ELENCO GENI PANNELLI CARRIER

1. PANNELLO NEXT CARRIER BASIC (5 GENES - FEMALE)

Gene	OMIM gene	Phenotype	Inheritance
CFTR	602421	Cystic fibrosis	Autosomal recessive
GJB2	121011	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6	Autosomal recessive; Digenic inheritance (GJB3, GJB6)
SMN1	600354	Spinal muscular atrophy	Autosomal recessive
FRAX-A		X-fragile	x-linked
DMD		Duchenne-Becker	

2. PANNELLO NEXT CARRIER BASIC (3 GENES – MALE)

Gene	OMIM gene	Phenotype	Inheritance
CFTR	602421	Cystic fibrosis	Autosomal recessive
GJB2	121011	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6	Autosomal recessive; Digenic inheritance (GJB3, GJB6)
SMN1	600354	Spinal muscular atrophy	Autosomal recessive

3. PANNELLO NEXT CARRIER STANDARD (312 GENES)

Gene	OMIM gene	Phenotype	Inheritance	OMIM phenotype
AAAS	605378	Triple-A syndrome (achalasia-addisonianism-alacrimia)	Autosomal recessive	231550
ABCA12	607800	Ichthyosis, congenital, autosomal recessive, type 4A; ICAR, type 4B (harlequin)	Autosomal recessive	601277; 242500
ABCA4	601691	Stargardt disease type 1; Cone-rod dystrophy type 3	Autosomal recessive	248200; 604116
ABCB11	603201	Cholestasis, benign recurrent intrahepatic, type 2; Cholestasis, progressive familial intrahepatic, type 2	Autosomal recessive	605479; 601847
ABCC8	600509	Hyperinsulinemic hypoglycemia, type 1 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive	256450; 606176
ABCD4	603214	Methylmalonic aciduria and homocystinuria, cblJ type	Autosomal recessive	614857
ACAD9	611103	Acyl-CoA dehydrogenase 9 deficiency (mitochondrial complex I deficiency, nuclear, type 20)	Autosomal recessive	611126
ACADM	607008	Medium-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive	201450
ACADS	606885	Short-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive	201470
ACADSB	600301	Short/branched-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive	610006
ACADVL	609575	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	Autosomal recessive	201475
ACAT1	607809	Alpha-methylacetoacetic aciduria (3-ketothiolase deficiency)	Autosomal recessive	203750
ACSF3	614245	Combined malonic and methylmalonic aciduria	Autosomal recessive	614265
ADA	608958	Severe combined immunodeficiency due to adenosine deaminase deficiency (ADA)	Autosomal recessive	102700
ADGRV1	602851	Usher syndrome, type 2C	Autosomal recessive, digenic inheritance (PDZD7 gene)	605472
ADK	102750	Hypermethioninemia due to adenosine kinase deficiency	Autosomal recessive	614300
AGL	610860	Glycogen storage disease, type 3	Autosomal recessive	232400
AGXT	604285	Hyperoxaluria, primary, type 1	Autosomal recessive	259900
AHI1	608894	Joubert syndrome, type 3	Autosomal recessive	608629
AIPL1	604392	Leber congenital amaurosis, type 4	Autosomal recessive	604393
AIRE	607358	Autoimmune polyendocrinopathy syndrome, type 1	Autosomal recessive	240300
ALDH4A1	606811	Hyperprolinemia, type 2	Autosomal recessive	239510
ALDOB	612724	Fructose intolerance, hereditary	Autosomal recessive	229600
ALG1	605907	Congenital disorder of glycosylation, type 1K	Autosomal recessive	608540
ALG6	604566	Congenital disorder of glycosylation, type 1C	Autosomal recessive	603147
ALMS1	606844	Alstr”m syndrome	Autosomal recessive	203800
ALPL	171760	Hypophosphatasia, infantile/childhood	Autosomal recessive	241500; 241510
ARL13B	608922	Joubert syndrome type 8	Autosomal recessive	612291
ARSA	607574	Metachromatic leukodystrophy	Autosomal recessive	250100
ARSB	611542	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)	Autosomal recessive	253200
ASL	608310	Argininosuccinic aciduria	Autosomal recessive	207900
ASPA	608034	Canavan disease	Autosomal recessive	271900
ASS1	603470	Citrullinemia, type 1	Autosomal recessive	215700
ATM	607585	Ataxia-telangiectasia	Autosomal recessive	208900
ATP7B	606882	Wilson disease	Autosomal recessive	277900
AUH	600529	3-methylglutaconic aciduria, type 1	Autosomal recessive	250950
B4GALT1	137060	Congenital disorder of glycosylation, type 2D	Autosomal recessive	607091
BBS1	209901	Bardet-Biedl syndrome, type 1	Autosomal recessive; Digenic inheritance (BBS genes, ARL6)	209900
BBS10	610148	Bardet-Biedl syndrome, type 10	Autosomal recessive; Digenic inheritance (BBS genes)	615987
BBS12	610683	Bardet-Biedl syndrome, type 12	Autosomal recessive; Digenic inheritance (BBS genes)	615989
BBS2	606151	Bardet-Biedl syndrome, type 2	Autosomal recessive; Digenic inheritance (BBS genes)	615981
BCKDHA	608348	Maple syrup urine disease, type 1A	Autosomal recessive	248600
BCKDHB	248611	Maple syrup urine disease, type 1B	Autosomal recessive	248600
BCS1L	603647	BCS1L-related disorders, including Leigh syndrome	Autosomal recessive	256000
BLM	604610	Bloom syndrome	Autosomal recessive	210900
BTD	609019	Biotinidase deficiency	Autosomal recessive	253260
CA2	611492	Osteopetrosis with renal tubular acidosis (osteopetrosis, autosomal recessive, type 3)	Autosomal recessive	259730
CAPN3	114240	Limb-girdle muscular dystrophy, type 1 (LGMD R1)	Autosomal recessive	253600
CASQ2	114251	Ventricular tachycardia, catecholaminergic polymorphic, type 2	Autosomal recessive	611938

Gene	OMIM gene	Phenotype	Inheritance	OMIM phenotype
CBS	613381	Homocystinuria due to cystathionine beta-synthase	Autosomal recessive	236200
CDH23	605516	Deafness, autosomal recessive, type 12; Usher syndrome, type 1D; Usher syndrome type 1D/F digenic	Autosomal recessive, Digenic recessive (PCDH15)	601386; 601067
CEP290	610142	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10	Autosomal recessive	611134; 610188; 611755
CERKL	608381	Retinitis pigmentosa, type 26	Autosomal recessive	608380
CFTR	602421	Cystic fibrosis	Autosomal recessive	219700
CHAT	118490	Myasthenic syndrome, congenital, type 6, presynaptic	Autosomal recessive	254210
CHRNE	100725	Myasthenic syndrome, congenital, type 4B, fast-channel; Myasthenic syndrome, congenital, type 4C, associated with acetylcholine receptor deficiency	Autosomal recessive	616324; 608931
CHRNG	100730	Multiple pterygium syndrome (MPS), Escobar type; MPS, lethal type	Autosomal recessive	265000; 253290
CHST6	605294	Macular corneal dystrophy	Autosomal recessive	217800
CLCN1	118425	Myotonia congenita, recessive	Autosomal recessive	255700
CLN3	607042	Ceroid lipofuscinosis, neuronal, type 3	Autosomal recessive	204200
CLRN1	606397	Usher syndrome, type 3A	Autosomal recessive	276902
CNGA1	123825	Retinitis pigmentosa type 49	Autosomal recessive	613756
CNGB3	605080	Achromatopsia, type 3	Autosomal recessive	262300
COL27A1	608461	Steel syndrome	Autosomal recessive	615155
COL4A3	120070	Alport syndrome, autosomal recessive, type 2	Autosomal recessive; Digenic inheritance (COL4A4 gene)	203780
COL4A4	120131	Alport syndrome, autosomal recessive, type 2	Autosomal recessive; Digenic inheritance (COL4A3 gene)	203780
COL7A1	120120	Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial	Autosomal recessive; Autosomal recessive; Autosomal recessive	226600; 604129; 131850
COX15	603646	Mitochondrial complex IV deficiency, nuclear type 6	Autosomal recessive	615119; 256000
CPS1	608307	Carbamoylphosphate synthetase 1 deficiency	Autosomal recessive	237300
CPT2	600650	Carnitine palmitoyltransferase type 2 deficiency, lethal neonatal; Carnitine palmitoyltransferase type 2 deficiency, infantile	Autosomal recessive	608836; 600649
CRB1	604210	Retinitis pigmentosa, type 12; Leber congenital amaurosis, type 8	Autosomal recessive	600105; 613835
CTH	607657	Cystathioninuria	Autosomal recessive	219500
CTNS	606272	Nephropathic cystinosis	Autosomal recessive	219800
CTSA	613111	Galactosialidosis	Autosomal recessive	256540
CTSC	602365	Haim-Munk syndrome; Papillon-Lefevre syndrome	Autosomal recessive	245010; 245000
CYP17A1	609300	17 alpha()-hydroxylase/17,20-lyase deficiency	Autosomal recessive	202110
CYP1B1	601771	Glaucoma, primary congenital, type 3A	Autosomal recessive	231300
CYP21A2	613815	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	Autosomal recessive	201910
CYP27A1	606530	Cerebrotendinous xanthomatosis	Autosomal recessive	213700
DBT	248610	Maple syrup urine disease, type 2	Autosomal recessive	248600
DHCR7	602858	Smith-Lemli-Opitz syndrome	Autosomal recessive	270400
DMD	300377	Duchenne/Becker muscular dystrophy	X-linked	310200; 300376
DNAH5	603335	Ciliary dyskinesia, primary, type 3, with or without situs inversus	Autosomal recessive	608644
DNAI1	604366	Ciliary dyskinesia, primary, type 1, with or without situs inversus	Autosomal recessive	244400
DNAJC12	606060	Hyperphenylalaninemia, mild, non-BH4-deficient	Autosomal recessive	617384
DOK7	610285	Fetal akinesia deformation sequence, type 3; Myasthenic syndrome, congenital, type 10	Autosomal recessive	618389; 254300
DOLK	610746	Congenital disorder of glycosylation, type 1M	Autosomal recessive	610768
DPAGT1	191350	Congenital disorder of glycosylation, type 1J; Myasthenic syndrome, congenital, type 13	Autosomal recessive	608093; 614750
DPM1	603503	Congenital disorder of glycosylation, type 1E	Autosomal recessive	608799
DPYD	612779	Dihydropyrimidine dehydrogenase deficiency	Autosomal recessive	274270
DUOX2	606759	Thyroid dyshormonogenesis, type 6	Autosomal recessive	607200
DUOXA2	612772	Thyroid dyshormonogenesis, type 5	Autosomal recessive	274900
DYSF	603009	Miyoshi muscular dystrophy, type 1; Limb-girdle muscular dystrophy, type 2 (LGMD R2)	Autosomal recessive	254130; 253601
EDA	300451	Ectodermal dysplasia, type 1, hypohidrotic, X-linked	X-linked	305100
EDAR	604095	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type	Autosomal recessive	224900
EIF2AK3	604032	Wolcott-Rallison syndrome	Autosomal recessive	226980
EIF2B5	603945	Leukoencephalopathy with vanishing white matter	Autosomal recessive	603896
ELP1	603722	Familial dysautonomia	Autosomal recessive	223900
ERCC2	126340	Trichothiodystrophy, type 1	Autosomal recessive	601675
ERCC3	133510	Trichothiodystrophy, type 2	Autosomal recessive	616390
ERCC6	609413	Cockayne syndrome, type B; Cerebrooculofacioskeletal syndrome, type 1	Autosomal recessive	133540; 214150
ESCO2	609353	Roberts syndrome	Autosomal recessive	268300
ETFDH	231675	Glutaric acidemia, type 2C	Autosomal recessive	231680
EVC	604831	Ellis-van Creveld syndrome	Autosomal recessive	225500
EVC2	607261	Ellis-van Creveld syndrome	Autosomal recessive	225500
EXOSC3	606489	Pontocerebellar hypoplasia, type 1B	Autosomal recessive	614678
EYS	612424	Retinitis pigmentosa, type 25	Autosomal recessive	602772
F11	264900	Factor XI deficiency	Autosomal recessive	612416
F2	176930	Prothrombin deficiency	Autosomal recessive	613679
F5	612309	Factor V deficiency	Autosomal recessive	227400
F8	300841	Hemophilia A	X-linked	306700
FAH	613871	Tyrosinemia, type 1	Autosomal recessive	276700
FAM161A	613596	Retinitis pigmentosa, type 28	Autosomal recessive	606068
FAM20C	611061	Raine syndrome	Autosomal recessive	259775
FANCA	607139	Fanconi anemia, complementation group A	Autosomal recessive	227650
FANCC	613899	Fanconi anemia, complementation group C	Autosomal recessive	227645
FANCG	602956	Fanconi anemia, complementation group G	Autosomal recessive	614082
FH	136850	Fumarase deficiency	Autosomal recessive	606812

Gene	OMIM gene	Phenotype	Inheritance	OMIM phenotype
FKRP	606596	Muscular dystrophy-dystroglycanopathy, type 5A (Walker-Warburg syndrome); Type 5B; Type 5C (limb-girdle muscular dystrophy, type 9 [LGMDR9])	Autosomal recessive	613153; 606612; 607155
FMR1	309550	Fragile X syndrome	X-linked	300624
FOXRED1	613622	Mitochondrial complex I deficiency, nuclear type 19	Autosomal recessive	618241
FRAS1	607830	Fraser syndrome, type 1	Autosomal recessive	219000
FTCD	606806	Glutamate formiminotransferase deficiency	Autosomal recessive	229100
G6PC1	613742	Glycogen storage disease, type 1A	Autosomal recessive	232200
G6PC3	611045	Dursun syndrome	Autosomal recessive	612541
G6PD	305900	Hemolytic anemia, G6PD deficient (favism)	X-linked	300908
GAA	606800	Glycogen storage disease, type 2	Autosomal recessive	232300
GALC	606890	Krabbe disease	Autosomal recessive	245200
GALNS	612222	Mucopolysaccharidosis, type 4A	Autosomal recessive	253000
GALT	606999	Galactosemia	Autosomal recessive	230400
GAMT	601240	Cerebral creatine deficiency syndrome, type 2	Autosomal recessive	612736
GBA	606463	Gaucher disease	Autosomal recessive	230800
GBE1	607839	Glycogen storage disease, type 4	Autosomal recessive	232500
GCDH	608801	Glutaricaciduria, type 1	Autosomal recessive	231670
GCH1	600225	Hyperphenylalaninemia, BH4-deficient, type B	Autosomal recessive	233910
GDAP1	606598	Charcot-Marie-Tooth disease, recessive intermediate, type A	Autosomal recessive	608340
GFM1	606639	Combined oxidative phosphorylation deficiency, type 1	Autosomal recessive	609060
GHRHR	139191	Growth hormone deficiency, isolated, type 1B	Autosomal recessive	612781
GJB2	121011	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6	Autosomal recessive; Digenic inheritance (GJB3, GJB6)	220290
GJB6	604418	Deafness, autosomal recessive, type 1B; Deafness, digenic GJB2/GJB6	Autosomal recessive; Digenic inheritance (GJB2 gene)	612645; 220290
GLB1	611458	GM1-gangliosidosis, types 1-3; Mucopolysaccharidosis, type 4B (Morquio)	Autosomal recessive	230500, 230600, 230650; 253010
GLDC	238300	Glycine encephalopathy	Autosomal recessive	605899
GLE1	603371	Lethal congenital contracture syndrome, type 1; Congenital arthrogryposis with anterior horn cell disease	Autosomal recessive	253310; 611890
GNE	603824	Inclusion body myopathy, type 2 (Nonaka myopathy)	Autosomal recessive	605820
GNMT	606628	Glycine N-methyltransferase deficiency	Autosomal recessive	606664
GNPTAB	607840	Mucolipidosis 2 alpha/beta; Mucolipidosis 3 alpha/beta	Autosomal recessive	252500; 252600
GNRHR	138850	Hypogonadotropic hypogonadism, type 7, without anosmia	Autosomal recessive	146110
GP1BB	138720	Bernard-Soulier syndrome, type B	Autosomal recessive	231200
GP9	173515	Bernard-Soulier syndrome, type C	Autosomal recessive	231200
GRHPR	604296	Hyperoxaluria, primary, type 2	Autosomal recessive	260000
GUCY2D	600179	Leber congenital amaurosis, type 1	Autosomal recessive	204000
GUSB	611499	Mucopolysaccharidosis, type 7	Autosomal recessive	253220
HADH	601609	3-hydroxyacyl-CoA dehydrogenase deficiency	Autosomal recessive	231530
HADHA	600890	Long-chain 3-hydroxyl-CoA dehydrogenase (LCHAD) deficiency; Mitochondrial trifunctional protein deficiency	Autosomal recessive	609016; 609015
HAX1	605998	Neutropenia, severe congenital, type 3, autosomal recessive	Autosomal recessive	610738
HBA1	141800	Thalassemia, alpha-	Autosomal recessive	604131
HBA2	141850	Thalassemia, alpha-	Autosomal recessive	604131
HBB	141900	HBB-related hemoglobinopathy	Autosomal recessive	603903
HEXA	606869	Tay-Sachs disease	Autosomal recessive	272800
HEXB	606873	Sandhoff disease, infantile, juvenile, and adult forms	Autosomal recessive	268800
HGD	607474	Alkaptonuria	Autosomal recessive	203500
HGSNAT	610453	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)	Autosomal recessive	252930
HMOX1	141250	Heme oxygenase-1 deficiency	Autosomal recessive	614034
HOGA1	613597	Hyperoxaluria, primary, type 3	Autosomal recessive	613616
HPS1	604982	Hermansky-Pudlak syndrome, type 1	Autosomal recessive	203300
HPS3	606118	Hermansky-Pudlak syndrome, type 3	Autosomal recessive	614072
HSD17B3	605573	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	Autosomal recessive	264300
HSD17B4	601860	D-bifunctional protein deficiency	Autosomal recessive	261515
HSPG2	142461	Dyssegmental dysplasia, Silverman-Handmaker type	Autosomal recessive	224410
HYLS1	610693	Hydrolethalus syndrome	Autosomal recessive	236680
IDH3B	604526	Retinitis pigmentosa, type 46	Autosomal recessive	612572
IDUA	252800	Mucopolysaccharidosis type 1	Autosomal recessive	607014; 607015; 607016
IGHMBP2	600502	Charcot-Marie-Tooth disease, axonal, type 2S	Autosomal recessive	616155
IVD	607036	Isovaleric acidemia	Autosomal recessive	243500
IYD	612025	Thyroid dyshormonogenesis, type 4	Autosomal recessive	274800
JAK3	600173	Severe Combined Immunodeficiency, autosomal recessive, T-negative/B-positive type	Autosomal recessive	600802
KCNJ11	600937	Hyperinsulinemic hypoglycemia, type 2 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive; Autosomal recessive	601820; 606176
LAMA2	156225	LAMA2-related muscular dystrophy	Autosomal recessive	607855; 618138
LAMA3	600805	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive	226700; 226650
LAMB3	150310	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive	226700; 226650
LARGE1	603590	Muscular dystrophy-dystroglycanopathy, type 6A and 6B	Autosomal recessive	613154; 608840
LHCGR	152790	Leydig cell hypoplasia	Autosomal recessive	238320
LIPA	613497	Lysosomal acid lipase deficiency	Autosomal recessive	278000
LIPH	607365	Hypotrichosis, type 7 or woolly hair, autosomal recessive, type 2, with or without hypotrichosis	Autosomal recessive	604379
LMBRD1	612625	Methylmalonic aciduria and homocystinuria, cblF type	Autosomal recessive	277380
LOXHD1	613072	Deafness, autosomal recessive, type 77	Autosomal recessive	613079
LPL	609708	Lipoprotein lipase deficiency	Autosomal recessive	238600
MAN2B1	609458	Alpha-mannosidosis	Autosomal recessive	248500
MCCC1	609010	3-Methylcrotonyl-CoA carboxylase deficiency, type 1	Autosomal recessive	210200
MCCC2	609014	3-Methylcrotonyl-CoA carboxylase deficiency, type 2	Autosomal recessive	210210
MCEE	608419	Methylmalonyl-CoA epimerase deficiency	Autosomal recessive	251120
MEFV	608107	Familial Mediterranean fever	Autosomal recessive	249100
MFSD8	611124	Ceroid lipofuscinosis, neuronal, type 7	Autosomal recessive	610951
MKS1	609883	Bardet-Biedl syndrome type 13; Meckel syndrome, type 1; Joubert syndrome, type 28	Autosomal recessive	615990; 249000; 617121
MLYCD	606761	Malonyl-CoA decarboxylase deficiency	Autosomal recessive	248360
MMACHC	609831	Methylmalonic aciduria and homocystinuria, cblC type	Autosomal recessive, digenic inheritance (PRDX1 gene)	277400

