Gene	Chr**	Mode of inheritance	Disease	Comments
AARS2	6	Autosomal recessive	Ovarioleukodystrophy; Combined oxidative phosphorylation defect type 8	
ABCA3	16	Autosomal recessive	Interstitial lung disease due to ABCA3 deficiency	
ABCB11	2	Autosomal recessive	Progressive familial intrahepatic cholestasis type 2	
ABCC6*	16	Autosomal recessive	Pseudoxanthoma elasticum; Generalized arterial calcification of infancy type 2	
ABCC8	11	Autosomal recessive	Permanent neonatal diabetes mellitus type 3; Hyperinsulinemic hypoglycemia familial type 1	
ABCD1*	X	X-linked	Adrenomyeloneuropathy	
ACADM	1	Autosomal recessive	Medium-chain acyl-CoA dehydrogenase deficiency	
ACADVL	17	Autosomal recessive	Very long-chain acyl-CoA dehydrogenase deficiency	
ADGRV1	5	Autosomal recessive	Usher syndrome type 2	
AGA	4	Autosomal recessive	Aspartylglucosaminuria	
AGL	1	Autosomal recessive	Glycogen storage disease type 3A/3B	
AGXT	2	Autosomal recessive	Primary hyperoxaluria type 1	
AIFM1	X	X-linked	AIFM1 related disorders	
AIRE	21	Autosomal recessive	Autoimmune polyendocrinopathy type 1	
ALDH5A1	6	Autosomal recessive	Succinic semialdehyde dehydrogenase deficiency	

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Ronda Guillermo Marconi, 11-A , 1°22B
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ALDH7A1	5	Autosomal recessive	Pyridoxine-dependent epilepsy type 4	
ALDOB	9	Autosomal recessive	Hereditary fructose intolerance	
ALMS1*	2	Autosomal recessive	Alström syndrome	
ALOX12B	17	Autosomal recessive	Congenital ichthyosis type 2	
ALOXE3	17	Autosomal recessive	Congenital ichthyosis type 3	
ALPL	1	Autosomal recessive	Infantile hypophosphatasia	
AR	X	X-linked	Androgen insensitivity syndrome	The current testing method does not assess CAG trinucleotide repeats in exon 1 in this gene.
ARSA	22	Autosomal recessive	Metachromatic leukodystrophy	
ARSB	5	Autosomal recessive	Mucopolysaccharidose type 6	
ASL	7	Autosomal recessive	Argininosuccinic aciduria	
ASPA	17	Autosomal recessive	Canavan disease	
ASPM	1	Autosomal recessive	Autosomal recessive primary microcephaly type 5	
ASS1	9	Autosomal recessive	Citrullinemia type 1	
ATM	11	Autosomal recessive	Ataxia-telangiectasia	
ATP7B	13	Autosomal recessive	Wilson disease	
ATR	3	Autosomal recessive	Seckel syndrome type 1	
ATRX	X	X-linked	ATR-X syndrome	
B3GLCT	13	Autosomal recessive	Peters plus syndrome	
BBS1	11	Autosomal recessive	Bardet-Biedl syndrome type 1	
BBS10	12	Autosomal recessive	Bardet-Biedl syndrome type 10	

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BBS2	16	Autosomal recessive	Retinitis pigmentosa type 74; Bardet-Biedl syndrome type 2	
BCKDHA	19	Autosomal recessive	Maple syrup urine disease type 1A	
BCKDHB	6	Autosomal recessive	Maple syrup urine disease type 1B	
BCS1L	2	Autosomal recessive	GRACILE syndrome; Björnstad syndrome; Isolated complex III deficiency	
BGN	X	X-linked	Meester-Loeys syndrome; X-linked spondyloepimetaphyseal dysplasia	
BRIP1	17	Autosomal recessive	Fanconi anemia group J	
BTD	3	Autosomal recessive	Biotinidase Deficiency	NM_001370658.1:c.1270G>C (p.Asp424His) variant is not reported, due to low disease penetrance and its association to reduced enzyme activity in the homozygous state.
BTK	X	X-linked	Isolated growth hormone deficiency type 3; X-linked agammaglobulinemia	
CAPN3	15	Autosomal recessive	Limb-girdle muscular dystrophy type 2A	
CASK	X	X-linked	FG syndrome type 4; X-linked intellectual developmental disorder, Najm type	
CBS	21	Autosomal recessive	Classical homocystinuria	
CC2D2A	4	Autosomal recessive	CC2D2A related disorders	
CCDC103	17	Autosomal recessive	Primary ciliary dyskinesia type 17	
CCDC40	17	Autosomal recessive	Primary ciliary dyskinesia type 15	
CD40LG	X	X-linked	Immunodeficiency with hyper-IgM type 1	
CDH23	10	Autosomal recessive	Usher syndrome type 1D; Deafness autosomal recessive type 12	

