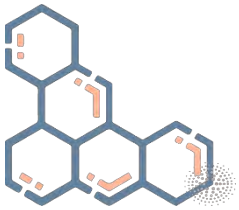
Organic Acidemias/Acidurias

Precision Panel



Overview

Organic Acidemias/Acidurias (Organic Acid Disorders, OADs) are an important group of inherited metabolic disorders that share a defect in intermediary metabolic pathways of carbohydrate, amino acids and fatty acid oxidation. These enzymatic defects lead to an accumulation of organic acids in tissues and their subsequent excretion in urine. As patients age, the natural progression of organic acidemias lead to intellectual difficulties, increased risk for neurologic complications such as stroke-like episodes and cardiac complications among others. All organic acidurias are inherited in an autosomal recessive pattern.

The Igenomix Organic Acidemias/Acidurias Precision Panel can be used to make an accurate and directed diagnosis as well as a differential diagnosis of hyperammonemia and high anion gap metabolic acidosis 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 Organic Acidemias/Acidurias Precision Panel is indicated for those patients with a clinical suspicion or diagnosis an organic acidemia with or without the following manifestations:

- Developmental delay
- Mental retardation
- Seizures
- Lethargy
- Coma
- Hypotonia
- Vomiting
- Failure to thrive
- Hepatomegaly
- Respiratory distress
- Cardiac dysfunction

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 of nutritional therapy, correction of fluid and electrolyte imbalances, adequate cerebral perfusion.
- 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**
ABCD4	Methylmalonic Aciduria And Homocystinuria	AR	100	8 of 8
ACAD8	Isobutyryl-CoA Dehydrogenase Deficiency	AR	100	35 of 35
ACAD9	Acyl-CoA Dehydrogenase Family	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	Shot-Chain Acyl-coa Dehydrogenase Deficiency	AR	100	84 of 84
ACADSB	2-Methylbutyryl-CoA Dehydrogenase Deficiency	AR	100	21 of 21
ACADVL	Very Long Chain Acyl-CoA Dehydrogenase Deficiency	AR	100	329 of 329
ACAT1	Alpha-Methylacetoacetic Aciduria, Beta-Ketothiolase Deficiency	AR	100	116 of 116
