MOx Metabólico

Desarrollado específicamente para pacientes con sospecha de un trastorno metabólico o que presentan síntomas complejos superpuestos, una crisis metabólica o afecciones neurológicas de etiología desconocida. Proporciona tiempos de respuesta cortos, y está dirigido a pacientes en estado crítico en la UCIN/UCIP. Aprovecha un enfoque multiómico al incluir pruebas de actividad enzimática cuando corresponda, así como una selección patentada de biomarcadores que se actualiza continuamente.

Nº de genes:	206
Entrega:	25 días
Cobertura:	≥99,5% ≥20x Cobertura media con profundidad ≥150 x
Detalles:	Análisis de CNV incluido Pruebas bioquímicas complementarias mediante biomarcadores patentados y ensayos de actividad enzimática, si corresponde

SÍNDROMES Y TRASTORNOS COMUNES CUBIERTOS

Trastornos metabólicos más comunes

Resumen de genes y enfermedades asociadas (OMIM) incluidos en este panel:

Genes	OMIM (gen)	Enfermedades asociadas (OMIM)	Herencia
ABCA1	600046	HDL deficiency, familial, 1;Tangier disease	AD, AR
ABCB4	171060	Gallbladder disease 1;Cholestasis, progressive familial intrahepatic 3;Cholestasis, intrahepatic, of pregnancy, 3	AD, AR, AR
ABCC2	601107	Dubin-Johnson syndrome	AR
ABCD1	300371	Adrenoleukodystrophy;Adrenomyeloneuropathy, adult	XLR

ABCD4	603214	Methylmalonic aciduria and homocystinuria, cblJ type	AR
ABCG5	605459	Sitosterolemia 2	AR
ABCG8	605460	Gallbladder disease 4;Sitosterolemia 1	AR
ACAT1	607809	Alpha-methylacetoacetic aciduria	AR
ADA	608958	Adenosine deaminase deficiency, partial;Severe combined immunodeficiency due to ADA deficiency	AR, SM
AGA	613228	Aspartylglucosaminuria	AR
AGL	610860	Glycogen storage disease IIIb;Glycogen storage disease IIIa	AR
AGPS	603051	Rhizomelic chondrodysplasia punctata, type 3	AR
AGXT	604285	Hyperoxaluria, primary, type 1	AR
ALAD	125270	Porphyria, acute hepatic;Lead poisoning, susceptibility to	AR
ALAS2	301300	Protoporphyria, erythropoietic, XL;Anemia, sideroblastic, 1	XL, XLR
ALDH4A1	606811	Hyperprolinemia, type II	AR
ALDOA	103850	Glycogen storage disease XII	AR
ALDOB	612724	Fructose intolerance, hereditary	AR
ALG3	608750	Congenital disorder of glycosylation, type Id	AR
ALPL	171760	Hypophosphatasia, infantile;Odontohypophosphatasia;Hypophosphatasia, childhood;Hypophosphatasia, adult	AR, AD, AR
ANTXR2	608041	Hyaline fibromatosis syndrome	AR
APOA2	107670	Hypercholesterolemia, familial, modifier of	AD, AR

APOA5	606368	Hyperchylomicronemia, late-onset;Hypertriglyceridemia, susceptibility to	AD
APOB	107730	Hypobetalipoproteinemia;Hypercholesterolemia, familial, 2	AR, AD
APOC2	608083	Hyperlipoproteinemia, type Ib	AR
APOE	107741	Sea-blue histiocyte disease;Lipoprotein glomerulopathy;?Alzheimer disease, protection against, due to APOE3-Christchurch;Hyperlipoproteinemia, type III;Coronary artery disease, severe, susceptibility to;?Macular degeneration, age-related;Alzheimer disease 2	AR, AD
ARG1	608313	Argininemia	AR
ARSA	607574	Metachromatic leukodystrophy	AR
ARSB	611542	Mucopolysaccharidosis type VI (Maroteaux-Lamy)	AR
ASAH1	613468	Spinal muscular atrophy with progressive myoclonic epilepsy;Farber lipogranulomatosis	AR
ASL	608310	Argininosuccinic aciduria	AR
ASS1	603470	Citrullinemia	AR
ATP7A	300011	Menkes disease;Occipital horn syndrome;Spinal muscular atrophy, distal, XL 3	XLR
ATP7B	606882	Wilson disease	AR
BCKDHA	608348	Maple syrup urine disease, type Ia	AR
BCKDHB	248611	Maple syrup urine disease, type Ib	AR
BTD	609019	Biotinidase deficiency	AR
CBS	613381	Thrombosis, hyperhomocysteinemic;Homocystinuria, B6-responsive and nonresponsive types	AR