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CEP152	15	Autosomal recessive	Autosomal recessive primary microcephaly type 9; Seckel syndrome type 5	
CEP290*	12	Autosomal recessive	CEP290 related disorders	Deep intronic variant NM_025114.4:c.2991+1655A>G not included.
CFI	4	Autosomal recessive	Complement factor I deficiency	
CFTR	7	Autosomal recessive	Cystic fibrosis	Only variants associated with classical Cystic Fibrosis are reported. Intron 8 polymorphic region in CFTR gene (5T allele) is only reported when the NM_000492.4:c.350G>A (p.Arg117His) variant is detected.
CHM	X	X-linked	Choroideremia	
CHRNE	17	Autosomal recessive	Congenital myasthenic syndrome type 4A, 4B, 4C	
CHRNG	2	Autosomal recessive	Lethal multiple pterygium syndrome; Escobar syndrome	
CLCN1	7	Autosomal recessive	Myotonia congenita	
CLCN5	X	X-linked	CLCN5 related disorders	
CLRN1	3	Autosomal recessive	Usher syndrome type 3A	
CNGA3	2	Autosomal recessive	Achromatopsia type 2	
CNGB3	8	Autosomal recessive	Achromatopsia type 3	
COL18A1	21	Autosomal recessive	Knobloch syndrome type 1	
COL4A3	2	Autosomal recessive	Alport syndrome type 3B	
COL4A5	X	X-linked	Alport syndrome type 1	
COL6A2	21	Autosomal recessive	Ullrich congenital muscular dystrophy type 1B; Bethlem muscular dystrophy type 1B	

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COL6A3	2	Autosomal recessive	Bethlem muscular dystrophy type 1C; Ullrich congenital muscular dystrophy type 1C; Primary dystonia, DYT27 type	
COL7A1	3	Autosomal recessive	Dystrophic epidermolysis bullosa COL7A1-Related	
COLQ	3	Autosomal recessive	Congenital myasthenic syndrome type 5	
COQ8A	1	Autosomal recessive	Primary coenzyme Q10 deficiency type 4	
CPLANE1	5	Autosomal recessive	Orofaciodigital syndrome type 6; Joubert syndrome type 17	
CPS1	2	Autosomal recessive	Carbamoyl-phosphate synthetase 1 deficiency	
CPT2	1	Autosomal recessive	Carnitine palmitoyltransferase deficiency type 2	
CRB1	1	Autosomal recessive	Leber congenital amaurosis type 8; Retinitis pigmentosa type 12	
CRB2	9	Autosomal recessive	Focal segmental glomerulosclerosis type 9; Ventriculomegaly-cystic kidney disease	
CTC1	17	Autosomal recessive	Coats plus syndrome	
CTNS	17	Autosomal recessive	Nephropathic cystinosis	
CUL4B	X	X-linked	X-linked intellectual developmental disorder, Cabezas type	
CYBB	X	X-linked	Immunodeficiency 34; Chronic granulomatous disease	
CYP1B1	2	Autosomal recessive	Congenital glaucoma type 3A; Anterior segment developmental anomaly	

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CYP21A2	6	Autosomal recessive	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	Whole gene analysis is not performed. Included variants: NM_000500.9:c.293-13C/A>G, NM_000500.9:c.332_339del, NM_000500.9:c.518T>A, NM_000500.9:c.710T>A, NM_000500.9:c.713T>A, NM_000500.9:c.719T>A, NM_000500.9:c.923dup, NM_000500.9:c.955C>T, NM_000500.9:c.1069C>T, 30kb deletion, Large gene conversion. NM_000500.9:c.955C>T variant is only reported when a CYP21A2 gene duplication is not detected. c.955C>T variant along witht the gene duplication has been rerported to be frequently found on the same chromosome (in cis), which results in the presence of two functional copies of the gene. In such cases, the individual is not considered a carrier for Congenital Adrenal Hyperplasia (Parajes et al., 2008; Kleinle et al., 2009).
CYP27A1	2	Autosomal recessive	Cerebrotendinous xanthomatosis	
CYP7B1	8	Autosomal recessive	Spastic paraplegia type 5A; Congenital bile acid synthesis defect type 3	
DCX	X	X-linked	Lissencephaly type 1; Subcortical band heterotopia	
DDX11	12	Autosomal recessive	Warsaw breakage syndrome	
DGAT1	8	Autosomal recessive	Congenital chronic diarrhea with protein-losing enteropathy	
DHCR7	11	Autosomal recessive	Smith-Lemli-Opitz syndrome	

