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chrom	OMIM (gene)	Gene	Previous symbol	OMIM (phen)	DISEASE	MOI
2	607800	ABCA12		601277; 242500	Ichthyosis, congenital, autosomal recessive, type 4A; ICAR, type 4B (harlequin)	Autosomal recessive
2	603201	ABCB11		605479; 601847	Cholestasis, benign recurrent intrahepatic, type 2; Cholestasis, progressive familial intrahepatic, type 2	Autosomal recessive
11	600509	ABCC8		256450*; 606176*	Hyperinsulinemic hypoglycemia, type 1 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive*
X	300371	ABCD1		300100	Adrenoleukodystrophy	X-linked
3	611103	ACAD9		611126	Acyl-CoA dehydrogenase 9 deficiency (mitochondrial complex I deficiency, nuclear, type 20)	Autosomal recessive
1	607008	ACADM		201450	Medium-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
12	606885	ACADS		201470	Short-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
17	609575	ACADVL		201475	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	Autosomal recessive
11	607809	ACAT1		203750	Alpha-methylacetoaceti c aciduria (3-ketothiolase deficiency)	Autosomal recessive
17	609751	ACOX1		264470	Peroxisomal acyl-CoA oxidase deficiency	Autosomal recessive
20	608958	ADA		102700	Severe combined immunodeficiency due to adenosine deaminase deficiency (ADA)	Autosomal recessive

Maternal inheritance of a mutation in the UBE3A gene causes Angelman syndrome*
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5	604539	ADAMTS2		225410	Ehlers-Danlos syndrome, dermatosparaxis type	Autosomal recessive
4	613228	AGA		208400	Aspartylglucosaminuria (glycosylasparaginase deficiency)	Autosomal recessive
1	610860	AGL		232400	Glycogen storage disease, type 3	Autosomal recessive
2	603051	AGPS		600121	Rhizomelic chondrodysplasia punctata, type 3	Autosomal recessive
2	604285	AGXT		259900	Hyperoxaluria, primary, type 1	Autosomal recessive
6	608894	AHI1		608629	Joubert syndrome, type 3	Autosomal recessive
21	607358	AIRE		240300*	Autoimmune polyendocrinopathy syndrome, type 1	Autosomal recessive*
17	609523	ALDH3A2		270200	Sjogren-Larsson syndrome	Autosomal recessive
9	612724	ALDOB		229600	Fructose intolerance, hereditary	Autosomal recessive
16	605907	ALG1		608540	Congenital disorder of glycosylation, type 1K	Autosomal recessive
1	604566	ALG6		603147	Congenital disorder of glycosylation, type 1C	Autosomal recessive
2	606844	ALMS1		203800	Alström syndrome	Autosomal recessive
1	171760	ALPL		241500; 241510	Hypophosphatasia, infantile/childhood	Autosomal recessive
3	238310	AMT		605899	Glycine encephalopathy	Autosomal recessive
X	313700	AR		300068	Androgen insensitivity syndrome, complete	X-linked
6	608313	ARG1		207800	Argininemia (arginase deficiency)	Autosomal recessive
22	607574	ARSA		250100	Metachromatic leukodystrophy	Autosomal recessive
5	611542	ARSB		253200	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)	Autosomal recessive

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X	300180	ARSL	ARSE	302950	Chondrodysplasia punctata, brachytelephalangic	X-linked
7	608310	ASL		207900	Argininosuccinic aciduria	Autosomal recessive
17	608034	ASPA		271900	Canavan disease	Autosomal recessive
9	603470	ASS1		215700	Citrullinemia, type 1	Autosomal recessive
11	607585	ATM		208900	Ataxia-telangiectasia	Autosomal recessive
X	300011	ATP7A		309400; 304150	Menkes disease; Occipital horn syndrome	X-linked
13	606882	ATP7B		277900	Wilson disease	Autosomal recessive
18	602397	ATP8B1		211600; 243300	Cholestasis, progressive familial intrahepatic, type 1; Cholestasis, benign recurrent intrahepatic, type 1	Autosomal recessive
9	137060	B4GALT1		607091	Congenital disorder of glycosylation, type 2D	Autosomal recessive
11	209901	BBS1		209900	Bardet-Biedl syndrome, type 1	Autosomal recessive
12	610148	BBS10		615987	Bardet-Biedl syndrome, type 10	Autosomal recessive
16	606151	BBS2		615981	Bardet-Biedl syndrome, type 2	Autosomal recessive
19	608348	BCKDHA		248600	Maple syrup urine disease, type 1A	Autosomal recessive
6	248611	BCKDHB		248600	Maple syrup urine disease, type 1B	Autosomal recessive
2	603647	BCS1L		256000	BCS1L-related disorders, including Leigh syndrome	Autosomal recessive
15	604610	BLM		210900	Bloom syndrome	Autosomal recessive
1	606412	BSND		602522	Bartter syndrome, type 4A	Autosomal recessive
3	609019	BTD		253260	Biotinidase deficiency	Autosomal recessive
X	300300	BTK		300755	Agammaglobulinemia X-linked, type 1	X-linked

