# qCarrier® Plus · Enfermedades

qgenomics
Genomics for Human Health

El qCarrier Plus 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 +9.000 mutaciones descritas en las bases de datos como causa de **+300 enfermedades recesivas**. El qCarrier Plus supone una mejora en cuanto a su antecesor en tanto que **analiza regiones codificantes completas**, evitando la necesidad de realizar en la mayoría de los casos un test genético distinto para determinar la compatibilidad entre individuos. La NGS nos permite abordar, en un sólo análisis, el estudio de distintos tipos de mutaciones: de nucleótido único (SNVs), pequeñas inserciones/deleciones, variantes de número de copia y ciertos reordenamientos.

| | CONDITION | OMIM | GENE | |
|---|---|---|---|---|
| ● | 17-beta-hydroxysteroid dehydrogenase X deficiency | 300438 | HSD17B10 | XL |
| ● | 2-methylbutyrylglycinuria | 610006 | ACADSB | AR |
| ● | 3-Methylcrotonyl-CoA carboxylase 1 deficiency | 210200 | MCCC1 | AR |
| ● | 3-Methylcrotonyl-CoA carboxylase 2 deficiency | 210210 | MCCC2 | AR |
| ● | Aarskog-Scott syndrome; Mental retardation, X-linked 16 | 305400 | FGD1 | XL |
| ● | Achondrogenesis Ib | 600972 | SLC26A2 | AR |
| ● | Achromatopsia-3 | 262300 | CNGB3 | AR |
| ● | Acyl-CoA dehydrogenase, medium chain, deficiency of | 201450 | ACADM | AR |
| ● | Acyl-CoA dehydrogenase, short-chain, deficiency of | 201470 | ACADS | AR |
| ● | Adrenal hyperplasia, congenital, due to 17-alpha-hydroxylase deficiency | 202110 | CYP17A1 | AR |
| O | Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency | 201910 | CYP21A2 | AR |
| ● | Adrenoleukodystrophy | 300100 | ABCD1 | XL |
| ● | Alkaptonuria | 203500 | HGD | AR |
| ● | Allan-Herndon-Dudley syndrome | 300523 | SLC16A2 | XL |
| ● | Alpha-methylacetoacetic aciduria | 203750 | ACAT1 | AR |
| ● | Alpha-thalassemia/mental retardation syndrome | 301040 | ATRX | XL |
| ● | Alport syndrome, autosomal recessive | 203780 | COL4A4 | AR |
| ● | Anauxetic dysplasia | 607095 | RMRP | AR |
| ● | Androgen insensitivity | 300068 | AR | XL |
| ● | Argininemia | 207800 | ARG1 | AR |
| ● | Argininosuccinic aciduria | 207900 | ASL | AR |
| ● | Arts syndrome | 301835 | PRPS1 | XL |
| ● | Aspartylglucosaminuria | 208400 | AGA | AR |
| ● | Ataxia with isolated vitamin E deficiency | 277460 | TTPA | AR |
| ● | Ataxia-telangiectasia | 208900 | ATM | AR |
| ● | Auditory neuropathy, autosomal recessive, 1 | 601071 | OTOF | AR |
| ● | Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia | 240300 | AIRE | AR |
| ● | Autosomal Recessive Polycystic Kidney Disease | 263200 | PKHD1 | AR |
| ● | Bardet-Biedl syndrome 1 | 209900 | BBS1 | AR |
| ● | Bardet-Biedl syndrome 10 | 615987 | BBS10 | AR |
| ● | Bardet-Biedl syndrome 14; Joubert syndrome 5; Meckel syndrome 4; Senior-Loken syndrome 6 | 615991; 610188; 611134; 610189 | CEP290 | AR |
| ● | Bardet-Biedl syndrome 2 | 615981 | BBS2 | AR |
| ● | Bartter syndrome, type 4a | 602522 | BSND | AR |
| ● | Biotinidase deficiency | 253260 | BTD | AR |
| ● | Bjornstad syndrome | 262000 | BCS1L | AR |
| ● | Canavan disease | 271900 | ASPA | AR |
| ● | Carbamoylphosphate synthetase I deficiency | 237300 | CPS1 | AR |
