qCarrier Test · Disease list

qgenomics
Genomics for Human Health

El qCarrier Test es un test de laboratorio desarrollado por qGenomics, en colaboración con Dexeus, que emplea tecnología de **secuenciación masiva** (next generation sequencing - NGS) para la detección de +4000 mutaciones descritas en las bases de datos como causa de **+200 enfermedades recesivas**. El qCarrier combina el análisis dirigido de mutaciones puntuales prevalentes (cuando estas existen) en ciertos genes (138, de los cuales, 17 en el cromosoma X); con el barrido de regiones codificantes completas (72 genes, 2 en el X). El NGS también nos permite abordar el estudio de distintos tipos de mutaciones: de nucleótido único, pequeñas inserciones/deleciones, variantes de número de copia y ciertos reordenamientos

Los siguientes listados no tienen ningún valor contractual, y la información en ellos contenida es para uso EXCLUSIVO de profesionales sanitarios con formación específica en el área de la genética humana.

High impact

	CONDITION	OMIM	GENE	
●	3-Methylcrotonyl-CoA carboxylase 1 deficiency	210200	MCCC1	
●	3-Methylcrotonyl-CoA carboxylase 2 deficiency	210210	MCCC2	
O	Abetalipoproteinemia	200100	MTTP	
●	Acyl-CoA dehydrogenase, medium chain, deficiency of	201450	ACADM	
O	Adrenoleukodystrophy	300100	ABCD1	X
●	Alpha-methylacetoacetic aciduria	203750	ACAT1	
O	Alport syndrome, autosomal recessive, COL4A3 related	203780	COL4A3	
O	Alport syndrome, autosomal recessive, COL4A4 related	203780	COL4A4	
O	Alport syndrome, X-linked	301050	COL4A5	X
O	Andermann Syndrome / Agenesis of the corpus callosum with peripheral neuropathy	218000	SLC12A6	
O	Androgen insensitivity	300068	AR	X
●	Argininosuccinic aciduria	207900	ASL	
O	Aspartylglucosaminuria	208400	AGA	
O	Ataxia with isolated vitamin E deficiency	277460	TTPA	
O	Autosomal Recessive Polycystic Kidney Disease (ARPKD)	263200	PKHD1	
O	Bardet-Biedl syndrome 1	209900	BBS1	
O	Bardet-Biedl syndrome 10	209900	BBS10	
O	Bardet-Biedl syndrome 2	209900	BBS2	
O	Bare Lymphocyte Syndrome, type II	209920	CIITA	
O	Bartter syndrome, type 4a	602522	BSND	
●	Biotinidase deficiency	253260	BTD	
●	Carnitine deficiency, systemic primary	212140	SLC22A5	
O	Cartilage-hair hypoplasia	250250	RMRP	
O	Cerebrotendinous xanthomatosis	213700	CYP27A1	
O	Ceroid lipofuscinosis, neuronal, 5	256731	CLN5	
O	Ceroid lipofuscinosis, neuronal, 8	600143	CLN8	
O	Ceroid lipofuscinosis, neuronal, type 1	256730	PPT1	
O	Ceroid lipofuscinosis, neuronal, type 2	204500	TPP1	
O	Charcot-Marie-Tooth Disease with Deafness: X-Linked: PRPS1 Related (CMTX5)	311070	PRPS1	X
O	Charcot-Marie-Tooth neuropathy, X-linked	302800	GJB1	X
O	Cholestasis, progressive	601847	ABCB11	
O	Chronic granulomatous disease, X-linked	306400	CYBB	X
●	Citrullinemia	215700	ASS1	
O	Congenital disorder of glycosylation, type Ib	602579	MPI	
O	Congenital disorder of glycosylation, type Ic	603147	ALG6	
O	Congenital disorder of glycosylation, type Ia	212065	PMM2	
O	Corneal endothelial dystrophy and sensorineural deafness (CDPD)	217400	SLC4A11	
●	CPT I (Carnitine Palmitoyltransferase IA) deficiency, hepatic, type IA	255120	CPT1A	
●	CPT II (Carnitine Palmitoyltransferase) deficiency, myopathy due to	608836	CPT2	
O	Crigler-Najjar syndrome, type I	218800	UGT1A1	
●	Cystic fibrosis	219700	CFTR	
O	Cystinosis, nephropathic	219800	CTNS	
O	Diastrophic dysplasia	222600	SLC26A2	