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8	611492	CA2		259730	Osteopetrosis with renal tubular acidosis (osteopetrosis, autosomal recessive, type 3)	Autosomal recessive
15	114240	CAPN3		253600	Limb-girdle muscular dystrophy, type 1 (LGMD R1)	Autosomal recessive
21	613381	CBS		236200	Homocystinuria due to cystathionine beta-synthase	Autosomal recessive
X	300386	CD40LG		308230	Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)	X-linked
10	605516	CDH23		601386; 601067	Deafness, autosomal recessive, type 12; Usher syndrome, type 1D	Autosomal recessive
12	610142	CEP290		611134; 610188; 611755	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10	Autosomal recessive
2	608381	CERKL		608380	Retinitis pigmentosa, type 26	Autosomal recessive
7	602421	CFTR		219700	Cystic fibrosis	Autosomal recessive
10	118490	CHAT		254210	Myasthenic syndrome, congenital, type 6, presynaptic	Autosomal recessive
X	300390	CHM		303100	Choroideremia	X-linked
17	100725	CHRNE		616324; 608931	Myasthenic syndrome, congenital, type 4B, fast-channel; Myasthenic syndrome, congenital, type 4C, associated with acetylcholine receptor deficiency	Autosomal recessive
16	607042	CLN3		204200	Ceroid lipofuscinosis, neuronal, type 3	Autosomal recessive

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13	608102	CLN5		256731	Ceroid lipofuscinosis, neuronal, type 5	Autosomal recessive
15	606725	CLN6		601780	Ceroid lipofuscinosis, neuronal, type 6	Autosomal recessive
8	607837	CLN8		600143	Ceroid lipofuscinosis, neuronal, type 8	Autosomal recessive
3	606397	CLRN1		276902	Usher syndrome, type 3A	Autosomal recessive
8	605080	CNGB3		262300	Achromatopsia, type 3	Autosomal recessive
2	120070	COL4A3		203780	Alport syndrome, autosomal recessive, type 2	Autosomal recessive
2	120131	COL4A4		203780	Alport syndrome, autosomal recessive, type 2	Autosomal recessive
3	120120	COL7A1		226600; 604129*; 131850*	Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial	Autosomal recessive; Autosomal recessive*; Autosomal recessive*
3	603033	COLQ		603034	Myasthenic syndrome, congenital, type 5	Autosomal recessive
2	608307	CPS1		237300	Carbamoylphosphate synthetase 1 deficiency	Autosomal recessive
11	600528	CPT1A		255120	Carnitine palmitoyltransferase type 1A deficiency, hepatic	Autosomal recessive
1	600650	CPT2		608836; 600649	Carnitine palmitoyltransferase type 2 deficiency, lethal neonatal; Carnitine palmitoyltransferase type 2 deficiency, infantile	Autosomal recessive
1	604210	CRB1		600105; 613835	Retinitis pigmentosa, type 12; Leber congenital amaurosis, type 8	Autosomal recessive