| ● | Carnitine deficiency, systemic primary | 212140 | SLC22A5 | AR |
| ● | Carnitine-acylcarnitine translocase deficiency | 212138 | SLC25A20 | AR |
| ● | Cerebrotendinous xanthomatosis | 213700 | CYP27A1 | AR |
| ● | Ceroid lipofuscinosis, neuronal, 10 | 610127 | CTSD | AR |
| ● | Ceroid lipofuscinosis, neuronal, 2 | 204500 | TPP1 | AR |
| ● | Ceroid lipofuscinosis, neuronal, 3 | 204200 | CLN3 | AR |
| ● | Ceroid lipofuscinosis, neuronal, 5 | 256731 | CLN5 | AR |
| ● | Ceroid lipofuscinosis, neuronal, 6 | 601780 | CLN6 | AR |
| ● | Ceroid lipofuscinosis, neuronal, 7 | 610951 | MFSD8 | AR |
| ● | Ceroid lipofuscinosis, neuronal, 8 | 600143 | CLN8 | AR |
| ● | Ceroid lipofuscinosis, neuronal, type 1 | 256730 | PPT1 | AR |
| ● | Charcot-Marie-Tooth disease, type 4B1 | 601382 | MTMR2 | AR |
| ● | Charcot-Marie-Tooth disease, type 4C | 601596 | SH3TC2 | AR |
| ● | Charcot-Marie-Tooth disease, type 4D | 601455 | NDRG1 | AR |
| ● | Charcot-Marie-Tooth Neuropathy Type 4A | 214400 | GDAP1 | AR |
| ● | Cholestasis, benign recurrent intrahepatic, 2 | 601847 | ABCB11 | AR |
| ● | Citrullinemia | 215700 | ASS1 | AR |
| ● | Citrullinemia, adult-onset type II; type II, neonatal-onset | 603471; 605814 | SLC25A13 | AR |
| ● | Coffin-Lowry syndrome | 303600 | RPS6KA3 | XL |
| ● | Combined malonic and methylmalonic acidemia | 614265 | ACSF3 | AR |
| ● | Cone-rod dystrophy 3 | 604116 | ABCA4 | AR |
| ● | Cone-rod dystrophy, 604393 (Congenital Leber Amaurosis, 4) | 604393 | AIPL1 | AR |
| O | Cone-rod dystrophy, X-linked, 1 | 304020 | RPGR | XL |
| ● | Congenital disorder of glycosylation, type Ia | 212065 | PMM2 | AR |
| ● | Corneal endothelial dystrophy and sensorineural deafness (CDPD) | 217400 | SLC4A11 | AR |
| ● | CPT deficiency, hepatic, type IA | 255120 | CPT1A | AR |
| ● | CPT II deficiency, lethal neonatal | 608836 | CPT2 | AR |
| ● | Cystathioninuria | 219500 | CTH | AR |
| ● | Cystic Fibrosis; Congenital bilateral absence of vas deferens | 219700; 277180 | CFTR | AR |
| ● | Cystinosis, atypical nephropathic | 219800 | CTNS | AR |
| ● | Cystinuria | 220100 | SLC3A1 | AR |
| ● | Cystinuria | 220100 | SLC7A9 | AR |
| ● | Deafness, autosomal recessive 12 | 601386 | CDH23 | AR |
| ● | Deafness, autosomal recessive 18A | 602092 | USH1C | AR |
| ● | Deafness, autosomal recessive 1A (DFNB1A); Deafness, digenic (GJB2/GJB3, GJB2/GJB6) | 220290 | GJB2 | AR, DD |
| ● | Deafness, autosomal recessive 1B (DFNB1B); Deafness, digenic GJB2/GJB6 | 612645; 220290 | GJB6 | AR, DD |
| ● | Deafness, autosomal recessive 23 | 609533 | PCDH15 | AR |
| ● | Deafness, autosomal recessive 4, with enlarged vestibular aqueduct | 600791 | SLC26A4 | AR |
| ● | Deafness, digenic, GJB2/GJB3 | 220290 | GJB3 | AR, DD |
| ● | Deafness, X-linked 2 | 304400 | POU3F4 | XL |
| ● | Dent disease 2 | 300555 | OCRL | XL |
| ● | Dihydrolipoamide dehydrogenase deficiency | 246900 | DLD | AR |
| ● | Duchenne muscular dystrophy; Becker muscular dystrophy | 310200; 300376 | DMD | XL |
| ● | Dysprothrombinemia | 613679 | F2 | AR |
| ● | Ehlers-Danlos syndrome, type VI | 225400 | PLOD1 | AR |
| ● | Ellis-van Creveld syndrome | 225500 | EVC2 | AR |
