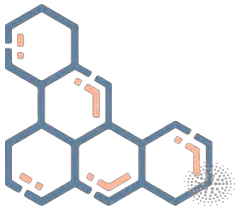
Urea Cycle Disorder

Precision Panel



Overview

The Urea Cycle is the metabolic pathway that transforms nitrogen from peripheral (muscle) and enteral sources (protein ingestion) into urea that is water soluble and can be excreted. Deficiency of an enzyme in the pathway causes a urea cycle disorder (UCD). Examples of UCDs include carabmyl phosphate synthetase I deficiency, ornithine transcarbamylase deficiency or arginase deficiency amongst others. All UCDs except for arginase deficiency, result in hyperammonaemia and life-threatening metabolic decompensations in infancy. If the patient survives, there is usually a severe neurologic injury. The inheritance of the UCDs is autosomal recessive except for ornithine transcarbamylase (OTC) deficiency which is X-linked.

The Igenomix Urea Cycle Disorder 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 Urea Cycle Disorder Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations during the newborn period:

- Decreased level of consciousness
- Altered mental status
- Abnormal motor function
- Seizures
- Vomiting
- Poor feeding
- Diarrhea
- Nausea
- Constipation
- Somnolence
- Inability to maintain body temperature
- Poor feeding
- Hyperammonemia and hyperventilation
- Lethargy
- Coma

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 in the form of rehydration and maintenance of good urine output without overhydration, remove nitrogen (ammonia) from the body with medications and/or haemodialysis, decrease protein intake and minimize catabolism, stimulate anabolism and uptake of nitrogen precursors by muscle.
- 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**
ACADM	Acyl-Coa Dehydrogenase Deficiency	AR	99.98	181 of 181
ACADS	Acyl-Coa Dehydrogenase Deficiency	AR	100	84 of 84
ACADVL	Acyl-Coa Dehydrogenase Deficiency	AR	100	329 of 329
ALDH18A1	Cutis Laxa, Mental Retardation, Spastic Paraplegia, De Barsy Syndrome	AD,AR	100	39 of 40
ARG1	Argininemia	AR	100	66 of 68
ASL	Argininosuccinic Aciduria	AR	100	170 of 170
ASS1	Citrullinemia	AR	100	150 of 153
BCKDHA	Maple Syrup Urine Disease	AR	98.41	96 of 97
BCKDHB	Maple Syrup Urine Disease	AR	99.99	122 of 123
CA5A	Carbonic Anhydrase VA Deficiency, Hyperammonemia	AR	100	5 of 6
CPS1	Carbamoyl Phosphate Synthetase I Deficiency, Hyperammonemia	AR	100	274 of 278
CPT1A	Carnitine Palmitoyltransferase I Deficiency	AR	100	50 of 50
CPT2	Carnitine Palmitoyltransferase Ii Deficiency, Encephalopathy	AD,AR	99.99	116 of 116
DBT	Maple Syrup Urine Disease	AR	100	73 of 75
DLD	Dihydrolipoamide Dehydrogenase Deficiency, Pyruvate Dehydrogenase E3 Deficiency	AR	100	26 of 26
ETFA	Acyl-Coa Dehydrogenase Deficiency	AR	92.33	32 of 32
ETFB	Acyl-Coa Dehydrogenase Deficiency	AR	100	21 of 21
ETFDH	Acyl-Coa Dehydrogenase Deficiency	AR	100	221 of 222
GLUD1	Hyperinsulinemic Hypoglycemia, Hyperammonemia	AD	99.98	39 of 39
GLUL	Glutamine Deficiency	AR	100	4 of 4
HADHA	3-Hydroxyacyl-Coa Dehydrogenase Deficiency, Trifunctional Protein Deficiency, 3-Hydroxyacyl-Coa Dehydrogenase Deficiency	AR	100	75 of 75
HADHB	Trifunctional Protein Deficiency	AR	99.99	66 of 68
HCFC1	Methylmalonic Acidemia, Homocysteinemia, Intellectual Disability	X,XR,G	99.81	-
HLCS	Holocarboxylase Synthetase Deficiency	AR	100	47 of 47
HMGCL	3-Hydroxy-3-Methylglutaric Aciduria	AR	100	54 of 54
HMGCS2	3-Hydroxy-3-Methylglutaryl-Coa Synthase-2 Deficiency , 3-Hydroxy-3-Methylglutaryl-Coa Synthase Deficiency	AR	100	37 of 37
IVD	Isovaleric Acidemia	AR	100	105 of 105

MCCC1	3-Methylcrotonyl-Coa Carboxylase 1 Deficiency	AR	100	110 of 111
MCCC2	3-Methylcrotonyl-Coa Carboxylase Deficiency	AR	99.98	120 of 120
MMAA	Methylmalonic Aciduria	AR	99.98	77 of 77
MMAB	Methylmalonic Aciduria	AR	99.52	43 of 43
MMACHC	Methylmalonic Aciduria, Homocystinuria	AR	99.97	105 of 105
MMADHC	Methylmalonic Aciduria, Homocystinuria	AR	99.63	20 of 20
MMUT	Methylmalonic Aciduria, Methylmalonyl-Coa Mutase Deficiency, Methylmalonic Acidemia	AR	99.97	-
NAGS	N-Acetylglutamate Synthase Deficiency, Hyperammonemia	AR	99.45	48 of 50
NBAS	Infantile Liver Failure Syndrome, Short Stature, Optic Nerve Atrophy, Pelger-Huet Anomaly	AR	99.98	60 of 61
OAT	Ornithine Aminotransferase Deficiency, Gyrate Atrophy Of Choroid And Retina	AR	100	72 of 73
OTC	Ornithine Transcarbamylase Deficiency, Hyperammonemia	X,XR,G	99.97	-
PC	Pyruvate Carboxylase Deficiency	AR	100	48 of 48
PCCA	Propionic Acidemia	AR	100	137 of 137
PCCB	Propionic Acidemia	AR	99.95	136 of 138
SLC22A5	Carnitine Deficiency	AR	100	161 of 162
SLC25A13	Citrullinemia, Neonatal Intrahepatic Cholestasis	AR	100	108 of 110
SLC25A15	Hyperornithinemia, Hyperammonemia, Homocitrullinuria	AR	100	41 of 41
SLC25A20	Carnitine-Acylcarnitine Translocase Deficiency	AR	100	39 of 39
SLC7A7	Lysinuric Protein Intolerance	AR	100	61 of 61
SUCLA2	Mitochondrial Dna Depletion Syndrome, Methylmalonic Aciduria	AR	100	27 of 27
SUCLG1	Mitochondrial Dna Depletion Syndrome, Methylmalonic Aciduria	AR	100	34 of 34
TMEM70	Mitochondrial Complex V Deficiency, Encephalocardiomyopathy	AR	100	22 of 24
UMPS	Orotic Aciduria	AR	100	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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