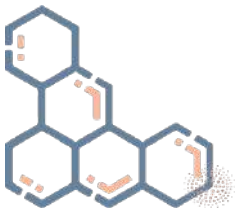
Lipodystrophy

Precision Panel



Overview

Lipodystrophy is a congenital or acquired disorder characterized by either complete or partial lack of adipose tissue. In some of these disorders, there is also the apparent accumulation of fat in other regions of the body. The primary defect is the loss of functional adipocytes, leading to ectopic steatosis, insulin resistance and severe dyslipidemia. The main cause of insulin resistance is the fact that the excess energy cannot be stored in adipose tissue, which is secondary to either the near total lack of adipocyte expandability in patients with generalized lipodystrophy, or a limited capacity to expand in partial lipodystrophy.

Abnormal appearance due to the absence or abnormal distribution of subcutaneous fat is noted within the first two years of life. During childhood, these patients manifest a high appetite, accelerated linear growth and advanced bone age. The onset of protuberant abdomen due to hepatomegaly caused by fatty infiltration of the liver is also common, which leads to severe cirrhosis. The mode of inheritance is autosomal recessive.

The Igenomix Lipodystrophy Precision Panel can be used to make an accurate and directed diagnosis 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 Lipodystrophy Precision Panel is indicated for those patients with a clinical diagnosis or suspicion presenting with or without the following manifestations:

- Loss of fat from upper extremities.
- Accumulation of fat into lower bodies.
- High appetite.
- Velvety dark skin.
- Disproportionate size of hands and feet.
- Irregular or absence period.

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 multidisciplinary treatment including pharmacological treatment with thiazolidinediones/PPARG modulators to increase adiponectin levels, and metformin in combination with insulin to stabilize glucose levels in blood. A proper low-fat diet may help reduce the symptoms, although it will not reverse lipoatrophy.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

The Igenomix Lipodystrophy Precision Panel includes 24 genes related to this condition.

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
AGPAT2	Lipodystrophy, Berardinelli-Seip Congenital Lipodystrophy	AR	100	42 of 43
AKT2	Lipodystrophy	AD	94.99	5 of 6
ATP2A1	Brody Myopathy	AR	100	20 of 20
BSCL2	Lipodystrophy, Berardinelli-Seip Congenital Lipodystrophy	AD,AR	99.83	60 of 61
CAV1	Lipodystrophy, Berardinelli-Seip Congenital Lipodystrophy	AD,AR	100	18 of 18
CAVIN1	Lipodystrophy, Berardinelli-Seip Congenital Lipodystrophy	AR	99.82	-
CIDEC	Lipodystrophy	AR	100	1 of 1
COL3A1	Acrogeria	AD,AR	100	676 of 676
FBN1	Stiff Skin Syndrome, Marfanoid-Progeroid-Lipodystrophy Syndrome	AD	100	2836 of 2845
GLRA1	Lipodystrophy	AD,AR	99.6	71 of 72
KCNJ6	Keppen-Lubinsky Syndrome	AD	99.94	3 of 3
LIPE	Abdominal Obesity-Metabolic Syndrome, Lipodystrophy	AR	97.08	4 of 4
LMNA	Atypical Werner Syndrome, Autosomal Recessive Emery-Dreifuss Muscular Dystrophy, Lipodystrophic Laminopathy, Lipodystrophy	AD,AR	100	619 of 620
LMNB2	Lipodystrophy	AD,AR	95.03	5 of 5
MTX2	Lipodystrophy	AR	95.5	-
OTULIN	Lipodystrophy	AR	87.67	-
PCYT1A	Lipodystrophy	AR	99.98	22 of 22
PIK3R1	Short Syndrome	AD,AR	99.89	29 of 29
PLIN1	Lipodystrophy	AD	97.47	8 of 8
POLD1	Lipodystrophy	AD	100	40 of 41
POLR3A	Neonatal Progeroid Syndrome	AR	100	122 of 122
PPARG	Lipodystrophy, Berardinelli-Seip Congenital Lipodystrophy	AD,AR,MU,P	99.94	53 of 53
PSMB8	Proteasome-Associated Autoinflammatory Syndrome	AR	100	11 of 11
ZMPSTE24	Lipodystrophy	AR	100	35 of 36

*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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