CGT Panels

chrom	OMIM (gene)	gene	previous	OMIM (phen)	disease name (phenotype)	inheritance
12	605378	AAAS		231550	Triple-A syndrome (achalasia-addisonianism-alacrimia)	Autosomal recessive
2	607800	ABCA12		601277; 242500	Ichthyosis, congenital, autosomal recessive, type 4A; ICAR, type 4B (harlequin)	Autosomal recessive
1	601691	ABCA4		248200; 604116	Stargardt disease type 1; Cone-rod dystrophy type 3	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
14	603214	ABCD4		614857	Methylmalonic aciduria and homocystinuria, cblJ type	Autosomal recessive
11	604773	ACAD8		611283	Isobutyryl-CoA dehydrogenase deficiency	Autosomal recessive
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
10	600301	ACADSB		610006	Short/branched-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-methylacetoacetic aciduria (3-ketothiolase deficiency)	Autosomal recessive
17	609751	ACOX1		264470	Peroxisomal acyl-CoA oxidase deficiency	Autosomal recessive
16	614245	ACSF3		614265	Combined malonic and methylmalonic aciduria	Autosomal recessive
20	608958	ADA		102700	Severe combined immunodeficiency due to adenosine deaminase deficiency (ADA)	Autosomal recessive
5	604539	ADAMTS2		225410	Ehlers-Danlos syndrome, dermatosparaxis type	Autosomal recessive
16	604110	ADGRG1	GPR56	606854	Polymicrogyria, bilateral frontoparietal	Autosomal recessive
5	602851	ADGRV1	GPR98	605472	Usher syndrome, type 2C	Autosomal recessive, digenic inheritance (PDZD7 gene)
10	102750	ADK		614300	Hypermethioninemia due to adenosine kinase deficiency	Autosomal recessive
4	613228	AGA		208400	Aspartylglucosaminuria (glycosylasparaginase deficiency)	Autosomal recessive

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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
20	180960	AHCY		613752	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	Autosomal recessive
6	608894	AHI1		608629	Joubert syndrome, type 3	Autosomal recessive
17	604392	AIPL1		604393	Leber congenital amaurosis, type 4	Autosomal recessive
21	607358	AIRE		240300*	Autoimmune polyendocrinopathy syndrome, type 1	Autosomal recessive*
17	609523	ALDH3A2		270200	Sjogren-Larsson syndrome	Autosomal recessive
1	606811	ALDH4A1		239510	Hyperprolinemia, type 2	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	300629	AP1S2		304340	Mental retardation, X-linked, syndromic, type 5 (Pettigrew syndrome)	X-linked
12	107777	AQP2		125800*	Diabetes insipidus, nephrogenic, type 2	Autosomal recessive*
X	313700	AR		300068	Androgen insensitivity syndrome, complete	X-linked
6	608313	ARG1		207800	Argininemia (arginase deficiency)	Autosomal recessive
3	608922	ARL13B		612291	Joubert syndrome type 8	Autosomal recessive
22	607574	ARSA		250100	Metachromatic leukodystrophy	Autosomal recessive
5	611542	ARSB		253200	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)	Autosomal recessive
X	300180	ARSL	ARSE	302950	Chondrodysplasia punctata, brachytelephalangic	X-linked
X	300382	ARX		308350; 300215; 309510	Epileptic encephalopathy, early infantile, type 1; ARX-related developmental disorders	X-linked
7	608310	ASL		207900	Argininosuccinic aciduria	Autosomal recessive
7	108370	ASNS		615574	Asparagine synthetase deficiency	Autosomal recessive

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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
2	192132	ATP6V1B1		267300	Renal tubular acidosis with deafness	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
X	300504	ATRX		309580; 301040	Mental retardation-hypotonic facies syndrome, X-linked; Alpha-thalassemia/ mental retardation syndrome	X-linked
9	600529	AUH		250950	3-methylglutaconic aciduria, 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
4	610683	BBS12		615989	Bardet-Biedl syndrome, type 12	Autosomal recessive
16	606151	BBS2		615981	Bardet-Biedl syndrome, type 2	Autosomal recessive
3	177400	BCHE		617936	Butyrylcholinesterase deficiency	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
X	300553	BRWD3		300659	Mental retardation, X-linked, type 93	X-linked
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
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
1	114251	CASQ2		611938	Ventricular tachycardia, catecholaminergic polymorphic, type 2	Autosomal recessive

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21	613381	CBS		236200	Homocystinuria due to cystathionine beta-synthase	Autosomal recessive
6	603400	CCN6	WISP3	208230	Arthropathy, progressive pseudorheumatoid, of childhood	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
2	100730	CHRNG		265000; 253290	Multiple pterygium syndrome (MPS), Escobar type; MPS, lethal type	Autosomal recessive
16	605294	CHST6		217800	Macular corneal dystrophy	Autosomal recessive
16	600005	CIITA		209920	Bare lymphocyte syndrome, type 2, complementation group A	Autosomal recessive
7	118425	CLCN1		255700	Myotonia congenita, recessive	Autosomal recessive
16	607042	CLN3		204200	Ceroid lipofuscinosis, neuronal, type 3	Autosomal recessive
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
4	123825	CNGA1		613756	Retinitis pigmentosa type 49	Autosomal recessive
16	600724	CNGB1		613767	Retinitis pigmentosa type 45	Autosomal recessive
8	605080	CNGB3		262300	Achromatopsia, type 3	Autosomal recessive
9	608461	COL27A1		615155	Steel syndrome	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