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Gene	OMIM gene	Phenotype	Inheritance	OMIM phenotype
MMUT	609058	Methylmalonic aciduria, mut(0) type	Autosomal recessive	251000
MOGS	601336	Congenital disorder of glycosylation, type 2B	Autosomal recessive	606056
MPI	154550	Congenital disorder of glycosylation, type 1B	Autosomal recessive	602579
MPL	159530	Thrombocytopenia, congenital amegakaryocytic	Autosomal recessive	604498
MPV17	137960	Mitochondrial DNA depletion syndrome type 6 (hepatocerebral); Charcot-Marie-Tooth disease, axonal, type 2EE	Autosomal recessive	256810; 618400
MTMR2	603557	Charcot-Marie-Tooth disease, type 4B1	Autosomal recessive	601382
MTR	156570	Homocystinuria-megaloblastic anemia, cblG complementation type	Autosomal recessive	250940
MVK	251170	Mevalonic aciduria	Autosomal recessive	610377
MYO7A	276903	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2	Autosomal recessive	276900; 600060
NADK2	615787	2,4-dienoyl-CoA reductase deficiency	Autosomal recessive	616034
NAGLU	609701	Mucopolysaccharidosis, type 3B (Sanfilippo B)	Autosomal recessive	252920
NBN	602667	Nijmegen breakage syndrome	Autosomal recessive	251260
NCF1	608512	Chronic granulomatous disease, type 1	Autosomal recessive	233700
NDUFV1	161015	Mitochondrial complex I deficiency, nuclear type 4	Autosomal recessive	618225
NEB	161650	Nemaline myopathy type 2	Autosomal recessive	256030
NHP2	606470	Dyskeratosis congenita, autosomal recessive type 2	Autosomal recessive	613987
NLRP7	609661	Hydatidiform mole, recurrent, type 1	Autosomal recessive	231090
NOP10	606471	Dyskeratosis congenita, autosomal recessive type 1	Autosomal recessive	224230
NPC1	607623	Niemann-Pick disease, type C1	Autosomal recessive	257220
NPHP1	607100	Joubert syndrome type 4	Autosomal recessive	609583
NPHS1	602716	Nephrotic syndrome, type 1	Autosomal recessive	256300
NPHS2	604766	Nephrotic syndrome, type 2	Autosomal recessive	600995
NR2E3	604485	Enhanced S-cone syndrome (Goldmann-Favre); Retinitis pigmentosa, type 37	Autosomal recessive; Autosomal recessive	268100; 611131
OCA2	611409	Oculocutaneous albinism type 2	Autosomal recessive	203200
OTOF	603681	Deafness, autosomal recessive, type 9	Autosomal recessive	601071
P3H1	610339	Osteogenesis imperfecta, type 8	Autosomal recessive	610915
PAH	612349	Phenylketonuria	Autosomal recessive	261600
PC	608786	Pyruvate carboxylase deficiency	Autosomal recessive	266150
PCDH15	605514	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic	Autosomal recessive; Digenic inheritance (CDH23 gene)	609533; 601067
PDE6A	180071	Retinitis pigmentosa type 43	Autosomal recessive	613810
PEX1	602136	Heimler syndrome type 1	Autosomal recessive	234580
PEX6	601498	Peroxisome biogenesis disorder, type 4A (Zellweger syndrome); Peroxisome biogenesis disorder, type 4B; Heimler syndrome 2	Autosomal recessive; Autosomal recessive; Autosomal recessive	614862; 616617; 614863
PEX7	601757	Rhizomelic chondrodysplasia punctata, type 1	Autosomal recessive	215100
PHGDH	606879	Neu-Laxova syndrome, type 1; Phosphoglycerate dehydrogenase deficiency	Autosomal recessive	256520; 601815
PKHD1	606702	Polycystic kidney disease type 4	Autosomal recessive	263200
PLA2G6	603604	Infantile neuroaxonal dystrophy type 1	Autosomal recessive	256600
PLOD1	153454	Ehlers-Danlos syndrome, kyphoscoliotic type, 1	Autosomal recessive	225400
PMM2	601785	Congenital disorder of glycosylation, type 1A	Autosomal recessive	212065
POLG	174763	POLG-related disorders	Autosomal recessive	203700; 613662; 607459
POMGNT1	606822	Muscular dystrophy-dystroglycanopathy, type 3A (Walker-Warburg syndrome); Type 3B; Type 3C (limb-girdle muscular dystrophy, type 15 [LGMDR15])	Autosomal recessive	253280; 613151; 613157
POMT1	607423	Muscular dystrophy-dystroglycanopathy, type 1A (Walker-Warburg syndrome); Type 1B; Type 1C (limb-girdle muscular dystrophy, type 11 [LGMD R11])	Autosomal recessive	236670; 613155; 609308
POR	124015	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	Autosomal recessive	201750
POU1F1	173110	Pituitary hormone deficiency, combined, type 1	Autosomal recessive	613038
PPT1	600722	Ceroid lipofuscinosis, neuronal, type 1	Autosomal recessive	256730
PRF1	170280	Hemophagocytic lymphohistiocytosis, familial, type 2	Autosomal recessive	603553
PROP1	601538	Pituitary hormone deficiency, combined, type 2	Autosomal recessive	262600
PTS	612719	Hyperphenylalaninemia, BH4-deficient, type A	Autosomal recessive	261640
PYGM	608455	McArdle disease	Autosomal recessive	232600
RAG1	179615	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	Autosomal recessive	603554; 601457
RAPSN	601592	Fetal akinesia deformation sequence, type 2; Myasthenic syndrome, congenital, type 11, associated with AChR deficiency	Autosomal recessive	208150; 616326
RARS2	611524	Pontocerebellar hypoplasia, type 6	Autosomal recessive	611523
RAX	601881	Isolated microphthalmia, type 3	Autosomal recessive	611038
RDH12	608830	Leber congenital amaurosis, type 13	Autosomal recessive	612712
RMRP	157660	Anauxetic dysplasia, type 1	Autosomal recessive	607095
RPE65	180069	RPE65-related Leberÿcongenitalÿamaurosis/early-onset severe retinal dystrophyÿ	Autosomal recessive	204100; 613794
RPGRIP1L	610937	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome	Autosomal recessive	611560; 611561; 619113
SACS	604490	Spastic ataxia, Charlevoix-Saguenay, type	Autosomal recessive	270550
SAMHD1	606754	Aicardi-Goutieres syndrome, type 5	Autosomal recessive	612952
SBDS	607444	Shwachman-Diamond syndrome	Autosomal recessive	260400
SERPINA1	107400	Alpha-1 antitrypsin deficiency	Autosomal recessive	613490
SGCA	600119	Limb-girdle muscular dystrophy, type 3 (LGMD R3)	Autosomal recessive	608099
SGCB	600900	Limb-girdle muscular dystrophy, type 4 (LGMD R4)	Autosomal recessive	604286
SGSH	605270	Mucopolysaccharidosis, type 3A (Sanfilippo A)	Autosomal recessive	252900
SH3TC2	608206	Charcot-Marie-Tooth disease, type 4C	Autosomal recessive	601596
SLC12A3	600968	Gitelman syndrome	Autosomal recessive	263800

Gene	OMIM gene	Phenotype	Inheritance	OMIM phenotype
SLC17A5	604322	Salla disease	Autosomal recessive	604369
SLC19A2	603941	Thiamine-responsive megaloblastic anemia syndrome	Autosomal recessive	249270
SLC22A5	603377	Carnitine deficiency, systemic primary	Autosomal recessive	212140
SLC25A13	603859	Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset	Autosomal recessive	605814; 603471
SLC26A2	606718	Achondrogenesis, type 1B (diastrophic dysplasia)	Autosomal recessive	600972
SLC26A4	605646	Deafness, autosomal recessive, type 4; Pendred syndrome	Autosomal recessive; Digenic inheritance (KCNJ10 gene)	600791; 274600
SLC35A1	605634	Congenital disorder of glycosylation, type 2F	Autosomal recessive	603585
SLC37A4	602671	Glycogen storage disease, type 1B	Autosomal recessive	232220
SLC39A4	607059	Acrodermatitis enteropathica	Autosomal recessive	201100
SLC3A1	104614	Cystinuria	Autosomal recessive	220100
SLC45A2	606202	Albinism, oculocutaneous, type 4	Autosomal recessive	606574
SLC5A5	601843	Thyroid dyshormonogenesis, type 1	Autosomal recessive	274400
SLC6A19	608893	Hartnup disorder	Autosomal recessive	234500
SLC7A7	603593	Lysinuric protein intolerance	Autosomal recessive	222700
SLC7A9	604144	Cystinuria	Autosomal recessive	220100
SMARCAL1	606622	Schimke immunoosseous dysplasia	Autosomal recessive	242900
SMN1	600354	Spinal muscular atrophy	Autosomal recessive	253300
SMPD1	607608	Niemann-Pick disease, type A; Niemann-Pick disease, type B	Autosomal recessive	257200; 607616
SPG11	610844	Amyotrophic lateral sclerosis, type 5, juvenile	Autosomal recessive	602099
SPG7	602783	Spastic paraplegia, type 7, autosomal recessive	Autosomal recessive	607259
SURF1	185620	Charcot-Marie-Tooth disease, type 4K; Leigh syndrome, due to COX IV deficiency	Autosomal recessive	616684; 256000
TCIRG1	604592	Osteopetrosis, autosomal recessive, type 1	Autosomal recessive	259700
TFR2	604720	Hemochromatosis, type 3	Autosomal recessive	604250
TG	188450	Thyroid dyshormonogenesis, type 3	Autosomal recessive	274700
TGM1	190195	Ichthyosis, congenital, autosomal recessive, type 1	Autosomal recessive	242300
TMEM67	609884	Joubert syndrome, type 6; Meckel syndrome, type 3; COACH syndrome	Autosomal recessive	610688; 607361; 216360
TPO	606765	Thyroid dyshormonogenesis, type 2A	Autosomal recessive	274500
TPP1	607998	Ceroid lipofuscinosis, neuronal, type 2; Spinocerebellar ataxia, autosomal recessive, type 7	Autosomal recessive	204500; 609270
TRDN	603283	Ventricular tachycardia, catecholaminergic polymorphic, type 5, with or without muscle weakness	Autosomal recessive	615441
TREX1	606609	Aicardi-Goutieres syndrome, type 1	Autosomal recessive	225750
TRIM32	602290	Limb-girdle muscular dystrophy, type 8 (LGMD R8)	Autosomal recessive	254110
TSEN54	608755	Pontocerebellar hypoplasia, type 2A; Pontocerebellar hypoplasia, type 4	Autosomal recessive	277470; 225753
TSFM	604723	Combined oxidative phosphorylation deficiency, type 3	Autosomal recessive	610505
TSHB	188540	Hypothyroidism, congenital, nongoitrous, type 4	Autosomal recessive	275100
TSHR	603372	Hypothyroidism, congenital, nongoitrous, type 1	Autosomal recessive	275200
TTC37	614589	Trichohepatoenteric syndrome, type 1 (diarrhea, syndromic)	Autosomal recessive	222470
TTPA	600415	Ataxia with isolated vitamin E deficiency	Autosomal recessive	277460
TYMP	131222	Mitochondrial DNA depletion syndrome, type 1 (MNGIE type)	Autosomal recessive	603041
TYR	606933	Oculocutaneous albinism (OCA) type 1A; OCA type 1B; Waardenburg syndrome/albinism, digenic	Autosomal recessive, digenic inheritance (MITF gene)	203100; 606952
TYRP1	115501	Albinism, oculocutaneous, type 3	Autosomal recessive	203290
UGT1A1	191740	Crigler-Najjar syndrome, type 1; Crigler-Najjar syndrome, type 2	Autosomal recessive	606785; 218800
UNC13D	608897	Hemophagocytic lymphohistiocytosis, familial, type 3	Autosomal recessive	608898
USH1C	605242	Usher syndrome, type 1C; Deafness, autosomal recessive, type 18A	Autosomal recessive	276904; 602092
USH2A	608400	Usher syndrome, type 2A	Autosomal recessive	276901
VPS13A	605978	Choreoacanthocytosis	Autosomal recessive	200150
VPS13B	607817	Cohen syndrome	Autosomal recessive	216550
WHRN	607928	Usher syndrome, type 2D; Deafness, autosomal recessive, type 31	Autosomal recessive	611383; 607084
WNT10A	606268	Odontoonychodermal dysplasia	Autosomal recessive	257980
WRN	604611	Werner syndrome	Autosomal recessive	277700
XPC	613208	Xeroderma pigmentosum, group C	Autosomal recessive	278720
ZFYVE26	612012	Spastic paraplegia, type 15, autosomal recessive	Autosomal recessive	270700

4. PANNELLO NEXT CARRIER COMPLETE (695 GENES - FEMALE)

Gene	OMIM gene	Phenotype PATOLOGIA EN	Inheritance
AAAS	605378	Triple-A syndrome (achalasia-addisonianism-alacrimia)	Autosomal recessive
AARS1	601065	Epileptic encephalopathy, early infantile, type 29	Autosomal recessive
AARS2	612035	Combined oxidative phosphorylation deficiency 8; Leukoencephalopathy, progressive, with ovarian failure	Autosomal recessive
ABCA12	607800	Ichthyosis, congenital, autosomal recessive, type 4A; ICAR, type 4B (harlequin)	Autosomal recessive
ABCA3	601615	Surfactant metabolism dysfunction, pulmonary, type 3	Autosomal recessive
ABCA4	601691	Stargardt disease type 1; Cone-rod dystrophy type 3	Autosomal recessive
ABCB11	603201	Cholestasis, benign recurrent intrahepatic, type 2; Cholestasis, progressive familial intrahepatic, type 2	Autosomal recessive
ABCB4	171060	Cholestasis, progressive familial intrahepatic, type 3	Autosomal recessive
ABCB7	300135	Anemia sideroblastic with ataxia	X-linked
ABCC6	603234	Pseudoxanthoma elasticum; Generalized arterial calcification of infancy, type 2	Autosomal recessive
ABCC8	600509	Hyperinsulinemic hypoglycemia, type 1 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive
ABCD1	300371	Adrenoleukodystrophy	X-linked
ABCD4	603214	Methylmalonic aciduria and homocystinuria, cblJ type	Autosomal recessive
ACAD8	604773	Isobutyryl-CoA dehydrogenase deficiency	Autosomal recessive
ACAD9	611103	Acyl-CoA dehydrogenase 9 deficiency (mitochondrial complex I deficiency, nuclear, type 20)	Autosomal recessive
ACADM	607008	Medium-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
ACADS	606885	Short-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
ACADSB	600301	Short/branched-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
ACADVL	609575	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	Autosomal recessive
ACAT1	607809	Alpha-methylacetoacetic aciduria (3-ketothiolase deficiency)	Autosomal recessive
ACO2	100850	Infantile cerebellar-retinal degeneration	Autosomal recessive
ACOX1	609751	Peroxisomal acyl-CoA oxidase deficiency	Autosomal recessive
ACP5	171640	Spondyloenchondrodysplasia with immune dysregulation	Autosomal recessive
ACSF3	614245	Combined malonic and methylmalonic aciduria	Autosomal recessive
ADA	608958	Severe combined immunodeficiency due to adenosine deaminase deficiency (ADA)	Autosomal recessive
ADAMTS10	608990	Weill-Marchesani syndrome, type 1, recessive	Autosomal recessive
ADAMTS2	604539	Ehlers-Danlos syndrome, dermatosparaxis type	Autosomal recessive
ADAMTSL2	612277	Geleophysic dysplasia type 1	Autosomal recessive
ADAT3	615302	Mental retardation, autosomal recessive 36	Autosomal recessive
ADGRG1	604110	Polymicrogyria, bilateral frontoparietal	Autosomal recessive
ADGRG6	612243	Lethal congenital contracture syndrome 9	Autosomal recessive
ADGRV1	602851	Usher syndrome, type 2C	Autosomal recessive, digenic inheritance (PDZD7 gene)
ADK	102750	Hypermethioninemia due to adenosine kinase deficiency	Autosomal recessive
AGA	613228	Aspartylglucosaminuria (glycosylasparaginase deficiency)	Autosomal recessive
AGL	610860	Glycogen storage disease, type 3	Autosomal recessive
AGPS	603051	Rhizomelic chondrodysplasia punctata, type 3	Autosomal recessive
AGXT	604285	Hyperoxaluria, primary, type 1	Autosomal recessive
AHCY	180960	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	Autosomal recessive
AHI1	608894	Joubert syndrome, type 3	Autosomal recessive
AIFM1	300169	Combined oxidative phosphorylation deficiency 6,Cowchock syndrome,Deafness X-linked 5,Spondyloepimetaphyseal dysplasia X-linked with hypomyelinating leukodystrophy	X-linked
AIMP1	603605	Leukodystrophy, hypomyelinating, type 3	Autosomal recessive
AIMP2	600859	Leukodystrophy, hypomyelinating, type 17	Autosomal recessive
AIPL1	604392	Leber congenital amaurosis, type 4	Autosomal recessive
AIRE	607358	Autoimmune polyendocrinopathy syndrome, type 1	Autosomal recessive
ALAS2	301300	Anemia sideroblastic 1,Protoporphyria erythropoietic X-linked	X-linked
ALDH3A2	609523	Sjogren-Larsson syndrome	Autosomal recessive
ALDH4A1	606811	Hyperprolinemia, type 2	Autosomal recessive
ALDH7A1	107323	Epilepsy, pyridoxine-dependent	Autosomal recessive
ALDOA	103850	Glycogen storage disease type 12	Autosomal recessive
ALDOB	612724	Fructose intolerance, hereditary	Autosomal recessive
ALG1	605907	Congenital disorder of glycosylation, type 1K	Autosomal recessive
ALG11	613666	Congenital disorder of glycosylation, type 1P	Autosomal recessive
ALG12	607144	Congenital disorder of glycosylation, type 1G	Autosomal recessive
ALG2	607905	Myasthenic syndrome, congenital, type 14, with tubular aggregates	Autosomal recessive
ALG3	608750	Congenital disorder of glycosylation, type 1D	Autosomal recessive
ALG6	604566	Congenital disorder of glycosylation, type 1C	Autosomal recessive
ALMS1	606844	Alstr"m syndrome	Autosomal recessive
ALPL	171760	Hypophosphatasia, infantile/childhood	Autosomal recessive
AMH	600957	Persistent Mullerian duct syndrome, type 1	Autosomal recessive
AMHR2	600956	Persistent Mullerian duct syndrome, type II	Autosomal recessive
AMPD2	102771	Pontocerebellar hypoplasia, type 9	Autosomal recessive
AMT	238310	Glycine encephalopathy	Autosomal recessive
ANO10	613726	Spinocerebellar ataxia, autosomal recessive, type 10	Autosomal recessive
ANOS1	300836	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1)	X-linked
AP1S1	603531	MEDNIK syndrome	Autosomal recessive
AP1S2	300629	Mental retardation, X-linked, syndromic, type 5 (Pettigrew syndrome)	X-linked
AP4B1	607245	Spastic paraplegia, type 47, autosomal recessive	Autosomal recessive
AP4E1	607244	Spastic paraplegia, type 51, autosomal recessive	Autosomal recessive
AP4M1	602296	Spastic paraplegia, type 50, autosomal recessive	Autosomal recessive
AP4S1	607243	Spastic paraplegia, type 52, autosomal recessive	Autosomal recessive
AP5Z1	613653	Spastic paraplegia, type 48, autosomal recessive	Autosomal recessive
AQP2	107777	Diabetes insipidus, nephrogenic, type 2	Autosomal recessive
AR	313700	Androgen insensitivity syndrome, complete	X-linked