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DHDDS	1	Autosomal recessive	Retinitis pigmentosa type 59	
DLD	7	Autosomal recessive	Pyruvate dehydrogenase E3 deficiency	
DLG3	X	X-linked	Intellectual developmental disorder type 90	
DMD	X	X-linked	Duchenne muscular dystrophy; Becker muscular dystrophy	
DNAAF1	16	Autosomal recessive	Primary ciliary dyskinesia type 13	
DNAH11*	7	Autosomal recessive	Primary ciliary dyskinesia type 7	
DNAH5	5	Autosomal recessive	Primary ciliary dyskinesia type 3	
DNAI1	9	Autosomal recessive	Primary ciliary dyskinesia type 1	
DNAJC12	10	Autosomal recessive	Hyperphenylalaninemia due to DNAJC12 deficiency	
DOCK6	19	Autosomal recessive	Adams-Oliver syndrome type 2	
DOCK8	9	Autosomal recessive	Combined immunodeficiency due to DOCK8 deficiency	
DOK7	4	Autosomal recessive	Fetal akinesia deformation sequence; Congenital myasthenic syndrome type 10	
DPYD	1	Autosomal recessive	Dihydropyrimidine dehydrogenase deficiency	
DYNC2H1	11	Autosomal recessive	Short rib-polydactyly syndrome type 3	
DYSF	2	Autosomal recessive	Limb-girdle muscular dystrophy type 2B; Miyoshi myopathy; Distal myopathy with anterior tibial onset	
EDA	X	X-linked	Hypohidrotic ectodermal dysplasia type 1	
EIF2B5	3	Autosomal recessive	Leukoencephalopathy with vanishing white matter type 5	

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ELP1	9	Autosomal recessive	Familial dysautonomia	
EMD	X	X-linked	Emery-Dreifuss muscular dystrophy type 1	
ERCC2	19	Autosomal recessive	Xeroderma pigmentosum group D; Trichothiodystrophy type 1	
ERCC6*	10	Autosomal recessive	Cockayne syndrome type B; Cerebrooculofacioskeletal syndrome type 1; UV-sensitive syndrome type 1	
ETFDH	4	Autosomal recessive	Glutaric aciduria type 2C	
EVC	4	Autosomal recessive	Ellis Van Creveld syndrome	
EYS*	6	Autosomal recessive	Retinitis pigmentosa type 25	
F8*	X	X-linked	Hemophilia A	Detection of intron 22 inversion in the F8 gene is also included
F9	X	X-linked	Hemophilia B	
FANCA	16	Autosomal recessive	Fanconi anemia group A	
FANCC	9	Autosomal recessive	Fanconi anemia group C	
FANCI	15	Autosomal recessive	Fanconi anemia group I	
FH	1	Autosomal recessive	Fumarase deficiency	
FHL1*	X	X-linked	FHL1 related disorders	
FKRP	19	Autosomal recessive	Muscular dystrophy-dystroglycanopathy type 5A, 5B, 5C	
FKTN	9	Autosomal recessive	Muscular dystrophy-dystroglycanopathy type 4A, 4B, 4C	
FLNA	X	X-linked	FLNA related disorders	