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chrom	OMIM (gene)	Gene	Previous symbol	OMIM (phen)	DISEASE	MOI
3	605497	CRTAP		610682	Osteogenesis imperfecta, type 7	Autosomal recessive
17	606272	CTNS		219800	Nephropathic cystinosis	Autosomal recessive
11	116840	CTSD		610127	Ceroid lipofuscinosis, neuronal, type 10	Autosomal recessive
1	601105	CTSK		265800	Pycnodysostosis	Autosomal recessive
16	608508	CYBA		233690	Chronic granulomatous disease, type 4	Autosomal recessive
X	300481	CYBB		306400	Chronic granulomatous disease, X-linked	X-linked
2	601771	CYP1B1		231300	Glaucoma, primary congenital, type 3A	Autosomal recessive
2	606530	CYP27A1		213700	Cerebrotendinous xanthomatosis	Autosomal recessive
1	248610	DBT		248600	Maple syrup urine disease, type 2	Autosomal recessive
10	605988	DCLRE1C		603554; 602450	Omenn syndrome; Severe combined immunodeficiency, Athabascan type	Autosomal recessive
11	600811	DDB2		278740	Xeroderma pigmentosum, complementation group E	Autosomal recessive
11	602858	DHCR7		270400	Smith-Lemli-Opitz syndrome	Autosomal recessive
1	608172	DHDDS		613861	Retinitis pigmentosa, type 59	Autosomal recessive
7	238331	DLD		246900	Dihydrolipoamide dehydrogenase deficiency	Autosomal recessive
4	610285	DOK7		618389; 254300	Fetal akinesia deformation sequence, type 3; Myasthenic syndrome, congenital, type 10	Autosomal recessive
9	610746	DOLK		610768	Congenital disorder of glycosylation, type 1M	Autosomal recessive
11	191350	DPAGT1		608093; 614750	Congenital disorder of glycosylation, type 1J; Myasthenic syndrome, congenital, type 13	Autosomal recessive

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20	603503	DPM1		608799	Congenital disorder of glycosylation, type 1E	Autosomal recessive
1	612779	DPYD		274270	Dihydropyrimidine dehydrogenase deficiency	Autosomal recessive
2	603009	DYSF		254130; 253601	Miyoshi muscular dystrophy, type 1; Limb-girdle muscular dystrophy, type 2 (LGMD R2)	Autosomal recessive
X	300451	EDA		305100	Ectodermal dysplasia, type 1, hypohidrotic, X-linked	X-linked
3	603945	EIF2B5		603896	Leukoencephalopathy with vanishing white matter	Autosomal recessive
X	300384	EMD		310300	Emery-Dreifuss muscular dystrophy, type 1, X-linked	X-linked
19	126340	ERCC2		601675	Trichothiodystrophy, type 1	Autosomal recessive
2	133510	ERCC3		616390	Trichothiodystrophy, type 2	Autosomal recessive
13	133530	ERCC5		616570	Cerebrooculofacioskeletal syndrome, type 3	Autosomal recessive
8	609353	ESCO2		268300	Roberts syndrome	Autosomal recessive
15	608053	ETFA		231680	Glutaric acidemia, type 2A	Autosomal recessive
19	130410	ETFB		231680	Glutaric acidemia, type 2B	Autosomal recessive
4	231675	ETFDH		231680	Glutaric acidemia, type 2C	Autosomal recessive
19	608451	ETHE1		602473	Ethylmalonic encephalopathy	Autosomal recessive
6	612424	EYS		602772	Retinitis pigmentosa, type 25	Autosomal recessive
X	300841	F8		306700	Hemophilia A	X-linked
X	300746	F9		306900	Hemophilia B	X-linked
15	613871	FAH		276700	Tyrosinemia, type 1	Autosomal recessive
7	611061	FAM20C		259775	Raine syndrome	Autosomal recessive

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16	607139	FANCA		227650	Fanconi anemia, complementation group A	Autosomal recessive
9	613899	FANCC		227645	Fanconi anemia, complementation group C	Autosomal recessive
9	602956	FANCG		614082	Fanconi anemia, complementation group G	Autosomal recessive
1	136850	FH		606812	Fumarase deficiency	Autosomal recessive
19	606596	FKRP		613153; 606612; 607155	Muscular dystrophy-dystroglycan opathy, type 5A (Walker-Warburg syndrome); Type 5B; Type 5C (limb-girdle muscular dystrophy, type 9 [LGMDR9])	Autosomal recessive
9	607440	FKTN		253800; 613152; 611588	Muscular dystrophy-dystroglycan opathy, type 4A (Walker-Warburg syndrome); Type 4B; Type 4C (limb-girdle muscular dystrophy, type 13 [LGMD R13])	Autosomal recessive
X	309550	FMR1		300624	Fragile X syndrome	X-linked
4	607830	FRAS1		219000	Fraser syndrome, type 1	Autosomal recessive
17	613742	G6PC		232200	Glycogen storage disease, type 1A	Autosomal recessive
17	611045	G6PC3		612541	Dursun syndrome	Autosomal recessive
X	305900	G6PD		300908	Hemolytic anemia, G6PD deficient (favism)	X-linked
17	606800	GAA		232300	Glycogen storage disease, type 2	Autosomal recessive
14	606890	GALC		245200	Krabbe disease	Autosomal recessive
17	604313	GALK1		230200	Galactokinase deficiency with cataracts	Autosomal recessive

