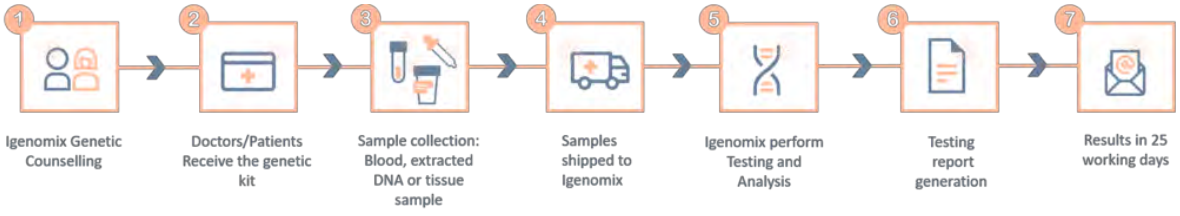
- The genetic and molecular diagnosis for an accurate clinical diagnosis and improve prognosis.
- Early initiation of treatment with a multidisciplinary team to maintain lung function, administering nutritional therapy to maintain adequate growth and manage complications.
- Risk assessment and genetic counselling of asymptomatic family members to identify the likelihood of the individual being affected by the disease or being a carrier.
- Factors to consider when deciding whether to have CF genetic testing.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
CA12	Hyperchlorhidrosis	AR	100	4 of 4
CFTR	Bronchiectasis, Congenital Bilateral Absence Of Vas Deferens, Cystic Fibrosis, Hereditary Pancreatitis	AD,AR	95.45	1615 of 1730
CLCA4	Cystic Fibrosis	-	97.66	NA of NA
DCTN4	Cystic Fibrosis	-	100	1 of 1
FCGR2A	Cystic Fibrosis	AD,AR	93.97	NA of NA
SCNN1A	Bronchiectasis With Or Without Elevated Sweat Chloride, Idiopathic Bronchiectasis	AD,AR	99.95	46 of 46
SCNN1B	Bronchiectasis	AD,AR	100	56 of 56
SCNN1G	Bronchiectasis With Or Without Elevated Sweat Chloride, Idiopathic Bronchiectasis	AD,AR	100	28 of 28
STX1A	Cystic Fibrosis	-	97	3 of 3
TGFB1	Cystic Fibrosis	AD,AR	99.75	24 of 24

* Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial
** HGMD: 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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