ACSF3	Combined Malonic And Methylmalonic Aciduria	AR	100	27 of 27
ADK	Hypermethioninemia Due To Adenosine Kinase Deficiency	AR	100	15 of 16
AGK	Congenital Cataract-Hypertrophic Cardiomyopathy-Mitochondrial Myopathy Syndrome	AR	99.98	33 of 33
AGXT	Primary Hyperoxaluria Type 1	AR	99.99	210 of 211
AHCY	Hypermethioninemia With S-Adenosylhomocysteine Hydrolase Deficiency	AR	100	11 of 11
ALDH4A1	Hyperprolinemia Type II	AR	100	7 of 7
ALDH5A1	Succinic Semialdehyde Dehydrogenase Deficiency	AR	95.41	65 of 69
ALDOB	Hereditary Fructose Intolerance	AR	100	61 of 65
AMN	Imerslund-Grasbeck Syndrome	AR	94.02	31 of 32
AMT	Glycine Encephalopathy	AR	99.98	94 of 96
ARG1	Argininemia	AR	100	66 of 68
ASL	Argininosuccinic Aciduria	AR	100	170 of 170
ASPA	Canavan Disease	AR	99.56	93 of 94
ASS1	Classic Citrullinemia	AR	100	150 of 153
ATAD3A	Harel-Yoon Syndrome, Pontocerebellar Hypoplasia, Hypotonia, And Respiratory Insufficiency Syndrome	AD,AR	90.98	6 of 8
ATP5F1D	Mitochondrial Complex V (ATP Synthase) Deficiency Nuclear Type 5	AR	98.19	NA of NA
ATP5F1E	Mitochondrial Complex V (ATP Synthase) Deficiency Nuclear Type 3	AR	100	NA of NA
ATP6	Leber Optic Atrophy, Neuropathy, Ataxia, And Retinitis Pigmentosa, Mitochondrial DNA-Associated Leigh Syndrome, Narp Syndrome	MI	na	na
ATP7B	Wilson Disease	AR	99.97	989 of 1000
ATPAF2	ATPase Deficiency Nuclear-Encoded	AR	100	2 of 2
AUH	3-Methylglutaconic Aciduria Type I	AR	99.99	11 of 11
BCKDHA	Maple Syrup Urine Disease	AR	98.41	96 of 97
BCKDHB	Maple Syrup Urine Disease	AR	99.99	122 of 123
BCS1L	Bjornstad Syndrome, Gracile Syndrome, Leigh Syndrome, Mitochondrial Complex III Deficiency	AR,MI	99.96	40 of 42
BTD	Biotinidase Deficiency Multiple Carboxylase Deficiency	AR	100	261 of 262
CAMKMT	2p21 Microdeletion Syndrome	-	99.99	NA of NA
CASR	Neonatal Severe Primary Hyperparathyroidism, Familial Hypocalciuric Hypercalcemia	AD,AR	100	445 of 446
CBS	Classic Homocystinuria	AR	99.98	192 of 194
CCN6	Progressive Pseudorheumatoid Arthropathy Of Childhood	AR	100	NA of NA
CD320	Methylmalonic Aciduria Due To Transcobalamin Receptor Defect	AR	89	2 of 2

