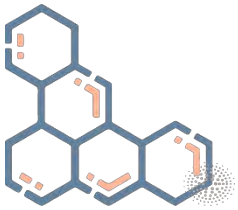
Lysosomal Storage Diseases

Precision Panel



Overview

Lysosomal Storage Diseases (LSD) are a group of dozens of inherited disorders that result from the accumulation of undigested or partially processed macromolecules inside organelles called lysosomes. Lysosomes are responsible for the physiologic turnover and digestion of cell constituents and do so with the help of catabolic enzymes. The accumulation of products inside the lysosomes results in cellular dysfunction and clinical abnormalities. Organomegaly, connective-tissue, ocular pathology and central nervous system dysfunction. It is transmitted in an autosomal recessive pattern.

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

- Intellectual disability
- Delayed physical development
- Seizures
- Facial and bone deformities
- Joint stiffness and pain
- Difficulty breathing
- Vision and hearing difficulties
- Anemia, nosebleeds and easy bleeding or bruising
- Enlarged liver or spleen

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 in the form enzyme replacement therapy, substrate reduction therapy, chaperone therapy and nutritional recommendations and frequent consultations to monitor possible complications.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ASAH1	Farber Lipogranulomatosis, Spinal Muscular Atrophy	AR	99.98	69 of 70
ATP13A2	Kufor-Rakeb Syndrome, Spastic Paraplegia, Neuronal Ceroid Lipofuscinosis	AR	99.97	53 of 53
CA2	Osteopetrosis, Tubular Acidosis	AR	100	36 of 36
CLCN7	Hypopigmentation, Osteopetrosis	AD,AR	99.85	109 of 111
CLN3	Ceroid Lipofuscinosis	AR	99.93	73 of 75
CLN5	Ceroid Lipofuscinosis	AR	99.56	52 of 55
CLN6	Ceroid Lipofuscinosis	AR	99.94	98 of 99
CLN8	Ceroid Lipofuscinosis, Progressive Epilepsy-Intellectual Disability Syndrome	AR	100	44 of 45
CTSD	Ceroid Lipofuscinosis	AR	100	18 of 18
CTSF	Ceroid Lipofuscinosis	AR	92.18	12 of 12
DNAJC5	Ceroid Lipofuscinosis	AD	100	2 of 2
GALNS	Morquio Syndrome	AR	100	344 of 348
GBA	Dementia, Gaucher Disease, Parkinson Disease	AD,AR	100	469 of 471
GLA	Fabry Disease	X,XR,G	98	-
GLB1	Gm1-Gangliosidosis, Morquio Syndrome	AR	100	242 of 243
GNPTAB	Mucolipidosis	AR	100	279 of 280
GRN	Ceroid Lipofuscinosis, Frontotemporal Lobar Degeneration, Semantic Dementia	AD,AR	100	220 of 229
IDS	Mucopolysaccharidosis	X,XR,G	99.86	-
IDUA	Hurler Syndrome, Scheie Syndrome	AR	99.73	287 of 292
KCTD7	Epilepsy	AR	99.99	40 of 40
LAMP2	Danon Disease, Glycogen Storage Disease	X,XD,G	99.96	-
LIPA	Cholesteryl Ester Storage Disease, Wolman Disease	AR	99.91	103 of 104
LMBRD1	Methylmalonic Aciduria, Methylmalonic Acidemia, Homocystinuria	AR	99.88	8 of 8
MAN2B1	Mannosidosis	AR	100	149 of 149
MANBA	Mannosidosis	AR	99.98	20 of 20
MFSD8	Ceroid Lipofuscinosis, Macular Dystrophy	AR	100	63 of 63
MYO5A	Griscelli Syndrome, Neuroectodermal Melanolysosomal Disease	AR	100	10 of 10
NPC1	Niemann-Pick Disease	AR	97	503 of 505
NPC2	Niemann-Pick Disease	AR	100	27 of 27
OCRL	Dent Disease, Lowe Oculocerebrorenal Syndrome	X,XR,G	100	-
PPT1	Ceroid Lipofuscinosis	AR	100	81 of 81
PSAP	Combined Saposin Deficiency, Gaucher Disease, Krabbe Disease, Metachromatic Leukodystrophy, Encephalopathy	AR	100	33 of 33
SCARB2	Action Myoclonus-Renal Failure Syndrome, Gaucher Disease, Unverricht-Lundborg Disease	AR	99.95	29 of 29

SMPD1	Niemann-Pick Disease	AR	99.98	258 of 258
TNFRSF11B	Paget Disease Of Bone, Calcium Pyrophosphate Deposition	AR	99.98	16 of 16
TPP1	Ceroid Lipofuscinosis, Spinocerebellar Ataxia	AR	100	147 of 147
TPP2	Autoimmune Hemolytic Anemia-Autoimmune Thrombocytopenia-Primary Immunodeficiency Syndrome	-	99.84	11 of 11

*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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