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FMR1	X	X-linked	Fragile X syndrome	5' UTR CGG trinucleotide repeats are analysed. Only alleles in the range of full mutation (>200 repeats) and premutation (55-200 repeats) are reported. Mosaicism, including gonadal mosaicism, may not be detected.
FOXP3	X	X-linked	IPEX syndrome	
FRAS1	4	Autosomal recessive	Fraser syndrome type 1	
G6PC1	17	Autosomal recessive	Glycogen storage disease type 1A	
GAA	17	Autosomal recessive	Pompe disease	
GALC	14	Autosomal recessive	Krabbe disease	
GALNS	16	Autosomal recessive	Mucopolysaccharidosis type 4A	
GALT	9	Autosomal recessive	Galactosemia	
GATM	15	Autosomal recessive	Cerebral creatine deficiency syndrome type 3	
GBE1	3	Autosomal recessive	Glycogen storage disease type 4; Adult polyglucosan body disease	
GCDH	19	Autosomal recessive	Glutaricaciduria type 1	
GDF1	19	Autosomal recessive	Ivemark syndrome	
GFPT1	2	Autosomal recessive	Congenital myasthenic syndrome type 12	
GJB2	13	Autosomal recessive	Deafness autosomal recessive type 1A	Variants associated with a mild phenotype are not reported.
GJB3	1	Autosomal recessive	Erythrokeratodermia variabilis type 1	Variants associated with a mild phenotype are not reported.
GJB6	13	Autosomal recessive	Deafness autosomal recessive type 1B	Variants associated with a mild phenotype are not reported.
GLA	X	X-linked	Fabry disease	

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GLB1	3	Autosomal recessive	GM1 gangliosidosis type 1,2,3; Mucopolysaccharidosis type 4B	
GLDC	9	Autosomal recessive	Glycine encephalopathy type 1	
GLE1	9	Autosomal recessive	Lethal congenital contracture syndrome type 1; Arthrogryposis-anterior horn cell disease syndrome	
GMPPB	3	Autosomal recessive	Muscular dystrophy due to dystroglycanopathy type 14A, 14B ,14C	
GPR143	X	X-linked	X-linked recessive ocular albinism; Congenital Nystagmus type 6	
GUCY2D	17	Autosomal recessive	Leber congenital amaurosis type 1; Cone rod dystrophy type 6; Congenital stationary night blindness type 1I	
HADHA	2	Autosomal recessive	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency; Mitochondrial trifunctional protein deficiency	
HBA1-2	16	Autosomal recessive	Alpha-thalassemia	Whole gene analysis is not performed. Included variants: --MED ; --SEA ; --THAI ; - α3.7 ; - α4.2 ; - α20.5 ; --FIL; Hb Constant Spring (NM_000517.4:c.427T>C)
HBB	11	Autosomal recessive	Beta-thalassemia; Sickle cell disease	
HERC2	15	Autosomal recessive	Developmental delay with autism spectrum disorder and gait instability	
HEXA	15	Autosomal recessive	Tay-Sachs disease	
HEXB	5	Autosomal recessive	Sandhoff disease	

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HMGCL	1	Autosomal recessive	HMG-CoA lyase deficiency	
HPRT1	X	X-linked	Kelley-Seegmiller syndrome; Lesch-Nyhan syndrome	
HUWE1	X	X-linked	X-linked intellectual developmental disorder, Tuner type	
HYLS1	11	Autosomal recessive	Hydrolethalus Syndrome	
IDS*	X	X-linked	Mucopolysaccharidosis type 2	
IDUA	4	Autosomal recessive	Mucopolysaccharidosis type 1	
IL10RA	11	Autosomal recessive	IL10-related early-onset inflammatory bowel disease type 28	
IL1RAPL1	X	X-linked	X-linked intellectual developmental disorder type 21	
IL2RG	X	X-linked	X-linked combined immunodeficiency	
IQSEC2	X	X-linked	X-linked intellectual developmental disorder type 1	
ITGB4	17	Autosomal recessive	Junctional epidermolysis bullosa with pyloric atresia type 5B; Intermediate generalized junctional epidermolysis bullosa type 5A	
IVD	15	Autosomal recessive	Isovaleric acidemia	
KCNQ1	11	Autosomal recessive	Jervell and Lange-Nielsen syndrome	
KDM5C	X	X-linked	KDM5C-related syndromic X-linked intellectual developmental disorder	
L1CAM	X	X-linked	MASA syndrome; Hydrocephalus with stenosis of the aqueduct of Sylvius	
LAMA1	18	Autosomal recessive	Poretti-Boltshauser syndrome	