CLCN5	Dent Disease, X-linked Recessive Hypophosphatemic Rickets, Nephrolithiasis With Renal Failure , Low Molecular Weight Proteinuria With Hypercalciuria And Nephrocalcinosis	X,XR,G	99.39	NA of NA
CLPB	3-Methylglutaconic Aciduria Type 7	AR	96	26 of 26
COA3	Mitochondrial Complex IV Deficiency	AR,MI	100	2 of 2
COA8	Mitochondrial Complex IV Deficiency, Non-Progressive Predominantly Posterior Cavitating Leukoencephalopathy With Peripheral Neuropathy	AR,MI	86.26	4 of 5
COX10	Leigh Syndrome, Mitochondrial Complex IV Deficiency	AR,MI	100	13 of 13
COX14	Mitochondrial Complex IV Deficiency	AR,MI	100	1 of 1
COX20	Mitochondrial Complex IV Deficiency	AR,MI	99.88	5 of 5
COX6B1	Mitochondrial Complex IV Deficiency	AR,MI	100	3 of 3
COX8A	Mitochondrial Complex IV Deficiency	AR,MI	100	1 of 1
CPS1	Carbamoyl Phosphate Synthetase I Deficiency	AR	100	274 of 278
CPT2	Infantile Carnitine Palmitoyltransferase II	AD,AR	99.99	116 of 116
CTH	Cystathioninuria	AR	100	8 of 8
CTNS	Cystinosis Adult Nonnephropathic, Nephropathic Infantile Cystinosis	AR	100	148 of 153
CUBN	Megaloblastic Anemia	AR	100	53 of 54
CYP27B1	Hypocalcemic Vitamin D-Dependent Rickets	AR	98.45	80 of 80
CYP2R1	Hypocalcemic Vitamin D-Dependent Rickets	AR	99.22	6 of 6
D2HGDH	D-2-Hydroxyglutaric Aciduria	AR	100	42 of 42
DBT	Maple Syrup Urine Disease	AR	100	73 of 75
DDB2	Xeroderma Pigmentosum Complementation Group E	AR	100	17 of 17
DGUOK	Progressive External Ophthalmoplegia With Mitochondrial DNA Deletions	AR	100	68 of 70
DHTKD1	2-Aminoadipic 2-Oxoadipic Aciduria, Charcot-Marie-Tooth Disease, Axonal Type 2q	AD,AR	99.94	25 of 25
DLD	Dihydrolipoamide Dehydrogenase Deficiency, Pyruvate Dehydrogenase E3 Deficiency	AR	100	26 of 26
DMGDH	Dimethylglycine Dehydrogenase Deficiency	AR	99.77	2 of 2
DNAJC19	3-Methylglutaconic Aciduria Type V, Dilated Cardiomyopathy With Ataxia	AR	100	6 of 6
EHHADH	Fanconi Renotubular Syndrome	AD	99.96	1 of 1
ERCC2	Cerebrooculofacioskeletal Syndrome, Xeroderma Pigmentosum Complementation Group D	AR	100	102 of 102
ERCC3	Photosensitive Trichothiodystrophy, Xeroderma Pigmentosum Complementation Group B	AR	99.98	24 of 24
ERCC4	Fanconi Anemia Complementation Group Q, Xeroderma Pigmentosum Complementation Group F, Cockayne Syndrome Type 1	AR	99.68	69 of 72
ERCC5	Cerebrooculofacioskeletal Syndrome, Xeroderma Pigmentosum, Complementation Group G	AR	99.94	58 of 58
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
ETHE1	Ethylmalonic Encephalopathy	AR	100	32 of 33
FAH	Tyrosinemia Type 1	AR	100	107 of 108
FARS2	Combined Oxidative Phosphorylation Deficiency	AR	99.98	23 of 23
FH	Fumarase Deficiency	AD,AR	100	229 of 232
FLAD1	Lipid Storage Myopathy Due To Flavin Adenine Dinucleotide Synthetase Deficiency	AR	97.13	13 of 14
FTCD	Formiminotransferase Deficiency	AR	99.83	17 of 17
GALE	Galactose Epimerase Deficiency	AR	100	29 of 29
GALT	Classic Galactosemia	AR	100	350 of 350
GATM	Cerebral Creatine Deficiency Syndrome, Fanconi Renotubular Syndrome	AD,AR	99.98	21 of 21
GCDH	Glutaric Acidemia, Glutaryl-CoA Dehydrogenase Deficiency	AR	88.74	254 of 254
GCLC	Gamma-Glutamyl-Cysteine Synthetase Deficiency	AR	99.97	7 of 7
GCSH	Glycine Encephalopathy	AR	93.52	1 of 1
GEMIN4	Neurodevelopmental Disorder With Microcephaly, Cataracts, And Renal Abnormalities	AR	99	2 of 2
GIF	Autosomal Recessive Intrinsic Factor Deficiency	-	100	NA of NA
GLDC	Glycine Encephalopathy	AR	98.69	359 of 367
GLYCTK	D-glyceric Aciduria	AR	100	6 of 6
GNMT	Glycine N-Methyltransferase Deficiency	AR	100	5 of 5
GPHN	Hyperekplexia, Hereditary , Molybdenum Cofactor Deficiency, Complementation Group C	AD,AR	99.2	6 of 6

