Idiopathic Pulmonary Fibrosis
Precision Panel

Overview

Idiopathic Pulmonary Fibrosis (IPF) is a specific form of chronic, progressive lung disease defined as the presence of progressive lung scarring in the form of fibrosing interstitial pneumonia of unknown cause with the histopathological finding of usual interstitial pneumonia (UIP). Although the etiology is unknown, there probably is an effect of endogenous and exogenous micro-environmental factors in subjects together with genetic predisposition. All of this causes repetitive micro-injury to the lung tissue and vasculature, triggering and inflammatory response and ultimately fibrosis. It occurs primarily in older adults, and the progressive lung scarring over time results in reduced oxygen intake.

The Igenomix Idiopathic Pulmonary Fibrosis Precision Panel can be used 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.

Indications

The Igenomix Idiopathic Pulmonary Fibrosis Precision Panel is indicated in those cases where there is a clinical suspicion of IPF with or without the following manifestations during at least six months:

- Weight loss
- Low-grade fevers
- Fatigue
- Arthralgias (articular pain)
- Myalgias (muscular pain)
- Gradual onset shortness of breath with exertion
- Non-productive cough

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 for treatment of comorbid medical conditions as well as initiate early supportive treatment, surgical treatment and regular surveillance of pulmonary function.

- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ABCA3	Surfactant Metabolism Dysfunction, Pulmonary, Idiopathic Pulmonary Fibrosis, Infant Acute Respiratory Distress Syndrome	AR	100	286 of 289
AP3B1	Hermansky-Pudlak Syndrome	AR	100	34 of 35
ATP11A	Idiopathic Pulmonary Fibrosis		99.97	NA of NA
DKC1	X-linked Dyskeratosis Congenita, Hoyeraal-Hreidarsson Syndrome	X,XR,G	100	NA of NA
DPP9	Idiopathic Pulmonary Fibrosis		93.97	1 of 1
DSP	Carvajal Syndrome, Idiopathic Pulmonary Fibrosis	AD,AR	99.91	366 of 369
FAM13A	Idiopathic Pulmonary Fibrosis		99.91	NA of NA
HPS1	Hermansky-Pudlak Syndrome	AR	99.98	68 of 68
HPS4	Hermansky-Pudlak Syndrome	AR	99.7	30 of 30
MUC5B	Idiopathic Pulmonary Fibrosis	AD	99.89	12 of 12
NKX2-1	Brain-Lung-Thyroid Syndrome	AD	97.04	115 of 123
PARN	Autosomal Recessive Dyskeratosis Congenita, Pulmonary Fibrosis And/Or Bone Marrow Failure, Hoyeraal-Hreidarsson Syndrome, Idiopathic Pulmonary Fibrosis	AD,AR	99.98	33 of 33
RTEL1	Autosomal Recessive Dyskeratosis Congenita, Pulmonary Fibrosis And/Or Bone Marrow Failure, Hoyeraal-Hreidarsson Syndrome, Idiopathic Pulmonary Fibrosis	AD,AR	99.73	127 of 131
SFTPA1	Idiopathic Pulmonary Fibrosis		100	4 of 4
SFTPA2	Idiopathic Pulmonary Fibrosis	AD	99.98	6 of 6
SFTPC	Idiopathic Pulmonary Fibrosis, Surfactant Metabolism Dysfunction, Infant Acute Respiratory Distress Syndrome	AD	99.84	83 of 83
STN1	Idiopathic Pulmonary Fibrosis	AR	99.87	NA of NA
TERC	Autosomal Dominant Dyskeratosis Congenita, Pulmonary Fibrosis And/Or Bone Marrow Failure, Idiopathic Pulmonary Fibrosis	AD	NA	NA
TERT	Autosomal Dominant Dyskeratosis Congenita, Pulmonary Fibrosis And/Or Bone Marrow Failure, Idiopathic Pulmonary Fibrosis, Hoyeraal-Hreidarsson Syndrome	AD,AR	99.09	194 of 197
TINF2	Autosomal Dominant Dyskeratosis Congenita, Revesz Syndrome, Hoyeraal-Hreidarsson Syndrome	AD	99.94	47 of 47

* 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

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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