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LAMA2	6	Autosomal recessive	Laminin subunit alpha 2-related congenital muscular dystrophy	
LIPA	10	Autosomal recessive	Cholesteryl ester storage disease; Wolman disease	
LMNA	1	Autosomal recessive	Mandibuloacral dysplasia; Emery-Dreifuss muscular dystrophy type 3; Charcot-Marie-Tooth disease type 2B1	
LOXHD1	18	Autosomal recessive	Deafness autosomal recessive type 77	
MBTPS2	X	X-linked	Keratosis follicularis spinulosa decalvans; Ichthyosis follicularis-alopecia-photophobia syndrome; Osteogenesis imperfecta type 19	
MCOLN1	19	Autosomal recessive	Mucolipidosis type 4	
MCPH1	8	Autosomal recessive	Autosomal recessive primary microcephaly type 1	
MECP2	X	X-linked	RETT related disorders	
MKS1	17	Autosomal recessive	Bardet-Biedl syndrome; Joubert syndrome; Meckel syndrome	
MMACHC	1	Autosomal recessive	Methylmalonic acidemia with homocystinuria type cblC	
MMUT	6	Autosomal recessive	Methylmalonic acidemia type mut0	
MPL	1	Autosomal recessive	Congenital amegakaryocytic thrombocytopenia type 1	
MRE11	11	Autosomal recessive	Ataxia-telangiectasia-like disorder type 1	
MTM1	X	X-linked	X-linked centronuclear myopathy	

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MVK	12	Autosomal recessive	Hyperimmunoglobulinemia D syndrome; Mevalonic aciduria	
MYMK	9	Autosomal recessive	Carey-Fineman-Ziter syndrome	
MYO15A	17	Autosomal recessive	Deafness autosomal recessive type 3	
MYO7A	11	Autosomal recessive	Usher syndrome type 1B; Deafness autosomal recessive type 2	
NAGA	22	Autosomal recessive	Alpha-N-acetylgalactosaminidase deficiency type 1,2,3	
NAGLU	17	Autosomal recessive	Sanfilippo syndrome type B	
NBAS	2	Autosomal recessive	Short stature-optic atrophy-Pelger-Huët anomaly syndrome; Fever-associated acute infantile liver failure syndrome	
NDP	X	X-linked	Familial exudative vitreoretinopathy; Norrie disease	
NEB*	2	Autosomal recessive	Nemaline myopathy type 2; Arthrogryposis multiplex congenita type 6	
NEXMIF	X	X-linked	X-linked intellectual developmental disorder type 98	
NFU1	2	Autosomal recessive	Multiple mitochondrial dysfunctions syndrome type 1; Spastic paraplegia type 93	
NMNAT1	1	Autosomal recessive	Leber congenital amaurosis type 9; SHILCA syndrome	
NPC1	18	Autosomal recessive	Niemann-Pick disease type C1/D	

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NPHP1	2	Autosomal recessive	Senior-Loken syndrome type 1; Nephronophthisis type 1; Joubert syndrome type 4	Deletions in this gene will not be reported, as the low reported penetrance limits their clinical utility.
NPHP3	3	Autosomal recessive	Nephronophthisis type 3; Renal-hepatic-pancreatic dysplasia type 1; Meckel syndrome type 7	
NPHS1	19	Autosomal recessive	Nephrotic syndrome type 1	
NPHS2	1	Autosomal recessive	Nephrotic syndrome type 2	
NR2E3	15	Autosomal recessive	Goldmann-Favre syndrome; Retinitis pigmentosa type 37	
NYX	X	X-linked	Congenital stationary night blindness type 1A	
OCA2	15	Autosomal recessive	Oculocutaneous albinism type 2	
OCRL	X	X-linked	Dent disease type 2; Lowe syndrome	
OFD1	X	X-linked	Joubert syndrome type 10	
OPA1	3	Autosomal recessive	Behr syndrome	
OTC	X	X-linked	Ornithine transcarbamylase deficiency	
PAH	12	Autosomal recessive	Phenylketonuria	
PANK2	20	Autosomal recessive	Pantothenate Kinase-Associated Neurodegeneration type 1	
PCCB	3	Autosomal recessive	Propionic acidemia	
PCDH15	10	Autosomal recessive	Usher syndrome type 1F; Deafness autosomal recessive type 23	