| ● | Emphysema due to Alpha1 Anti-Trypsin deficiency | 613490 | SERPINA1 | AR |
| ● | Epidermolysis bullosa dystrophica, AR | 226600 | COL7A1 | AR |
| ● | Epidermolysis bullosa, junctional, Herlitz type; non-Herlitz type | 226700; 226650 | LAMB3 | AR |
| ● | Epidermolysis bullosa, junctional, Herlitz type; non-Herlitz type | 226700; 226650 | LAMC2 | AR |
| ● | Epilepsy, X-linked, with variable learning disabilities and behavior disorders | 300491 | SYN1 | XL |
| ● | Epileptic encephalopathy, early infantile, 1 | 308350 | ARX | XL |
| ● | Ethylmalonic encephalopathy | 602473 | ETHE1 | AR |
| ● | Fabry disease | 301500 | GLA | XL |
| ● | Factor V deficiency | 227400 | F5 | AR |

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| | CONDITION | OMIM | GENE | |
|---|---|---|---|---|
| ● | Factor XI deficiency, autosomal dominant & recessive | 612416 | F11 | AR, AD |
| ● | Familial Mediterranean fever, AR | 249100 | MEFV | AR |
| ● | Fanconi anemia | 227650 | FANCA | AR |
| ● | Fanconi anemia, complementation group C | 227645 | FANCC | AR |
| ● | Favism | 134700 | G6PD | XL |
| ● | Folate malabsorption, hereditary | 229050 | SLC46A1 | AR |
| ● | Fragile X syndrome | 300624 | FMR1 | XL |
| ● | Friedreich ataxia with retained reflexes | 229300 | FXN | AR |
| ● | Fructose intolerance | 229600 | ALDOB | AR |
| ● | Fumarase deficiency | 606812 | FH | AR |
| ● | Galactokinase deficiency with cataracts | 230200 | GALK1 | AR |
| ● | Galactose epimerase deficiency | 230350 | GALE | AR |
| ● | Galactosemia | 230400 | GALT | AR |
| ● | Gaucher disease, perinatal lethal | 608013 | GBA | AR |
| ● | Glutamate formiminotransferase deficiency | 229100 | FTCD | AR |
| ● | Glutaric acidemia IIA | 231680 | ETFA | AR |
| ● | Glutaric acidemia IIB | 231680 | ETFB | AR |
| ● | Glutaric acidemia IIC | 231680 | ETFDH | AR |
| ● | Glutaricaciduria, type I | 231670 | GCDH | AR |
| ● | Glycine encephalopathy | 605899 | AMT | AR |
| ● | Glycine encephalopathy | 605899 | GLDC | AR |
| ● | Glycine N-methyltransferase deficiency | 606664 | GNMT | AR |
| ● | Glycogen storage disease Ia | 232200 | G6PC | AR |
| ● | Glycogen storage disease Ib | 232220 | SLC37A4 | AR |
| ● | Glycogen storage disease II / Pompe Disease | 232300 | GAA | AR |
| ● | Glycogen storage disease IIIa | 232400 | AGL | AR |
| ● | Glycogen storage disease IV | 232500 | GBE1 | AR |
| ● | GM1-gangliosidosis, types I, II and III | 230500; 230600; 230650 | GLB1 | AR |
| ● | Goldmann-Favre syndrome | 268100 | NR2E3 | AR |
| ● | HARP syndrome | 607236 | PANK2 | AR |
| ● | Hartnup disorder | 234500 | SLC6A19 | AR |
| ● | Heimler syndrome, type 2 | 616617 | PEX6 | AR |
| ● | Hemochromatosis, type 3 | 604250 | TFR2 | AR |
| ● | Hemochromatosis: Type 2A; HFE2 Related | 602390 | HFE2 | AR |
| O | Hemophilia A | 306700 | F8 | XL |
| ● | Hemophilia B | 306900 | F9 | XL |
| ● | Histidinemia | 235800 | HAL | AR |
| ● | HMG-CoA lyase deficiency | 246450 | HMGCL | AR |
| ● | Holocarboxylase synthetase deficiency | 253270 | HLCS | AR |
| ● | Homocystinuria due to MTHFR deficiency | 236250 | MTHFR | AR |
| ● | Homocystinuria-megaloblastic anemia, cbl E type | 236270 | MTRR | AR |