Gene	OMIM gene	Phenotype PATOLOGIA EN	Inheritance
ARFGEF2	605371	Periventricular heterotopia with microcephaly	Autosomal recessive
ARG1	608313	Argininemia (arginase deficiency)	Autosomal recessive
ARHGEF9	300429	Developmental and epileptic encephalopathy 8	X-linked
ARL13B	608922	Joubert syndrome type 8	Autosomal recessive
ARSA	607574	Metachromatic leukodystrophy	Autosomal recessive
ARSB	611542	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)	Autosomal recessive
ARSL	300180	Chondrodysplasia punctata, brachytelephalangic	X-linked
ARV1	611647	Epileptic encephalopathy, early infantile, 38	Autosomal recessive
ARX	300382	Epileptic encephalopathy, early infantile, type 1; ARX-related developmental disorders	X-linked
ASL	608310	Argininosuccinic aciduria	Autosomal recessive
ASNS	108370	Asparagine synthetase deficiency	Autosomal recessive
ASPA	608034	Canavan disease	Autosomal recessive
ASPM	605481	Primary microcephaly type 5, autosomal recessive	Autosomal recessive
ASS1	603470	Citrullinemia, type 1	Autosomal recessive
ATM	607585	Ataxia-telangiectasia	Autosomal recessive
ATP6AP2	300556	Parkinsonism with spasticity X-linked,Congenital disorder of glycosylation type IIr,Intellectual developmental disorder X-linked syndromic Hedera type	X-linked
ATP6V1B1	192132	Renal tubular acidosis with deafness	Autosomal recessive
ATP7A	300011	Menkes disease; Occipital horn syndrome	X-linked
ATP7B	606882	Wilson disease	Autosomal recessive
ATP8B1	602397	Cholestasis, progressive familial intrahepatic, type 1; Cholestasis, benign recurrent intrahepatic, type 1	Autosomal recessive
ATR	601215	Seckel syndrome, type 1	Autosomal recessive
ATRX	300032	Mental retardation-hypotonic facies syndrome, X-linked; Alpha-thalassemia/mental retardation syndrome	X-linked
AUH	600529	3-methylglutaconic aciduria, type 1	Autosomal recessive
AVPR2	300538	Diabetes insipidus nephrogenic 1,Nephrogenic syndrome of inappropriate antidiuresis	X-linked
B4GALT1	137060	Congenital disorder of glycosylation, type 2D	Autosomal recessive
BBS1	209901	Bardet-Biedl syndrome, type 1	Autosomal recessive; Digenic inheritance (BBS genes, ARL6)
BBS10	610148	Bardet-Biedl syndrome, type 10	Autosomal recessive; Digenic inheritance (BBS genes)
BBS12	610683	Bardet-Biedl syndrome, type 12	Autosomal recessive; Digenic inheritance (BBS genes)
BBS2	606151	Bardet-Biedl syndrome, type 2	Autosomal recessive; Digenic inheritance (BBS genes)
BBS4	600374	Bardet-Biedl syndrome, type 4	Autosomal recessive; Digenic inheritance (BBS genes)
BBS9	607968	Bardet-Biedl syndrome, type 9	Autosomal recessive; Digenic inheritance (BBS genes)
BCAP31	300398	Deafness dystonia and cerebral hypomyelination	X-linked
BCHE	177400	Butyrylcholinesterase deficiency	Autosomal recessive
BCKDHA	608348	Maple syrup urine disease, type 1A	Autosomal recessive
BCKDHB	248611	Maple syrup urine disease, type 1B	Autosomal recessive
BCS1L	603647	BCS1L-related disorders, including Leigh syndrome	Autosomal recessive
BLM	604610	Bloom syndrome	Autosomal recessive
BRAT1	614506	Rigidity and multifocal seizure syndrome, lethal neonatal; Neurodevelopmental disorder with cerebellar atrophy and with or without seizures	Autosomal recessive
BRIP1	605882	Fanconi anemia, complementation group J	Autosomal recessive
BRWD3	300553	Mental retardation, X-linked, type 93	X-linked
BSND	606412	Bartter syndrome, type 4A	Autosomal recessive
BTD	609019	Biotinidase deficiency	Autosomal recessive
BTK	300300	Agammaglobulinemia X-linked, type 1	X-linked
BUB1B	602860	Mosaic variegated aneuploidy syndrome 1	Autosomal recessive
C2CD3	615944	Orofaciodigital syndrome, type 14	Autosomal recessive
CA2	611492	Osteopetrosis with renal tubular acidosis (osteopetrosis, autosomal recessive, type 3)	Autosomal recessive
CA8	114815	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3	Autosomal recessive
CABP2	607314	Deafness, autosomal recessive, type 93	Autosomal recessive
CACNA1F	300110	Aland Island eye disease,Cone-rod dystrophy X-linked 3,Night blindness congenital stationary (incomplete) 2A X-linked	X-linked
CAD	114010	Epileptic encephalopathy, early infantile, 50	Autosomal recessive
CANT1	613165	Desbuquois dysplasia, type 1; Epiphyseal dysplasia, multiple, type 7	Autosomal recessive
CAPN3	114240	Limb-girdle muscular dystrophy, type 1 (LGMD R1)	Autosomal recessive
CASK	300172	FG syndrome 4,Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia,Intellectual developmental disorder with or without nystagmus	X-linked
CASQ2	114251	Ventricular tachycardia, catecholaminergic polymorphic, type 2	Autosomal recessive
CBS	613381	Homocystinuria due to cystathionine beta-synthase	Autosomal recessive
CC2D1A	610055	Mental retardation, autosomal recessive, type 3	Autosomal recessive
CC2D2A	612013	Joubert syndrome, type 9; Meckel syndrome, type 6	Autosomal recessive
CCDC88C	611204	Hydrocephalus, congenital, type 1	Autosomal recessive
CCN6	603400	Arthropathy, progressive pseudorheumatoid, of childhood	Autosomal recessive
CD40LG	300386	Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)	X-linked
CDC14A	603504	Deafness, autosomal recessive, type 105	Autosomal recessive
CDC45	603465	Meier-Gorlin syndrome 7	Autosomal recessive
CDH23	605516	Deafness, autosomal recessive, type 12; Usher syndrome, type 1D; Usher syndrome type 1D/F digenic	Autosomal recessive, Digenic recessive (PCDH15)
CDH3	114021	Ectodermal dysplasia, ectrodactyly, and macular dystrophy	Autosomal recessive
CDK5RAP2	608201	Primary microcephaly type 3, autosomal recessive	Autosomal recessive
CENPJ	609279	Primary microcephaly type 6, autosomal recessive	Autosomal recessive
CEP290	610142	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10	Autosomal recessive
CERKL	608381	Retinitis pigmentosa, type 26	Autosomal recessive
CFP	300383	Properdin deficiency X-linked	X-linked
CFTR	602421	Cystic fibrosis	Autosomal recessive
CHAT	118490	Myasthenic syndrome, congenital, type 6, presynaptic	Autosomal recessive
CHM	300390	Choroideremia	X-linked
CHRDL1	300350	Megalocornea 1 X-linked	X-linked
CHRNE	100725	Myasthenic syndrome, congenital, type 4B, fast-channel; Myasthenic syndrome, congenital, type 4C, associated with acetylcholine receptor deficiency	Autosomal recessive

Gene	OMIM gene	Phenotype PATOLOGIA EN	Inheritance
CHRNG	100730	Multiple pterygium syndrome (MPS), Escobar type; MPS, lethal type	Autosomal recessive
CHST3	603799	Spondyloepiphyseal dysplasia with congenital joint dislocations	Autosomal recessive
CHST6	605294	Macular corneal dystrophy	Autosomal recessive
CIITA	600005	Bare lymphocyte syndrome, type 2, complementation group A	Autosomal recessive
CISD2	611507	Wolfram syndrome 2	Autosomal recessive
CIT	605629	Microcephaly 17, primary, autosomal recessive	Autosomal recessive
CLCN1	118425	Myotonia congenita, recessive	Autosomal recessive
CLCN2	600570	Leukoencephalopathy with ataxia	Autosomal recessive
CLCN5	300008	Dent disease 1,Hypophosphatemic rickets,Nephrolithiasis type I,Proteinuria low molecular weight with hypercalciuric nephrocalcinosis	X-linked
CLN3	607042	Ceroid lipofuscinosis, neuronal, type 3	Autosomal recessive
CLN5	608102	Ceroid lipofuscinosis, neuronal, type 5	Autosomal recessive
CLN6	606725	Ceroid lipofuscinosis, neuronal, type 6	Autosomal recessive
CLN8	607837	Ceroid lipofuscinosis, neuronal, type 8	Autosomal recessive
CLP1	608757	Pontocerebellar hypoplasia, type 10	Autosomal recessive
CLRN1	606397	Usher syndrome, type 3A	Autosomal recessive
CNGA1	123825	Retinitis pigmentosa type 49	Autosomal recessive
CNGA3	600053	Achromatopsia, type 2	Autosomal recessive
CNGB1	600724	Retinitis pigmentosa type 45	Autosomal recessive
CNGB3	605080	Achromatopsia, type 3	Autosomal recessive
CNKSR2	300724	Intellectual developmental disorder X-linked syndromic Houge type	X-linked
CNPY3	610774	Epileptic encephalopathy, early infantile, type 60	Autosomal recessive
COA8	616003	Mitochondrial complex IV deficiency, nuclear type 17	Autosomal recessive
COL11A2	120290	Otospondylomegaepiphyseal dysplasia, autosomal recessive	Autosomal recessive
COL27A1	608461	Steel syndrome	Autosomal recessive
COL4A3	120070	Alport syndrome, autosomal recessive, type 2	Autosomal recessive; Digenic inheritance (COL4A4 gene)
COL4A4	120131	Alport syndrome, autosomal recessive, type 2	Autosomal recessive; Digenic inheritance (COL4A3 gene)
COL4A5	303630	Alport syndrome, X-linked	X-linked
COL7A1	120120	Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial	Autosomal recessive; Autosomal recessive; Autosomal recessive
COLQ	603033	Myasthenic syndrome, congenital, type 5	Autosomal recessive
COQ2	609825	Primary coenzyme Q10 deficiency, type 1	Autosomal recessive
COX10	602125	Mitochondrial complex IV deficiency, nuclear type 3	Autosomal recessive
COX15	603646	Mitochondrial complex IV deficiency, nuclear type 6	Autosomal recessive
COX6B1	124089	Mitochondrial complex IV deficiency, nuclear type 7	Autosomal recessive
CPS1	608307	Carbamoylphosphate synthetase 1 deficiency	Autosomal recessive
CPT1A	600528	Carnitine palmitoyltransferase type 1A deficiency, hepatic	Autosomal recessive
CPT2	600650	Carnitine palmitoyltransferase type 2 deficiency, lethal neonatal; Carnitine palmitoyltransferase type 2 deficiency, infantile	Autosomal recessive
CRB1	604210	Retinitis pigmentosa, type 12; Leber congenital amaurosis, type 8	Autosomal recessive
CRTAP	605497	Osteogenesis imperfecta, type 7	Autosomal recessive
CTH	607657	Cystathioninuria	Autosomal recessive
CTNS	606272	Nephropathic cystinosis	Autosomal recessive
CTSA	613111	Galactosialidosis	Autosomal recessive
CTSC	602365	Haim-Munk syndrome; Papillon-Lefevre syndrome	Autosomal recessive
CTSD	116840	Ceroid lipofuscinosis, neuronal, type 10	Autosomal recessive
CTSK	601105	Pycnodysostosis	Autosomal recessive
CUL4B	300304	Mental retardation, X-linked, syndromic, type 15 (Cabezas type)	X-linked
CWF19L1	616120	Spinocerebellar ataxia, autosomal recessive, type 17	Autosomal recessive
CYBA	608508	Chronic granulomatous disease, type 4	Autosomal recessive
CYBB	300481	Chronic granulomatous disease, X-linked	X-linked
CYP11A1	118485	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency	Autosomal recessive
CYP11B1	610613	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	Autosomal recessive
CYP11B2	124080	Hypoaldosteronism, congenital, due to CMO I deficiency	Autosomal recessive
CYP17A1	609300	17 alpha()-hydroxylase/17,20-lyase deficiency	Autosomal recessive
CYP19A1	107910	Aromatase deficiency	Autosomal recessive
CYP1B1	601771	Glaucoma, primary congenital, type 3A	Autosomal recessive
CYP21A2	613815	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	Autosomal recessive
CYP26B1	605207	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies	Autosomal recessive
CYP27A1	606530	Cerebrotendinous xanthomatosis	Autosomal recessive
CYP27B1	609506	Vitamin D-dependent rickets, type 1	Autosomal recessive
DARS1	603084	Hypomyelination with brainstem and spinal cord involvement and leg spasticity	Autosomal recessive
DBT	248610	Maple syrup urine disease, type 2	Autosomal recessive
DCLRE1C	605988	Omenn syndrome; Severe combined immunodeficiency, Athabascan type	Autosomal recessive
DCX	300121	Lissencephaly, X-linked, type 1	X-linked
DDB2	600811	Xeroderma pigmentosum, complementation group E	Autosomal recessive
DDX3X	300160	Intellectual developmental disorder X-linked syndrome Snijders Blok type	X-linked
DHCR7	602858	Smith-Lemli-Opitz syndrome	Autosomal recessive
DHDDS	608172	Retinitis pigmentosa, type 59	Autosomal recessive
DHODH	126064	Miller syndrome	Autosomal recessive
DKC1	300126	Dyskeratosis congenita, X-linked	X-linked
DLD	238331	Dihydrolipoamide dehydrogenase deficiency	Autosomal recessive
DLG3	300189	Mental retardation, X-linked, type 90	X-linked
DMD	300377	Duchenne/Becker muscular dystrophy	X-linked
DMXL2	612186	Developmental and epileptic encephalopathy, type 81	Autosomal recessive
DNAH5	603335	Ciliary dyskinesia, primary, type 3, with or without situs inversus	Autosomal recessive
DNAI1	604366	Ciliary dyskinesia, primary, type 1, with or without situs inversus	Autosomal recessive
DNAI2	605483	Ciliary dyskinesia, primary, type 9, with or without situs inversus	Autosomal recessive

Gene	OMIM gene	Phenotype PATOLOGIA EN	Inheritance
DNAJC12	606060	Hyperphenylalaninemia, mild, non-BH4-deficient	Autosomal recessive
DNAL1	610062	Ciliary dyskinesia, primary, type 16	Autosomal recessive
DNM1L	603850	Encephalopathy due to defective mitochondrial and peroxisomal fission, type 1	Autosomal recessive
DNM2	602378	Lethal congenital contracture syndrome, type 5	Autosomal recessive
DOK7	610285	Fetal akinesia deformation sequence, type 3; Myasthenic syndrome, congenital, type 10	Autosomal recessive
DOLK	610746	Congenital disorder of glycosylation, type 1M	Autosomal recessive
DPAGT1	191350	Congenital disorder of glycosylation, type 1J; Myasthenic syndrome, congenital, type 13	Autosomal recessive
DPM1	603503	Congenital disorder of glycosylation, type 1E	Autosomal recessive
DPYD	612779	Dihydropyrimidine dehydrogenase deficiency	Autosomal recessive
DST	113810	Epidermolysis bullosa simplex, autosomal recessive, type 2	Autosomal recessive
DUOX2	606759	Thyroid dyshormonogenesis, type 6	Autosomal recessive
DUOXA2	612772	Thyroid dyshormonogenesis, type 5	Autosomal recessive
DYNC2H1	603297	Short-rib thoracic dysplasia, type 3, with or without polydactyly	Autosomal recessive; Digenic inheritance (NEK1 gene)
DYSF	603009	Miyoshi muscular dystrophy, type 1; Limb-girdle muscular dystrophy, type 2 (LGMD R2)	Autosomal recessive
EBP	300205	Chondrodysplasia punctata X-linked dominant,MEND syndrome	X-linked
EDA	300451	Ectodermal dysplasia, type 1, hypohidrotic, X-linked	X-linked
EDAR	604095	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type	Autosomal recessive
EIF2AK3	604032	Wolcott-Rallison syndrome	Autosomal recessive
EIF2B5	603945	Leukoencephalopathy with vanishing white matter	Autosomal recessive
ELP1	603722	Familial dysautonomia	Autosomal recessive
EMC1	616846	Cerebellar atrophy, visual impairment, and psychomotor retardation	Autosomal recessive
EMD	300384	Emery-Dreifuss muscular dystrophy, type 1, X-linked	X-linked
EPRS1	138295	Leukodystrophy, hypomyelinating, type 15	Autosomal recessive
ERCC2	126340	Trichothiodystrophy, type 1	Autosomal recessive
ERCC3	133510	Trichothiodystrophy, type 2	Autosomal recessive
ERCC4	133520	Fanconi anemia, complementation group Q	Autosomal recessive
ERCC5	133530	Cerebrooculofacioskeletal syndrome, type 3	Autosomal recessive
ERCC6	609413	Cockayne syndrome, type B; Cerebrooculofacioskeletal syndrome, type 1	Autosomal recessive
ERCC8	609412	Cockayne syndrome, type A	Autosomal recessive
ESCO2	609353	Roberts syndrome	Autosomal recessive
ETFA	608053	Glutaric acidemia, type 2A	Autosomal recessive
ETFB	130410	Glutaric acidemia, type 2B	Autosomal recessive
ETFDH	231675	Glutaric acidemia, type 2C	Autosomal recessive
ETHE1	608451	Ethylmalonic encephalopathy	Autosomal recessive
EVC	604831	Ellis-van Creveld syndrome	Autosomal recessive
EVC2	607261	Ellis-van Creveld syndrome	Autosomal recessive
EXOSC3	606489	Pontocerebellar hypoplasia, type 1B	Autosomal recessive
EYS	612424	Retinitis pigmentosa, type 25	Autosomal recessive
F11	264900	Factor XI deficiency	Autosomal recessive
F2	176930	Prothrombin deficiency	Autosomal recessive
F5	612309	Factor V deficiency	Autosomal recessive
F8	300841	Hemophilia A	X-linked
F9	300746	Hemophilia B	X-linked
FAH	613871	Tyrosinemia, type 1	Autosomal recessive
FAM161A	613596	Retinitis pigmentosa, type 28	Autosomal recessive
FAM20C	611061	Raine syndrome	Autosomal recessive
FANCA	607139	Fanconi anemia, complementation group A	Autosomal recessive
FANCB	300515	Fanconi anemia complementation group B	X-linked
FANCC	613899	Fanconi anemia, complementation group C	Autosomal recessive
FANCD2	613984	Fanconi anemia, complementation group D2	Autosomal recessive
FANCG	602956	Fanconi anemia, complementation group G	Autosomal recessive
FANCL	608111	Fanconi anemia, complementation group L	Autosomal recessive
FGD1	300546	Aarskog-Scott syndrome; Mental retardation, X-linked syndromic, type 16	X-linked
FH	136850	Fumarase deficiency	Autosomal recessive
FHL1	300163	Uruguay faciocardiomusculoskeletal syndrome,Emery-Dreifuss muscular dystrophy 6 X-linked,Myopathy X-linked with postural muscle atrophy,Reducing body myopathy X-linked 1a severe infantile or early childhood onset,Reducing body myopathy X-linked 1b with late childhood or adult onset,Scapuloperoneal myopathy X-linked dominant	X-linked
FKRP	606596	Muscular dystrophy-dystroglycanopathy, type 5A (Walker-Warburg syndrome); Type 5B; Type 5C (limb-girdle muscular dystrophy, type 9 [LGMDR9])	Autosomal recessive
FKTN	607440	Muscular dystrophy-dystroglycanopathy, type 4A (Walker-Warburg syndrome); Type 4B; Type 4C (limb-girdle muscular dystrophy, type 13 [LGMD R13])	Autosomal recessive
FMO3	136132	Trimethylaminuria	Autosomal recessive
FMR1	309550	Fragile X syndrome	X-linked
FOXP3	300292	Immunodysregulation polyendocrinopathy and enteropathy X-linked	X-linked
FOXRED1	613622	Mitochondrial complex I deficiency, nuclear type 19	Autosomal recessive
FRAS1	607830	Fraser syndrome, type 1	Autosomal recessive
FRMD7	300628	Nystagmus 1 congenital X-linked,Nystagmus infantile periodic alternating X-linked	X-linked
FRMPD4	300838	Intellectual developmental disorder X-linked 104	X-linked
FTCD	606806	Glutamate formiminotransferase deficiency	Autosomal recessive
FTSJ1	300499	Mental retardation, X-linked 44	X-linked
FUCA1	612280	Fucosidosis	Autosomal recessive
G6PC1	613742	Glycogen storage disease, type 1A	Autosomal recessive
G6PC3	611045	Dursun syndrome	Autosomal recessive
G6PD	305900	Hemolytic anemia, G6PD deficient (favism)	X-linked
GAA	606800	Glycogen storage disease, type 2	Autosomal recessive
GALC	606890	Krabbe disease	Autosomal recessive
GALE	606953	Galactose epimerase deficiency	Autosomal recessive