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PCNT	21	Autosomal recessive	Microcephalic osteodysplastic primordial dwarfism type 2	
PDHA1	X	X-linked	Pyruvate dehydrogenase E1-alpha deficiency	
PDHB	3	Autosomal recessive	Pyruvate dehydrogenase E1-beta deficiency	
PDZD7	10	Autosomal recessive	Usher syndrome type 2	
PEX1	7	Autosomal recessive	Zellweger syndrome 1A/B; Heimler syndrome 1	
PFKM	12	Autosomal recessive	Glycogen storage disease type 7	
PIGA*	X	X-linked	Multiple congenital anomalies-hypotonia-seizures syndrome type 2; Ferro-cerebro-cutaneous syndrome	
PKHD1	6	Autosomal recessive	Polycystic kidney disease type 4	
PLA2G6	22	Autosomal recessive	Infantile neuroaxonal dystrophy 1; Dystonia-parkinsonism type Paisan-Ruiz; Neurodegeneration with brain iron accumulation type 2B	
PLOD1	1	Autosomal recessive	Ehlers-Danlos syndrome type 6	
PMM2	16	Autosomal recessive	Congenital disorder of glycosylation type 1A	
PNKP	19	Autosomal recessive	Ataxia-oculomotor apraxia type 4; Early infantile epileptic encephalopathy	
PNPLA6	19	Autosomal recessive	Spastic paraplegia type 39; Oliver-McFarlane syndrome; Boucher-Neuhäuser syndrome	
POLG	15	Autosomal recessive	POLG related disorders	
POLR1C	6	Autosomal recessive	Hypomyelinating leukodystrophy type 11; Treacher-Collins syndrome type 3	

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POLR3A	10	Autosomal recessive	Wiedemann-Rautenstrauch syndrome; Hypomyelinating leukodystrophy type 7	
POMGNT1	1	Autosomal recessive	Muscular dystrophy-dystroglycanopathy type 3A, 3B, 3C; Retinitis pigmentosa type 76	
PPA2	4	Autosomal recessive	Infantile sudden cardiac failure	
PPT1	1	Autosomal recessive	Neuronal ceroid lipofuscinosis type 1	
PQBP1	X	X-linked	Renpenning syndrome	
PRF1	10	Autosomal recessive	Familial hemophagocytic lymphohistiocytosis type 2	
PRPS1*	X	X-linked	PRPS1 related disorders	
RAD50	5	Autosomal recessive	Nijmegen breakage syndrome-like disorder	
RAG1	11	Autosomal recessive	Omenn syndrome; Combined immunodeficiency with granulomatosis; Severe combined immunodeficiency due to complete RAG1/2 deficiency	
RAPSN	11	Autosomal recessive	Fetal akinesia deformation sequence type 2; Congenital myasthenic syndrome type 11	
RARS2	6	Autosomal recessive	Pontocerebellar hypoplasia type 6	
RBM10	X	X-linked	TARP syndrome	

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**Chromosome

Juno Genetics España
Ronda Guillermo Marconi, 11-A , 1°22B
46980-Paterna, Valencia, Spain
www.junogenetics.es

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Gene	Chr**	Mode of inheritance	Disease	Comments
RDH12	14	Autosomal recessive	Leber congenital amaurosis type 13	
RECQL4	8	Autosomal recessive	Baller-Gerold syndrome; Rothmund-Thomson syndrome type 2; RAPADILINO syndrome	
RMRP	9	Autosomal recessive	Anauxetic dysplasia type 1; Metaphyseal dysplasia without hypotrichosis; Cartilage-hair hypoplasia	
RNASEH2B	13	Autosomal recessive	Aicardi-Goutières syndrome type 2	
RPGRIP1	14	Autosomal recessive	Cone rod dystrophy type 13; Leber congenital amaurosis type 6	
RPGRIP1L	16	Autosomal recessive	Meckel syndrome type 5; Joubert syndrome type 7	
RS1	X	X-linked	Retinoschisis	
RTEL1	20	Autosomal recessive	Dyskeratosis congenita type 5	
SAMD9	7	Autosomal recessive	Familial normophosphatemic tumoral calcinosis	
SBDS*	7	Autosomal recessive	Shwachman-Diamond syndrome type 1	
SCNN1B	16	Autosomal recessive	Pseudohypoaldosteronism type 1B2	
SCO2	22	Autosomal recessive	Myopia type 6	
SGCA	17	Autosomal recessive	Limb-girdle muscular dystrophy type 3	
SGSH	17	Autosomal recessive	Mucopolysaccharidosis type 3	
SH3TC2	5	Autosomal recessive	Charcot-Marie-Tooth disease type 4C	