| ● | Homocystinuria, B6-responsive and nonresponsive types | 236200 | CBS | AR |
| ● | Homocystinuria, cblD type, variant 1 / Methylmalonic aciduria and homocystinuria, cblD type / Methylmalonic aciduria, cblD type, variant 2 | 277410; 277410; 611935 | MMADHC | AR |
| ● | Hyper-IgD syndrome; Mevalonic aciduria | 260920; 610377 | MVK | AR |
| ● | Hypercholesterolemia, familial | 143890 | LDLR | AR, CD |
| ● | Hypercholesterolemia, familial, autosomal recessive | 603813 | LDLRAP1 | AR |
| ● | Hyperinsulinemic hypoglycemia, familial, Type 2 | 601820 | KCNJ11 | AD, AR |
| ● | Hypermethioninemia due to adenosine kinase deficiency | 614300 | ADK | AR |
| ● | Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase | 613752 | AHCY | AR |
| ● | Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency | 250850 | MAT1A | AR |
| ● | Hyperoxaluria, primary, type 1 | 259900 | AGXT | AR |
| ● | Hyperoxaluria, primary, type II | 260000 | GRHPR | AR |
| ● | Hyperoxaluria, primary, type III | 613616 | HOGA1 | AR |
| ● | Hyperphenylalaninemia, BH4-deficient, A | 261640 | PTS | AR |
| ● | Hyperphenylalaninemia, BH4-deficient, C | 261630 | QDPR | AR |
| ● | Hyperphenylalaninemia, BH4-deficient, D | 264070 | PCBD1 | AR |
| ● | Hyperprolinemia, type II | 239510 | ALDH4A1 | AR |
| ● | Hypogonadotropic hypogonadism 7 without anosmia | 146110 | GNRHR | AR |
| ● | Hypothryoidism, congenital, nongoitrous 4 | 275100 | TSHB | AR |
| ● | Hypothyroidism, congenital, nongoitrous, 1 | 275200 | TSHR | AR |
| ● | Ichthyosis, congenital, autosomal recessive 1 | 242300 | TGM1 | AR |
| ● | Immunodeficiency, X-linked, with hyper-IgM | 308230 | CD40LG | XL |
| ● | Isovaleric acidemia | 243500 | IVD | AR |
| ● | Joubert syndrome 2 | 608091 | TMEM216 | AR |
| ● | Joubert syndrome 3 | 608629 | AHI1 | AR |
| ● | Joubert syndrome 4 | 609583 | NPHP1 | AR |
| ● | Joubert syndrome 8 | 612291 | ARL13B | AR |
| ● | Krabbe disease | 245200 | GALC | AR |
| ● | LCHAD deficiency | 609016 | HADHA | AR |
| ● | Leber congenital amaurosis 1 | 204000 | GUCY2D | AR |
| ● | Leber congenital amaurosis 13 | 612712 | RDH12 | AR |
| ● | Leber congenital amaurosis 2 | 204100 | RPE65 | AR |
| ● | Leber congenital amaurosis 8 | 613835 | CRB1 | AR |
| ● | Leigh syndrome, due to COX deficiency | 256000 | SURF1 | AR |
| ● | Leigh syndrome, French-Canadian type | 220111 | LRPPRC | AR |
| ● | limb-girdle muscular dystrophy type 2B | 253601 | DYSF | AR |
| ● | Lipoid adrenal hyperplasia | 201710 | STAR | AR |
| ● | Lissencephaly, X-linked | 300067 | DCX | XL |
| ● | Macular corneal dystrophy | 217800 | CHST6 | AR |
| ● | Malonyl-CoA decarboxylase deficiency | 248360 | MLYCD | AR |
| ● | Mannosidosis, alpha-, types I and II | 248500 | MAN2B1 | AR |
| ● | Maple syrup urine disease, type Ia | 248600 | BCKDHA | AR |
| ● | Maple syrup urine disease, type Ib | 248600 | BCKDHB | AR |
| ● | Maple syrup urine disease, type II | 248600 | DBT | AR |
| ● | MASA syndrome / CRASH syndrome | 303350 | L1CAM | XL |
| ● | McArdle disease / Glycogen Storage Disease: Type V | 232600 | PYGM | AR |
| ● | Meckel syndrome 1 | 249000 | MKS1 | AR |