●	Duchenne muscular dystrophy	310200	DMD	X
O	Ellis-van Creveld Syndrome	225500	EVC2	
O	Emery-Dreifuss muscular dystrophy 1, X-linked	310300	EMD	X
O	Enhanced S-cone syndrome (Retinitis pigmentosa 37)	268100	NR2E3	
O	Epidermolysis bullosa dystrophica, AR	226600	COL7A1	
O	Epidermolysis bullosa, junctional, Herlitz type	226700	LAMB3	
O	Ethylmalonic encephalopathy	602473	ETHE1	
O	Fabry disease	301500	GLA	X
●	Familial Mediterranean fever, autosomal recessive, including mild form	249100	MEFV	
O	Fibular hypoplasia and complex brachydactyly / Du pan syndrome	228900	GDF5	
O	Fructose intolerance	229600	ALDOB	
O	Fumarase deficiency	606812	FH	
●	Glutaric aciduria, type I	231670	GCDH	
O	Glycogen storage disease Ib	232220	SLC37A4	
O	Glycogen storage disease II / Pompe disease	232300	GAA	
O	Glycogen storage disease IV	232500	GBE1	
O	GM1-gangliosidosis, types I, II	230500	GLB1	
O	GRACILE Syndrome	603358	BCS1L	
O	Hemochromatosis, type 3	604250	TFR2	
O	Hemochromatosis: Type 2A: HFE2 Related	602390	HFE2	
O	Hemophilia A, factor VIII deficiency, X-linked	306700	F8	X
O	Hemophilia B, factor IX deficiency	306900	F9	X
O	Hermansky-Pudlak syndrome 3	614072	HPS3	
●	HMG-CoA lyase deficiency	246450	HMGCL	
O	Holocarboxylase synthetase deficiency	253270	HLCS	
●	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	613752	AHCY	
O	Hyperoxaluria III	613616	HOGA1	
O	Hyperoxaluria, primary, type I	259900	AGXT	
O	Hyperoxaluria, primary, type II	260000	GRHPR	
O	Hypohidrotic Ectodermal Dysplasia, X-Linked	305100	EDA	X
O	Hypophosphatasia, infantile	241500	ALPL	
●	Isovaleric acidemia	243500	IVD	
O	Krabbe disease	245200	GALC	
●	LCHAD deficiency	609016	HADHA	
O	Leber congenital amaurosis 5	604537	LCA5	
O	Limb-Girdle Muscular Dystrophy: Type 2I; Muscular dystrophy-dystroglycanopathy	607155	FKRP	
O	Lipoid adrenal hyperplasia	201710	STAR	
●	Maple syrup urine disease, type II	248600	DBT	
●	Mevalonic aciduria	610377	MVK	
O	Mucolipidosis III alpha/beta, and type II	252500, 252600	GNPTAB	
O	Mucopolysaccharidosis Ih / Hurler Syndrome	607014	IDUA	
O	Mucopolysaccharidosis II / Hunter Syndrome: X-linked	309900	IDS	X
O	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies)	253280	POMGNT1	
O	Muscular dystrophy, limb-girdle, type 2E	604286	SGCB	
O	Myopathy due to myoadenylate deaminase deficiency	612874	AMPD1	
O	Myotubular myopathy, X-linked	310400	MTM1	X
O	Neutropenia, severe congenital 3, autosomal recessive	610738	HAX1	
O	Niemann-Pick Disease, Type C2	607625	NPC2	
O	Niemann-Pick Disease: Type C1	257220	NPC1	
O	OTC deficiency / Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	238970	SLC25A15	
●	Pendred syndrome	274600	SLC26A4	
O	Peroxisomal acyl-CoA oxidase deficiency	264470	ACOX1	
●	Phenylketonuria	261600	PAH	
●	Propionic acidemia	606054	PCCA	
●	Propionic acidemia	606054	PCCB	
O	Pycnodysostosis	265800	CTSK	
O	Pyruvate dehydrogenase E1-alpha deficiency: X-linked	312170	PDHA1	X
O	Pyruvate dehydrogenase E1-beta deficiency	614111	PDHB	
O	Rhizomelic chondrodysplasia punctata, type 1	215100	PEX7	
O	Salla disease	604369	SLC17A5	
O	Segawa syndrome, recessive (tyrosine hydroxylase deficiency)	605407	TH	
O	Severe combined immunodeficiency, X-linked	300400	IL2RG	X
O	Sjogren-Larsson syndrome	270200	ALDH3A2	
O	Smith-Lemli-Opitz syndrome	270400	DHCR7	
●	Spinal muscle atrophy (several types)	253300, 253550, 253400, 271150	SMN1, (SMN2)	

