Surfactant Metabolism Dysfunction Precision Panel

Overview

Surfactant Metabolism Dysfunction is a condition characterized by insufficient pulmonary surfactant needed for appropriate respiration. Pulmonary surfactant is responsible for maintaining surface tension at the liquid-air interphase in the alveoli, so a deficiency in this component results in easily collapsing alveoli immediately after expiration. Surfactant is composed of phospholipids and proteins and so a deficient metabolism can lead to an imbalance in the amount of these components resulting in a non-functioning surfactant. Genetic disorders of the surfactant homeostasis genes can lead to multiple lung diseases in neonates, children and adults, including neonatal respiratory distress syndrome, interstitial pneumonia, pulmonary alveolar proteinosis and pulmonary fibrosis.

The Igenomix Surfactant Metabolism Dysfunction 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 Surfactant Metabolism Dysfunction Precision Panel is indicated in those cases where there is a clinical suspicion of surfactant metabolism dysfunction with or without the following manifestations:

- Abnormally rapid breathing (tachypnea)
- Low concentration of oxygen in blood (hypoxemia)
- Failure to thrive
- Premature birth

Clinical Utility

The clinical utility of this panel is:

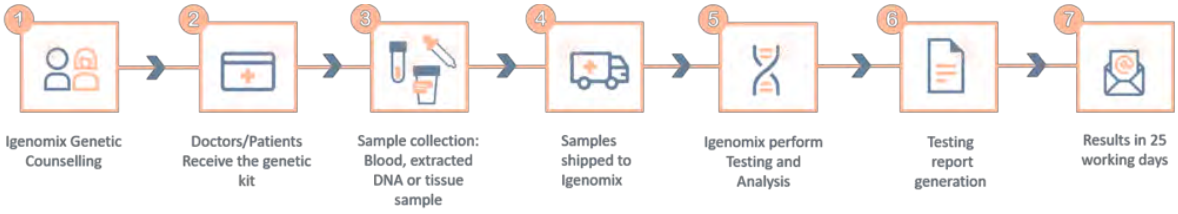
- The genetic and molecular diagnosis for an accurate clinical diagnosis and improve prognosis.
- Early initiation of treatment with a multidisciplinary team to maintain lung function, early surfactant replacement therapy, and manage complications.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ABCA3	Pulmonary Surfactant Metabolism Dysfunction, Idiopathic Pulmonary Fibrosis, Infant Acute Respiratory Distress Syndrome	AR	100	286 of 289
CSF2RA	Pulmonary Surfactant Metabolism Dysfunction, Hereditary Pulmonary Alveolar Proteinosis	X	96.75	NA of NA
CSF2RB	Pulmonary Surfactant Metabolism Dysfunction, Hereditary Pulmonary Alveolar Proteinosis	AR	99.98	6 of 6
MARS1	Interstitial Lung And Liver Disease	AD,AR	99.98	19 of 19
SFTPA1	Idiopathic Pulmonary Fibrosis	-	100	4 of 4
SFTPB	Pulmonary Surfactant Metabolism Dysfunction, Infant Acute Respiratory Distress Syndrome, Neonatal Acute Respiratory Distress Due To SP-B Deficiency	AR	100	27 of 27
SFTPC	Idiopathic Pulmonary Fibrosis, Pulmonary Surfactant Metabolism Dysfunction, Infant Acute Respiratory Distress Syndrome	AD	99.84	83 of 83
SFTPD	Pulmonary Alveolar Proteinosis, Pulmonary Fibrosis, Intersitial Lung Disease	-	99.98	1 of 1

* 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

1. McFetridge, L., McMorrow, A., Morrison, P. J., & Shields, M. D. (2009). Surfactant Metabolism Dysfunction and Childhood Interstitial Lung Disease (chILD). *The Ulster medical journal*, *78*(1), 7–9.
2. Al-Saiedy, M., Pratt, R., Lai, P., Kerek, E., Joyce, H., Prenner, E., Green, F., Ling, C. C., Veldhuizen, R., Ghandorah, S., & Amrein, M. (2018). Dysfunction of pulmonary surfactant mediated by phospholipid oxidation is cholesterol-dependent. *Biochimica et biophysica acta. General subjects*, *1862*(4), 1040–1049. https://doi.org/10.1016/j.bbagen.2018.01.008
3. Nkadi, P. O., Merritt, T. A., & Pillers, D. A. (2009). An overview of pulmonary surfactant in the neonate: genetics, metabolism, and the role of surfactant in health and disease. *Molecular genetics and metabolism*, *97*(2), 95–101. https://doi.org/10.1016/j.ymgme.2009.01.015
4. Nogee L. M. (2004). Genetic mechanisms of surfactant deficiency. *Biology of the neonate*, *85*(4), 314–318. https://doi.org/10.1159/000078171