GRHPR	Primary Hyperoxaluria Type 2	AR	88	51 of 51
GSS	Glutathione Synthetase Deficiency	AR	100	36 of 38
GUCY2D	Central Areolar Choroidal Dystrophy, Cone-Rod Dystrophy, Leber Congenital Amaurosis Type I, Congenital Stationary Night Blindness	AD,AR	99.98	248 of 248
HADH	3-Hydroxyacyl-CoA Dehydrogenase Deficiency	AR	96.71	26 of 27
HAL	Histidinemia	-	100	9 of 9
HCFC1	Methylmalonic Acidemia And Homocysteinemia	X,XR,G	99.81	NA of NA
HGD	Alkaptonuria	AR	100	158 of 159
HIBCH	3-Hydroxyisobutyryl-CoA Hydrolase Deficiency	AR	96.47	27 of 27
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	AR	100	37 of 37
HNF4A	Fanconi Renotubular Syndrome With Maturity-Onset Diabetes Of The Young, Hnf1b-Related Autosomal Dominant Tubulointerstitial Kidney Disease	AD	100	172 of 174
HOGA1	Primary Hyperoxaluria Type 3	AR	100	42 of 42
HPD	Hawkinsinuria, Tyrosinemia Type III	AD,AR	100	10 of 10
HTRA2	3-Methylglutaconic Aciduria Type VIII, Young-Onset Parkinson Disease	AD,AR	99.81	18 of 18
IDH2	D-2-Hydroxyglutaric Aciduria, Maffucci Syndrome, Ollier Disease	AD	99.99	4 of 4
IVD	Isovaleric Acidemia	AR	100	105 of 105
KIF1B	Charcot-Marie-Tooth Disease Axonal Type 2a1	AD	99.89	17 of 17
KYNU	Hydroxykynureninuria, Vertebral, Cardiac, Renal, And Limb Defects Syndrome	AR	99.98	7 of 7
L2HGDH	L-2-Hydroxyglutaric Aciduria	AR	100	72 of 73
LDHD	Lactic Aciduria Due To D-Lactic Acid	AR	100	3 of 3
LMBRD1	Methylmalonic Acidemia With Homocystinuria Type Cblf	AR	99.88	8 of 8
MARS1	Charcot-Marie-Tooth Disease Axonal Type 2u	AD,AR	99.98	19 of 19
MCCC1	3-Methylcrotonyl-CoA Carboxylase Deficiency	AR	100	110 of 111
MCCC2	3-Methylcrotonyl-CoA Carboxylase Deficiency	AR	99.98	120 of 120
MCEE	Methylmalonyl-CoA Epimerase Deficiency	AR	100	5 of 6
MICOS13	Combined Oxidative Phosphorylation Deficiency, 3-Methylglutaconic Aciduria Type 3	AR	na	na
MLYCD	Malonyl-CoA Decarboxylase Deficiency	AR	93.84	32 of 40
MMAA	Methylmalonic Aciduria Cbla Type	AR	99.98	77 of 77
MMAB	Methylmalonic Aciduria Cblb Type	AR	99.52	43 of 43
MMACHC	Methylmalonic Acidemia With Homocystinuria Type Cblc	AR	99.97	105 of 105
MMADHC	Methylmalonic Acidemia With Homocystinuria Type Cbld	AR	99.63	20 of 20
MMUT	Methylmalonic Aciduria Due To Methylmalonyl-CoA Mutase Deficiency	AR	99.97	NA of NA
MOCS1	Molybdenum Cofactor Deficiency Complementation Group A	AR	100	36 of 37
MOCS2	Molybdenum Cofactor Deficiency Complementation Group B	AR	100	32 of 32
MPC1	Mitochondrial Pyruvate Carrier Deficiency	AR	100	2 of 2
MRPS2	Combined Oxidative Phosphorylation Deficieny	AR	100	3 of 3
MTHFR	Homocystinuria Due To Deficiency Of N(5,10)-Methylene Tetrahydrofolate Reductase Activity	AD,AR	100	122 of 122
MTR	Methylcobalamin Deficiency	AR	99.94	42 of 45
MTRR	Homocystinuria-Megaloblastic Anemia Due To Defect In Cobalamin Metabolism CBLE Complementation Type	AR	100	39 of 40
MVK	Hyper-IgD Syndrome, Mevalonic Aciduria	AD,AR	100	180 of 181
NADK2	2,4-Dienoyl-CoA Reductase Deficiency, Progressive Encephalopathy With Leukodystrophy Due To Decr Deficiency	AR	95.37	3 of 3
NAGA	Kanzaki Disease, Schindler Disease Type I, Alpha-n-Acetylgalactosaminidase Deficiency Type 1 , Alpha-n-Acetylgalactosaminidase Deficiency Type 2, Alpha-n-Acetylgalactosaminidase Deficiency Type 3	AR	100	12 of 12
ND1	Leber Optic Atrophy, Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes, Isolated Complex I Deficiency, Mitochondrial DNA-Associated Leigh Syndrome	MI	na	na
ND2	Leber Optic Atrophy, Isolated Complex I Deficiency, Leber Hereditary Optic Neuropathy, Mitochondrial DNA-Associated Leigh Syndrome	MI	85.56	NA of NA
ND3	Isolated Complex I Deficiency, Mitochondrial DNA-Associated Leigh Syndrome	-	99.99	NA of NA