Gene	OMIM gene	Phenotype PATOLOGIA EN	Inheritance
GALK1	604313	Galactokinase deficiency with cataracts	Autosomal recessive
GALNS	612222	Mucopolysaccharidosis, type 4A	Autosomal recessive
GALNT3	601756	Tumoral calcinosis, hyperphosphatemic, familial, type 1	Autosomal recessive
GALT	606999	Galactosemia	Autosomal recessive
GAMT	601240	Cerebral creatine deficiency syndrome, type 2	Autosomal recessive
GATA1	305371	Anemia X-linked with/without neutropenia and/or platelet abnormalities,Thrombocytopenia with beta-thalassemia X-linked,Thrombocytopenia X-linked with or without dyserythropoietic anemia	X-linked
GBA	606463	Gaucher disease	Autosomal recessive
GBE1	607839	Glycogen storage disease, type 4	Autosomal recessive
GCDH	608801	Glutaricaciduria, type 1	Autosomal recessive
GCH1	600225	Hyperphenylalaninemia, BH4-deficient, type B	Autosomal recessive
GCSH	238330	Glycine encephalopathy	Autosomal recessive
GDAP1	606598	Charcot-Marie-Tooth disease, recessive intermediate, type A	Autosomal recessive
GDF5	601146	Chondrodysplasia, Grebe type	Autosomal recessive
GFM1	606639	Combined oxidative phosphorylation deficiency, type 1	Autosomal recessive
GH1	139250	Growth hormone deficiency, isolated, type 1A; Kowarski syndrome	Autosomal recessive
GHRHR	139191	Growth hormone deficiency, isolated, type 1B	Autosomal recessive
GJB1	304040	Charcot-Marie-Tooth neuropathy, X-linked dominant, type 1	X-linked
GJB2	121011	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6	Autosomal recessive; Digenic inheritance (GJB3, GJB6)
GJB3	603324	Deafness autosomal dominant 2B,Deafness autosomal dominant with peripheral neuropathy,Deafness autosomal recessive,Deafness digenic GJB2/GJB3,Erythrokeratodermia variabilis et progressiva 1	Autosomal recessive; Digenic inheritance (GJB2 gene)
GJB6	604418	Deafness, autosomal recessive, type 1B; Deafness, digenic GJB2/GJB6	Autosomal recessive; Digenic inheritance (GJB2 gene)
GK	300474	Glycerol kinase deficiency	X-linked
GLA	300644	Fabry disease	X-linked
GLB1	611458	GM1-gangliosidosis, types 1-3; Mucopolysaccharidosis, type 4B (Morquio)	Autosomal recessive
GLDC	238300	Glycine encephalopathy	Autosomal recessive
GLE1	603371	Lethal congenital contracture syndrome, type 1; Congenital arthrogryposis with anterior horn cell disease	Autosomal recessive
GNE	603824	Inclusion body myopathy, type 2 (Nonaka myopathy)	Autosomal recessive
GNMT	606628	Glycine N-methyltransferase deficiency	Autosomal recessive
GNPTAB	607840	Mucolipidosis 2 alpha/beta; Mucolipidosis 3 alpha/beta	Autosomal recessive
GNPTG	607838	Mucolipidosis III gamma	Autosomal recessive
GNRHR	138850	Hypogonadotropic hypogonadism, type 7, without anosmia	Autosomal recessive
GNS	607664	Mucopolysaccharidosis, type 3D (Sanfilippo syndrome D)	Autosomal recessive
GORAB	607983	Geroderma osteodysplasticum	Autosomal recessive
GOT2	138150	Epileptic encephalopathy, early infantile, 82	Autosomal recessive
GP1BA	606672	Bernard-Soulier syndrome, type A1	Autosomal recessive
GP1BB	138720	Bernard-Soulier syndrome, type B	Autosomal recessive
GP9	173515	Bernard-Soulier syndrome, type C	Autosomal recessive
GPC3	300037	Simpson-Golabi-Behmel syndrome type 1	X-linked
GPR143	300808	Ocular albinism, type 1 (Nettleship-Falls type)	X-linked
GPSM2	609245	Chudley-McCullough syndrome	Autosomal recessive
GRHPR	604296	Hyperoxaluria, primary, type 2	Autosomal recessive
GRIA3	305915	Intellectual developmental disorder X-linked syndromic Wu type	X-linked
GRIP1	604597	Fraser syndrome 3	Autosomal recessive
GRN	138945	Ceroid lipofuscinosis, neuronal, type 11	Autosomal recessive
GSS	601002	Glutathione synthetase deficiency	Autosomal recessive
GUCY2D	600179	Leber congenital amaurosis, type 1	Autosomal recessive
GUSB	611499	Mucopolysaccharidosis, type 7	Autosomal recessive
HADH	601609	3-hydroxyacyl-CoA dehydrogenase deficiency	Autosomal recessive
HADHA	600890	Long-chain 3-hydroxyl-CoA dehydrogenase (LCHAD) deficiency; Mitochondrial trifunctional protein deficiency	Autosomal recessive
HADHB	143450	Mitochondrial trifunctional protein deficiency	Autosomal recessive
HAX1	605998	Neutropenia, severe congenital, type 3, autosomal recessive	Autosomal recessive
HBA1	141800	Thalassemia, alpha-	Autosomal recessive
HBA2	141850	Thalassemia, alpha-	Autosomal recessive
HBB	141900	HBB-related hemoglobinopathy	Autosomal recessive
HCFC1	300019	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type)	X-linked
HEXA	606869	Tay-Sachs disease	Autosomal recessive
HEXB	606873	Sandhoff disease, infantile, juvenile, and adult forms	Autosomal recessive
HFE	613609	Hemochromatosis,Transferrin serum level QTL2,Alzheimer disease susceptibility to,Microvascular complications of diabetes 7,Porphyria cutanea tarda susceptibility to,Porphyria variegata susceptibility to	Autosomal recessive
HGD	607474	Alkaptonuria	Autosomal recessive
HGSNAT	610453	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)	Autosomal recessive
HJV	608374	Hemochromatosis, type 2A	Autosomal recessive
HLCS	609018	Holocarboxylase synthetase deficiency	Autosomal recessive
HMGCL	613898	HMG-CoA lyase deficiency	Autosomal recessive
HMOX1	141250	Heme oxygenase-1 deficiency	Autosomal recessive
HOGA1	613597	Hyperoxaluria, primary, type 3	Autosomal recessive
HPD	609695	Tyrosinemia, type 3	Autosomal recessive
HPRT1	308000	Lesch-Nyhan syndrome	X-linked
HPS1	604982	Hermansky-Pudlak syndrome, type 1	Autosomal recessive
HPS3	606118	Hermansky-Pudlak syndrome, type 3	Autosomal recessive
HPS4	606682	Hermansky-Pudlak syndrome, type 4	Autosomal recessive
HSD17B10	300256	HSD10 mitochondrial disease	X-linked
HSD17B3	605573	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	Autosomal recessive
HSD17B4	601860	D-bifunctional protein deficiency	Autosomal recessive
HSD3B2	613890	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	Autosomal recessive
HSPG2	142461	Dyssegmental dysplasia, Silverman-Handmaker type	Autosomal recessive
HUWE1	300697	Intellectual developmental disorder X-linked Turner type	X-linked

Gene	OMIM gene	Phenotype PATOLOGIA EN	Inheritance
HYAL1	607071	Mucopolysaccharidosis, type 9	Autosomal recessive
HYLS1	610693	Hydrolethalus syndrome	Autosomal recessive
IDH3B	604526	Retinitis pigmentosa, type 46	Autosomal recessive
IDS	300823	Mucopolysaccharidosis, type 2	X-linked
IDUA	252800	Mucopolysaccharidosis type 1	Autosomal recessive
IGHMBP2	600502	Charcot-Marie-Tooth disease, axonal, type 2S	Autosomal recessive
IL1RAPL1	300206	Mental retardation, X-linked, type 21/34	X-linked
IL2RG	308380	Severe combined immunodeficiency, X-linked	X-linked
IMPA1	602064	Mental retardation, autosomal recessive 59	Autosomal recessive
ITGB3	173470	Glanzmann thrombasthenia	Autosomal recessive
ITPA	147520	Epileptic encephalopathy, early infantile, type 35	Autosomal recessive
IVD	607036	Isovaleric acidemia	Autosomal recessive
IYD	612025	Thyroid dyshormonogenesis, type 4	Autosomal recessive
JAK3	600173	Severe Combined Immunodeficiency, autosomal recessive, T-negative/B-positive type	Autosomal recessive
KATNB1	602703	Lissencephaly 6, with microcephaly	Autosomal recessive
KCNJ11	600937	Hyperinsulinemic hypoglycemia, type 2 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive; Autosomal recessive
KDM5C	314690	Mental retardation, X-linked, syndromic, Claes-Jensen type	X-linked
KIAA0586	610178	Short-rib thoracic dysplasia 14 with polydactyly	Autosomal recessive
L1CAM	308840	L1 Syndrome	X-linked
LAMA2	156225	LAMA2-related muscular dystrophy	Autosomal recessive
LAMA3	600805	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive
LAMB3	150310	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive
LAMC2	150292	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive
LARGE1	603590	Muscular dystrophy-dystroglycanopathy, type 6A and 6B	Autosomal recessive
LCA5	611408	Leber congenital amaurosis, type 5	Autosomal recessive
LDLR	606945	Hypercholesterolemia familial 1,LDL cholesterol level QTL2	Autosomal recessive
LDLRAP1	605747	Hypercholesterolemia, familial, autosomal recessive	Autosomal recessive
LHCGR	152790	Leydig cell hypoplasia	Autosomal recessive
LHX3	600577	Pituitary hormone deficiency, combined, type 3	Autosomal recessive
LIFR	151443	Stuve-Wiedemann syndrome / Schwartz-Jampel type 2 syndrome	Autosomal recessive
LIPA	613497	Lysosomal acid lipase deficiency	Autosomal recessive
LIPH	607365	Hypotrichosis, type 7 or woolly hair, autosomal recessive, type 2, with or without hypotrichosis	Autosomal recessive
LMBRD1	612625	Methylmalonic aciduria and homocystinuria, cblF type	Autosomal recessive
LOXHD1	613072	Deafness, autosomal recessive, type 77	Autosomal recessive
LPL	609708	Lipoprotein lipase deficiency	Autosomal recessive
LRP2	600073	Donnai-Barrow syndrome	Autosomal recessive
LRPPRC	607544	Leigh syndrome, French-Canadian type	Autosomal recessive
LYST	606897	Chediak-Higashi syndrome	Autosomal recessive
MAGT1	300715	Congenital disorder of glycosylation type Icc,Immunodeficiency X-linked with magnesium defect Epstein-Barr virus infection and neoplasia	X-linked
MAMLD1	300120	Hypospadias 2 X-linked	X-linked
MAN2B1	609458	Alpha-mannosidosis	Autosomal recessive
MANBA	609489	Mannosidosis, beta	Autosomal recessive
MAOA	309850	Brunner syndrome,Antisocial behavior	X-linked
MAT1A	610550	Methionine adenosyltransferase deficiency, autosomal recessive	Autosomal recessive
MBTPS2	300294	Olmsted syndrome X-linked,IFAP syndrome with or without BRESHECK syndrome,Keratosis follicularis spinulosa decalvans X-linked,Osteogenesis imperfecta type XIX	X-linked
MCCC1	609010	3-Methylcrotonyl-CoA carboxylase deficiency, type 1	Autosomal recessive
MCCC2	609014	3-Methylcrotonyl-CoA carboxylase deficiency, type 2	Autosomal recessive
MCEE	608419	Methylmalonyl-CoA epimerase deficiency	Autosomal recessive
MCOLN1	605248	Mucolipidosis type 4	Autosomal recessive
MCPH1	607117	Microcephaly type 1, primary, autosomal recessive	Autosomal recessive
MECP2	300005	Encephalopathy, neonatal severe; Rett syndrome	X-linked
MED12	300188	Hardikar syndrome,Lujan-Fryns syndrome,Ohdo syndrome X-linked,Opitz-Kaveggia syndrome	X-linked
MED17	603810	Microcephaly, postnatal progressive, with seizures and brain atrophy	Autosomal recessive
MEFV	608107	Familial Mediterranean fever	Autosomal recessive
MESP2	605195	Spondylocostal dysostosis, type 2, autosomal recessive	Autosomal recessive
MFSD8	611124	Ceroid lipofuscinosis, neuronal, type 7	Autosomal recessive
MID1	300552	Opitz GBBB syndrome	X-linked
MKKS	604896	Bardet-Biedl syndrome type 6	Autosomal recessive
MKS1	609883	Bardet-Biedl syndrome type 13; Meckel syndrome, type 1; Joubert syndrome, type 28	Autosomal recessive
MLC1	605908	Megalencephalic leukoencephalopathy with subcortical cysts	Autosomal recessive
MLYCD	606761	Malonyl-CoA decarboxylase deficiency	Autosomal recessive
MMAA	607481	Methylmalonic aciduria, vitamin B12-responsive	Autosomal recessive
MMAB	607568	Methylmalonic aciduria, vitamin B12-responsive, type cblB	Autosomal recessive
MMACHC	609831	Methylmalonic aciduria and homocystinuria, cblC type	Autosomal recessive, digenic inheritance (PRDX1 gene)
MMADHC	611935	Homocystinuria, cblD type, variant 1	Autosomal recessive
MMUT	609058	Methylmalonic aciduria, mut(0) type	Autosomal recessive
MOCS1	603707	Molybdenum cofactor deficiency A	Autosomal recessive
MOGS	601336	Congenital disorder of glycosylation, type 2B	Autosomal recessive
MPI	154550	Congenital disorder of glycosylation, type 1B	Autosomal recessive
MPL	159530	Thrombocytopenia, congenital amegakaryocytic	Autosomal recessive
MPV17	137960	Mitochondrial DNA depletion syndrome type 6 (hepatocerebral); Charcot-Marie-Tooth disease, axonal, type 2EE	Autosomal recessive
MRE11	600814	Ataxia-telangiectasia-like disorder 1	Autosomal recessive
MTHFR	607093	Homocystinuria due to MTHFR deficiency	Autosomal recessive