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chrom	OMIM (gene)	Gene	Previous symbol	OMIM (phen)	DISEASE	MOI
16	612222	GALNS		253000	Mucopolysaccharidosis, type 4A	Autosomal recessive
9	606999	GALT		230400	Galactosemia	Autosomal recessive
19	601240	GAMT		612736	Cerebral creatine deficiency syndrome, type 2	Autosomal recessive
1	606463	GBA		230800	Gaucher disease	Autosomal recessive
3	607839	GBE1		232500	Glycogen storage disease, type 4	Autosomal recessive
19	608801	GCDH		231670	Glutaricaciduria, type 1	Autosomal recessive
14	600225	GCH1		233910	Hyperphenylalaninemia, BH4-deficient, type B	Autosomal recessive
3	606639	GFM1		609060	Combined oxidative phosphorylation deficiency, type 1	Autosomal recessive
13	121011	GJB2		220290	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6	Autosomal recessive; Digenic inheritance (GJB6 gene)
X	300644	GLA		301500	Fabry disease	X-linked
3	611458	GLB1		230500, 230600, 230650; 253010	GM1-gangliosidosis, types 1-3; Mucopolysaccharidosis, type 4B (Morquio)	Autosomal recessive
9	238300	GLDC		605899	Glycine encephalopathy	Autosomal recessive
9	603371	GLE1		253310; 611890	Lethal congenital contracture syndrome, type 1; Congenital arthrogryposis with anterior horn cell disease	Autosomal recessive
9	603824	GNE		605820	Inclusion body myopathy, type 2 (Nonaka myopathy)	Autosomal recessive
12	607840	GNPTAB		252500; 252600	Mucolipidosis 2 alpha/beta; Mucolipidosis 3 alpha/beta	Autosomal recessive
12	607664	GNS		252940	Mucopolysaccharidosis, type 3D (Sanfilippo syndrome D)	Autosomal recessive

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X	300808	GPR143		300500	Ocular albinism, type 1 (Nettleship-Falls type)	X-linked
9	604296	GRHPR		260000	Hyperoxaluria, primary, type 2	Autosomal recessive
7	611499	GUSB		253220	Mucopolysaccharidosis, type 7	Autosomal recessive
2	600890	HADHA		609016; 609015	Long-chain 3-hydroxyl-CoA dehydrogenase (LCHAD) deficiency; Mitochondrial trifunctional protein deficiency	Autosomal recessive
2	143450	HADHB		609015	Mitochondrial trifunctional protein deficiency	Autosomal recessive
1	605998	HAX1		610738	Neutropenia, severe congenital, type 3, autosomal recessive	Autosomal recessive
16	141800	HBA1		604131	Thalassemia, alpha-	Autosomal recessive
16	141850	HBA2		604131	Thalassemia, alpha-	Autosomal recessive
11	141900	HBB		603903	HBB-related hemoglobinopathy	Autosomal recessive
15	606869	HEXA		272800	Tay-Sachs disease	Autosomal recessive
5	606873	HEXB		268800	Sandhoff disease, infantile, juvenile, and adult forms	Autosomal recessive
3	607474	HGD		203500	Alkaptonuria	Autosomal recessive
8	610453	HGSNAT		252930	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)	Autosomal recessive
21	609018	HLCS		253270	Holocarboxylase synthetase deficiency	Autosomal recessive
1	613898	HMGCL		246450	HMG-CoA lyase deficiency	Autosomal recessive
10	604982	HPS1		203300	Hermansky-Pudlak syndrome, type 1	Autosomal recessive
3	606118	HPS3		614072	Hermansky-Pudlak syndrome, type 3	Autosomal recessive
5	601860	HSD17B4		261515	D-bifunctional protein deficiency	Autosomal recessive

