Hyperlipidemia

Precision Panel

Overview

Hyperlipidemia is a set of metabolic disorders that can be genetic or acquired that are characterized by excess lipids in the blood which can include cholesterol and/or triglycerides. This excess causes increased fatty acid deposition which leads to blockages in the arteries. This, in turn can lead to the development of atherosclerotic plaques throughout the body and subsequent vascular disease. Genetic hyperlipidemia is mostly inherited in an autosomal dominant manner but can also be inherited in an autosomal recessive pattern. Hyperlipidemia is a general term used to identify a disease associated with excess lipids and/or fats in the body and hypercholesterolemia is one of the most common forms of hyperlipidemia.

The Igenomix Hyperlipidemia Precision Panel can be used to make a directed and accurate differential diagnosis of excess lipids ultimately leading to a better management and prognosis of the disease and its outcomes. 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 Hyperlipidemia Precision Panel is indicated for those patients with clinical suspicion of inherited hyperlipidemia presenting with the following manifestations:

- High cholesterol
- High blood pressure
- Symptoms of peripheral artery disease: leg discomfort, leg pain or cramps when walking, pain in the ball of the foot, foot ulcers.
- Symptoms of stroke: sudden severe headache, weakness, numbness, loss of movement in one arm, partial vision loss, inability to speak clearly.
- Symptoms of heart attack: chest pain, shortness of breath, sweating.

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 pharmacologic therapy, dietary modifications, and primary prevention to reduce comorbidities associated with hyperlipidemia.
- Risk assessment of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ABCA1	Apolipoprotein A-I Deficiency, Familial HDL Deficiency, Tangier Disease	AD,AR	100%	238 of 241
ABCG5	Homozygous Familial Hypercholesterolemia	-	99.81%	57 of 57
ABCG8	Gallbladder Disease, Homozygous Familial Hypercholesterolemia, Sitosterolemia	AR,MU,P	100%	64 of 64
ALMS1	Alstrom Syndrome	AR	99.92%	302 of 305
APOA1	Familial Visceral Amyloidosis, Apolipoprotein A-I Deficiency	AD	99.89%	68 of 70
APOA5	Hyperlipoproteinemia Type V, Familial Hypertriglyceridemia	AD	99.88%	66 of 66
APOB	Homozygous Familial Hypercholesterolemia, Autosomal Dominant Hypercholesterolemia Type B, Familial Hypobetalipoproteinemia	AD,AR	99.62%	369 of 375
APOC2	Apolipoprotein C-II Deficiency	AR	100%	22 of 23
APOC3	Cholesterol-Ester Transfer Protein Deficiency, Hyperalphalipoproteinemia	-	100%	8 of 8
APOE	Alzheimer Disease, Dysbetalipoproteinemia, Lipoprotein Glomerulopathy, Age-Related Macular Degeneration, Sea-Blue Histiocyte Disease	AD,AR	99.53%	65 of 68
ATF6	Achromatopsia, Cone Rod Dystrophy	AR	99.98%	16 of 16
CETP	Cholesterol-Ester Transfer Protein Deficiency, Hyperalphalipoproteinemia	AD	100%	52 of 54
CREB3L3	Spondyloepimetaphyseal Dysplasia, Sturdwick Type, Platyspondylic Lethal Skeletal Dysplasia, Torrance Type, Autosomal Recessive Otospondylomegaepiphyseal Dysplasia	-	100%	12 of 12
FABP2	Perinatal Necrotizing Enterocolitis, Acute Vascular Insufficiency of Intestine, Refractory Celiac Disease, Intestinal Atresia, Ileitis	-	99.69%	NA of NA
FOLH1	Prostate Cancer, Canavan Disease	-	99.63%	NA of NA
GPIHBP1	Hyperlipoproteinemia Type Id	AR	100%	35 of 36
LCAT	Fish-Eye Disease, Lecithin:Cholesterol Acyltransferase Deficiency	AR	90%	110 of 110
LDLR	Homozygous Familial Hypercholesterolemia	AD	99.89%	1921 of 1996
LDLRAP1	Homozygous Familial Hypercholesterolemia	AR	91.83%	18 of 27
LEPR	Leptin Receptor Deficiency	AR	97.92%	49 of 49
LIPA	Cholesteryl Ester Storage Disease, Lysosomal Acid Lipase Deficiency, Wolman Disease	AR	99.91%	103 of 104
LMF1	Combined Lipase Deficiency	AR	99.80%	34 of 36
LPL	Familial Combined Hyperlipidemia, Hyperlipoproteinemia Type I	AD,AR	100%	220 of 222
MADD	Multiple Acyl-CoA Dehydrogenation Deficiency	-	99.98%	9 of 9
NR1H3	Multiple Sclerosis, Fatty Liver Disease	-	99.87%	1 of 1
PCSK9	Homozygous Familial Hypercholesterolemia	AD	100%	96 of 98
PLTP	Hyperalphalipoproteinemia 1, Tangier Disease	-	100%	1 of 1
PPARG	Berardinelli-Seip Congenital Lipodystrophy, Carotid Intimal Medial Thickness, Noninsulin-Dependent Diabetes Mellitus, Familial Partial Lipodystrophy Type 3, Obesity, PPARG-related Familial Partial Lipodystrophy	AD,AR,MU,P	99.94%	53 of 53
SLC25A40	Congenital Myasthenic Syndrome	-	99.98%	2 of 2
USF1	Familial Combined Hyperlipidemia, Cowden Syndrome	-	100%	1 of 1

*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

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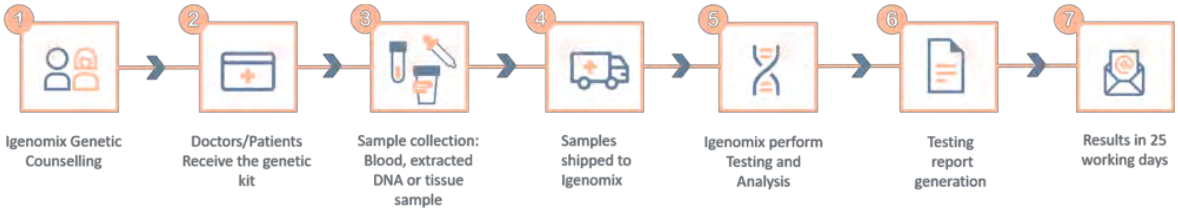
Methodology



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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