

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

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

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<b>BBS2</b>	16	Autosomal recessive	Retinitis pigmentosa type 74; Bardet-Biedl syndrome type 2	
<b>BCKDHA</b>	19	Autosomal recessive	Maple syrup urine disease type 1A	
<b>BCKDHB</b>	6	Autosomal recessive	Maple syrup urine disease type 1B	
<b>BCS1L</b>	2	Autosomal recessive	GRACILE syndrome; Björnstad syndrome; Isolated complex III deficiency	
<b>BGN</b>	X	X-linked	Meester-Loeys syndrome; X-linked spondyloepimetaphyseal dysplasia	
<b>BRIP1</b>	17	Autosomal recessive	Fanconi anemia group J	
<b>BTD</b>	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.
<b>BTK</b>	X	X-linked	Isolated growth hormone deficiency type 3; X-linked agammaglobulinemia	
<b>CAPN3</b>	15	Autosomal recessive	Limb-girdle muscular dystrophy type 2A	
<b>CASK</b>	X	X-linked	FG syndrome type 4; X-linked intellectual developmental disorder, Najm type	
<b>CBS</b>	21	Autosomal recessive	Classical homocystinuria	
<b>CC2D2A</b>	4	Autosomal recessive	CC2D2A related disorders	
<b>CCDC103</b>	17	Autosomal recessive	Primary ciliary dyskinesia type 17	
<b>CCDC40</b>	17	Autosomal recessive	Primary ciliary dyskinesia type 15	
<b>CD40LG</b>	X	X-linked	Immunodeficiency with hyper-IgM type 1	
<b>CDH23</b>	10	Autosomal recessive	Usher syndrome type 1D; Deafness autosomal recessive type 12	

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<b>CEP152</b>	15	Autosomal recessive	Autosomal recessive primary microcephaly type 9; Seckel syndrome type 5	
<b>CEP290*</b>	12	Autosomal recessive	CEP290 related disorders	Deep intronic variant NM_025114.4:c.2991+1655A>G not included.
<b>CFI</b>	4	Autosomal recessive	Complement factor I deficiency	
<b>CFTR</b>	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.
<b>CHM</b>	X	X-linked	Choroideremia	
<b>CHRNE</b>	17	Autosomal recessive	Congenital myasthenic syndrome type 4A, 4B, 4C	
<b>CHRNA3</b>	2	Autosomal recessive	Lethal multiple pterygium syndrome; Escobar syndrome	
<b>CLCN1</b>	7	Autosomal recessive	Myotonia congenita	
<b>CLCN5</b>	X	X-linked	CLCN5 related disorders	
<b>CLRN1</b>	3	Autosomal recessive	Usher syndrome type 3A	
<b>CNGA3</b>	2	Autosomal recessive	Achromatopsia type 2	
<b>CNGB3</b>	8	Autosomal recessive	Achromatopsia type 3	
<b>COL18A1</b>	21	Autosomal recessive	Knobloch syndrome type 1	
<b>COL4A3</b>	2	Autosomal recessive	Alport syndrome type 3B	
<b>COL4A5</b>	X	X-linked	Alport syndrome type 1	
<b>COL6A2</b>	21	Autosomal recessive	Ullrich congenital muscular dystrophy type 1B; Bethlem muscular dystrophy type 1B	

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

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<b>CYP21A2</b>	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 with the gene duplication has been reported 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).
<b>CYP27A1</b>	2	Autosomal recessive	Cerebrotendinous xanthomatosis	
<b>CYP7B1</b>	8	Autosomal recessive	Spastic paraplegia type 5A; Congenital bile acid synthesis defect type 3	
<b>DCX</b>	X	X-linked	Lissencephaly type 1; Subcortical band heterotopia	
<b>DDX11</b>	12	Autosomal recessive	Warsaw breakage syndrome	
<b>DGAT1</b>	8	Autosomal recessive	Congenital chronic diarrhea with protein-losing enteropathy	
<b>DHCR7</b>	11	Autosomal recessive	Smith-Lemli-Opitz syndrome	