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1	142461	HSPG2		224410	Dyssegmental dysplasia, Silverman-Handmaker type	Autosomal recessive
11	610693	HYLS1		236680	Hydrolethalus syndrome	Autosomal recessive
X	300823	IDS		309900	Mucopolysaccharidosis, type 2	X-linked
4	252800	IDUA		607014; 607015; 607016	Mucopolysaccharidosis type 1	Autosomal recessive
11	600502	IGHMBP2		616155	Charcot-Marie-Tooth disease, axonal, type 2S	Autosomal recessive
X	308380	IL2RG		300400	Severe combined immunodeficiency, X-linked	X-linked
15	607036	IVD		243500	Isovaleric acidemia	Autosomal recessive
11	600937	KCNJ11		601820; 606176*	Hyperinsulinemic hypoglycemia, type 2 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive; Autosomal recessive*
X	308840	L1CAM		307000; 303350; 304100	L1 Syndrome	X-linked
6	156225	LAMA2		607855; 618138	LAMA2-related muscular dystrophy	Autosomal recessive
18	600805	LAMA3		226700; 226650	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive
1	150310	LAMB3		226700; 226650	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive
1	150292	LAMC2		226700; 226650	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive

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22	603590	LARGE1	LARGE	613154; 608840	Muscular dystrophy-dystroglycan opathy, type 6A and 6B	Autosomal recessive
9	600577	LHX3		221750	Pituitary hormone deficiency, combined, type 3	Autosomal recessive
5	151443	LIFR		601559	Stuve-Wiedemann syndrome / Schwartz-Jampel type 2 syndrome	Autosomal recessive
10	613497	LIPA		278000	Lysosomal acid lipase deficiency	Autosomal recessive
18	613072	LOXHD1		613079	Deafness, autosomal recessive, type 77	Autosomal recessive
2	607544	LRPPRC		220111	Leigh syndrome, French-Canadian type	Autosomal recessive
1	606897	LYST		214500	Chediak-Higashi syndrome	Autosomal recessive
19	609458	MAN2B1		248500	Alpha-mannosidosis	Autosomal recessive
5	609014	MCCC2		210210	3-Methylcrotonyl-CoA carboxylase deficiency, type 2	Autosomal recessive
19	605248	MCOLN1		252650	Mucolipidosis type 4	Autosomal recessive
X	300005	MECP2		300673; 312750	Encephalopathy, neonatal severe; Rett syndrome	X-linked
16	608107	MEFV		249100	Familial Mediterranean fever	Autosomal recessive
4	611124	MFSD8		610951	Ceroid lipofuscinosis, neuronal, type 7	Autosomal recessive
17	609883	MKS1		615990; 249000; 617121	Bardet-Biedl syndrome type 13; Meckel syndrome, type 1; Joubert syndrome, type 28	Autosomal recessive
22	605908	MLC1		604004	Megalencephalic leukoencephalopathy with subcortical cysts	Autosomal recessive
4	607481	MMAA		251100	Methylmalonic aciduria, vitamin B12-responsive	Autosomal recessive

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12	607568	MMAB		251110	Methylmalonic aciduria, vitamin B12-responsive, type cblB	Autosomal recessive
1	609831	MMACHC		277400	Methylmalonic aciduria and homocystinuria, cblC type	Autosomal recessive, digenic inheritance (PRDX1 gene)
2	611935	MMADHC		277410	Homocystinuria, cblD type, variant 1	Autosomal recessive
6	609058	MMUT	MUT	251000	Methylmalonic aciduria, mut(0) type	Autosomal recessive
2	601336	MOGS		606056	Congenital disorder of glycosylation, type 2B	Autosomal recessive
15	154550	MPI		602579	Congenital disorder of glycosylation, type 1B	Autosomal recessive
1	159530	MPL		604498	Thrombocytopenia, congenital amegakaryocytic	Autosomal recessive
2	137960	MPV17		256810; 618400	Mitochondrial DNA depletion syndrome type 6 (hepatocerebral); Charcot-Marie-Tooth disease, axonal, type 2EE	Autosomal recessive
X	300415	MTM1		310400	Myotubular myopathy, X-linked	X-linked
1	156570	MTR		250940	Homocystinuria-megaloblastic anemia, cblG complementation type	Autosomal recessive
5	602568	MTRR		236270	Homocystinuria-megaloblastic anemia, cbl E type	Autosomal recessive
4	157147	MTTP		200100	Abetalipoproteinemia	Autosomal recessive
11	276903	MYO7A		276900; 600060	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2	Autosomal recessive
17	609701	NAGLU		252920	Mucopolysaccharidosis, type 3B (Sanfilippo B)	Autosomal recessive
17	608300	NAGS		237310	N-acetylglutamate synthase deficiency	Autosomal recessive