● Patologia cuyos exones están completamente cubiertos. O Patología en qué sólo se cubren determinadas mutaciones puntuales **§ Alguna de las variantes analizadas presenta una frecuencia especialmente elevada en alguna población mundial. X Gen localizado en el cromosoma sexual homónimo.**

High impact

	CONDITION	OMIM	GENE	
●	Tyrosinemia, type I	276700	FAH	
●	VLCAD deficiency	201475	ACADVL	
O	Wilson disease	277900	ATP7B	
O	Zellweger syndrome	214100	PEX10	
O	Zellweger syndrome-1	214100	PEX1	
O	Ataxia-telangiectasia	208900	ATM	
O	Autoimmune polyendocrinopathy syndrome, type I	240300	AIRE	§
O	Bloom Syndrome	210900	BLM	§
O	Canavan disease	271900	ASPA	§
●	Deafness, autosomal recessive 1A (DFNB1-related)	220290	GJB2	§
O	Dihydropyrimidine dehydrogenase deficiency	274270	DPYD	§
O	Dysautonomia, familial	223900	IKBKAP	§
O	Fanconi anemia, complementation group C	227645	FANCC	§
O	Fragile X syndrome	300624	FMR1	§,X
O	Gaucher disease, types I and II	230800, 230900	GBA	§
O	Glycogen storage disease Ia	232200	G6PC	§
O	Hyperinsulinemic hypoglycemia, familial, type 2	601820	KCNJ11	§
O	Hypoaldosteronism , congenital dur to Corticosterone Methvloxidase II Deficiencv	610600	CYP11B2	§
O	Inclusion Body Myopathy, type 2	600737	GNE	§
O	Joubert syndrome 2	608091	TMEM216	§
O	Leigh syndrome, French-Canadian type	220111	LRPPRC	§
●	Maple syrup urine disease, type Ia	248600	BCKDHA	§
●	Maple syrup urine disease, type Ib	248600	BCKDHB	§
●	Maple syrup urine disease, type III	248600	DLD	§
O	Metachromatic leukodystrophy	250100	ARSA	§
O	Mucolipidosis IV	252650	MCOLN1	§
O	Muscular dystrophy, limb-girdle, type 2D	608099	SGCA	§
O	Nephrotic syndrome, type 1 (Finnish Type)	256300	NPHS1	§
O	Niemann-Pick disease, type A	257200	SMPD1	§
O	Niemann-Pick disease, type B	607616	SMPD1	§
O	Spastic ataxia, Charlevoix-Saguenay type (ARSACS)	270550	SACS	§
O	Tay-Sachs disease	272800	HEXA	§
●	Thalassemia, alpha-	604131	HBA1	§
●	Thalassemia, alpha-	604131	HBA2	§
●	Thalassemias, beta- (Sickle Cell Anemia)	613985	HBB	§
O	Thrombocytopenia, congenital amegakaryocytic	604498	MPL	§
O	Usher syndrome, type 1F	602083	PCDH15	§
O	Usher syndrome, type 3A	276902	CLRN1	§