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

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

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<b>FMR1</b>	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.
<b>FOXP3</b>	X	X-linked	IPEX syndrome	
<b>FRAS1</b>	4	Autosomal recessive	Fraser syndrome type 1	
<b>G6PC1</b>	17	Autosomal recessive	Glycogen storage disease type 1A	
<b>GAA</b>	17	Autosomal recessive	Pompe disease	
<b>GALC</b>	14	Autosomal recessive	Krabbe disease	
<b>GALNS</b>	16	Autosomal recessive	Mucopolysaccharidosis type 4A	
<b>GALT</b>	9	Autosomal recessive	Galactosemia	
<b>GATM</b>	15	Autosomal recessive	Cerebral creatine deficiency syndrome type 3	
<b>GBE1</b>	3	Autosomal recessive	Glycogen storage disease type 4; Adult polyglucosan body disease	
<b>GCDH</b>	19	Autosomal recessive	Glutaricaciduria type 1	
<b>GDF1</b>	19	Autosomal recessive	Ivemark syndrome	
<b>GFPT1</b>	2	Autosomal recessive	Congenital myasthenic syndrome type 12	
<b>GJB2</b>	13	Autosomal recessive	Deafness autosomal recessive type 1A	Variants associated with a mild phenotype are not reported.
<b>GJB3</b>	1	Autosomal recessive	Erythrokeratoderma variabilis type 1	Variants associated with a mild phenotype are not reported.
<b>GJB6</b>	13	Autosomal recessive	Deafness autosomal recessive type 1B	Variants associated with a mild phenotype are not reported.
<b>GLA</b>	X	X-linked	Fabry disease	

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<b>GLB1</b>	3	Autosomal recessive	GM1 gangliosidosis type 1,2,3; Mucopolysaccharidosis type 4B	
<b>GLDC</b>	9	Autosomal recessive	Glycine encephalopathy type 1	
<b>GLE1</b>	9	Autosomal recessive	Lethal congenital contracture syndrome type 1; Arthrogryposis-anterior horn cell disease syndrome	
<b>GMPPB</b>	3	Autosomal recessive	Muscular dystrophy due to dystroglycanopathy type 14A, 14B ,14C	
<b>GPR143</b>	X	X-linked	X-linked recessive ocular albinism; Congenital Nystagmus type 6	
<b>GUCY2D</b>	17	Autosomal recessive	Leber congenital amaurosis type 1; Cone rod dystrophy type 6; Congenital stationary night blindness type 11	
<b>HADHA</b>	2	Autosomal recessive	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency; Mitochondrial trifunctional protein deficiency	
<b>HBA1-2</b>	16	Autosomal recessive	Alpha-thalassemia	Whole gene analysis is not performed. Included variants: - -MED ; --SEA ; --THAI ; - $\alpha$ 3.7 ; - $\alpha$ 4.2 ; - $\alpha$ 20.5 ; --FIL; Hb Constant Spring (NM_000517.4:c.427T>C)
<b>HBB</b>	11	Autosomal recessive	Beta-thalassemia; Sickle cell disease	
<b>HERC2</b>	15	Autosomal recessive	Developmental delay with autism spectrum disorder and gait instability	
<b>HEXA</b>	15	Autosomal recessive	Tay-Sachs disease	
<b>HEXB</b>	5	Autosomal recessive	Sandhoff disease	

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

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

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

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<b>NPHP1</b>	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.
<b>NPHP3</b>	3	Autosomal recessive	Nephronophthisis type 3; Renal-hepatic-pancreatic dysplasia type 1; Meckel syndrome type 7	
<b>NPHS1</b>	19	Autosomal recessive	Nephrotic syndrome type 1	
<b>NPHS2</b>	1	Autosomal recessive	Nephrotic syndrome type 2	
<b>NR2E3</b>	15	Autosomal recessive	Goldmann-Favre syndrome; Retinitis pigmentosa type 37	
<b>NYX</b>	X	X-linked	Congenital stationary night blindness type 1A	
<b>OCA2</b>	15	Autosomal recessive	Oculocutaneous albinism type 2	
<b>OCRL</b>	X	X-linked	Dent disease type 2; Lowe syndrome	
<b>OFD1</b>	X	X-linked	Joubert syndrome type 10	
<b>OPA1</b>	3	Autosomal recessive	Behr syndrome	
<b>OTC</b>	X	X-linked	Ornithine transcarbamylase deficiency	
<b>PAH</b>	12	Autosomal recessive	Phenylketonuria	
<b>PANK2</b>	20	Autosomal recessive	Pantothenate Kinase-Associated Neurodegeneration type 1	
<b>PCCB</b>	3	Autosomal recessive	Propionic acidemia	
<b>PCDH15</b>	10	Autosomal recessive	Usher syndrome type 1F; Deafness autosomal recessive type 23	