Gene	OMIM gene	Phenotype PATOLOGIA EN	Inheritance
MTM1	300415	Myotubular myopathy, X-linked	X-linked
MTMR2	603557	Charcot-Marie-Tooth disease, type 4B1	Autosomal recessive
MTR	156570	Homocystinuria-megaloblastic anemia, cblG complementation type	Autosomal recessive
MTRR	602568	Homocystinuria-megaloblastic anemia, cbl E type	Autosomal recessive
MTTP	157147	Abetalipoproteinemia	Autosomal recessive
MVK	251170	Mevalonic aciduria	Autosomal recessive
MYO15A	602666	Deafness, autosomal recessive, type 3	Autosomal recessive
MYO7A	276903	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2	Autosomal recessive
NAA10	300013	Microphthalmia syndromic 1,Ogden syndrome	X-linked
NADK2	615787	2,4-dienoyl-CoA reductase deficiency	Autosomal recessive
NAGA	104170	Schindler disease, type I	Autosomal recessive
NAGLU	609701	Mucopolysaccharidosis, type 3B (Sanfilippo B)	Autosomal recessive
NAGS	608300	N-acetylglutamate synthase deficiency	Autosomal recessive
NBN	602667	Nijmegen breakage syndrome	Autosomal recessive
NCF1	608512	Chronic granulomatous disease, type 1	Autosomal recessive
NCF2	608515	Chronic granulomatous disease, type 2	Autosomal recessive
NDP	300658	Norrie disease	X-linked
NDRG1	605262	Charcot-Marie-Tooth disease, type 4D	Autosomal recessive
NDUFA1	300078	Mitochondrial complex I deficiency nuclear type 12	X-linked
NDUFAF2	609653	Mitochondrial complex I deficiency, nuclear type 10	Autosomal recessive
NDUFAF5	612360	Mitochondrial complex I deficiency, nuclear type 16	Autosomal recessive
NDUFS4	602694	Mitochondrial complex I deficiency, nuclear type 1	Autosomal recessive
NDUFS6	603848	Mitochondrial complex I deficiency, nuclear type 9	Autosomal recessive
NDUFS7	601825	Mitochondrial complex I deficiency, nuclear type 3	Autosomal recessive
NDUFV1	161015	Mitochondrial complex I deficiency, nuclear type 4	Autosomal recessive
NEB	161650	Nemaline myopathy type 2	Autosomal recessive
NEU1	608272	Sialidosis, type 1 and type 2	Autosomal recessive
NHP2	606470	Dyskeratosis congenita, autosomal recessive type 2	Autosomal recessive
NLRP7	609661	Hydatidiform mole, recurrent, type 1	Autosomal recessive
NOP10	606471	Dyskeratosis congenita, autosomal recessive type 1	Autosomal recessive
NPC1	607623	Niemann-Pick disease, type C1	Autosomal recessive
NPC2	601015	Niemann-pick disease, type C2	Autosomal recessive
NPHP1	607100	Joubert syndrome type 4	Autosomal recessive
NPHS1	602716	Nephrotic syndrome, type 1	Autosomal recessive
NPHS2	604766	Nephrotic syndrome, type 2	Autosomal recessive
NR0B1	300473	Adrenal hypoplasia, congenital	X-linked
NR2E3	604485	Enhanced S-cone syndrome (Goldmann-Favre); Retinitis pigmentosa, type 37	Autosomal recessive; Autosomal recessive
NSDHL	300275	CHILD syndrome,CK syndrome	X-linked
NTRK1	191315	Insensitivity to pain, congenital, with anhidrosis	Autosomal recessive
NYX	300278	Night blindness congenital stationary (complete) 1A X-linked	X-linked
OAT	613349	Gyrate atrophy of choroid and retina	Autosomal recessive
OCA2	611409	Oculocutaneous albinism type 2	Autosomal recessive
OCRL	300535	Lowe Syndrome; Dent disease type 2	X-linked
OFD1	300170	Retinitis pigmentosa 23,Joubert syndrome 10,Orofaciodigital syndrome I,Simpson-Golabi-Behmel syndrome type 2	X-linked
OPA3	606580	3-methylglutaconic aciduria, type 3	Autosomal recessive
OPHN1	300127	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	X-linked
OSTM1	607649	Osteopetrosis, autosomal recessive type 5	Autosomal recessive
OTC	300461	Ornithine transcarbamylase deficiency	X-linked
OTOF	603681	Deafness, autosomal recessive, type 9	Autosomal recessive
P3H1	610339	Osteogenesis imperfecta, type 8	Autosomal recessive
PAH	612349	Phenylketonuria	Autosomal recessive
PAK3	300142	Mental retardation, X-linked, type 30	X-linked
PANK2	606157	Neurodegeneration with brain iron accumulation type 1	Autosomal recessive
PC	608786	Pyruvate carboxylase deficiency	Autosomal recessive
PCBD1	126090	Hyperphenylalaninemia, BH4-deficient, type D	Autosomal recessive
PCCA	232000	Propionic acidemia	Autosomal recessive
PCCB	232050	Propionic acidemia	Autosomal recessive
PCDH15	605514	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic	Autosomal recessive; Digenic inheritance (CDH23 gene)
PCDH19	300460	Developmental and epileptic encephalopathy 9	X-linked, female restricted
PDE6A	180071	Retinitis pigmentosa type 43	Autosomal recessive
PDHA1	300502	Pyruvate dehydrogenase E1-alpha deficiency	X-linked
PDHB	179060	Pyruvate dehydrogenase E1-beta deficiency	Autosomal recessive
PEX1	602136	Heimler syndrome type 1	Autosomal recessive
PEX10	602859	Peroxisome biogenesis disorder, type 6A (Zellweger syndrome); Peroxisome biogenesis disorder, type 6B	Autosomal recessive
PEX12	601758	Peroxisome biogenesis disorder type 3A (Zellweger)	Autosomal recessive
PEX2	170993	Peroxisome biogenesis disorder type 5A (Zellweger)	Autosomal recessive
PEX26	608666	Peroxisome biogenesis disorder type 7A (Zellweger)	Autosomal recessive
PEX5	600414	Peroxisome biogenesis disorder type 2A (Zellweger)	Autosomal recessive
PEX6	601498	Peroxisome biogenesis disorder, type 4A (Zellweger syndrome); Peroxisome biogenesis disorder, type 4B; Heimler syndrome 2	Autosomal recessive; Autosomal recessive; Autosomal recessive
PEX7	601757	Rhizomelic chondrodysplasia punctata, type 1	Autosomal recessive
PFKM	610681	Glycogen storage disease, type 7	Autosomal recessive
PGK1	311800	Phosphoglycerate kinase 1 deficiency	X-linked
PHF6	300414	Borjeson-Forssman-Lehmann syndrome	X-linked
PHF8	300560	Mental retardation syndrome, X-linked, Siderius type	X-linked

Gene	OMIM gene	Phenotype PATOLOGIA EN	Inheritance
PHGDH	606879	Neu-Laxova syndrome, type 1; Phosphoglycerate dehydrogenase deficiency	Autosomal recessive
PHKA1	311870	Muscle glycogenosis	X-linked
PHKA2	300798	Glycogen storage disease type IXa1,Glycogen storage disease type IXa2	X-linked
PIGA	311770	Multiple congenital anomalies-hypotonia-seizures syndrome 2,Neurodevelopmental disorder with epilepsy and hemochromatosis	X-linked
PKHD1	606702	Polycystic kidney disease type 4	Autosomal recessive
PLA2G6	603604	Infantile neuroaxonal dystrophy type 1	Autosomal recessive
PLOD1	153454	Ehlers-Danlos syndrome, kyphoscoliotic type, 1	Autosomal recessive
PLP1	300401	Pelizaeus-Merzbacher disease	X-linked
PMM2	601785	Congenital disorder of glycosylation, type 1A	Autosomal recessive
PNPO	603287	Pyridoxamine 5'-phosphate oxidase deficiency	Autosomal recessive
POLG	174763	POLG-related disorders	Autosomal recessive
POLR1C	610060	Leukodystrophy, hypomyelinating, type 11; Treacher Collins syndrome 3	Autosomal recessive
POMGNT1	606822	Muscular dystrophy-dystroglycanopathy, type 3A (Walker-Warburg syndrome); Type 3B; Type 3C (limb-girdle muscular dystrophy, type 15 [LGMDR15])	Autosomal recessive
POMT1	607423	Muscular dystrophy-dystroglycanopathy, type 1A (Walker-Warburg syndrome); Type 1B; Type 1C (limb-girdle muscular dystrophy, type 11 [LGMD R11])	Autosomal recessive
POMT2	607439	Muscular dystrophy-dystroglycanopathy, type 2A (Walker-Warburg syndrome); Type 2B; Type 2C (limb-girdle muscular dystrophy, type 14 [LGMD R14])	Autosomal recessive
POR	124015	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	Autosomal recessive
POU1F1	173110	Pituitary hormone deficiency, combined, type 1	Autosomal recessive
POU3F4	300039	Deafness, X-linked, type 2	X-linked
PPM1K	611065	Maple syrup urine disease, mild variant	Autosomal recessive
PPT1	600722	Ceroid lipofuscinosis, neuronal, type 1	Autosomal recessive
PQBP1	300463	Renpenning syndrome	X-linked
PRDX1	176763	Methylmalonic aciduria and homocystinuria, cblC type, digenic	Autosomal recessive, digenic inheritance (MMACHC gene)
PRF1	170280	Hemophagocytic lymphohistiocytosis, familial, type 2	Autosomal recessive
PRODH	606810	Hyperprolinemia, type 1	Autosomal recessive
PROP1	601538	Pituitary hormone deficiency, combined, type 2	Autosomal recessive
PRPS1	311850	PRPS1-related disoders	X-linked
PSAP	176801	Combined SAP deficiency	Autosomal recessive
PTS	612719	Hyperphenylalaninemia, BH4-deficient, type A	Autosomal recessive
PUS1	608109	Myopathy, lactic acidosis, and sideroblastic anemia, type 1	Autosomal recessive
PYGM	608455	McArdle disease	Autosomal recessive
QDPR	612676	Hyperphenylalaninemia, BH4-deficient, type C	Autosomal recessive
RAB23	606144	Carpenter syndrome	Autosomal recessive
RAB39B	300774	Intellectual developmental disorder X-linked 72,Waisman syndrome	X-linked
RAG1	179615	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	Autosomal recessive
RAG2	179616	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	Autosomal recessive
RAPSN	601592	Fetal akinesia deformation sequence, type 2; Myasthenic syndrome, congenital, type 11, associated with AChR deficiency	Autosomal recessive
RARS2	611524	Pontocerebellar hypoplasia, type 6	Autosomal recessive
RAX	601881	Isolated microphthalmia, type 3	Autosomal recessive
RBM10	300080	TARP syndrome	X-linked
RDH12	608830	Leber congenital amaurosis, type 13	Autosomal recessive
RLBP1	180090	Bothnia retinal dystrophy; Fundus albipunctatus	Autosomal recessive; Autosomal recessive
RMRP	157660	Anauxetic dysplasia, type 1	Autosomal recessive
RNASEH2B	610326	Aicardi-Goutieres syndrome, type 2	Autosomal recessive
RNASEH2C	610330	Aicardi-Goutieres syndrome, type 3	Autosomal recessive
RP2	300757	Retinitis pigmentosa, type 2, X-linked	X-linked
RPE65	180069	RPE65-related Leberÿcongenitalÿamaurosis/early-onset severe retinal dystrophyÿ	Autosomal recessive
RPGR	312610	Retinitis pigmentosa, type 3, X-linked; Cone-rod dystrophy, X-linked, 1	X-linked
RPGRIP1L	610937	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome	Autosomal recessive
RPL10	312173	Intellectual developmental disorder X-linked syndromic 35,Autism susceptibility to X-linked 5	X-linked
RS1	300839	Retinoschisis	X-linked
RTEL1	608833	Dyskeratosis congenita, autosomal recessive type 5	Autosomal recessive
SACS	604490	Spastic ataxia, Charlevoix-Saguenay, type	Autosomal recessive
SAG	181031	Oguchi disease, type 1	Autosomal recessive
SAMHD1	606754	Aicardi-Goutieres syndrome, type 5	Autosomal recessive
SBDS	607444	Shwachman-Diamond syndrome	Autosomal recessive
SCO2	604272	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1	Autosomal recessive
SEPSECS	613009	Pontocerebellar hypoplasia, type 2D	Autosomal recessive
SERPINA1	107400	Alpha-1 antitrypsin deficiency	Autosomal recessive
SGCA	600119	Limb-girdle muscular dystrophy, type 3 (LGMD R3)	Autosomal recessive
SGCB	600900	Limb-girdle muscular dystrophy, type 4 (LGMD R4)	Autosomal recessive
SGCD	601411	Limb-girdle muscular dystrophy, type 6 (LGMD R6)	Autosomal recessive
SGCG	608896	Limb-girdle muscular dystrophy, type 5 (LGMD R5)	Autosomal recessive
SGSH	605270	Mucopolysaccharidosis, type 3A (Sanfilippo A)	Autosomal recessive
SH2D1A	300490	Lymphoproliferative syndrome, X-linked, type 1	X-linked
SH3TC2	608206	Charcot-Marie-Tooth disease, type 4C	Autosomal recessive
SHOX	312865	Langer mesomelic dysplasia,Leri-Weill dyschondrosteosis,Short stature idiopathic familial,Langer mesomelic dysplasia,Leri-Weill dyschondrosteosis,Short stature idiopathic familial	X-linked
SHROOM4	300579	Intellectual developmental disorder X-linked syndromic Stocco dos Santos type	X-linked
SLC12A3	600968	Gitelman syndrome	Autosomal recessive
SLC12A6	604878	Agenesis of the corpus callosum with peripheral neuropathy	Autosomal recessive
SLC16A2	300095	Allan-Herndon-Dudley syndrome	X-linked
SLC17A5	604322	Salla disease	Autosomal recessive
SLC19A2	603941	Thiamine-responsive megaloblastic anemia syndrome	Autosomal recessive
SLC19A3	606152	Thiamine metabolism dysfunction syndrome, type 2 (biotin- or thiamine-responsive encephalopathy type)	Autosomal recessive
SLC22A5	603377	Carnitine deficiency, systemic primary	Autosomal recessive
SLC25A13	603859	Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset	Autosomal recessive
SLC25A15	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	Autosomal recessive
SLC25A20	613698	Carnitine-acylcarnitine translocase deficiency	Autosomal recessive

Gene	OMIM gene	Phenotype PATOLOGIA EN	Inheritance
SLC26A2	606718	Achondrogenesis, type 1B (diastrophic dysplasia)	Autosomal recessive
SLC26A3	126650	Diarrhea 1, secretory chloride, congenital	Autosomal recessive
SLC26A4	605646	Deafness, autosomal recessive, type 4; Pendred syndrome	Autosomal recessive; Digenic inheritance (KCNJ10 gene)
SLC35A1	605634	Congenital disorder of glycosylation, type 2F	Autosomal recessive
SLC35A3	605632	Arthrogryposis, mental retardation, and seizures	Autosomal recessive
SLC35C1	605881	Congenital disorder of glycosylation, type 2C	Autosomal recessive
SLC35D1	610804	Schneckenbecken dysplasia	Autosomal recessive
SLC37A4	602671	Glycogen storage disease, type 1B	Autosomal recessive
SLC39A4	607059	Acrodermatitis enteropathica	Autosomal recessive
SLC3A1	104614	Cystinuria	Autosomal recessive
SLC45A2	606202	Albinism, oculocutaneous, type 4	Autosomal recessive
SLC46A1	611672	Folate malabsorption, hereditary	Autosomal recessive
SLC4A11	610206	Corneal endothelial dystrophy, autosomal recessive	Autosomal recessive
SLC5A5	601843	Thyroid dyshormonogenesis, type 1	Autosomal recessive
SLC6A19	608893	Hartnup disorder	Autosomal recessive
SLC6A8	300036	Cerebral creatine deficiency syndrome, type 1	X-linked
SLC7A7	603593	Lysinuric protein intolerance	Autosomal recessive
SLC7A9	604144	Cystinuria	Autosomal recessive
SLC9A6	300231	Intellectual developmental disorder X-linked syndromic Christianson type	X-linked
SMARCAL1	606622	Schimke immunoosseous dysplasia	Autosomal recessive
SMN1	600354	Spinal muscular atrophy	Autosomal recessive
SMPD1	607608	Niemann-Pick disease, type A; Niemann-Pick disease, type B	Autosomal recessive
SMPX	300226	Deafness X-linked 4,Myopathy distal 7 adult-onset X-linked	X-linked
SMS	300105	Intellectual developmental disorder X-linked syndromic Snyder-Robinson type	X-linked
SOX3	313430	Intellectual developmental disorder X-linked with isolated growth hormone deficiency,Panhypopituitarism X-linked	X-linked
SPG11	610844	Amyotrophic lateral sclerosis, type 5, juvenile	Autosomal recessive
SPG7	602783	Spastic paraplegia, type 7, autosomal recessive	Autosomal recessive
SRD5A2	607306	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency (pseudovaginal perineoscrotal hypospadias)	Autosomal recessive
ST3GAL5	604402	Salt and pepper developmental regression syndrome	Autosomal recessive
STAR	600617	Lipoid adrenal hyperplasia	Autosomal recessive
STRC	606440	Deafness, autosomal recessive, type 16	Autosomal recessive
STS	300747	Ichthyosis X-linked	X-linked
SUMF1	607939	Multiple sulfatase deficiency	Autosomal recessive
SURF1	185620	Charcot-Marie-Tooth disease, type 4K; Leigh syndrome, due to COX IV deficiency	Autosomal recessive
SYN1	313440	Epilepsy, X-linked, with variable learning disabilities and behavior disorders	X-linked
SYP	313475	Intellectual developmental disorder X-linked 96	X-linked
TAF1	313650	Dystonia-Parkinsonism X-linked,Intellectual developmental disorder X-linked syndromic 33	X-linked
TAT	613018	Tyrosinemia, type 2	Autosomal recessive
TBX22	300307	Abruzzo-Erickson syndrome,Cleft palate with ankyloglossia	X-linked
TCIRG1	604592	Osteopetrosis, autosomal recessive, type 1	Autosomal recessive
TECPR2	615000	Spastic paraplegia, type 49, autosomal recessive	Autosomal recessive
TF	190000	Atransferrinemia	Autosomal recessive
TFR2	604720	Hemochromatosis, type 3	Autosomal recessive
TG	188450	Thyroid dyshormonogenesis, type 3	Autosomal recessive
TGM1	190195	Ichthyosis, congenital, autosomal recessive, type 1	Autosomal recessive
TH	191290	Segawa syndrome, recessive	Autosomal recessive
THOC2	300395	Mental retardation, X-linked 12	X-linked
THRB	190160	Thyroid hormone resistance, autosomal recessive	Autosomal recessive
TIMM8A	300356	Mohr-Tranebjaerg syndrome	X-linked
TMC1	606706	Deafness, autosomal recessive, type 7	Autosomal recessive
TMEM216	613277	Joubert syndrome, type 2; Meckel syndrome, type 2	Autosomal recessive
TMEM67	609884	Joubert syndrome, type 6; Meckel syndrome, type 3; COACH syndrome	Autosomal recessive
TMPRSS3	605511	Deafness, autosomal recessive, type 8/10	Autosomal recessive
TNXB	600985	Ehlers-Danlos syndrome, classic-like	Autosomal recessive
TPO	606765	Thyroid dyshormonogenesis, type 2A	Autosomal recessive
TPP1	607998	Ceroid lipofuscinosis, neuronal, type 2; Spinocerebellar ataxia, autosomal recessive, type 7	Autosomal recessive
TRAPPC2	300202	Spondyloepiphyseal dysplasia tarda	X-linked
TRDN	603283	Ventricular tachycardia, catecholaminergic polymorphic, type 5, with or without muscle weakness	Autosomal recessive
TREX1	606609	Aicardi-Goutieres syndrome, type 1	Autosomal recessive
TRIM32	602290	Limb-girdle muscular dystrophy, type 8 (LGMD R8)	Autosomal recessive
TRIM37	605073	Mulibrey nanism	Autosomal recessive
TRMU	610230	Liver failure, transient infantile	Autosomal recessive
TSEN54	608755	Pontocerebellar hypoplasia, type 2A; Pontocerebellar hypoplasia, type 4	Autosomal recessive
TSFM	604723	Combined oxidative phosphorylation deficiency, type 3	Autosomal recessive
TSHB	188540	Hypothyroidism, congenital, nongoitrous, type 4	Autosomal recessive
TSHR	603372	Hypothyroidism, congenital, nongoitrous, type 1	Autosomal recessive
TSPAN7	300096	Intellectual developmental disorder X-linked 58	X-linked
TSPYL1	604714	Sudden infant death with dysgenesis of the testes syndrome	X-linked
TTC37	614589	Trichohepatoenteric syndrome, type 1 (diarrhea, syndromic)	Autosomal recessive
TTN	188840	Limb-girdle muscular dystrophy type 10 (LGMDR10); Early-onset myopathy with fatal cardiomyopathy (Salih myopathy)	Autosomal recessive
TTPA	600415	Ataxia with isolated vitamin E deficiency	Autosomal recessive