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chrom	OMIM (gene)	Gene	Previous symbol	OMIM (phen)	DISEASE	MOI
8	602667	NBN		251260	Nijmegen breakage syndrome	Autosomal recessive
7	608512	NCF1		233700	Chronic granulomatous disease, type 1	Autosomal recessive
1	608515	NCF2		233710	Chronic granulomatous disease, type 2	Autosomal recessive
8	605262	NDRG1		601455	Charcot-Marie-Tooth disease, type 4D	Autosomal recessive
2	161650	NEB		256030	Nemaline myopathy type 2	Autosomal recessive
5	606470	NHP2		613987	Dyskeratosis congenita, autosomal recessive type 2	Autosomal recessive
15	606471	NOP10		224230	Dyskeratosis congenita, autosomal recessive type 1	Autosomal recessive
18	607623	NPC1		257220	Niemann-Pick disease, type C1	Autosomal recessive
14	601015	NPC2		607625	Niemann-pick disease, type C2	Autosomal recessive
19	602716	NPHS1		256300	Nephrotic syndrome, type 1	Autosomal recessive
1	604766	NPHS2		600995	Nephrotic syndrome, type 2	Autosomal recessive
X	300473	NR0B1		300200	Adrenal hypoplasia, congenital	X-linked
15	604485	NR2E3		268100; 611131*	Enhanced S-cone syndrome (Goldmann-Favre); Retinitis pigmentosa, type 37	Autosomal recessive; Autosomal recessive*
1	191315	NTRK1		256800	Insensitivity to pain, congenital, with anhidrosis	Autosomal recessive
10	613349	OAT		258870	Gyrate atrophy of choroid and retina	Autosomal recessive
15	611409	OCA2		203200	Oculocutaneous albinism type 2	Autosomal recessive
X	300535	OCRL		309000; 300555	Lowe Syndrome; Dent disease type 2	X-linked

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19	606580	OPA3		258501	3-methylglutaconic aciduria, type 3	Autosomal recessive
6	607649	OSTM1		259720	Osteopetrosis, autosomal recessive type 5	Autosomal recessive
X	300461	OTC		311250	Ornithine transcarbamylase deficiency	X-linked
1	610339	P3H1	LEPRE1	610915	Osteogenesis imperfecta, type 8	Autosomal recessive
12	612349	PAH		261600	Phenylketonuria	Autosomal recessive
11	608786	PC		266150	Pyruvate carboxylase deficiency	Autosomal recessive
13	232000	PCCA		606054	Propionic acidemia	Autosomal recessive
3	232050	PCCB		606054	Propionic acidemia	Autosomal recessive
10	605514	PCDH15		609533; 601067	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic	Autosomal recessive
X	300502	PDHA1		312170	Pyruvate dehydrogenase E1-alpha deficiency	X-linked
7	602136	PEX1		234580	Heimler syndrome type 1	Autosomal recessive
8	170993	PEX2		614866	Peroxisome biogenesis disorder type 5A (Zellweger)	Autosomal recessive
12	600414	PEX5		214110	Peroxisome biogenesis disorder type 2A (Zellweger)	Autosomal recessive
6	601498	PEX6		614862; 616617*; 614863	Peroxisome biogenesis disorder, type 4A (Zellweger syndrome); Peroxisome biogenesis disorder, type 4B; Heimler syndrome 2	Autosomal recessive; Autosomal recessive*; Autosomal recessive
6	601757	PEX7		215100	Rhizomelic chondrodysplasia punctata, type 1	Autosomal recessive