ND4	Leber Optic Atrophy, Leber Hereditary Optic Neuropathy, Mitochondrial DNA-Associated Leigh Syndrome	MI	na	na
ND5	Leber Optic Atrophy, Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes, Mitochondrial DNA-Associated Leigh Syndrome	MI	99.89	NA of NA
ND6	Leber Optic Atrophy, Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes, Mitochondrial DNA-Associated Leigh Syndrome	MI	100	NA of NA
NDUFAF6	Fanconi Renotubular Syndrome, Mitochondrial Complex I Deficiency Nuclear Type 17, Leigh Syndrome With Leukodystrophy	AR	99.4	12 of 13
NEU1	Neuraminidase Deficiency, Congenital Sialidosis Type 2, Sialidosis Type 1	AR	100	68 of 68
NGLY1	Congenital Disorder Of Glycosylation, Type IV, Alacrimia-Choreoathetosis-Liver Dysfunction Syndrome	AR	99.8	28 of 28
OAT	Ornithine Aminotransferase Deficiency, Gyrate Atrophy Of Choroid And Retina	AR	100	72 of 73
OCRL	Dent Disease, Lowe Oculocerebrorenal Syndrome	X,XR,G	100	NA of NA
OGDH	Alpha-Ketoglutarate Dehydrogenase Deficiency, Oxoglutaric Aciduria	AR	100	1 of 1
OPA3	3-Methylglutaconic Aciduria Type III, Autosomal Dominant Optic Atrophy And Cataract	AD,AR	100	18 of 18
OPLAH	5-Oxoprolinase Deficiency	AD,AR	99.98	29 of 30
OTC	Ornithine Transcarbamylase Deficiency	X,XR,G	99.97	NA of NA
OXCT1	Succinyl CoA-3-Oxoacid CoA Transferase Deficiency ,	AR	100	33 of 33
PCCA	Propionic Acidemia	AR	100	137 of 137
PCCB	Propionic Acidemia	AR	99.95	136 of 138
PEPD	Prolidase Deficiency	AR	95	34 of 34
PET100	Mitochondrial Complex IV Deficiency, Leigh Syndrome With Leukodystrophy	AR,MI	98	2 of 2
PEX1	Peroxisome Biogenesis Disorder, Zellweger Syndrome, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	97.02	126 of 134
PEX5	Neonatal Adrenoleukodystrophy, Cerebrohepatorenal Syndrome, Infantile Refsum Disease, Zellweger Syndrome	AR	100	12 of 12
PLOD2	Bruck Syndrome	AR	99.97	29 of 29
POLG	Mitochondrial Dna Depletion Syndrome, Sensory Ataxic Neuropathy, Dysarthria, And Ophthalmoparesis, Alpers-Huttenlocher Syndrome	AD,AR	99.92	325 of 326
PPM1B	2p21 Microdeletion Syndrome	-	99.61	1 of 1
PRDX1	Methylmalonic Aciduria And Homocystinuria Cblc Type	AR	100	3 of 3
PREPL	Congenital Myasthenic Syndrome, 2p21 Microdeletion Syndrome, Hypotonia-Cystinuria Syndrome	AR	99.92	7 of 12
PRODH	Hyperprolinemia Type 1	AD,AR	98.57	5 of 5
RET	Multiple Endocrine Neoplasia Type IIa, Multiple Endocrine Neoplasia Type IIb, Haddad Syndrome, Bilateral Renal Agenesis	AD	100	453 of 454
RRM2B	Mitochondrial DNA Depletion Syndrome 8a (Encephalomyopathic Type Withrenal Tubulopathy), Kearns-Sayre Syndrome	AD,AR	92.38	46 of 46
SARDH	Sarcosinemia		98.61	7 of 7
SCO1	Mitochondrial Complex IV Deficiency	AR,MI	100	6 of 6
SERAC1	3-Methylglutaconic Aciduria With Deafness, Encephalopathy, And Leigh-like Syndrome	AR	99.93	53 of 53
SLC19A2	Thiamine-Responsive Megaloblastic Anemia Syndrome	AR	99.99	67 of 68
SLC1A1	Dicarboxylicamino Aciduria	AR	100	7 of 7
SLC25A1	Combined D-2- And L-2-Hydroxyglutaric Aciduria	AR	90	23 of 25
SLC25A15	Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	AR	100	41 of 41
SLC25A19	Thiamine Metabolism Dysfunction Syndrome (Bilateral Striatal Degenerationand Progressive Polyneuropathy Type)	AR	97.13	10 of 10
SLC25A20	Carnitine-Acylcarnitine Translocase Deficiency	AR	100	39 of 39
SLC25A21	Mitochondrial DNA Depletion Syndrome	AR	99.95	1 of 1
SLC25A4	Mitochondrial DNA Depletion Syndrome (Cardiomyopathic Type)	AD,AR	99.84	16 of 16
SLC26A1	Calcium Oxalate Nephrolithiasis	AR	99.94	4 of 4
SLC2A2	Fanconi-Bickel Syndrome	AD,AR	100	75 of 79
SLC34A1	Fanconi Renotubular Syndrome, Hypophosphatemic Nephrolithiasis/Osteoporosis	AD,AR	100	39 of 39