Gene	OMIM gene	Phenotype PATOLOGIA EN	Inheritance
TYMP	131222	Mitochondrial DNA depletion syndrome, type 1 (MNGIE type)	Autosomal recessive
TYR	606933	Oculocutaneous albinism (OCA) type 1A; OCA type 1B; Waardenburg syndrome/albinism, digenic	Autosomal recessive, digenic inheritance (MITF gene)
TYRP1	115501	Albinism, oculocutaneous, type 3	Autosomal recessive
UBA1	314370	Spinal muscular atrophy X-linked 2 infantile	X-linked
UBE2A	312180	Intellectual developmental disorder X-linked syndromic Nascimento type	X-linked
UBE3A	601623	Angelman syndrome	Autosomal dominant
UGT1A1	191740	Crigler-Najjar syndrome, type 1; Crigler-Najjar syndrome, type 2	Autosomal recessive
UNC13D	608897	Hemophagocytic lymphohistiocytosis, familial, type 3	Autosomal recessive
UPF3B	300298	Mental retardation, X-linked, syndromic, type 14	X-linked
USH1C	605242	Usher syndrome, type 1C; Deafness, autosomal recessive, type 18A	Autosomal recessive
USH1G	607696	Usher syndrome, type 1G	Autosomal recessive
USH2A	608400	Usher syndrome, type 2A	Autosomal recessive
USP9X	300072	Intellectual developmental disorder X-linked 99,Intellectual developmental disorder X-linked 99 syndromic female-restricted	X-linked
VPS13A	605978	Choreoacanthocytosis	Autosomal recessive
VPS13B	607817	Cohen syndrome	Autosomal recessive
VPS45	610035	Neutropenia, severe congenital, type 5	Autosomal recessive
VPS53	615850	Pontocerebellar hypoplasia, type 2E	Autosomal recessive
VRK1	602168	Pontocerebellar hypoplasia, type 1A	Autosomal recessive
VSX2	142993	Microphthalmia with coloboma 3; Isolated microphthalmia 2	Autosomal recessive
WAS	300392	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked	X-linked
WHRN	607928	Usher syndrome, type 2D; Deafness, autosomal recessive, type 31	Autosomal recessive
WNT10A	606268	Odontoonychodermal dysplasia	Autosomal recessive
WRN	604611	Werner syndrome	Autosomal recessive
XK	314850	McLeod syndrome with or without chronic granulomatous disease	X-linked
XPA	611153	Xeroderma pigmentosum, group A	Autosomal recessive
XPC	613208	Xeroderma pigmentosum, group C	Autosomal recessive
ZDHHC9	300646	Mental retardation, X-linked syndromic, Raymond type	X-linked
ZFYVE26	612012	Spastic paraplegia, type 15, autosomal recessive	Autosomal recessive
ZIC3	300265	Congenital heart defects nonsyndromic 1 X-linked,Heterotaxy visceral 1 X-linked,VACTERL association X-linked	X-linked
ZNF711	314990	Mental retardation, X-linked, type 97	X-linked

5. PANNELLO NEXT CARRIER COMPLETE MALE (618 GENI - MALE)

gene	OMIM	Phenotype PATOLOGIA EN	Inheritance
AAAS	605378	Triple-A syndrome (achalasia-addisonianism-alacrimia)	Autosomal recessive
AARS1	601065	Epileptic encephalopathy, early infantile, type 29	Autosomal recessive
AARS2	612035	Combined oxidative phosphorylation deficiency 8; Leukoencephalopathy, progressive, with ovarian failure	Autosomal recessive
ABCA12	607800	Ichthyosis, congenital, autosomal recessive, type 4A; ICAR, type 4B (harlequin)	Autosomal recessive
ABCA3	601615	Surfactant metabolism dysfunction, pulmonary, type 3	Autosomal recessive
ABCA4	601691	Stargardt disease type 1; Cone-rod dystrophy type 3	Autosomal recessive
ABCB11	603201	Cholestasis, benign recurrent intrahepatic, type 2; Cholestasis, progressive familial intrahepatic, type 2	Autosomal recessive
ABCB4	171060	Cholestasis, progressive familial intrahepatic, type 3	Autosomal recessive
ABCC6	603234	Pseudoxanthoma elasticum; Generalized arterial calcification of infancy, type 2	Autosomal recessive
ABCC8	600509	Hyperinsulinemic hypoglycemia, type 1 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive
ABCD1	300371	Adrenoleukodystrophy	X-linked
ABCD4	603214	Methylmalonic aciduria and homocystinuria, cblJ type	Autosomal recessive
ACAD8	604773	Isobutyryl-CoA dehydrogenase deficiency	Autosomal recessive
ACAD9	611103	Acyl-CoA dehydrogenase 9 deficiency (mitochondrial complex I deficiency, nuclear, type 20)	Autosomal recessive
ACADM	607008	Medium-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
ACADS	606885	Short-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
ACADSB	600301	Short/branched-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
ACADVL	609575	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	Autosomal recessive
ACAT1	607809	Alpha-methylacetoacetic aciduria (3-ketothiolase deficiency)	Autosomal recessive
ACO2	100850	Infantile cerebellar-retinal degeneration	Autosomal recessive
ACOX1	609751	Peroxisomal acyl-CoA oxidase deficiency	Autosomal recessive
ACP5	171640	Spondyloenchondrodysplasia with immune dysregulation	Autosomal recessive
ACSF3	614245	Combined malonic and methylmalonic aciduria	Autosomal recessive
ADA	608958	Severe combined immunodeficiency due to adenosine deaminase deficiency (ADA)	Autosomal recessive
ADAMTS1	608990	Weill-Marchesani syndrome, type 1, recessive	Autosomal recessive
ADAMTS2	604539	Ehlers-Danlos syndrome, dermatosparaxis type	Autosomal recessive
ADAMTSL	612277	Geleophysic dysplasia type 1	Autosomal recessive
ADAT3	615302	Mental retardation, autosomal recessive 36	Autosomal recessive
ADGRG1	604110	Polymicrogyria, bilateral frontoparietal	Autosomal recessive
ADGRG6	612243	Lethal congenital contracture syndrome 9	Autosomal recessive
ADGRV1	602851	Usher syndrome, type 2C	Autosomal recessive, di
ADK	102750	Hypermethioninemia due to adenosine kinase deficiency	Autosomal recessive
AGA	613228	Aspartylglucosaminuria (glycosylasparaginase deficiency)	Autosomal recessive
AGL	610860	Glycogen storage disease, type 3	Autosomal recessive
AGPS	603051	Rhizomelic chondrodysplasia punctata, type 3	Autosomal recessive
AGXT	604285	Hyperoxaluria, primary, type 1	Autosomal recessive
AHCY	180960	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	Autosomal recessive
AHI1	608894	Joubert syndrome, type 3	Autosomal recessive
AIMP1	603605	Leukodystrophy, hypomyelinating, type 3	Autosomal recessive
AIMP2	600859	Leukodystrophy, hypomyelinating, type 17	Autosomal recessive
AIPL1	604392	Leber congenital amaurosis, type 4	Autosomal recessive
AIRE	607358	Autoimmune polyendocrinopathy syndrome, type 1	Autosomal recessive
ALDH3A2	609523	Sjogren-Larsson syndrome	Autosomal recessive
ALDH4A1	606811	Hyperprolinemia, type 2	Autosomal recessive
ALDH7A1	107323	Epilepsy, pyridoxine-dependent	Autosomal recessive
ALDOA	103850	Glycogen storage disease type 12	Autosomal recessive
ALDOB	612724	Fructose intolerance, hereditary	Autosomal recessive
ALG1	605907	Congenital disorder of glycosylation, type 1K	Autosomal recessive
ALG11	613666	Congenital disorder of glycosylation, type 1P	Autosomal recessive
ALG12	607144	Congenital disorder of glycosylation, type 1G	Autosomal recessive
ALG2	607905	Myasthenic syndrome, congenital, type 14, with tubular aggregates	Autosomal recessive
ALG3	608750	Congenital disorder of glycosylation, type 1D	Autosomal recessive
ALG6	604566	Congenital disorder of glycosylation, type 1C	Autosomal recessive
ALMS1	606844	Alstr”m syndrome	Autosomal recessive
ALPL	171760	Hypophosphatasia, infantile/childhood	Autosomal recessive
AMH	600957	Persistent Mullerian duct syndrome, type 1	Autosomal recessive
AMHR2	600956	Persistent Mullerian duct syndrome, type II	Autosomal recessive
AMPD2	102771	Pontocerebellar hypoplasia, type 9	Autosomal recessive

gene	OMIM ger	Phenotype PATOLOGIA EN	Inheritance
AMT	238310	Glycine encephalopathy	Autosomal recessive
ANO10	613726	Spinocerebellar ataxia, autosomal recessive, type 10	Autosomal recessive
AP1S1	603531	MEDNIK syndrome	Autosomal recessive
AP1S2	300629	Mental retardation, X-linked, syndromic, type 5 (Pettigrew syndrome)	X-linked
AP4B1	607245	Spastic paraplegia, type 47, autosomal recessive	Autosomal recessive
AP4E1	607244	Spastic paraplegia, type 51, autosomal recessive	Autosomal recessive
AP4M1	602296	Spastic paraplegia, type 50, autosomal recessive	Autosomal recessive
AP4S1	607243	Spastic paraplegia, type 52, autosomal recessive	Autosomal recessive
AP5Z1	613653	Spastic paraplegia, type 48, autosomal recessive	Autosomal recessive
AQP2	107777	Diabetes insipidus, nephrogenic, type 2	Autosomal recessive
AR	313700	Androgen insensitivity syndrome, complete	X-linked
ARFGEF2	605371	Periventricular heterotopia with microcephaly	Autosomal recessive
ARG1	608313	Argininemia (arginase deficiency)	Autosomal recessive
ARL13B	608922	Joubert syndrome type 8	Autosomal recessive
ARSA	607574	Metachromatic leukodystrophy	Autosomal recessive
ARSB	611542	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)	Autosomal recessive
ARV1	611647	Epileptic encephalopathy, early infantile, 38	Autosomal recessive
ARX	300382	Epileptic encephalopathy, early infantile, type 1; ARX-related developmental disorders	X-linked
ASL	608310	Argininosuccinic aciduria	Autosomal recessive
ASNS	108370	Asparagine synthetase deficiency	Autosomal recessive
ASPA	608034	Canavan disease	Autosomal recessive
ASPM	605481	Primary microcephaly type 5, autosomal recessive	Autosomal recessive
ASS1	603470	Citrullinemia, type 1	Autosomal recessive
ATM	607585	Ataxia-telangiectasia	Autosomal recessive
ATP6V1B1	192132	Renal tubular acidosis with deafness	Autosomal recessive
ATP7B	606882	Wilson disease	Autosomal recessive
ATP8B1	602397	Cholestasis, progressive familial intrahepatic, type 1; Cholestasis, benign recurrent intrahepatic, type 1	Autosomal recessive
ATR	601215	Seckel syndrome, type 1	Autosomal recessive
ATRX	300032	Mental retardation-hypotonic facies syndrome, X-linked; Alpha-thalassemia/mental retardation syndrome	X-linked
AUH	600529	3-methylglutaconic aciduria, type 1	Autosomal recessive
B4GALT1	137060	Congenital disorder of glycosylation, type 2D	Autosomal recessive
BBS1	209901	Bardet-Biedl syndrome, type 1	Autosomal recessive; D
BBS10	610148	Bardet-Biedl syndrome, type 10	Autosomal recessive; D
BBS12	610683	Bardet-Biedl syndrome, type 12	Autosomal recessive; D
BBS2	606151	Bardet-Biedl syndrome, type 2	Autosomal recessive; D
BBS4	600374	Bardet-Biedl syndrome, type 4	Autosomal recessive; D
BBS9	607968	Bardet-Biedl syndrome, type 9	Autosomal recessive; D
BCHE	177400	Butyrylcholinesterase deficiency	Autosomal recessive
BCKDHA	608348	Maple syrup urine disease, type 1A	Autosomal recessive
BCKDHB	248611	Maple syrup urine disease, type 1B	Autosomal recessive
BCS1L	603647	BCS1L-related disorders, including Leigh syndrome	Autosomal recessive
BLM	604610	Bloom syndrome	Autosomal recessive
BRAT1	614506	Rigidity and multifocal seizure syndrome, lethal neonatal; Neurodevelopmental disorder with cerebellar atrophy and with or without seizures	Autosomal recessive
BRIP1	605882	Fanconi anemia, complementation group J	Autosomal recessive
BRWD3	300553	Mental retardation, X-linked, type 93	X-linked
BSND	606412	Bartter syndrome, type 4A	Autosomal recessive
BTD	609019	Biotinidase deficiency	Autosomal recessive
BUB1B	602860	Mosaic variegated aneuploidy syndrome 1	Autosomal recessive
C2CD3	615944	Orofaciodigital syndrome, type 14	Autosomal recessive
CA2	611492	Osteopetrosis with renal tubular acidosis (osteopetrosis, autosomal recessive, type 3)	Autosomal recessive
CA8	114815	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3	Autosomal recessive

gene	OMIM ger	Phenotype PATOLOGIA EN	Inheritance
CABP2	607314	Deafness, autosomal recessive, type 93	Autosomal recessive
CAD	114010	Epileptic encephalopathy, early infantile, 50	Autosomal recessive
CANT1	613165	Desbuquois dysplasia, type 1; Epiphyseal dysplasia, multiple, type 7	Autosomal recessive
CAPN3	114240	Limb-girdle muscular dystrophy, type 1 (LGMD R1)	Autosomal recessive
CASK	300172	FG syndrome 4,Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia,Intellectual developmental disorder with or without nystagmus	X-linked
CASQ2	114251	Ventricular tachycardia, catecholaminergic polymorphic, type 2	Autosomal recessive
CBS	613381	Homocystinuria due to cystathionine beta-synthase	Autosomal recessive
CC2D1A	610055	Mental retardation, autosomal recessive, type 3	Autosomal recessive
CC2D2A	612013	Joubert syndrome, type 9; Meckel syndrome, type 6	Autosomal recessive
CCDC88C	611204	Hydrocephalus, congenital, type 1	Autosomal recessive
CCN6	603400	Arthropathy, progressive pseudorheumatoid, of childhood	Autosomal recessive
CD40LG	300386	Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)	X-linked
CDC14A	603504	Deafness, autosomal recessive, type 105	Autosomal recessive
CDC45	603465	Meier-Gorlin syndrome 7	Autosomal recessive
CDH23	605516	Deafness, autosomal recessive, type 12; Usher syndrome, type 1D; Usher syndrome type 1D/F digenic	Autosomal recessive, Di
CDH3	114021	Ectodermal dysplasia, ectrodactyly, and macular dystrophy	Autosomal recessive
CDK5RAP2	608201	Primary microcephaly type 3, autosomal recessive	Autosomal recessive
CENPJ	609279	Primary microcephaly type 6, autosomal recessive	Autosomal recessive
CEP290	610142	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10	Autosomal recessive
CERKL	608381	Retinitis pigmentosa, type 26	Autosomal recessive
CFTR	602421	Cystic fibrosis	Autosomal recessive
CHAT	118490	Myasthenic syndrome, congenital, type 6, presynaptic	Autosomal recessive
CHRNE	100725	Myasthenic syndrome, congenital, type 4B, fast-channel; Myasthenic syndrome, congenital, type 4C, associated with acetylcholine receptor deficiency	Autosomal recessive
CHRNG	100730	Multiple pterygium syndrome (MPS), Escobar type; MPS, lethal type	Autosomal recessive
CHST3	603799	Spondyloepiphyseal dysplasia with congenital joint dislocations	Autosomal recessive
CHST6	605294	Macular corneal dystrophy	Autosomal recessive
CIITA	600005	Bare lymphocyte syndrome, type 2, complementation group A	Autosomal recessive
CISD2	611507	Wolfram syndrome 2	Autosomal recessive
CIT	605629	Microcephaly 17, primary, autosomal recessive	Autosomal recessive
CLCN1	118425	Myotonia congenita, recessive	Autosomal recessive
CLCN2	600570	Leukoencephalopathy with ataxia	Autosomal recessive
CLN3	607042	Ceroid lipofuscinosis, neuronal, type 3	Autosomal recessive
CLN5	608102	Ceroid lipofuscinosis, neuronal, type 5	Autosomal recessive
CLN6	606725	Ceroid lipofuscinosis, neuronal, type 6	Autosomal recessive
CLN8	607837	Ceroid lipofuscinosis, neuronal, type 8	Autosomal recessive
CLP1	608757	Pontocerebellar hypoplasia, type 10	Autosomal recessive
CLRN1	606397	Usher syndrome, type 3A	Autosomal recessive
CNGA1	123825	Retinitis pigmentosa type 49	Autosomal recessive
CNGA3	600053	Achromatopsia, type 2	Autosomal recessive
CNGB1	600724	Retinitis pigmentosa type 45	Autosomal recessive
CNGB3	605080	Achromatopsia, type 3	Autosomal recessive
CNPY3	610774	Epileptic encephalopathy, early infantile, type 60	Autosomal recessive
COA8	616003	Mitochondrial complex IV deficiency, nuclear type 17	Autosomal recessive
COL11A2	120290	Otospondylomegaepiphyseal dysplasia, autosomal recessive	Autosomal recessive
COL27A1	608461	Steel syndrome	Autosomal recessive
COL4A3	120070	Alport syndrome, autosomal recessive, type 2	Autosomal recessive; D
COL4A4	120131	Alport syndrome, autosomal recessive, type 2	Autosomal recessive; D

gene	OMIM ger	Phenotype PATOLOGIA EN	Inheritance
COL7A1	120120	Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial	Autosomal recessive; A
COLQ	603033	Myasthenic syndrome, congenital, type 5	Autosomal recessive
COQ2	609825	Primary coenzyme Q10 deficiency, type 1	Autosomal recessive
COX10	602125	Mitochondrial complex IV deficiency, nuclear type 3	Autosomal recessive
COX15	603646	Mitochondrial complex IV deficiency, nuclear type 6	Autosomal recessive
COX6B1	124089	Mitochondrial complex IV deficiency, nuclear type 7	Autosomal recessive
CPS1	608307	Carbamoylphosphate synthetase 1 deficiency	Autosomal recessive
CPT1A	600528	Carnitine palmitoyltransferase type 1A deficiency, hepatic	Autosomal recessive
CPT2	600650	Carnitine palmitoyltransferase type 2 deficiency, lethal neonatal; Carnitine palmitoyltransferase type 2 deficiency, infantile	Autosomal recessive
CRB1	604210	Retinitis pigmentosa, type 12; Leber congenital amaurosis, type 8	Autosomal recessive
CRTAP	605497	Osteogenesis imperfecta, type 7	Autosomal recessive
CTH	607657	Cystathioninuria	Autosomal recessive
CTNS	606272	Nephropathic cystinosis	Autosomal recessive
CTSA	613111	Galactosialidosis	Autosomal recessive
CTSC	602365	Haim-Munk syndrome; Papillon-Lefevre syndrome	Autosomal recessive
CTSD	116840	Ceroid lipofuscinosis, neuronal, type 10	Autosomal recessive
CTSK	601105	Pycnodysostosis	Autosomal recessive
CUL4B	300304	Mental retardation, X-linked, syndromic, type 15 (Cabezas type)	X-linked
CWF19L1	616120	Spinocerebellar ataxia, autosomal recessive, type 17	Autosomal recessive
CYBA	608508	Chronic granulomatous disease, type 4	Autosomal recessive
CYP11A1	118485	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency	Autosomal recessive
CYP11B1	610613	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	Autosomal recessive
CYP11B2	124080	Hypoaldosteronism, congenital, due to CMO I deficiency	Autosomal recessive
CYP17A1	609300	17 alpha()-hydroxylase/17,20-lyase deficiency	Autosomal recessive
CYP19A1	107910	Aromatase deficiency	Autosomal recessive
CYP1B1	601771	Glaucoma, primary congenital, type 3A	Autosomal recessive
CYP21A2	613815	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	Autosomal recessive
CYP26B1	605207	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies	Autosomal recessive
CYP27A1	606530	Cerebrotendinous xanthomatosis	Autosomal recessive
CYP27B1	609506	Vitamin D-dependent rickets, type 1	Autosomal recessive
DARS1	603084	Hypomyelination with brainstem and spinal cord involvement and leg spasticity	Autosomal recessive
DBT	248610	Maple syrup urine disease, type 2	Autosomal recessive
DCLRE1C	605988	Omenn syndrome; Severe combined immunodeficiency, Athabascan type	Autosomal recessive
DCX	300121	Lissencephaly, X-linked, type 1	X-linked
DDB2	600811	Xeroderma pigmentosum, complementation group E	Autosomal recessive
DHCR7	602858	Smith-Lemli-Opitz syndrome	Autosomal recessive
DHDDS	608172	Retinitis pigmentosa, type 59	Autosomal recessive
DHODH	126064	Miller syndrome	Autosomal recessive
DLD	238331	Dihydrolipoamide dehydrogenase deficiency	Autosomal recessive
DLG3	300189	Mental retardation, X-linked, type 90	X-linked
DMD	300377	Duchenne/Becker muscular dystrophy	X-linked
DMXL2	612186	Developmental and epileptic encephalopathy, type 81	Autosomal recessive
DNAH5	603335	Ciliary dyskinesia, primary, type 3, with or without situs inversus	Autosomal recessive
DNAI1	604366	Ciliary dyskinesia, primary, type 1, with or without situs inversus	Autosomal recessive
DNAI2	605483	Ciliary dyskinesia, primary, type 9, with or without situs inversus	Autosomal recessive
DNAJC12	606060	Hyperphenylalaninemia, mild, non-BH4-deficient	Autosomal recessive
DNAL1	610062	Ciliary dyskinesia, primary, type 16	Autosomal recessive
DNM1L	603850	Encephalopathy due to defective mitochondrial and peroxisomal fission, type 1	Autosomal recessive
DNM2	602378	Lethal congenital contracture syndrome, type 5	Autosomal recessive
DOK7	610285	Fetal akinesia deformation sequence, type 3; Myasthenic syndrome, congenital, type 10	Autosomal recessive
DOLK	610746	Congenital disorder of glycosylation, type 1M	Autosomal recessive
DPAGT1	191350	Congenital disorder of glycosylation, type 1J; Myasthenic syndrome, congenital, type 13	Autosomal recessive
DPM1	603503	Congenital disorder of glycosylation, type 1E	Autosomal recessive
DPYD	612779	Dihydropyrimidine dehydrogenase deficiency	Autosomal recessive
DST	113810	Epidermolysis bullosa simplex, autosomal recessive, type 2	Autosomal recessive
DUOX2	606759	Thyroid dyshormonogenesis, type 6	Autosomal recessive
DUOXA2	612772	Thyroid dyshormonogenesis, type 5	Autosomal recessive
DYNC2H1	603297	Short-rib thoracic dysplasia, type 3, with or without polydactyly	Autosomal recessive; D
DYSF	603009	Miyoshi muscular dystrophy, type 1; Limb-girdle muscular dystrophy, type 2 (LGMD R2)	Autosomal recessive
EDAR	604095	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type	Autosomal recessive
EIF2AK3	604032	Wolcott-Rallison syndrome	Autosomal recessive
EIF2B5	603945	Leukoencephalopathy with vanishing white matter	Autosomal recessive

