Gene	Chr**	Mode of inheritance	Disease	Comments
ABCA3	16	Autosomal recessive	Interstitial lung disease due to ABCA3 deficiency	
ABCC6*	16	Autosomal recessive	Pseudoxanthoma elasticum; Generalized arterial calcification of infancy type 2	
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	
AGA	4	Autosomal recessive	Aspartylglucosaminuria	
AIFM1	X	X-linked	AIFM1 related disorders	
ALDOB	9	Autosomal recessive	Hereditary fructose intolerance	
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.
ASL	7	Autosomal recessive	Argininosuccinic aciduria	
ASPA	17	Autosomal recessive	Canavan disease	
ATP7B	13	Autosomal recessive	Wilson disease	
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.

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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	
CEP290*	12	Autosomal recessive	CEP290 related disorders	Deep intronic variant NM_025114.4:c.2991+1655A>G not included.
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	
CLCN1	7	Autosomal recessive	Myotonia congenita	
CNGA3	2	Autosomal recessive	Achromatopsia type 2	
CNGB3	8	Autosomal recessive	Achromatopsia type 3	
COL4A3	2	Autosomal recessive	Alport syndrome type 3B	
COL4A5	X	X-linked	Alport syndrome type 1	
COL7A1	3	Autosomal recessive	Dystrophic epidermolysis bullosa COL7A1-Related	
CPT2	1	Autosomal recessive	Carnitine palmitoyltransferase deficiency type 2	

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CRB1	1	Autosomal recessive	Leber congenital amaurosis type 8; Retinitis pigmentosa type 12	
CTC1	17	Autosomal recessive	Coats plus syndrome	
CYP1B1	2	Autosomal recessive	Congenital glaucoma type 3A; Anterior segment developmental anomaly	
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).
DCX	X	X-linked	Lissencephaly type 1; Subcortical band heterotopia	
DDX11	12	Autosomal recessive	Warsaw breakage syndrome	

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DHCR7	11	Autosomal recessive	Smith-Lemli-Opitz syndrome	
DLD	7	Autosomal recessive	Pyruvate dehydrogenase E3 deficiency	
DMD	X	X-linked	Duchenne muscular dystrophy; Becker muscular dystrophy	
DNAH5	5	Autosomal recessive	Primary ciliary dyskinesia type 3	
DPYD	1	Autosomal recessive	Dihydropyrimidine dehydrogenase deficiency	
DYNC2H1	11	Autosomal recessive	Short rib-polydactyly syndrome type 3	
EDA	X	X-linked	Hypohidrotic ectodermal dysplasia type 1	
ELP1	9	Autosomal recessive	Familial dysautonomia	
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	
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	

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GAA	17	Autosomal recessive	Pompe disease	
GALC	14	Autosomal recessive	Krabbe disease	
GALT	9	Autosomal recessive	Galactosemia	
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	
GLE1	9	Autosomal recessive	Lethal congenital contracture syndrome type 1; Arthrogryposis-anterior horn cell disease syndrome	
GPR143	X	X-linked	X-linked recessive ocular albinism; Congenital Nystagmus type 6	
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)

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HBB	11	Autosomal recessive	Beta-thalassemia; Sickle cell disease	
HEXA	15	Autosomal recessive	Tay-Sachs disease	
HYLS1	11	Autosomal recessive	Hydrolethalus Syndrome	
IDS*	X	X-linked	Mucopolysaccharidosis type 2	
IQSEC2	X	X-linked	X-linked intellectual developmental disorder 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	
MPL	1	Autosomal recessive	Congenital amegakaryocytic thrombocytopenia type 1	
MTM1	X	X-linked	X-linked centronuclear myopathy	
MYO15A	17	Autosomal recessive	Deafness autosomal recessive type 3	
NPHS1	19	Autosomal recessive	Nephrotic syndrome type 1	
OCA2	15	Autosomal recessive	Oculocutaneous albinism type 2	
OFD1	X	X-linked	Joubert syndrome type 10	
OTC	X	X-linked	Ornithine transcarbamylase deficiency	
PAH	12	Autosomal recessive	Phenylketonuria	
PKHD1	6	Autosomal recessive	Polycystic kidney disease type 4	
PMM2	16	Autosomal recessive	Congenital disorder of glycosylation type 1A	

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POLG	15	Autosomal recessive	POLG related disorders	
POLR3A	10	Autosomal recessive	Wiedemann-Rautenstrauch syndrome; Hypomyelinating leukodystrophy type 7	
PRPS1*	X	X-linked	PRPS1 related disorders	
RMRP	9	Autosomal recessive	Anauxetic dysplasia type 1; Metaphyseal dysplasia without hypotrichosis; Cartilage-hair hypoplasia	
RNASEH2B	13	Autosomal recessive	Aicardi-Goutières syndrome type 2	
RS1	X	X-linked	Retinoschisis	
SBDS*	7	Autosomal recessive	Shwachman-Diamond syndrome type 1	
SCO2	22	Autosomal recessive	Myopia type 6	
SLC22A5	5	Autosomal recessive	Systemic primary carnitine deficiency	
SLC25A13	7	Autosomal recessive	Citrullinemia type 2	
SLC26A4	7	Autosomal recessive	Pendred syndrome; Deafness autosomal recessive type 4	
SLC45A2	5	Autosomal recessive	Oculocutaneous albinism type 4	
SLC6A8*	X	X-linked	Cerebral creatine deficiency syndrome type 1	

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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	
SYN1	X	X-linked	X-linked epilepsy-learning disabilities-behavior disorders syndrome; X-linked intellectual developmental disorder type 50	
TAFAZZIN	X	X-linked	Barth syndrome	
TMEM67	8	Autosomal recessive	TMEM67 related disorders	
TRIM37	17	Autosomal recessive	Mulibrey nanism syndrome	
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.
UGT1A1	2	Autosomal recessive	Crigler-Najjar syndrome type 1 and 2	Variants in the UGT1A1 gene associated with Gilbert syndrome are not reported.
USH2A	1	Autosomal recessive	Usher syndrome type 2A; Retinitis pigmentosa type 39	

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WAS	X	X-linked	Wiskott-Aldrich syndrome; X-linked severe congenital neutropenia; X-linked thrombocytopenia	
XIAP*	X	X-linked	X-linked lymphoproliferative syndrome type 2	
ZIC3	X	X-linked	X-linked heterotaxia; VACTERL with hydrocephalus	

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