| ● | Mental retardation and microcephaly with pontine and cerebellar hypoplasia | 300749 | CASK | XL |
| ● | Mental retardation syndrome, X-linked, Siderius type | 300263 | PHF8 | XL |
| ● | Mental retardation, X-linked | 300486 | OPHN1 | XL |
| ● | Mental retardation, X-linked 1/78 | 309530 | IQSEC2 | XL |
| ● | Mental retardation, X-linked 12/35 | 300957 | THOC2 | XL |
| ● | Mental retardation, X-linked 21/34 | 300143 | IL1RAPL1 | XL |
| ● | Mental retardation, X-linked 30/47 | 300558 | PAK3 | XL |
| ● | Mental retardation, X-linked 41 | 300849 | GDI1 | XL |
| ● | Mental retardation, X-linked 58 | 300210 | TSPAN7 | XL |
| ● | Mental retardation, X-linked 63 | 300387 | ACSL4 | XL |
| ● | Mental retardation, X-linked 9 | 309549 | FTSJ1 | XL |
| ● | Mental retardation, X-linked 90 | 300850 | DLG3 | XL |
| ● | Mental retardation, X-linked 94 | 300699 | GRIA3 | XL |
| ● | Mental retardation, X-linked 97 | 300803 | ZNF711 | XL |
| ● | Mental retardation, X-linked 99 | 300919 | USP9X | XL |
| ● | Mental retardation, X-linked syndromic 5 | 304340 | AP1S2 | XL |
| ● | Mental retardation, X-linked syndromic, Raymond type | 300799 | ZDHHC9 | XL |
| ● | Mental retardation, X-linked syndromic, Turner type | 300706 | HUWE1 | XL |
| ● | Mental retardation, X-linked, Asperger syndrome susceptibility, X-linked | 300427 | NLGN4X | XL |
| ● | Mental retardation, X-linked, FRAXE type | 309548 | AFF2 | XL |
| ● | Mental retardation, X-linked, syndromic 13 | 300055 | MECP2 | XL |
| ● | Mental retardation, X-linked, syndromic 14 | 300676 | UPF3B | XL |
| ● | Mental retardation, X-linked, syndromic 15 | 300354 | CUL4B | XL |
| ● | Mental retardation, X-linked, syndromic, Claes-Jensen type | 300534 | KDM5C | XL |
| ● | Metachromatic leukodystrophy | 250100 | ARSA | AR |
| ● | Methylmalonic aciduria and homocystinuria, cblC type | 277400 | MMACHC | AR |
| ● | Methylmalonic aciduria and homocystinuria, cblF type | 277380 | LMBRD1 | AR |
| ● | Methylmalonic aciduria and homocystinuria, cblJ type | 614857 | ABCD4 | AR |
| ● | Methylmalonic aciduria, mut(0) type | 251000 | MUT | AR |
| ● | Methylmalonic aciduria, vitamin B12-responsive | 251100 | MMAA | AR |

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| | CONDITION | OMIM | GENE | |
|---|---|---|---|---|
| ● | Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type | 251110 | MMAB | AR |
| ● | Methylmalonyl-CoA epimerase deficiency | 251120 | MCEE | AR |
| ● | Microphthalmia, isolated 3 | 611038 | RAX | AR |
| ● | Mucolipidosis II and III, alpha/beta | 252500; 252600 | GNPTAB | AR |
| ● | Mucolipidosis IV | 252650 | MCOLN1 | AR |
| ● | Mucopolysaccharidisis type IIIA (Sanfilippo A) | 252900 | SGSH | AR |
| ● | Mucopolysaccharidosis Ih | 607014 | IDUA | AR |
| ● | Mucopolysaccharidosis II | 309900 | IDS | XL |
| ● | Mucopolysaccharidosis IVA | 253000 | GALNS | AR |
| ● | Mucopolysaccharidosis type IIIB (Sanfilippo B) | 252920 | NAGLU | AR |
| ● | Mucopolysaccharidosis type IIIC (Sanfilippo C) | 252930 | HGSNAT | AR |
| ● | Mucopolysaccharidosis type IIID | 252940 | GNS | AR |
| ● | Mucopolysaccharidosis type VI (Maroteaux-Lamy) | 253200 | ARSB | AR |
| ● | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1 | 236670 | POMT1 | AR |
| ● | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2 | 613150 | POMT2 | AR |
| ● | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3 | 253280 | POMGNT1 | AR |
| ● | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5 | 613153 | FKRP | AR |
| ● | Muscular dystrophy, limb-girdle, type 2A | 253600 | CAPN3 | AR |
| ● | Muscular dystrophy, limb-girdle, type 2D | 608099 | SGCA | AR |
| ● | Muscular dystrophy, limb-girdle, type 2E | 604286 | SGCB | AR |
| ● | Myotonia congenita, dominant; recessive | 160800; 255700 | CLCN1 | AR, AD |
| O | Nemaline myopathy 2, autosomal recessive | 256030 | NEB | AR |
| ● | Nephrotic syndrome, type 1 | 256300 | NPHS1 | AR |
| ● | Neutropenia, severe congenital 3, autosomal recessive | 610738 | HAX1 | AR |
| ● | Niemann-Pick disease, type A | 257200 | SMPD1 | AR |
| ● | Niemann-Pick disease, type C1 | 257220 | NPC1 | AR |
| ● | Niemann-pick disease, type C2 | 607625 | NPC2 | AR |
| ● | Nijmegen Breakage Syndrome (Ataxia telangectasia, type 1) | 251260 | NBN | AR |
| ● | Norrie disease | 310600 | NDP | XL |
| ● | Nystagmus 6, congenital, X-linked | 300814 | GPR143 | XL |
| ● | Ornithine transcarbamylase deficiency | 311250 | OTC | XL |
| ● | Osteogenesis imperfecta, type VIII | 610915 | P3H1 | AR |
| ● | Pelizaeus-Merzbacher disease | 312080 | PLP1 | XL |
| ● | Peroxisomal acyl-CoA oxidase deficiency | 264470 | ACOX1 | AR |
| ● | Peroxisome biogenesis disorder 1A (Zellweger) | 214100 | PEX1 | AR |
| ● | Peroxisome biogenesis disorder 6A (Zellweger) | 614870 | PEX10 | AR |
| ● | Peroxisome biogenesis disorder 9B; Rhizomelic chondroplasia punctata, type I | 614879; 215100 | PEX7 | AR |
| ● | Phenylketonuria | 261600 | PAH | AR |
| ● | Phosphoglycerate kinase 1 deficiency | 300653 | PGK1 | XL |
| ● | Pituitary hormone deficiency, combined, 2 | 262600 | PROP1 | AR |
| ● | Primary ciliary dyskinesia | 608644 | DNAH5 | AR |
| ● | Propionic acidemia | 606054 | PCCA | AR |
| ● | Propionic acidemia | 606054 | PCCB | AR |
| ● | Pyruvate carboxylase deficiency | 266150 | PC | AR |
| ● | Pyruvate dehydrogenase E1-beta deficiency | 614111 | PDHB | AR |
| ● | Renpenning syndrome | 309500 | PQBP1 | XL |
| ● | Retinitis pigmentosa 2 | 312600 | RP2 | XL |
| ● | Retinitis pigmentosa 25 | 602772 | EYS | AR |
| ● | Retinitis pigmentosa 26 | 608380 | CERKL | AR |
| ● | Retinitis pigmentosa 39 | 613809 | USH2A | AR |
| ● | Retinitis pigmentosa 43 | 613810 | PDE6A | AR |
| ● | Retinitis pigmentosa 45 | 613767 | CNGB1 | AR |
| ● | Retinitis pigmentosa 46 | 612572 | IDH3B | AR |
| ● | Retinitis pigmentosa 49 | 613756 | CNGA1 | AR |
| ● | Retinitis pigmentosa 59 | 613861 | DHDDS | AR |
| ● | Retinoschisis, X-linked | 312700 | RS1 | XL |
| ● | Rhizomelic chondrodysplasia punctata, type 3 | 600121 | AGPS | AR |
| ● | Sandhoff disease, infantile, juvenile, and adult forms | 268800 | HEXB | AR |
| ● | SCID, autosomal recessive, T-negative/B-positive type | 600802 | JAK3 | AR |
| ● | Segawa syndrome, recessive | 605407 | TH | AR |
| ● | Severe combined immunodeficiency due to ADA deficiency | 102700 | ADA | AR |