gene	OMIM ger	Phenotype PATOLOGIA EN	Inheritance
ELP1	603722	Familial dysautonomia	Autosomal recessive
EMC1	616846	Cerebellar atrophy, visual impairment, and psychomotor retardation	Autosomal recessive
EPRS1	138295	Leukodystrophy, hypomyelinating, type 15	Autosomal recessive
ERCC2	126340	Trichothiodystrophy, type 1	Autosomal recessive
ERCC3	133510	Trichothiodystrophy, type 2	Autosomal recessive
ERCC4	133520	Fanconi anemia, complementation group Q	Autosomal recessive
ERCC5	133530	Cerebrooculofacioskeletal syndrome, type 3	Autosomal recessive
ERCC6	609413	Cockayne syndrome, type B; Cerebrooculofacioskeletal syndrome, type 1	Autosomal recessive
ERCC8	609412	Cockayne syndrome, type A	Autosomal recessive
ESCO2	609353	Roberts syndrome	Autosomal recessive
ETFA	608053	Glutaric acidemia, type 2A	Autosomal recessive
ETFB	130410	Glutaric acidemia, type 2B	Autosomal recessive
ETFDH	231675	Glutaric acidemia, type 2C	Autosomal recessive
ETHE1	608451	Ethylmalonic encephalopathy	Autosomal recessive
EVC	604831	Ellis-van Creveld syndrome	Autosomal recessive
EVC2	607261	Ellis-van Creveld syndrome	Autosomal recessive
EXOSC3	606489	Pontocerebellar hypoplasia, type 1B	Autosomal recessive
EYS	612424	Retinitis pigmentosa, type 25	Autosomal recessive
F11	264900	Factor XI deficiency	Autosomal recessive
F2	176930	Prothrombin deficiency	Autosomal recessive
F5	612309	Factor V deficiency	Autosomal recessive
F8	300841	Hemophilia A	X-linked
F9	300746	Hemophilia B	X-linked
FAH	613871	Tyrosinemia, type 1	Autosomal recessive
FAM161A	613596	Retinitis pigmentosa, type 28	Autosomal recessive
FAM20C	611061	Raine syndrome	Autosomal recessive
FANCA	607139	Fanconi anemia, complementation group A	Autosomal recessive
FANCC	613899	Fanconi anemia, complementation group C	Autosomal recessive
FANCD2	613984	Fanconi anemia, complementation group D2	Autosomal recessive
FANCG	602956	Fanconi anemia, complementation group G	Autosomal recessive
FANCL	608111	Fanconi anemia, complementation group L	Autosomal recessive
FGD1	300546	Aarskog-Scott syndrome; Mental retardation, X-linked syndromic, type 16	X-linked
FH	136850	Fumarase deficiency	Autosomal recessive
FKRP	606596	Muscular dystrophy-dystroglycanopathy, type 5A (Walker-Warburg syndrome); Type 5B; Type 5C (limb-girdle muscular dystrophy, type 9 [LGMDR9])	Autosomal recessive
FKTN	607440	Muscular dystrophy-dystroglycanopathy, type 4A (Walker-Warburg syndrome); Type 4B; Type 4C (limb-girdle muscular dystrophy, type 13 [LGMD R13])	Autosomal recessive
FMO3	136132	Trimethylaminuria	Autosomal recessive
FMR1	309550	Fragile X syndrome	X-linked
FOXRED1	613622	Mitochondrial complex I deficiency, nuclear type 19	Autosomal recessive
FRAS1	607830	Fraser syndrome, type 1	Autosomal recessive
FTCD	606806	Glutamate formiminotransferase deficiency	Autosomal recessive
FTSJ1	300499	Mental retardation, X-linked 44	X-linked
FUCA1	612280	Fucosidosis	Autosomal recessive
G6PC1	613742	Glycogen storage disease, type 1A	Autosomal recessive
G6PC3	611045	Dursun syndrome	Autosomal recessive
G6PD	305900	Hemolytic anemia, G6PD deficient (favism)	X-linked
GAA	606800	Glycogen storage disease, type 2	Autosomal recessive
GALC	606890	Krabbe disease	Autosomal recessive
GALE	606953	Galactose epimerase deficiency	Autosomal recessive
GALK1	604313	Galactokinase deficiency with cataracts	Autosomal recessive
GALNS	612222	Mucopolysaccharidosis, type 4A	Autosomal recessive

gene	OMIM ger	Phenotype PATOLOGIA EN	Inheritance
GALNT3	601756	Tumoral calcinosis, hyperphosphatemic, familial, type 1	Autosomal recessive
GALT	606999	Galactosemia	Autosomal recessive
GAMT	601240	Cerebral creatine deficiency syndrome, type 2	Autosomal recessive
GBA	606463	Gaucher disease	Autosomal recessive
GBE1	607839	Glycogen storage disease, type 4	Autosomal recessive
GCDH	608801	Glutaricaciduria, type 1	Autosomal recessive
GCH1	600225	Hyperphenylalaninemia, BH4-deficient, type B	Autosomal recessive
GCSH	238330	Glycine encephalopathy	Autosomal recessive
GDAP1	606598	Charcot-Marie-Tooth disease, recessive intermediate, type A	Autosomal recessive
GDF5	601146	Chondrodysplasia, Grebe type	Autosomal recessive
GFM1	606639	Combined oxidative phosphorylation deficiency, type 1	Autosomal recessive
GH1	139250	Growth hormone deficiency, isolated, type 1A; Kowarski syndrome	Autosomal recessive
GHRHR	139191	Growth hormone deficiency, isolated, type 1B	Autosomal recessive
GJB2	121011	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6	Autosomal recessive; D
GJB3	603324	Deafness autosomal dominant 2B,Deafness autosomal dominant with peripheral neuropathy,Deafness autosomal recessive,Deafness digenic GJB2/GJB3,Erythrokeratodermia variabilis et progressiva 1	Autosomal recessive; D
GJB6	604418	Deafness, autosomal recessive, type 1B; Deafness, digenic GJB2/GJB6	Autosomal recessive; D
GLA	300644	Fabry disease	X-linked
GLB1	611458	GM1-gangliosidosis, types 1-3; Mucopolysaccharidosis, type 4B (Morquio)	Autosomal recessive
GLDC	238300	Glycine encephalopathy	Autosomal recessive
GLE1	603371	Lethal congenital contracture syndrome, type 1; Congenital arthrogryposis with anterior horn cell disease	Autosomal recessive
GNE	603824	Inclusion body myopathy, type 2 (Nonaka myopathy)	Autosomal recessive
GNMT	606628	Glycine N-methyltransferase deficiency	Autosomal recessive
GNPTAB	607840	Mucolipidosis 2 alpha/beta; Mucolipidosis 3 alpha/beta	Autosomal recessive
GNPTG	607838	Mucolipidosis III gamma	Autosomal recessive
GNRHR	138850	Hypogonadotropic hypogonadism, type 7, without anosmia	Autosomal recessive
GNS	607664	Mucopolysaccharidosis, type 3D (Sanfilippo syndrome D)	Autosomal recessive
GORAB	607983	Geroderma osteodysplasticum	Autosomal recessive
GOT2	138150	Epileptic encephalopathy, early infantile, 82	Autosomal recessive
GP1BA	606672	Bernard-Soulier syndrome, type A1	Autosomal recessive
GP1BB	138720	Bernard-Soulier syndrome, type B	Autosomal recessive
GP9	173515	Bernard-Soulier syndrome, type C	Autosomal recessive
GPR143	300808	Ocular albinism, type 1 (Nettleship-Falls type)	X-linked
GPSM2	609245	Chudley-McCullough syndrome	Autosomal recessive
GRHPR	604296	Hyperoxaluria, primary, type 2	Autosomal recessive
GRIA3	305915	Intellectual developmental disorder X-linked syndromic Wu type	X-linked
GRIP1	604597	Fraser syndrome 3	Autosomal recessive
GRN	138945	Ceroid lipofuscinosis, neuronal, type 11	Autosomal recessive
GSS	601002	Glutathione synthetase deficiency	Autosomal recessive
GUCY2D	600179	Leber congenital amaurosis, type 1	Autosomal recessive
GUSB	611499	Mucopolysaccharidosis, type 7	Autosomal recessive
HADH	601609	3-hydroxyacyl-CoA dehydrogenase deficiency	Autosomal recessive
HADHA	600890	Long-chain 3-hydroxyl-CoA dehydrogenase (LCHAD) deficiency; Mitochondrial trifunctional protein deficiency	Autosomal recessive
HADHB	143450	Mitochondrial trifunctional protein deficiency	Autosomal recessive
HAX1	605998	Neutropenia, severe congenital, type 3, autosomal recessive	Autosomal recessive
HBA1	141800	Thalassemia, alpha-	Autosomal recessive
HBA2	141850	Thalassemia, alpha-	Autosomal recessive
HBB	141900	HBB-related hemoglobinopathy	Autosomal recessive
HEXA	606869	Tay-Sachs disease	Autosomal recessive
HEXB	606873	Sandhoff disease, infantile, juvenile, and adult forms	Autosomal recessive
HFE	613609	Hemochromatosis,Transferrin serum level QTL2,Alzheimer disease susceptibility to,Microvascular complications of diabetes 7,Porphyria cutanea tarda susceptibility to,Porphyria variegata susceptibility to	Autosomal recessive
HGD	607474	Alkaptonuria	Autosomal recessive
HGSNAT	610453	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)	Autosomal recessive
HJV	608374	Hemochromatosis, type 2A	Autosomal recessive
HLCS	609018	Holocarboxylase synthetase deficiency	Autosomal recessive
HMGCL	613898	HMG-CoA lyase deficiency	Autosomal recessive
HMOX1	141250	Heme oxygenase-1 deficiency	Autosomal recessive
HOGA1	613597	Hyperoxaluria, primary, type 3	Autosomal recessive
HPD	609695	Tyrosinemia, type 3	Autosomal recessive
HPS1	604982	Hermansky-Pudlak syndrome, type 1	Autosomal recessive

gene	OMIM ger	Phenotype PATOLOGIA EN	Inheritance
HPS3	606118	Hermansky-Pudlak syndrome, type 3	Autosomal recessive
HPS4	606682	Hermansky-Pudlak syndrome, type 4	Autosomal recessive
HSD17B10	300256	HSD10 mitochondrial disease	X-linked
HSD17B3	605573	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	Autosomal recessive
HSD17B4	601860	D-bifunctional protein deficiency	Autosomal recessive
HSD3B2	613890	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	Autosomal recessive
HSPG2	142461	Dyssegmental dysplasia, Silverman-Handmaker type	Autosomal recessive
HUWE1	300697	Intellectual developmental disorder X-linked Turner type	X-linked
HYAL1	607071	Mucopolysaccharidosis, type 9	Autosomal recessive
HYLS1	610693	Hydrolethalus syndrome	Autosomal recessive
IDH3B	604526	Retinitis pigmentosa, type 46	Autosomal recessive
IDS	300823	Mucopolysaccharidosis, type 2	X-linked
IDUA	252800	Mucopolysaccharidosis type 1	Autosomal recessive
IGHMBP2	600502	Charcot-Marie-Tooth disease, axonal, type 2S	Autosomal recessive
IL1RAPL1	300206	Mental retardation, X-linked, type 21/34	X-linked
IL2RG	308380	Severe combined immunodeficiency, X-linked	X-linked
IMPA1	602064	Mental retardation, autosomal recessive 59	Autosomal recessive
ITGB3	173470	Glanzmann thrombasthenia	Autosomal recessive
ITPA	147520	Epileptic encephalopathy, early infantile, type 35	Autosomal recessive
IVD	607036	Isovaleric acidemia	Autosomal recessive
IYD	612025	Thyroid dyshormonogenesis, type 4	Autosomal recessive
JAK3	600173	Severe Combined Immunodeficiency, autosomal recessive, T-negative/B-positive type	Autosomal recessive
KATNB1	602703	Lissencephaly 6, with microcephaly	Autosomal recessive
KCNJ11	600937	Hyperinsulinemic hypoglycemia, type 2 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive; A
KDM5C	314690	Mental retardation, X-linked, syndromic, Claes-Jensen type	X-linked
KIAA0586	610178	Short-rib thoracic dysplasia 14 with polydactyly	Autosomal recessive
L1CAM	308840	L1 Syndrome	X-linked
LAMA2	156225	LAMA2-related muscular dystrophy	Autosomal recessive
LAMA3	600805	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive
LAMB3	150310	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive
LAMC2	150292	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive
LARGE1	603590	Muscular dystrophy-dystroglycanopathy, type 6A and 6B	Autosomal recessive
LCA5	611408	Leber congenital amaurosis, type 5	Autosomal recessive
LDLR	606945	Hypercholesterolemia familial 1,LDL cholesterol level QTL2	Autosomal recessive
LDLRAP1	605747	Hypercholesterolemia, familial, autosomal recessive	Autosomal recessive
LHCGR	152790	Leydig cell hypoplasia	Autosomal recessive
LHX3	600577	Pituitary hormone deficiency, combined, type 3	Autosomal recessive
LIFR	151443	Stuve-Wiedemann syndrome / Schwartz-Jampel type 2 syndrome	Autosomal recessive
LIPA	613497	Lysosomal acid lipase deficiency	Autosomal recessive
LIPH	607365	Hypotrichosis, type 7 or woolly hair, autosomal recessive, type 2, with or without hypotrichosis	Autosomal recessive
LMBRD1	612625	Methylmalonic aciduria and homocystinuria, cblF type	Autosomal recessive
LOXHD1	613072	Deafness, autosomal recessive, type 77	Autosomal recessive
LPL	609708	Lipoprotein lipase deficiency	Autosomal recessive
LRP2	600073	Donnai-Barrow syndrome	Autosomal recessive
LRPPRC	607544	Leigh syndrome, French-Canadian type	Autosomal recessive
LYST	606897	Chediak-Higashi syndrome	Autosomal recessive
MAN2B1	609458	Alpha-mannosidosis	Autosomal recessive
MANBA	609489	Mannosidosis, beta	Autosomal recessive
MAT1A	610550	Methionine adenosyltransferase deficiency, autosomal recessive	Autosomal recessive

gene	OMIM ger	Phenotype PATOLOGIA EN	Inheritance
MCCC1	609010	3-Methylcrotonyl-CoA carboxylase deficiency, type 1	Autosomal recessive
MCCC2	609014	3-Methylcrotonyl-CoA carboxylase deficiency, type 2	Autosomal recessive
MCEE	608419	Methylmalonyl-CoA epimerase deficiency	Autosomal recessive
MCOLN1	605248	Mucolipidosis type 4	Autosomal recessive
MCPH1	607117	Microcephaly type 1, primary, autosomal recessive	Autosomal recessive
MECP2	300005	Encephalopathy, neonatal severe; Rett syndrome	X-linked
MED17	603810	Microcephaly, postnatal progressive, with seizures and brain atrophy	Autosomal recessive
MEFV	608107	Familial Mediterranean fever	Autosomal recessive
MESP2	605195	Spondylocostal dysostosis, type 2, autosomal recessive	Autosomal recessive
MFSD8	611124	Ceroid lipofuscinosis, neuronal, type 7	Autosomal recessive
MKKS	604896	Bardet-Biedl syndrome type 6	Autosomal recessive
MKS1	609883	Bardet-Biedl syndrome type 13; Meckel syndrome, type 1; Joubert syndrome, type 28	Autosomal recessive
MLC1	605908	Megalencephalic leukoencephalopathy with subcortical cysts	Autosomal recessive
MLYCD	606761	Malonyl-CoA decarboxylase deficiency	Autosomal recessive
MMAA	607481	Methylmalonic aciduria, vitamin B12-responsive	Autosomal recessive
MMAB	607568	Methylmalonic aciduria, vitamin B12-responsive, type cblB	Autosomal recessive
MMACHC	609831	Methylmalonic aciduria and homocystinuria, cblC type	Autosomal recessive, di
MMADHC	611935	Homocystinuria, cblD type, variant 1	Autosomal recessive
MMUT	609058	Methylmalonic aciduria, mut(0) type	Autosomal recessive
MOCS1	603707	Molybdenum cofactor deficiency A	Autosomal recessive
MOGS	601336	Congenital disorder of glycosylation, type 2B	Autosomal recessive
MPI	154550	Congenital disorder of glycosylation, type 1B	Autosomal recessive
MPL	159530	Thrombocytopenia, congenital amegakaryocytic	Autosomal recessive
MPV17	137960	Mitochondrial DNA depletion syndrome type 6 (hepatocerebral); Charcot-Marie-Tooth disease, axonal, type 2EE	Autosomal recessive
MRE11	600814	Ataxia-telangiectasia-like disorder 1	Autosomal recessive
MTHFR	607093	Homocystinuria due to MTHFR deficiency	Autosomal recessive
MTMR2	603557	Charcot-Marie-Tooth disease, type 4B1	Autosomal recessive
MTR	156570	Homocystinuria-megaloblastic anemia, cblG complementation type	Autosomal recessive
MTRR	602568	Homocystinuria-megaloblastic anemia, cbl E type	Autosomal recessive
MTTP	157147	Abetalipoproteinemia	Autosomal recessive
MVK	251170	Mevalonic aciduria	Autosomal recessive
MYO15A	602666	Deafness, autosomal recessive, type 3	Autosomal recessive
MYO7A	276903	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2	Autosomal recessive
NADK2	615787	2,4-dienoyl-CoA reductase deficiency	Autosomal recessive
NAGA	104170	Schindler disease, type I	Autosomal recessive
NAGLU	609701	Mucopolysaccharidosis, type 3B (Sanfilippo B)	Autosomal recessive
NAGS	608300	N-acetylglutamate synthase deficiency	Autosomal recessive
NBN	602667	Nijmegen breakage syndrome	Autosomal recessive
NCF1	608512	Chronic granulomatous disease, type 1	Autosomal recessive
NCF2	608515	Chronic granulomatous disease, type 2	Autosomal recessive
NDP	300658	Norrie disease	X-linked
NDRG1	605262	Charcot-Marie-Tooth disease, type 4D	Autosomal recessive
NDUFAF2	609653	Mitochondrial complex I deficiency, nuclear type 10	Autosomal recessive
NDUFAF5	612360	Mitochondrial complex I deficiency, nuclear type 16	Autosomal recessive
NDUFS4	602694	Mitochondrial complex I deficiency, nuclear type 1	Autosomal recessive
NDUFS6	603848	Mitochondrial complex I deficiency, nuclear type 9	Autosomal recessive
NDUFS7	601825	Mitochondrial complex I deficiency, nuclear type 3	Autosomal recessive
NDUFV1	161015	Mitochondrial complex I deficiency, nuclear type 4	Autosomal recessive
NEB	161650	Nemaline myopathy type 2	Autosomal recessive
NEU1	608272	Sialidosis, type 1 and type 2	Autosomal recessive
NHP2	606470	Dyskeratosis congenita, autosomal recessive type 2	Autosomal recessive
NLRP7	609661	Hydatidiform mole, recurrent, type 1	Autosomal recessive
NOP10	606471	Dyskeratosis congenita, autosomal recessive type 1	Autosomal recessive
NPC1	607623	Niemann-Pick disease, type C1	Autosomal recessive
NPC2	601015	Niemann-pick disease, type C2	Autosomal recessive
NPHP1	607100	Joubert syndrome type 4	Autosomal recessive
NPHS1	602716	Nephrotic syndrome, type 1	Autosomal recessive
NPHS2	604766	Nephrotic syndrome, type 2	Autosomal recessive
NR2E3	604485	Enhanced S-cone syndrome (Goldmann-Favre); Retinitis pigmentosa, type 37	Autosomal recessive; A
NTRK1	191315	Insensitivity to pain, congenital, with anhidrosis	Autosomal recessive