CD320	606475	Methylmalonic aciduria, transient, due to transcobalamin receptor defect	
CETP	118470	Hyperalphalipoproteinemia;[High density lipoprotein cholesterol level QTL 10]	AD
CLN3	607042	Ceroid lipofuscinosis, neuronal, 3	AR
CLN5	608102	Ceroid lipofuscinosis, neuronal, 5	AR
CLN6	606725	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset;Ceroid lipofuscinosis, neuronal, 6	AR
CLN8	607837	Ceroid lipofuscinosis, neuronal, 8;Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant	AR
CPOX	612732	Harderoporphyria;Coproporphyria	AR, AD, AR
CPS1	608307	Carbamoylphosphate synthetase I deficiency;Pulmonary hypertension, neonatal, susceptibility to	AR
CPT1A	600528	CPT deficiency, hepatic, type IA	AR
CTNS	606272	Cystinosis, late-onset juvenile or adolescent nephropathic;Cystinosis, ocular nonnephropathic;Cystinosis, nephropathic;Cystinosis, atypical nephropathic	AR
CTSA	613111	Galactosialidosis	AR
CTSD	116840	Ceroid lipofuscinosis, neuronal, 10	AR
CTSK	601105	Pycnodysostosis	AR
CYP11B1	610613	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency;Aldosteronism, glucocorticoid-remediable	AR, AD

CYP17A1	609300	17,20-lyase deficiency, isolated;17-alpha-hydroxylase/17,20-lyase deficiency	AR
CYP19A1	107910	Aromatase excess syndrome;Aromatase deficiency	AD
CYP21A2	613815	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency;Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency	AR
DBT	248610	Maple syrup urine disease, type II	AR
DDC	107930	Aromatic L-amino acid decarboxylase deficiency	AR
DHCR7	602858	Smith-Lemli-Opitz syndrome	AR
DIABLO	605219	Deafness, AD 64	AD
DLX4	601911	?Orofacial cleft 15	AD
DNAJC5	611203	Ceroid lipofuscinosis, neuronal, 4, Parry type	AD
DPYD	612779	5-fluorouracil toxicity;Dihydropyrimidine dehydrogenase deficiency	AR
ENO3	131370	?Glycogen storage disease XIII	AR
ENPP1	173335	Arterial calcification, generalized, of infancy, 1;Obesity, susceptibility to;Hypophosphatemic rickets, AR, 2;Cole disease;Diabetes mellitus, non-insulin-dependent, susceptibility to	AR, AD, AR, mi, AD
EPHX2	132811	Hypercholesterolemia, familial, due to LDLR defect, modifier of	AD, AR
ETHE1	608451	Ethylmalonic encephalopathy	AR
FAH	613871	Tyrosinemia, type I	AR
FBP1	611570	Fructose-1,6-bisphosphatase deficiency	AR

FECH	612386	Protoporphyria, erythropoietic, 1	AR
FGF23	605380	Tumoral calcinosis, hyperphosphatemic, familial, 2;Hypophosphatemic rickets, AD	AR, AD
FUCA1	612280	Fucosidosis	AR
G6PC1	613742	Glycogen storage disease Ia	AR
G6PD	305900	Resistance to malaria due to G6PD deficiency;Hemolytic anemia, G6PD deficient (favism)	XLD
GAA	606800	Glycogen storage disease II	AR
GALC	606890	Krabbe disease	AR
GALE	606953	Galactose epimerase deficiency	AR
GALK1	604313	Galactokinase deficiency with cataracts	AR
GALNS	612222	Mucopolysaccharidosis IVA	AR
GALT	606999	Galactosemia	AR
GAMT	601240	Cerebral creatine deficiency syndrome 2	AR
GATM	602360	Cerebral creatine deficiency syndrome 3;Fanconi renotubular syndrome 1	AR, AD
GBA	606463	Lewy body dementia, susceptibility to;Gaucher disease, type IIIC;Parkinson disease, late-onset, susceptibility to;Gaucher disease, type II;Gaucher disease, type III;Gaucher disease, perinatal lethal;Gaucher disease, type I	AD, AR, AD, mi
GBE1	607839	Glycogen storage disease IV;Polyglucosan body disease, adult form	AR