High impact but variable clinical spectrum

	CONDITION	OMIM	GENE
●	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	201910	CYP21A2
●	Argininemia	207800	ARG1
●	Diabetes mellitus, neonatal, with congenital hypothyroidism	610199	GLIS3
●	Glutaric acidemia IIA	231680	ETFA
●	Glutaric acidemia IIB	231680	ETFB
●	Glutaric acidemia IIC	231680	ETFDH
O	Glycogen storage disease IIIa	232400	AGL
O	Herlitz Junctional Epidermolysis Bullosa: LAMC2 Related	226700	LAMC2
●	Hypermethioninemia due to Glycine N-methyltransferase deficiency	606664	GNMT
●	Hypermethioninemia, persistent, due to MAT1 deficiency	250850	MAT1A
●	Hypothryoidism, congenital, nongoitrous 4	275100	TSHB
●	Hypothyroidism congenital due to thyroid dysgenesis or hypoplasia	218700	PAX8
●	Hypothyroidism, congenital, nongoitrous 1	275200	TSHR
●	Methylmalonic aciduria and homocystinuria, cblC type	277400	MMACHC
●	Methylmalonic aciduria and homocystinuria, cblD type	277410	MMADHC
●	Methylmalonic aciduria, vitamin B12-responsive, cblB type	251110	MMAB
●	Methylmalonic aciduria, vitamin B12-responsive, cblA type	251100	MMAA
●	Methylmalonic aciduria, mut(0) type	251000	MUT
●	Methylmalonyl-CoA epimerase deficiency	251120	MCEE
●	Thyroid dyshormonogenesis 1	274400	SLC5A5

High impact but variable clinical spectrum

	CONDITION	OMIM	GENE
●	Thyroid dyshormonogenesis 2A	274500	TPO
●	Thyroid dyshormonogenesis 3	274700	TG
●	Thyroid dyshormonogenesis 4	274800	IYD
●	Thyroid dyshormonogenesis 5	274900	DUOXA2
●	Thryoid dyshormonogenesis 6	607200	DUOX2

Mild impact - variable clinical spectrum

	CONDITION	OMIM	GENE	
O	Factor XI deficiency, autosomal recessive	612416	F11	§
●	G6PD deficiency / Favism	134700	G6PD	§,X
O	Achromatopsia: CNGB3 Related	262300	CNGB3	
O	Acrodermatitis enteropathica	201100	SLC39A4	
●	Acyl-CoA dehydrogenase, short-chain, deficiency of	201470	ACADS	
●	Alkaptonuria	203500	HGD	
O	Aromatase deficiency	613546	CYP19A1	
●	Cystathioninuria	219500	CTH	
●	Cystinuria	220100	SLC3A1	
●	Cystinuria	220100	SLC7A9	
O	Diabetes Mellitus, permanent neonatal	606176	ABCC8	
O	Dubin-Johnson syndrome	237500	ABCC2	
O	Ehlers-Danlos syndrome, type VIIC	225410	ADAMTS2	
O	Emphysema due to Alpha1 Anti-Trypsin deficiency	613490	SERPINA1	
O	Factor V Deficiency	227400	F5	
O	Factor V Leiden Thrombophilia	188055	F5	
●	Galactokinase deficiency with cataracts	230200	GALK1	
●	Galactose epimerase deficiency	230350	GALE	
●	Galactosemia	230400	GALT	
O	Gitelman syndrome	263800	SLC12A3	
O	Glycogen storage disease VII	232800	PFKM	
●	Histidinemia	235800	HAL	
●	Homocystinuria, B6-responsive and nonresponsive types	236200	CBS	
O	Hyperinsulinemic hypoglycemia, familial, type 1	256450	ABCC8	
●	Hyperprolinemia, type II	239510	ALDH4A1	
O	Lipoprotein lipase deficiency	238600	LPL	
O	McArdle disease / Glycogen Storage Disease: Type V	232600	PYGM	
O	Prothrombin thrombophilia / Factor II deficiency	613679	F2	
O	Retinoschisis: X-linked	312700	RS1	X
O	von Willebrand disease types 2A, 2B, 2M and 2N	613554	VWF	

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Para más información puede contactarnos en:

http://www.qgenomics.com/
Quantitative Genomic Medicine Laboratories, s.l.
c/ Joan XXIII, 10 - 08950 Esplugues del Llobregat
Tel. 932 301 270

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