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SLC16A2	X	X-linked	Allan-Herndon-Dudley syndrome	
SLC17A5	6	Autosomal recessive	Salla disease; Free sialic acid storage disease, infantile form	
SLC1A4	2	Autosomal recessive	Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome	
SLC22A5	5	Autosomal recessive	Systemic primary carnitine deficiency	
SLC25A13	7	Autosomal recessive	Citrullinemia type 2	
SLC26A2	5	Autosomal recessive	SLC26A2 related disorders	
SLC26A3	7	Autosomal recessive	Congenital chloride diarrhea	
SLC26A4	7	Autosomal recessive	Pendred syndrome; Deafness autosomal recessive type 4	
SLC37A4	11	Autosomal recessive	Glycogen storage disease Ib, Ic	
SLC45A2	5	Autosomal recessive	Oculocutaneous albinism type 4	
SLC6A8*	X	X-linked	Cerebral creatine deficiency syndrome type 1	
SLC7A7	14	Autosomal recessive	Lysinuric protein intolerance	
SMN1	5	Autosomal recessive	Spinal Muscular Atrophy	Only deletion of the exon 7 in SMN1 gene is included. No sequencing or deletion/duplication analysis is conducted in other regions of this gene. This test does not detect “silent” carriers of SMA, who have two copies of the SMN1 gene on one chromosome and none on the other.
SMPD1	11	Autosomal recessive	Niemann-Pick disease type A/B	

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SPG11	15	Autosomal recessive	Juvenile amyotrophic lateral sclerosis type 5; Charcot-Marie-Tooth disease type 2X; Spastic paraplegia type 11	
SURF1	9	Autosomal recessive	Charcot-Marie-Tooth disease type 4K; Isolated cytochrome C oxidase deficiency	
SYN1	X	X-linked	X-linked epilepsy-learning disabilities-behavior disorders syndrome; X-linked intellectual developmental disorder type 50	
SYP	X	X-linked	Intellectual developmental disorder type 96	
TAFAZZIN	X	X-linked	Barth syndrome	
TGM1	14	Autosomal recessive	Congenital ichthyosis type 1	
TMEM216	11	Autosomal recessive	Meckel syndrome type 2; Joubert syndrome type 2; Retinitis pigmentaria type 98	
TMEM67	8	Autosomal recessive	TMEM67 related disorders	
TPP1	11	Autosomal recessive	Ceroid lipofuscinosis neuronal tipo 2; Spinocerebellar ataxia type 7	
TRIM37	17	Autosomal recessive	Mulibrey nanism syndrome	
TRIT1	1	Autosomal recessive	Combined oxidative phosphorylation deficiency type 35	
TSEN54	17	Autosomal recessive	Pontocerebellar hypoplasia type 2 and 4	
TSFM	12	Autosomal recessive	Combined oxidative phosphorylation deficiency type 3	
TYR*	11	Autosomal recessive	Oculocutaneous albinism type 1A/1B	NM_000372.5:c.1205G>A, p.(Arg402Gln) hypomorphic variant is associated with milder clinical manifestations is not reported.

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TYRP1	9	Autosomal recessive	Oculocutaneous albinism type 3	
UBA5*	3	Autosomal recessive	Non-specific early-onset epileptic encephalopathy	
UGT1A1	2	Autosomal recessive	Crigler-Najjar syndrome type 1 and 2	Variants in the UGT1A1 gene associated with Gilbert syndrome are not reported.
UPF3B	X	X-linked	Syndromic intellectual developmental disorder type 14	
USH2A	1	Autosomal recessive	Usher syndrome type 2A; Retinitis pigmentosa type 39	
USP9X	X	X-linked	Intellectual developmental disorder type 99	
VARS2	6	Autosomal recessive	Combined oxidative phosphorylation defect type 20	
VPS13B	8	Autosomal recessive	Cohen syndrome	
WAS	X	X-linked	Wiskott-Aldrich syndrome; X-linked severe congenital neutropenia; X-linked thrombocytopenia	
WFS1	4	Autosomal recessive	Wolfram syndrome type 1	
XIAP*	X	X-linked	X-linked lymphoproliferative syndrome type 2	
ZIC3	X	X-linked	X-linked heterotaxia; VACTERL with hydrocephalus	
ZNF711	X	X-linked	X-linked intellectual developmental disorder type 97	
ZNHIT3	17	Autosomal recessive	PEHO syndrome	

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