GHR	600946	Hypercholesterolemia, familial, modifier of;Laron dwarfism;Growth hormone insensitivity, partial;Increased responsiveness to growth hormone	AD, AR, AR, AD
GK	300474	Glycerol kinase deficiency	XLR
GLA	300644	Fabry disease;Fabry disease, cardiac variant	XL
GLB1	611458	GM1-gangliosidosis, type II;GM1-gangliosidosis, type I;Mucopolysaccharidosis type IVB (Morquio);GM1-gangliosidosis, type III	AR
GM2A	613109	GM2-gangliosidosis, AB variant	AR
GNPAT	602744	Rhizomelic chondrodysplasia punctata, type 2	AR
GNPTAB	607840	Mucolipidosis III alpha/beta;Mucolipidosis II alpha/beta	AR
GNPTG	607838	Mucolipidosis III gamma	AR
GNS	607664	Mucopolysaccharidosis type IIID	AR
GUSB	611499	Mucopolysaccharidosis VII	AR
GYG1	603942	Polyglucosan body myopathy 2;?Glycogen storage disease XV	AR
GYS1	138570	Glycogen storage disease 0, muscle	AR
GYS2	138571	Glycogen storage disease 0, liver	AR
HCFC1	300019	Mental retardation, XL 3 (methylmalonic acidemia and homocysteinemia, cblX type)	XLR
HEXA	606869	Tay-Sachs disease;[Hex A pseudodeficiency];GM2-gangliosidosis, several forms	AR
HEXB	606873	Sandhoff disease, infantile, juvenile, and adult forms	AR

HFE	613609	Porphyria variegata, susceptibility to;Alzheimer disease, susceptibility to;Hemochromatosis;[Transferrin serum level QTL2];Porphyria cutanea tarda, susceptibility to;Microvascular complications of diabetes 7	AD, AR, AD, AR
HJV	608374	Hemochromatosis, type 2A	AR
HGD	607474	Alkaptonuria	AR
HGSNAT	610453	Mucopolysaccharidosis type IIIC (Sanfilippo C);Retinitis pigmentosa 73	AR
HLCS	609018	Holocarboxylase synthetase deficiency	AR
HMBS	609806	Porphyria, acute intermittent;Porphyria, acute intermittent, nonerythroid variant	AD
HPD	609695	Hawkinsinuria;Tyrosinemia, type III	AD, AR
HPRT1	308000	Hyperuricemia, HRPT-related;Lesch-Nyhan syndrome	XLR
HSD3B2	613890	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	AR
HYAL1	607071	?Mucopolysaccharidosis type IX	AR
IDS	300823	Mucopolysaccharidosis II	XLR
IDUA	252800	Mucopolysaccharidosis Is;Mucopolysaccharidosis Ih/s;Mucopolysaccharidosis Ih	AR
ITIH4	600564		
IVD	607036	Isovaleric acidemia	AR
KHK	614058	[Fructosuria]	AR
LAMP2	309060	Danon disease	XLD
LCAT	606967	Norum disease;Fish-eye disease	AR
LDHA	150000	Glycogen storage disease XI	AR

