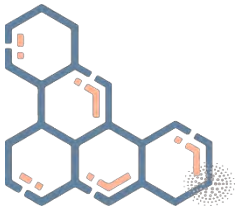
Fatty Acid Oxidation Disorders

Precision Panel



Overview

Fatty Acid Oxidation Disorders (FAODs) are inborn errors of metabolism resulting in failure of mitochondrial beta-oxidation or the carnitine-based transport of fatty acids into the mitochondria. Fatty acid oxidation takes place in the mitochondria and provides a major source of energy, especially during prolonged fasting and sub-maximal exercise. FAODs lead to deficient energy production and produce a wide range of clinical presentations ranging from mild hypotonia in adults to sudden death in infants and symptoms usually arise or exacerbate during catabolic situations, such as fasting, illness and exercise. The most common FAOD is medium-chain acyl-CoA dehydrogenase deficiency (MCADD). Typically, they are inherited in an autosomal recessive pattern.

The Igenomix Fatty Acid Oxidation Disorders 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 Fatty Acid Oxidation Disorders Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations during the newborn period:

- Hypoglycemia
- Hyperammonemia
- Liver disease and liver failure
- Cardiac and skeletal myopathy
- Rhabdomyolysis
- Retinal degeneration

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 emergency protocols and safe fasting times to prevent metabolic decompensation, dietary management, substrate (anaplerotic) therapy, maintenance of constant energy supply during times of catabolism
- 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**
ABCC8	Familial Hyperinsulinemic Hypoglycemia, Leucine-Sensitive Hypoglycemia Of Infancy, Autosomal Dominant Hyperinsulinism, Dend Syndrome	AD,AR	99.98	710 of 712
ABCD1	Adrenoleukodystrophy	X,XR,G	100	-
ACAD8	Isobutyryl-CoA Dehydrogenase Deficiency	AR	100	35 of 35
ACAD9	Acyl-CoA Dehydrogenase Deficiency	AR	100	62 of 62
ACADL	Long Chain Acyl-CoA Dehydrogenase Deficiency		100	1 of 1
ACADM	Medium Chain Acyl-CoA Dehydrogenase Deficiency	AR	99.98	181 of 181
ACADS	Short Chain Acyl-CoA Dehydrogenase Deficiency	AR	100	84 of 84
ACADSB	2-a Methylbutyryl-CoA Dehydrogenase Deficiency	AR	100	21 of 21
ACADVL	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	AR	100	329 of 329
ACBD5	Retinal Dystrophy With Leukodystrophy	AR	100	3 of 3
ALDH3A2	Sjogren-Larsson Syndrome	AR	96	119 of 119
ALDH5A1	Succinic Semialdehyde Dehydrogenase Deficiency	AR	95.41	65 of 69
COL7A1	Epidermolysis Bullosa Dystrophica	AD,AR	100	861 of 863
CPT1A	Carnitine Palmitoyltransferase I Deficiency	AR	100	50 of 50
CPT2	Carnitine Palmitoyltransferase II Deficiency	AD,AR	99.99	116 of 116
CTNS	Nephropathic Infantile Cystinosis	AR	100	148 of 153
DLD	Dihydrolipoamide Dehydrogenase Deficiency, Pyruvate Dehydrogenase E3 Deficiency	AR	100	26 of 26
ECHS1	Mitochondrial Short-Chain Enoyl-CoA Hydratase 1 Deficiency, Leigh Syndrome With Leukodystrophy	AR	100	39 of 39
ETFA	Multiple Acyl-CoA Dehydrogenase Deficiency	AR	92.33	32 of 32
ETFB	Multiple Acyl-CoA Dehydrogenase Deficiency	AR	100	21 of 21
ETFDH	Multiple Acyl-CoA Dehydrogenase Deficiency	AR	100	221 of 222
FA2H	Fatty Acid Hydroxylase-Associated Neurodegeneration	AR	88.77	60 of 62
GLUD1	Hyperinsulinism-Hyperammonemia Syndrome	AD	99.98	39 of 39
HADH	3-a Hydroxyacyl-CoA Dehydrogenase Deficiency	AR	96.71	26 of 27
HADHA	Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency, Trifunctional Protein Deficiency	AR	100	75 of 75
HADHB	Trifunctional Protein Deficiency	AR	99.99	66 of 68
HMGCL	3-a Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency, 3-Hydroxy-3-Methylglutaric Aciduria	AR	100	54 of 54
HMGCS2	3-Hydroxy-3-Methylglutaryl-CoA Synthase-2 Deficiency	AR	100	37 of 37
HNF1A	Insulin-Dependent Diabetes Mellitus	AD	100	529 of 538
HNF4A	Noninsulin-Dependent Diabetes Mellitus, Fanconi Renotubular Syndrome With Maturity-Onset Diabetes Of The Young, Hnf1b-Related Autosomal Dominant Tubulointerstitial Kidney Disease	AD	100	172 of 174
HSD17B10	Hydroxyacyl-CoA Dehydrogenase II Deficiency	X,XD,G	100	-
KCNJ1	Bartter Syndrome	AR	100	67 of 67
KCNJ11	Noninsulin-Dependent Diabetes Mellitus, Familial Hyperinsulinemic Hypoglycemia	AD,AR	100	190 of 191
LPIN1	Acute Recurrent Rhabdomyolysis	AR	99.98	31 of 31
LTC4S	Leukotriene C4 Synthase Deficiency	AR	99.69	4 of 4