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1	606879	PHGDH		256520; 601815	Neu-Laxova syndrome, type 1; Phosphoglycerate dehydrogenase deficiency	Autosomal recessive
6	606702	PKHD1		263200	Polycystic kidney disease type 4	Autosomal recessive
22	603604	PLA2G6		256600	Infantile neuroaxonal dystrophy type 1	Autosomal recessive
1	153454	PLOD1		225400	Ehlers-Danlos syndrome, kyphoscoliotic type, 1	Autosomal recessive
16	601785	PMM2		212065	Congenital disorder of glycosylation, type 1A	Autosomal recessive
17	603287	PNPO		610090	Pyridoxamine 5'-phosphate oxidase deficiency	Autosomal recessive
15	174763	POLG		203700; 613662; 607459	POLG-related disorders	Autosomal recessive
1	606822	POMGNT1		253280; 613151; 613157	Muscular dystrophy-dystroglycan opathy, type 3A (Walker-Warburg syndrome); Type 3B; Type 3C (limb-girdle muscular dystrophy, type 15 [LGMDR15])	Autosomal recessive
9	607423	POMT1		236670; 613155; 609308	Muscular dystrophy-dystroglycan opathy, type 1A (Walker-Warburg syndrome); Type 1B; Type 1C (limb-girdle muscular dystrophy, type 11 [LGMD R11])	Autosomal recessive
14	607439	POMT2		613150; 613156; 613158	Muscular dystrophy-dystroglycan opathy, type 2A (Walker-Warburg syndrome); Type 2B; Type 2C (limb-girdle muscular dystrophy, type 14 [LGMD R14])	Autosomal recessive

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3	173110	POU1F1		613038*	Pituitary hormone deficiency, combined, type 1	Autosomal recessive*
1	600722	PPT1		256730	Ceroid lipofuscinosis, neuronal, type 1	Autosomal recessive
10	170280	PRF1		603553	Hemophagocytic lymphohistiocytosis, familial, type 2	Autosomal recessive
5	601538	PROP1		262600	Pituitary hormone deficiency, combined, type 2	Autosomal recessive
10	176801	PSAP		611721	Combined SAP deficiency	Autosomal recessive
11	612719	PTS		261640	Hyperphenylalaninemia, BH4-deficient, type A	Autosomal recessive
11	608455	PYGM		232600	McArdle disease	Autosomal recessive
4	612676	QDPR		261630	Hyperphenylalaninemia, BH4-deficient, type C	Autosomal recessive
6	606144	RAB23		201000	Carpenter syndrome	Autosomal recessive
11	179615	RAG1		603554; 601457	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	Autosomal recessive
11	179616	RAG2		603554; 601457	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	Autosomal recessive
11	601592	RAPSN		208150; 616326	Fetal akinesia deformation sequence, type 2; Myasthenic syndrome, congenital, type 11, associated with AChR deficiency	Autosomal recessive
14	608830	RDH12		612712	Leber congenital amaurosis, type 13	Autosomal recessive
1	180069	RPE65		204100; 613794	RPE65-related Leber congenital amaurosis/early-onset severe retinal dystrophy	Autosomal recessive

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16	610937	RPGRIP1L		611560; 611561; 619113	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome	Autosomal recessive
20	608833	RTEL1		615190*	Dyskeratosis congenita, autosomal recessive type 5	Autosomal recessive*
13	604490	SACS		270550	Spastic ataxia, Charlevoix-Saguenay, type	Autosomal recessive
7	607444	SBDS		260400	Shwachman-Diamond syndrome	Autosomal recessive
17	600119	SGCA		608099	Limb-girdle muscular dystrophy, type 3 (LGMD R3)	Autosomal recessive
4	600900	SGCB		604286	Limb-girdle muscular dystrophy, type 4 (LGMD R4)	Autosomal recessive
13	608896	SGCG		253700	Limb-girdle muscular dystrophy, type 5 (LGMD R5)	Autosomal recessive
17	605270	SGSH		252900	Mucopolysaccharidosis, type 3A (Sanfilippo A)	Autosomal recessive
X	300490	SH2D1A		308240	Lymphoproliferative syndrome, X-linked, type 1	X-linked
16	600968	SLC12A3		263800	Gitelman syndrome	Autosomal recessive
15	604878	SLC12A6		218000	Agenesis of the corpus callosum with peripheral neuropathy	Autosomal recessive
6	604322	SLC17A5		604369	Salla disease	Autosomal recessive
5	603377	SLC22A5		212140	Carnitine deficiency, systemic primary	Autosomal recessive
7	603859	SLC25A13		605814; 603471	Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset	Autosomal recessive
13	603861	SLC25A15		238970	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	Autosomal recessive