LDLR	606945	LDL cholesterol level QTL2;Hypercholesterolemia, familial, 1	AD, AR
LDLRAP1	605747	Hypercholesterolemia, familial, 4	AR
LIPA	613497	Wolman disease;Cholesteryl ester storage disease	AR
LIPC	151670	[High density lipoprotein cholesterol level QTL 12];Hepatic lipase deficiency;Diabetes mellitus, noninsulin-dependent	AR, AD
LIPI	609252		
LMBRD1	612625	Methylmalonic aciduria and homocystinuria, cblF type	AR
LPA	152200	Coronary artery disease, susceptibility to;[LPA deficiency, congenital]	AD
LPL	609708	[High density lipoprotein cholesterol level QTL 11];Combined hyperlipidemia, familial;Lipoprotein lipase deficiency	AR, AD
MAN2B1	609458	Mannosidosis, alpha-, types I and II	AR
MANBA	609489	Mannosidosis, beta	AR
MCOLN1	605248	Mucolipidosis IV	AR
MFSD8	611124	Ceroid lipofuscinosis, neuronal, 7;Macular dystrophy with central cone involvement	AR
MMAA	607481	Methylmalonic aciduria, vitamin B12-responsive	AR
MMAB	607568	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type	AR
MMACHC	609831	Methylmalonic aciduria and homocystinuria, cblC type	AR

MMADHC	611935	Methylmalonic aciduria and homocystinuria, cblD type;Methylmalonic aciduria, cblD type, variant 2;Homocystinuria, cblD type, variant 1	AR
MMUT	609058	Methylmalonic aciduria, mut(0) type	AR
NAGA	104170	Schindler disease, type III;Kanzaki disease;Schindler disease, type I	AR
NAGLU	609701	?Charcot-Marie-Tooth disease, axonal, type 2V;Mucopolysaccharidosis type IIIB (Sanfilippo B)	AD, AR
NAGS	608300	N-acetylglutamate synthase deficiency	AR
NEU1	608272	Sialidosis, type I;Sialidosis, type II	AR
NPC1	607623	Niemann-Pick disease, type C1;Niemann-Pick disease, type D	AR
NPC2	601015	Niemann-pick disease, type C2	AR
OTC	300461	Ornithine transcarbamylase deficiency	XL
PAH	612349	Phenylketonuria;[Hyperphenylalaninemia, non-PKU mild]	AR
PCSK9	607786	Hypercholesterolemia, familial, 3;Low density lipoprotein cholesterol level QTL 1	AD
PDHB	179060	Pyruvate dehydrogenase E1-beta deficiency	AR
PEX1	602136	Peroxisome biogenesis disorder 1A (Zellweger);Peroxisome biogenesis disorder 1B (NALD/IRD);Heimler syndrome 1	AR
PEX10	602859	Peroxisome biogenesis disorder 6A (Zellweger);Peroxisome biogenesis disorder 6B	AR

PEX12	601758	Peroxisome biogenesis disorder 3B;Peroxisome biogenesis disorder 3A (Zellweger)	AR
PEX13	601789	Peroxisome biogenesis disorder 11B;Peroxisome biogenesis disorder 11A (Zellweger)	AR
PEX14	601791	Peroxisome biogenesis disorder 13A (Zellweger)	AR
PEX16	603360	Peroxisome biogenesis disorder 8A (Zellweger);Peroxisome biogenesis disorder 8B	AR
PEX19	600279	Peroxisome biogenesis disorder 12A (Zellweger)	AR
PEX2	170993	Peroxisome biogenesis disorder 5B;Peroxisome biogenesis disorder 5A (Zellweger)	AR
PEX26	608666	Peroxisome biogenesis disorder 7A (Zellweger);Peroxisome biogenesis disorder 7B	AR
PEX3	603164	Peroxisome biogenesis disorder 10A (Zellweger);?Peroxisome biogenesis disorder 10B	AR
PEX5	600414	Rhizomelic chondrodysplasia punctata, type 5;Peroxisome biogenesis disorder 2B;Peroxisome biogenesis disorder 2A (Zellweger)	AR
PEX6	601498	Peroxisome biogenesis disorder 4B;Peroxisome biogenesis disorder 4A (Zellweger);Heimler syndrome 2	AD, AR, AR
PEX7	601757	Peroxisome biogenesis disorder 9B;Rhizomelic chondrodysplasia punctata, type 1	AR
PFKM	610681	Glycogen storage disease VII	AR
PGAM2	612931	Glycogen storage disease X	AR