gene	OMIM ger	Phenotype PATOLOGIA EN	Inheritance
OAT	613349	Gyrate atrophy of choroid and retina	Autosomal recessive
OCA2	611409	Oculocutaneous albinism type 2	Autosomal recessive
OCRL	300535	Lowe Syndrome; Dent disease type 2	X-linked
OPA3	606580	3-methylglutaconic aciduria, type 3	Autosomal recessive
OPHN1	300127	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	X-linked
OSTM1	607649	Osteopetrosis, autosomal recessive type 5	Autosomal recessive
OTC	300461	Ornithine transcarbamylase deficiency	X-linked
OTOF	603681	Deafness, autosomal recessive, type 9	Autosomal recessive
P3H1	610339	Osteogenesis imperfecta, type 8	Autosomal recessive
PAH	612349	Phenylketonuria	Autosomal recessive
PAK3	300142	Mental retardation, X-linked, type 30	X-linked
PANK2	606157	Neurodegeneration with brain iron accumulation type 1	Autosomal recessive
PC	608786	Pyruvate carboxylase deficiency	Autosomal recessive
PCBD1	126090	Hyperphenylalaninemia, BH4-deficient, type D	Autosomal recessive
PCCA	232000	Propionic acidemia	Autosomal recessive
PCCB	232050	Propionic acidemia	Autosomal recessive
PCDH15	605514	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic	Autosomal recessive; D
PDE6A	180071	Retinitis pigmentosa type 43	Autosomal recessive
PDHB	179060	Pyruvate dehydrogenase E1-beta deficiency	Autosomal recessive
PEX1	602136	Heimler syndrome type 1	Autosomal recessive
PEX10	602859	Peroxisome biogenesis disorder, type 6A (Zellweger syndrome); Peroxisome biogenesis disorder, type 6B	Autosomal recessive
PEX12	601758	Peroxisome biogenesis disorder type 3A (Zellweger)	Autosomal recessive
PEX2	170993	Peroxisome biogenesis disorder type 5A (Zellweger)	Autosomal recessive
PEX26	608666	Peroxisome biogenesis disorder type 7A (Zellweger)	Autosomal recessive
PEX5	600414	Peroxisome biogenesis disorder type 2A (Zellweger)	Autosomal recessive
PEX6	601498	Peroxisome biogenesis disorder, type 4A (Zellweger syndrome); Peroxisome biogenesis disorder, type 4B; Heimler syndrome 2	Autosomal recessive; A
PEX7	601757	Rhizomelic chondrodysplasia punctata, type 1	Autosomal recessive
PFKM	610681	Glycogen storage disease, type 7	Autosomal recessive
PGK1	311800	Phosphoglycerate kinase 1 deficiency	X-linked
PHF8	300560	Mental retardation syndrome, X-linked, Siderius type	X-linked
PHGDH	606879	Neu-Laxova syndrome, type 1; Phosphoglycerate dehydrogenase deficiency	Autosomal recessive
PKHD1	606702	Polycystic kidney disease type 4	Autosomal recessive
PLA2G6	603604	Infantile neuroaxonal dystrophy type 1	Autosomal recessive
PLOD1	153454	Ehlers-Danlos syndrome, kyphoscoliotic type, 1	Autosomal recessive
PLP1	300401	Pelizaeus-Merzbacher disease	X-linked
PMM2	601785	Congenital disorder of glycosylation, type 1A	Autosomal recessive
PNPO	603287	Pyridoxamine 5'-phosphate oxidase deficiency	Autosomal recessive
POLG	174763	POLG-related disorders	Autosomal recessive
POLR1C	610060	Leukodystrophy, hypomyelinating, type 11; Treacher Collins syndrome 3	Autosomal recessive
POMGNT1	606822	Muscular dystrophy-dystroglycanopathy, type 3A (Walker-Warburg syndrome); Type 3B; Type 3C (limb-girdle muscular dystrophy, type 15 [LGMDR15])	Autosomal recessive
POMT1	607423	Muscular dystrophy-dystroglycanopathy, type 1A (Walker-Warburg syndrome); Type 1B; Type 1C (limb-girdle muscular dystrophy, type 11 [LGMD R11])	Autosomal recessive
POMT2	607439	Muscular dystrophy-dystroglycanopathy, type 2A (Walker-Warburg syndrome); Type 2B; Type 2C (limb-girdle muscular dystrophy, type 14 [LGMD R14])	Autosomal recessive
POR	124015	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	Autosomal recessive
POU1F1	173110	Pituitary hormone deficiency, combined, type 1	Autosomal recessive
POU3F4	300039	Deafness, X-linked, type 2	X-linked
PPM1K	611065	Maple syrup urine disease, mild variant	Autosomal recessive
PPT1	600722	Ceroid lipofuscinosis, neuronal, type 1	Autosomal recessive
PQBP1	300463	Renpenning syndrome	X-linked
PRDX1	176763	Methylmalonic aciduria and homocystinuria, cblC type, digenic	Autosomal recessive, di
PRF1	170280	Hemophagocytic lymphohistiocytosis, familial, type 2	Autosomal recessive
PRODH	606810	Hyperprolinemia, type 1	Autosomal recessive
PROP1	601538	Pituitary hormone deficiency, combined, type 2	Autosomal recessive
PRPS1	311850	PRPS1-related disoders	X-linked
PSAP	176801	Combined SAP deficiency	Autosomal recessive

gene	OMIM ger	Phenotype PATOLOGIA EN	Inheritance
PTS	612719	Hyperphenylalaninemia, BH4-deficient, type A	Autosomal recessive
PUS1	608109	Myopathy, lactic acidosis, and sideroblastic anemia, type 1	Autosomal recessive
PYGM	608455	McArdle disease	Autosomal recessive
QDPR	612676	Hyperphenylalaninemia, BH4-deficient, type C	Autosomal recessive
RAB23	606144	Carpenter syndrome	Autosomal recessive
RAG1	179615	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	Autosomal recessive
RAG2	179616	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	Autosomal recessive
RAPSN	601592	Fetal akinesia deformation sequence, type 2; Myasthenic syndrome, congenital, type 11, associated with AChR deficiency	Autosomal recessive
RARS2	611524	Pontocerebellar hypoplasia, type 6	Autosomal recessive
RAX	601881	Isolated microphthalmia, type 3	Autosomal recessive
RDH12	608830	Leber congenital amaurosis, type 13	Autosomal recessive
RLBP1	180090	Bothnia retinal dystrophy; Fundus albipunctatus	Autosomal recessive; A
RMRP	157660	Anauxetic dysplasia, type 1	Autosomal recessive
RNASEH2E	610326	Aicardi-Goutieres syndrome, type 2	Autosomal recessive
RNASEH2C	610330	Aicardi-Goutieres syndrome, type 3	Autosomal recessive
RP2	300757	Retinitis pigmentosa, type 2, X-linked	X-linked
RPE65	180069	RPE65-related Leberÿcongenitalÿamaurosis/early-onset severe retinal dystrophyÿ	Autosomal recessive
RPGR	312610	Retinitis pigmentosa, type 3, X-linked; Cone-rod dystrophy, X-linked, 1	X-linked
RPGRIP1L	610937	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome	Autosomal recessive
RS1	300839	Retinoschisis	X-linked
RTEL1	608833	Dyskeratosis congenita, autosomal recessive type 5	Autosomal recessive
SACS	604490	Spastic ataxia, Charlevoix-Saguenay, type	Autosomal recessive
SAG	181031	Oguchi disease, type 1	Autosomal recessive
SAMHD1	606754	Aicardi-Goutieres syndrome, type 5	Autosomal recessive
SBDS	607444	Shwachman-Diamond syndrome	Autosomal recessive
SCO2	604272	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1	Autosomal recessive
SEPSECS	613009	Pontocerebellar hypoplasia, type 2D	Autosomal recessive
SERPINA1	107400	Alpha-1 antitrypsin deficiency	Autosomal recessive
SGCA	600119	Limb-girdle muscular dystrophy, type 3 (LGMD R3)	Autosomal recessive
SGCB	600900	Limb-girdle muscular dystrophy, type 4 (LGMD R4)	Autosomal recessive
SGCD	601411	Limb-girdle muscular dystrophy, type 6 (LGMD R6)	Autosomal recessive
SGCG	608896	Limb-girdle muscular dystrophy, type 5 (LGMD R5)	Autosomal recessive
SGSH	605270	Mucopolysaccharidosis, type 3A (Sanfilippo A)	Autosomal recessive
SH3TC2	608206	Charcot-Marie-Tooth disease, type 4C	Autosomal recessive
SLC12A3	600968	Gitelman syndrome	Autosomal recessive
SLC12A6	604878	Agenesis of the corpus callosum with peripheral neuropathy	Autosomal recessive
SLC16A2	300095	Allan-Herndon-Dudley syndrome	X-linked
SLC17A5	604322	Salla disease	Autosomal recessive
SLC19A2	603941	Thiamine-responsive megaloblastic anemia syndrome	Autosomal recessive
SLC19A3	606152	Thiamine metabolism dysfunction syndrome, type 2 (biotin- or thiamine-responsive encephalopathy type)	Autosomal recessive
SLC22A5	603377	Carnitine deficiency, systemic primary	Autosomal recessive
SLC25A13	603859	Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset	Autosomal recessive
SLC25A15	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	Autosomal recessive
SLC25A20	613698	Carnitine-acylcarnitine translocase deficiency	Autosomal recessive
SLC26A2	606718	Achondrogenesis, type 1B (diastrophic dysplasia)	Autosomal recessive
SLC26A3	126650	Diarrhea 1, secretory chloride, congenital	Autosomal recessive
SLC26A4	605646	Deafness, autosomal recessive, type 4; Pendred syndrome	Autosomal recessive; D
SLC35A1	605634	Congenital disorder of glycosylation, type 2F	Autosomal recessive
SLC35A3	605632	Arthrogryposis, mental retardation, and seizures	Autosomal recessive
SLC35C1	605881	Congenital disorder of glycosylation, type 2C	Autosomal recessive
SLC35D1	610804	Schneckenbecken dysplasia	Autosomal recessive
SLC37A4	602671	Glycogen storage disease, type 1B	Autosomal recessive
SLC39A4	607059	Acrodermatitis enteropathica	Autosomal recessive
SLC3A1	104614	Cystinuria	Autosomal recessive
SLC45A2	606202	Albinism, oculocutaneous, type 4	Autosomal recessive

gene	OMIM ger	Phenotype PATOLOGIA EN	Inheritance
SLC46A1	611672	Folate malabsorption, hereditary	Autosomal recessive
SLC4A11	610206	Corneal endothelial dystrophy, autosomal recessive	Autosomal recessive
SLC5A5	601843	Thyroid dyshormonogenesis, type 1	Autosomal recessive
SLC6A19	608893	Hartnup disorder	Autosomal recessive
SLC7A7	603593	Lysinuric protein intolerance	Autosomal recessive
SLC7A9	604144	Cystinuria	Autosomal recessive
SMARCAL1	606622	Schimke immunoosseous dysplasia	Autosomal recessive
SMN1	600354	Spinal muscular atrophy	Autosomal recessive
SMPD1	607608	Niemann-Pick disease, type A; Niemann-Pick disease, type B	Autosomal recessive
SPG11	610844	Amyotrophic lateral sclerosis, type 5, juvenile	Autosomal recessive
SPG7	602783	Spastic paraplegia, type 7, autosomal recessive	Autosomal recessive
SRD5A2	607306	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency (pseudovaginal perineoscrotal hypospadias)	Autosomal recessive
ST3GAL5	604402	Salt and pepper developmental regression syndrome	Autosomal recessive
STAR	600617	Lipoid adrenal hyperplasia	Autosomal recessive
STRC	606440	Deafness, autosomal recessive, type 16	Autosomal recessive
SUMF1	607939	Multiple sulfatase deficiency	Autosomal recessive
SURF1	185620	Charcot-Marie-Tooth disease, type 4K; Leigh syndrome, due to COX IV deficiency	Autosomal recessive
SYN1	313440	Epilepsy, X-linked, with variable learning disabilities and behavior disorders	X-linked
TAT	613018	Tyrosinemia, type 2	Autosomal recessive
TCIRG1	604592	Osteopetrosis, autosomal recessive, type 1	Autosomal recessive
TECPR2	615000	Spastic paraplegia, type 49, autosomal recessive	Autosomal recessive
TF	190000	Atransferrinemia	Autosomal recessive
TFR2	604720	Hemochromatosis, type 3	Autosomal recessive
TG	188450	Thyroid dyshormonogenesis, type 3	Autosomal recessive
TGM1	190195	Ichthyosis, congenital, autosomal recessive, type 1	Autosomal recessive
TH	191290	Segawa syndrome, recessive	Autosomal recessive
THOC2	300395	Mental retardation, X-linked 12	X-linked
THRB	190160	Thyroid hormone resistance, autosomal recessive	Autosomal recessive
TMC1	606706	Deafness, autosomal recessive, type 7	Autosomal recessive
TMEM216	613277	Joubert syndrome, type 2; Meckel syndrome, type 2	Autosomal recessive
TMEM67	609884	Joubert syndrome, type 6; Meckel syndrome, type 3; COACH syndrome	Autosomal recessive
TMPRSS3	605511	Deafness, autosomal recessive, type 8/10	Autosomal recessive
TNXB	600985	Ehlers-Danlos syndrome, classic-like	Autosomal recessive
TPO	606765	Thyroid dyshormonogenesis, type 2A	Autosomal recessive
TPP1	607998	Ceroid lipofuscinosis, neuronal, type 2; Spinocerebellar ataxia, autosomal recessive, type 7	Autosomal recessive
TRDN	603283	Ventricular tachycardia, catecholaminergic polymorphic, type 5, with or without muscle weakness	Autosomal recessive
TREX1	606609	Aicardi-Goutieres syndrome, type 1	Autosomal recessive
TRIM32	602290	Limb-girdle muscular dystrophy, type 8 (LGMD R8)	Autosomal recessive
TRIM37	605073	Mulibrey nanism	Autosomal recessive
TRMU	610230	Liver failure, transient infantile	Autosomal recessive
TSEN54	608755	Pontocerebellar hypoplasia, type 2A; Pontocerebellar hypoplasia, type 4	Autosomal recessive
TSFM	604723	Combined oxidative phosphorylation deficiency, type 3	Autosomal recessive
TSHB	188540	Hypothyroidism, congenital, nongoitrous, type 4	Autosomal recessive
TSHR	603372	Hypothyroidism, congenital, nongoitrous, type 1	Autosomal recessive
TSPAN7	300096	Intellectual developmental disorder X-linked 58	X-linked
TTC37	614589	Trichohepatoenteric syndrome, type 1 (diarrhea, syndromic)	Autosomal recessive
TTN	188840	Limb-girdle muscular dystrophy type 10 (LGMDR10); Early-onset myopathy with fatal cardiomyopathy (Salih myopathy)	Autosomal recessive
TTPA	600415	Ataxia with isolated vitamin E deficiency	Autosomal recessive
TYMP	131222	Mitochondrial DNA depletion syndrome, type 1 (MNGIE type)	Autosomal recessive

gene	OMIM ger	Phenotype PATOLOGIA EN	Inheritance
TYR	606933	Oculocutaneous albinism (OCA) type 1A; OCA type 1B; Waardenburg syndrome/albinism, digenic	Autosomal recessive, di
TYRP1	115501	Albinism, oculocutaneous, type 3	Autosomal recessive
UBE3A	601623	Angelman syndrome	Autosomal dominant
UGT1A1	191740	Crigler-Najjar syndrome, type 1; Crigler-Najjar syndrome, type 2	Autosomal recessive
UNC13D	608897	Hemophagocytic lymphohistiocytosis, familial, type 3	Autosomal recessive
UPF3B	300298	Mental retardation, X-linked, syndromic, type 14	X-linked
USH1C	605242	Usher syndrome, type 1C; Deafness, autosomal recessive, type 18A	Autosomal recessive
USH1G	607696	Usher syndrome, type 1G	Autosomal recessive
USH2A	608400	Usher syndrome, type 2A	Autosomal recessive
USP9X	300072	Intellectual developmental disorder X-linked 99,Intellectual developmental disorder X-linked 99 syndromic female-restricted	X-linked
VPS13A	605978	Choreoacanthocytosis	Autosomal recessive
VPS13B	607817	Cohen syndrome	Autosomal recessive
VPS45	610035	Neutropenia, severe congenital, type 5	Autosomal recessive
VPS53	615850	Pontocerebellar hypoplasia, type 2E	Autosomal recessive
VRK1	602168	Pontocerebellar hypoplasia, type 1A	Autosomal recessive
VSX2	142993	Microphthalmia with coloboma 3; Isolated microphthalmia 2	Autosomal recessive
WHRN	607928	Usher syndrome, type 2D; Deafness, autosomal recessive, type 31	Autosomal recessive
WNT10A	606268	Odontoonychodermal dysplasia	Autosomal recessive
WRN	604611	Werner syndrome	Autosomal recessive
XPA	611153	Xeroderma pigmentosum, group A	Autosomal recessive
XPC	613208	Xeroderma pigmentosum, group C	Autosomal recessive
ZDHHC9	300646	Mental retardation, X-linked syndromic, Raymond type	X-linked
ZFYVE26	612012	Spastic paraplegia, type 15, autosomal recessive	Autosomal recessive
ZNF711	314990	Mental retardation, X-linked, type 97	X-linked