MMP1	Epidermolysis Bullosa Dystrophica	AR	100	4 of 4
NADK2	2,4-a Dienoyl-CoA Reductase Deficiency, Progressive Encephalopathy With Leukodystrophy Due To Decr Deficiency	AR	95.37	3 of 3
PEX1	Peroxisome Biogenesis Disorder, Zellweger Syndrome, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	97.02	126 of 134
PEX10	Peroxisome Biogenesis Disorder (Zellweger), Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	99.76	29 of 32
PEX11B	Peroxisome Biogenesis Disorder, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy, Zellweger Syndrome	AR	90.29	7 of 7
PEX12	Peroxisome Biogenesis Disorder (Zellweger), Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	100	38 of 38
PEX13	Peroxisome Biogenesis Disorder (Zellweger), Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	99.98	11 of 12
PEX14	Peroxisome Biogenesis Disorder (Zellweger), Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	100	4 of 4
PEX16	Peroxisome Biogenesis Disorder (Zellweger), Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	100	17 of 17
PEX19	Peroxisome Biogenesis Disorder (Zellweger), Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	100	5 of 5
PEX2	Peroxisome Biogenesis Disorder (Zellweger), Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	99.89	17 of 17
PEX26	Peroxisome Biogenesis Disorder (Zellweger), Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	100	29 of 29
PEX3	Peroxisome Biogenesis Disorder (Zellweger), Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	100	9 of 9
PEX5	Infantile Refsum Disease, Neonatal Adrenoleukodystrophy, Zellweger Syndrome	AR	100	12 of 12
PEX6	Heimler Syndrome, Peroxisome Biogenesis Disorder (Zellweger), Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AD,AR	99.94	105 of 108
PEX7	Peroxisome Biogenesis Disorder, Refsum Disease	AR	99.21	47 of 53
PHYH	Refsum Disease	AR	100	34 of 34
PLA2G4A	Gastrointestinal Ulceration, Recurrent, With Dysfunctional Platelets	AR	100	4 of 4
PNPLA2	Neutral Lipid Storage Myopathy	AR	100	53 of 53
PPARG	Berardinelli-Seip Congenital Lipodystrophy	AD,AR,MU,P	99.94	53 of 53
SLC12A1	Bartter Syndrome	AR	99	90 of 95
SLC22A5	Systemic Primary Carnitine Deficiency	AR	100	161 of 162
SLC25A20	Carnitine-Acylcarnitine Translocase Deficiency	AR	100	39 of 39
SLC52A1	Riboflavin Deficiency	AD	99.91	2 of 2
TANGO2	Recurrent Metabolic Encephalomyopathic Crises-Rhabdomyolysis-Cardiac Arrhythmia-Intellectual Disability Syndrome	AR	99.94	15 of 15
TAZ	Barth Syndrome, Familial Isolated Dilated Cardiomyopathy	X,XR,G	100	-
TRMU	Mitochondrial Myopathy With Reversible Cytochrome C Oxidase Deficiency	AR,MI	100	25 of 25
TRNE	Maternally-Inherited Diabetes And Deafness , Mitochondrial Myopathy With Reversible Cytochrome C Oxidase Deficiency	-	-	-
UCP2	Hyperinsulinism	-	100	7 of 7

*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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