PGK1	311800	Phosphoglycerate kinase 1 deficiency	XLR
PGM1	171900	Congenital disorder of glycosylation, type It	AR
PHKA1	311870	Muscle glycogenosis	XLR
PHKA2	300798	Glycogen storage disease, type IXa1;Glycogen storage disease, type IXa2	XLR
PHKB	172490	Phosphorylase kinase deficiency of liver and muscle, AR	AR
PHKG2	172471	Glycogen storage disease IXc	AR
PKLR	609712	Adenosine triphosphate, elevated, of erythrocytes;Pyruvate kinase deficiency	AD, AR
PNPO	603287	Pyridoxamine 5'-phosphate oxidase deficiency	AR
POR	124015	Disordered steroidogenesis due to cytochrome P450 oxidoreductase;Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	AR
PPOX	600923	Porphyria variegata	AD
PPP1R17	604088	Hypercholesterolemia, susceptibility to	AD, AR
PPT1	600722	Ceroid lipofuscinosis, neuronal, 1	AR
PRKAG2	602743	Glycogen storage disease of heart, lethal congenital;Cardiomyopathy, hypertrophic 6;Wolff-Parkinson-White syndrome	AD

PSAP	176801	Combined SAP deficiency;Gaucher disease, atypical;Krabbe disease, atypical;Parkinson disease 24, AD, susceptibility to;Metachromatic leukodystrophy due to SAP-b deficiency	AR, AD
PYGL	613741	Glycogen storage disease VI	AR
PYGM	608455	McArdle disease	AR
RBCK1	610924	Polyglucosan body myopathy 1 with or without immunodeficiency	AR
SGSH	605270	Mucopolysaccharidosis type IIIA (Sanfilippo A)	AR
SI	609845	Sucrase-isomaltase deficiency, congenital	AR
SLC17A5	604322	Sialic acid storage disorder, infantile;Salla disease	AR
SLC22A5	603377	Carnitine deficiency, systemic primary	AR
SLC25A13	603859	Citrullinemia, type II, neonatal-onset;Citrullinemia, adult-onset type II	AR
SLC25A15	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	AR
SLC25A20	613698	Carnitine-acylcarnitine translocase deficiency	AR
SLC25A36	616149		
SLC2A1	138140	GLUT1 deficiency syndrome 2, childhood onset;GLUT1 deficiency syndrome 1, infantile onset, severe;Epilepsy, idiopathic generalized, susceptibility to, 12;Stomatin-deficient cryohydrocytosis with neurologic defects;Dystonia 9	AD, AD, AR

SLC2A2	138160	Fanconi-Bickel syndrome;Diabetes mellitus, noninsulin-dependent	AR, AD
SLC2A3	138170		
SLC37A4	602671	Glycogen storage disease Ib;Congenital disorder of glycosylation, type IIw;Glycogen storage disease Ic	AR, AD
SLC3A1	104614	Cystinuria	AD, AR
SLC3A2	158070		
SLC40A1	604653	Hemochromatosis, type 4	AD
SLC6A19	608893	Iminoglycinuria, digenic;Hyperglycinuria;Hartnup disorder	AR, DR, AD, AR
SLC6A8	300036	Cerebral creatine deficiency syndrome 1	XLR
SLC7A7	603593	Lysinuric protein intolerance	AR
SLC7A9	604144	Cystinuria	AD, AR
SLCO1B1	604843	Hyperbilirubinemia, Rotor type, digenic	DR
SLCO1B3	605495	Hyperbilirubinemia, Rotor type, digenic	DR
SMPD1	607608	Niemann-Pick disease, type A;Niemann-Pick disease, type B	AR
SUMF1	607939	Multiple sulfatase deficiency	AR
TAT	613018	Tyrosinemia, type II	AR
TFR2	604720	Hemochromatosis, type 3	AR
TPP1	607998	Ceroid lipofuscinosis, neuronal, 2;Spinocerebellar ataxia, AR 7	AR
UGT1A1	191740	[Gilbert syndrome];Crigler-Najjar syndrome, type II;Crigler-Najjar syndrome, type I;Hyperbilirubinemia, familial transient neonatal;[Bilirubin, serum level of, QTL1]	AR
UMPS	613891	Orotic aciduria	AR
UROD	613521	Porphyria, hepatoerythropoietic;Porphyria cutanea tarda	AD, AR

UROS	606938	Porphyria, congenital erythropoietic	AR