Hereditary Pancreatitis

Precision Panel

Overview

Pancreatitis is a condition that occurs when the pancreas becomes inflamed leading to a state of systemic inflammation. Hereditary pancreatitis is caused by repeated episodes of pancreas inflammation. The pancreas produces enzymes that help digest food and it also produces insulin, a hormone that controls blood sugar levels in the body. Episodes of pancreatitis can lead to a permanent tissue damage and loss of pancreatic function. Symptoms usually begin within the first two decades but can start at any time. It is due in part to an autosomal dominant gain-of-function disorder related to mutations of the cationic trypsinogen gene with an 80% penetrance. Mutations in this gene cause a premature activation of trypsinogen to trypsin. Most cases are inherited in an autosomal dominant manner or due to a de novo gene change.

The Igenomix Hereditary Pancreatitis Precision Panel can be used to make a directed and accurate differential diagnosis of acute pancreatitis 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 Disorders of Sex Development Precision Panel is indicated for those patients with clinical suspicion of an intersex condition presenting with the following manifestations:

- Abdominal pain
- Nausea and vomiting
- Weight loss
- Diarrhea
- Flatulence and bloating
- Fat in stools
- Diabetes mellitus
- Fever

Clinical Utility

The clinical utility of this panel is:

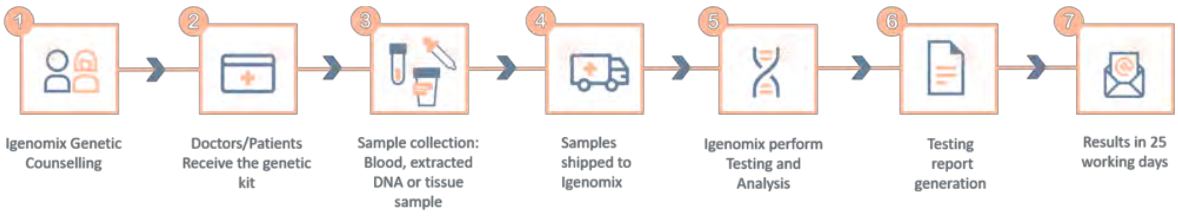
- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of treatment with a multidisciplinary team for early preventative strategies, surgical care and pharmacologic treatment.
- Risk assessment of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
CASR	Autosomal Dominant Hypocalcemia, Hereditary Chronic Pancreatitis, Hyperparathyroidism, Autosomal Dominant Neonatal Severe Primary Hypocalcemia, Familial Hypocalciuric Hypercalcemia, Neonatal Severe Primary Hyperparathyroidism	AD,AR	100%	445 of 446
CFTR	Bronchiectasis, Congenital Bilateral Absence Of Vas Deferens, Cystic Fibrosis, Hereditary Chronic Pancreatitis	AD,AR	95.45%	1615 of 1730
CLDN2	Male Infertility, Inflammatory Bowel Disease	-	99.94%	NA of NA
CTRC	Hereditary Chronic Pancreatitis	AD	100%	46 of 46
PRSS1	Hereditary Chronic Pancreatitis	AD	100%	52 of 55
PRSS2	Hereditary Chronic Pancreatitis	AD	na	na
SPINK1	Hereditary Chronic Pancreatitis	AD,AR	100%	45 of 50

*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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