SLC35A1	Congenital Disorder Of Glycosylation Type IIf	AR	100	6 of 6
SLC36A2	Glycinuria With Or Without Oxalate Urolithiasis, Iminoglycinuria	AD,AR	100	1 of 1
SLC3A1	Hypotonia-Cystinuria Syndrome	AD,AR	100	202 of 202
SLC52A1	Riboflavin Deficiency	AD	99.91	2 of 2
SLC52A2	Brown-Vialetto-Van Laere Syndrome, Autosomal Recessive Spinocerebellar Ataxia-blindness-deafness Syndrome	AR	100	31 of 32
SLC6A18	Iminoglycinuria	-	100	NA of NA
SLC6A19	Glycinuria With Or Without Oxalate Urolithiasis, Hartnup Disorder, Iminoglycinuria	AD,AR	100	25 of 25
SLC6A20	Glycinuria With Or Without Oxalate Urolithiasis, Iminoglycinuria	AD,AR	100	1 of 1
SLC7A7	Lysinuric Protein Intolerance	AR	100	61 of 61
SLC7A9	Cystinuria	AD,AR	100	144 of 144
SPINK5	Netherton Syndrome	AR	99.98	84 of 84
SUCLA2	Mitochondrial DNA Depletion Syndrome (Encephalomyopathic With Or Without Methylmalonic Aciduria)	AR	100	27 of 27
SUCLG1	Mitochondrial Dna Depletion Syndrome (Encephalomyopathic Type With Methylmalonic Aciduria)	AR	100	34 of 34
SUGCT	Glutaric Aciduria III	AR	99.85	7 of 7
TACO1	Mitochondrial Complex IV Deficiency, Leigh Syndrome With Leukodystrophy	AR,MI	100	3 of 3
TAT	Tyrosine Transaminase Deficiency, Tyrosinemia Type 2	AR	100	34 of 34
TAZ	Barth Syndrome	X,XR,G	100	NA of NA
TCN2	Transcobalamin Deficiency	AR	100	25 of 27
TIMM50	3-Methylglutaconic Aciduria Type IX	AR	91	7 of 7
TK2	Mitochondrial DNA Depletion Syndrome Myopathic Form, Progressive External Ophthalmoplegia With Mitochondrial Dna Deletions	AR	97.08	64 of 65
TMEM70	Mitochondrial Complex V (ATP Synthase) Deficiency Nuclear Type 2	AR	100	22 of 24
TNFRSF11A	Autosomal Recessive Osteopetrosis, Paget Disease Of Bone, Polyostotic Osteolytic Dysplasia	AD,AR	96.37	17 of 22
TNFRSF11B	Familial Calcium Pyrophosphate Deposition, Juvenile Paget Disease	AR	99.98	16 of 16
TRNK	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-lik Episodes, Myoclonic Epilepsy Associated With Ragged-Red Fibers, Mitochondrial DNA-Associated Leigh Syndrome	MI	na	na
TRNL1	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-lik Episodes, Myoclonic Epilepsy Associated With Ragged-Red Fibers, Kearns-Sayre Syndrome, Mitochondrial DNA-Associated Leigh Syndrome	MI	na	na
TRNN	Mitochondrial Complex IV Deficiency, Mitochondrial DNA-Related Progressive External Ophthalmoplegia	AR,MI	na	na
TRNS1	Deafness, Aminoglycoside-Induced, Mitochondrial Complex IV Deficiency, Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes	AR,MI	na	na
TRNT1	Retinitis Pigmentosa And Erythrocytic Microcytosis, Sideroblastic Anemia With B-Cell Immunodeficiency, Periodic Fevers,and Developmental Delay	AR	99.47	22 of 27
TRNV	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes, Mitochondrial DNA-Associated Leigh Syndrome	MI	na	na
TRNW	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes, Mitochondrial Myopathy, Episodic, With Optic Atrophy And Reversible Leukoencephalopathy, Mitochondrial DNA-Associated Leigh Syndrome	AR,MI	na	na
UMPS	Hereditary Orotic Aciduria	AR	100	11 of 11
UROC1	Urocanic Aciduria	AR	100	5 of 5
XPA	Xeroderma Pigmentosum Complementation Group A	AR	99.91	49 of 49
XPC	Xeroderma Pigmentosum Complementation Group C	AR	99.83	86 of 87

*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.

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