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

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\*\*Chromosome

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

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

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<b>SLC16A2</b>	X	X-linked	Allan-Herndon-Dudley syndrome	
<b>SLC17A5</b>	6	Autosomal recessive	Salla disease; Free sialic acid storage disease, infantile form	
<b>SLC1A4</b>	2	Autosomal recessive	Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome	
<b>SLC22A5</b>	5	Autosomal recessive	Systemic primary carnitine deficiency	
<b>SLC25A13</b>	7	Autosomal recessive	Citrullinemia type 2	
<b>SLC26A2</b>	5	Autosomal recessive	SLC26A2 related disorders	
<b>SLC26A3</b>	7	Autosomal recessive	Congenital chloride diarrhea	
<b>SLC26A4</b>	7	Autosomal recessive	Pendred syndrome; Deafness autosomal recessive type 4	
<b>SLC37A4</b>	11	Autosomal recessive	Glycogen storage disease Ib, Ic	
<b>SLC45A2</b>	5	Autosomal recessive	Oculocutaneous albinism type 4	
<b>SLC6A8*</b>	X	X-linked	Cerebral creatine deficiency syndrome type 1	
<b>SLC7A7</b>	14	Autosomal recessive	Lysinuric protein intolerance	
<b>SMN1</b>	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.
<b>SMPD1</b>	11	Autosomal recessive	Niemann-Pick disease type A/B	

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<b>SPG11</b>	15	Autosomal recessive	Juvenile amyotrophic lateral sclerosis type 5; Charcot-Marie-Tooth disease type 2X; Spastic paraplegia type 11	
<b>SURF1</b>	9	Autosomal recessive	Charcot-Marie-Tooth disease type 4K; Isolated cytochrome C oxidase deficiency	
<b>SYN1</b>	X	X-linked	X-linked epilepsy-learning disabilities-behavior disorders syndrome; X-linked intellectual developmental disorder type 50	
<b>SYP</b>	X	X-linked	Intellectual developmental disorder type 96	
<b>TAFAZZIN</b>	X	X-linked	Barth syndrome	
<b>TGM1</b>	14	Autosomal recessive	Congenital ichthyosis type 1	
<b>TMEM216</b>	11	Autosomal recessive	Meckel syndrome type 2; Joubert syndrome type 2; Retinitis pigmentaria type 98	
<b>TMEM67</b>	8	Autosomal recessive	TMEM67 related disorders	
<b>TPP1</b>	11	Autosomal recessive	Ceroid lipofuscinosis neuronal tipo 2; Spinocerebellar ataxia type 7	
<b>TRIM37</b>	17	Autosomal recessive	Mulibrey nanism syndrome	
<b>TRIT1</b>	1	Autosomal recessive	Combined oxidative phosphorylation deficiency type 35	
<b>TSEN54</b>	17	Autosomal recessive	Pontocerebellar hypoplasia type 2 and 4	
<b>TSFM</b>	12	Autosomal recessive	Combined oxidative phosphorylation deficiency type 3	
<b>TYR*</b>	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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<b>TYRP1</b>	9	Autosomal recessive	Oculocutaneous albinism type 3	
<b>UBA5*</b>	3	Autosomal recessive	Non-specific early-onset epileptic encephalopathy	
<b>UGT1A1</b>	2	Autosomal recessive	Crigler-Najjar syndrome type 1 and 2	Variants in the UGT1A1 gene associated with Gilbert syndrome are not reported.
<b>UPF3B</b>	X	X-linked	Syndromic intellectual developmental disorder type 14	
<b>USH2A</b>	1	Autosomal recessive	Usher syndrome type 2A; Retinitis pigmentosa type 39	
<b>USP9X</b>	X	X-linked	Intellectual developmental disorder type 99	
<b>VARs2</b>	6	Autosomal recessive	Combined oxidative phosphorylation defect type 20	
<b>VPS13B</b>	8	Autosomal recessive	Cohen syndrome	
<b>WAS</b>	X	X-linked	Wiskott-Aldrich syndrome; X-linked severe congenital neutropenia; X-linked thrombocytopenia	
<b>WFS1</b>	4	Autosomal recessive	Wolfram syndrome type 1	
<b>XIAP*</b>	X	X-linked	X-linked lymphoproliferative syndrome type 2	
<b>ZIC3</b>	X	X-linked	X-linked heterotaxia; VACTERL with hydrocephalus	
<b>ZNF711</b>	X	X-linked	X-linked intellectual developmental disorder type 97	
<b>ZNHIT3</b>	17	Autosomal recessive	PEHO syndrome	

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