| ● | Severe combined immunodeficiency, X-linked | 300400 | IL2RG | XL |
| ● | Smith-Lemli-Opitz syndrome | 270400 | DHCR7 | AR |
| ● | Spastic ataxia, Charlevoix-Saguenay type (ARSACS) | 270550 | SACS | AR |
| ● | Spastic paraplegia 11, autosomal recessive | 604360 | SPG11 | AR |
| ● | Spastic paraplegia 7, autosomal recessive | 607259 | SPG7 | AR |
| O | Spinal muscular atrophy, type I | 253300 | SMN1 | AR |
| ● | Tay-Sachs; GM2-gangliosidosis, several forms | 272800 | HEXA | AR |
| ● | Thalassemias, alpha- | 604131 | HBA1 | AR, AD |
| ● | Thalassemias, alpha- | 604131 | HBA2 | AR, AD |
| ● | Thalassemias, beta- | 613985 | HBB | AR |
| ● | Thrombocytopenia, congenital amegakaryocytic | 604498 | MPL | AR |
| ● | Thryoid dyshormonogenesis 6 | 607200 | DUOX2 | AR |
| ● | Thyroid dyshormonogenesis 1 | 274400 | SLC5A5 | AR |
| ● | Thyroid dyshormonogenesis 2A | 274500 | TPO | AR |
| ● | Thyroid dyshormonogenesis 3 | 274700 | TG | AR |
| ● | Thyroid dyshormonogenesis 4 | 274800 | IYD | AR |
| ● | Thyroid dyshormonogenesis 5 | 274900 | DUOXA2 | AR |
| ● | Thyroid hormone resistance | 274700 | THRB | AR |
| ● | Treacher Collins syndrome 3 | 248390 | POLR1C | AR |
| ● | Trifunctional protein deficiency | 609015 | HADHB | AR |
| ● | Tyrosinemia, type I | 276700 | FAH | AR |
| ● | Tyrosinemia, type II | 276600 | TAT | AR |
| ● | Usher syndrome, type 1B; Deafness, autosomal dominant 11 | 276900; 601317 | MYO7A | AR, AD |
| ● | Usher syndrome, type 1G | 606943 | USH1G | AR |
| ● | Usher syndrome, type 2D / Deafness, autosomal recessive 31 | 611383 | WHRN | AR |
| ● | Usher syndrome, type 3A | 276902 | CLRN1 | AR |
| ● | Ventricular tachycardia, catecholaminergic polymorphic, 2 | 611938 | CASQ2 | AR |
| ● | Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness | 615441 | TRDN | AR |
| ● | VLCAD deficiency | 201475 | ACADVL | AR |
| ● | Walker-Warburg syndrome, Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4 | 253800 | FKTN | AR |
| ● | Wilson disease | 277900 | ATP7B | AR |
| ● | Wolman disease (lysosomal acid lipase deficiency) | 278000 | LIPA | AR |
| ● | X-linked mental retardation (XLMR) associated with macrocephaly | 300659 | BRWD3 | XL |

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**Para más información puede contactarnos en:**

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**NOTA:** Estos listados CARECEN DE VALOR CONTRACTUAL y son sólo informativos, pudiendo ser modificados sin previo aviso por parte de la compañía. La información en ellos contenida es para uso EXCLUSIVO de profesionales sanitarios con formación específica en genética humana. Para obtener una versión actualizada del listado contactar siempre con el laboratorio.

El análisis se centra en exones procedentes de la anotación de referencia RefSeq. Ciertos exones de tránscritos alternativos de algunos genes no pueden ser anali-

● Patologia cuyos exones están completamente cubiertos. O Patología en qué sólo se cubren determinadas mutaciones. **AR: herencia autosómica recesiva; AD: herencia autosómica dominante; XL: herencia ligada al cromosoma X; DD: patron de herencia digénico; CD: herencia co-dominante**