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5	606718	SLC26A2		600972	Achondrogenesis, type 1B (diastrophic dysplasia)	Autosomal recessive
7	605646	SLC26A4		600791; 274600	Deafness, autosomal recessive, type 4; Pendred syndrome	Autosomal recessive
6	605634	SLC35A1		603585	Congenital disorder of glycosylation, type 2F	Autosomal recessive
1	605632	SLC35A3		615553	?Arthrogryposis, mental retardation, and seizures	Autosomal recessive
11	605881	SLC35C1		266265	Congenital disorder of glycosylation, type 2C	Autosomal recessive
1	610804	SLC35D1		269250	Schneckenbecken dysplasia	Autosomal recessive
11	602671	SLC37A4		232220	Glycogen storage disease, type 1B	Autosomal recessive
5	606202	SLC45A2		606574	Albinism, oculocutaneous, type 4	Autosomal recessive
20	610206	SLC4A11		217700	Corneal endothelial dystrophy, autosomal recessive	Autosomal recessive
X	300036	SLC6A8		300352	Cerebral creatine deficiency syndrome, type 1	X-linked
14	603593	SLC7A7		222700	Lysinuric protein intolerance	Autosomal recessive
5	600354	SMN1		253300	Spinal muscular atrophy	Autosomal recessive
11	607608	SMPD1		257200; 607616	Niemann-Pick disease, type A; Niemann-Pick disease, type B	Autosomal recessive
8	600617	STAR		201710	Lipoid adrenal hyperplasia	Autosomal recessive
3	607939	SUMF1		272200	Multiple sulfatase deficiency	Autosomal recessive
11	604592	TCIRG1		259700	Osteopetrosis, autosomal recessive, type 1	Autosomal recessive
7	604720	TFR2		604250	Hemochromatosis, type 3	Autosomal recessive

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14	190195	TGM1		242300	Ichthyosis, congenital, autosomal recessive, type 1	Autosomal recessive
11	191290	TH		605407	Segawa syndrome, recessive	Autosomal recessive
11	613277	TMEM216		608091; 603194	Joubert syndrome, type 2; Meckel syndrome, type 2	Autosomal recessive
11	607998	TPP1		204500; 609270	Ceroid lipofuscinosis, neuronal, type 2; Spinocerebellar ataxia, autosomal recessive, type 7	Autosomal recessive
17	608755	TSEN54		277470; 225753	Pontocerebellar hypoplasia, type 2A; Pontocerebellar hypoplasia, type 4	Autosomal recessive
12	604723	TSFM		610505	Combined oxidative phosphorylation deficiency, type 3	Autosomal recessive
8	600415	TTPA		277460	Ataxia with isolated vitamin E deficiency	Autosomal recessive
11	606933	TYR		203100; 606952	Oculocutaneous albinism (OCA) type 1A; OCA type 1B	Autosomal recessive
2	191740	UGT1A1		606785; 218800	Crigler-Najjar syndrome, type 1; Crigler-Najjar syndrome, type 2	Autosomal recessive
17	608897	UNC13D		608898	Hemophagocytic lymphohistiocytosis, familial, type 3	Autosomal recessive
11	605242	USH1C		276904; 602092	Usher syndrome, type 1C; Deafness, autosomal recessive, type 18A	Autosomal recessive
17	607696	USH1G		606943	Usher syndrome, type 1G	Autosomal recessive
1	608400	USH2A		276901	Usher syndrome, type 2A	Autosomal recessive

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9	605978	VPS13A		200150	Choreoacanthocytosis	Autosomal recessive
X	300392	WAS		301000; 313900	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked	X-linked
2	606268	WNT10A		257980	Odontoonychodermal dysplasia	Autosomal recessive
9	611153	XPA		278700	Xeroderma pigmentosum, group A	Autosomal recessive

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