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X	303630	COL4A5		301050	Alport syndrome, X-linked	X-linked
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
10	603646	COX15		615119; 256000	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 2; Leigh syndrome due to cytochrome c oxidase deficiency	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
3	605497	CRTAP		610682	Osteogenesis imperfecta, type 7	Autosomal recessive
1	607657	CTH		219500	Cystathioninuria	Autosomal recessive
17	606272	CTNS		219800	Nephropathic cystinosis	Autosomal recessive
20	613111	CTSA		256540	Galactosialidosis	Autosomal recessive
11	602365	CTSC		245010; 245000	Haim-Munk syndrome; Papillon-Lefevre syndrome	Autosomal recessive
11	116840	CTSD		610127	Ceroid lipofuscinosis, neuronal, type 10	Autosomal recessive
1	601105	CTSK		265800	Pycnodysostosis	Autosomal recessive
X	300304	CUL4B		300354	Mental retardation, X-linked, syndromic, type 15 (Cabezas type)	X-linked
16	608508	CYBA		233690	Chronic granulomatous disease, type 4	Autosomal recessive
X	300481	CYBB		306400	Chronic granulomatous disease, X-linked	X-linked
8	610613	CYP11B1		202010	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	Autosomal recessive
8	124080	CYP11B2		203400	Hypoaldosteronism, congenital, due to CMO I deficiency	Autosomal recessive
10	609300	CYP17A1		202110	17 alpha(α)-hydroxylase/17,20-lyase deficiency	Autosomal recessive
15	107910	CYP19A1		613546	Aromatase deficiency	Autosomal recessive
2	601771	CYP1B1		231300	Glaucoma, primary congenital, type 3A	Autosomal recessive
6	613815	CYP21A2		201910	Congenital adrenal hyperplasia due to	Autosomal

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					21-hydroxylase deficiency	recessive
2	606530	CYP27A1		213700	Cerebrotendinous xanthomatosis	Autosomal recessive
12	609506	CYP27B1		264700	Vitamin D-dependent rickets, type 1	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
X	300121	DCX		300067	Lissencephaly, X-linked, type 1	X-linked
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
X	300126	DKC1		305000	Dyskeratosis congenita, X-linked	X-linked
7	238331	DLD		246900	Dihydrolipoamide dehydrogenase deficiency	Autosomal recessive
X	300189	DLG3		300850	Mental retardation, X-linked, type 90	X-linked
X	300377	DMD		310200; 300376	Duchenne/Becker muscular dystrophy	X-linked
5	603335	DNAH5		608644	Ciliary dyskinesia, primary, type 3, with or without situs inversus	Autosomal recessive
9	604366	DNAI1		244400	Ciliary dyskinesia, primary, type 1, with or without situs inversus	Autosomal recessive
17	605483	DNAI2		612444	Ciliary dyskinesia, primary, type 9, with or without situs inversus	Autosomal recessive
10	606060	DNAJC12		617384	Hyperphenylalaninemia, mild, non-BH4-deficient	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
20	603503	DPM1		608799	Congenital disorder of glycosylation, type 1E	Autosomal recessive
1	612779	DPYD		274270	Dihydropyrimidine dehydrogenase deficiency	Autosomal recessive
15	606759	DUOX2		607200	Thyroid dyshormonogenesis, type 6	Autosomal recessive
15	612772	DUOXA2		274900	Thyroid dyshormonogenesis, type 5	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
2	604095	EDAR		224900	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type	Autosomal recessive

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2	604032	EIF2AK3		226980	Wolcott-Rallison syndrome	Autosomal recessive
3	603945	EIF2B5		603896	Leukoencephalopathy with vanishing white matter (VWM)	Autosomal recessive
9	603722	ELP1	IKBKAP	223900	Familial dysautonomia	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
10	609413	ERCC6		133540; 214150	Cockayne syndrome, type B; Cerebrooculofacioskeletal syndrome, type 1	Autosomal recessive
5	609412	ERCC8		216400	Cockayne syndrome, type A	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
4	604831	EVC		225500	Ellis-van Creveld syndrome	Autosomal recessive
4	607261	EVC2		225500	Ellis-van Creveld syndrome	Autosomal recessive
9	606489	EXOSC3		614678	Pontocerebellar hypoplasia, type 1B	Autosomal recessive
6	612424	EYS		602772	Retinitis pigmentosa, type 25	Autosomal recessive
4	264900	F11		612416*	Factor XI deficiency	Autosomal recessive*
11	176930	F2		613679	Prothrombin deficiency	Autosomal recessive
1	612309	F5		227400	Factor V deficiency	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
2	613596	FAM161A		606068	Retinitis pigmentosa, type 28	Autosomal recessive
7	611061	FAM20C		259775	Raine syndrome	Autosomal recessive
16	607139	FANCA		227650	Fanconi anemia, complementation group A	Autosomal recessive

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9	613899	FANCC		227645	Fanconi anemia, complementation group C	Autosomal recessive
9	602956	FANCG		614082	Fanconi anemia, complementation group G	Autosomal recessive
X	300546	FGD1		305400	Aarskog-Scott syndrome; Mental retardation, X-linked syndromic, type 16	X-linked
1	136850	FH		606812	Fumarase deficiency	Autosomal recessive
19	606596	FKRP		613153; 606612; 607155	Muscular dystrophy-dystroglycanopathy, 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-dystroglycanopathy, 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
11	613622	FOXRED1		618241	Mitochondrial complex I deficiency, nuclear type 19	Autosomal recessive
4	607830	FRAS1		219000	Fraser syndrome, type 1	Autosomal recessive
21	606806	FTCD		229100	Glutamate formiminotransferase deficiency	Autosomal recessive
X	300499	FTSJ1		309549	Mental retardation, X-linked 44	X-linked
1	612280	FUCA1		230000	Fucosidosis	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
1	606953	GALE		230350	Galactose epimerase deficiency	Autosomal recessive
17	604313	GALK1		230200	Galactokinase deficiency with cataracts	Autosomal recessive
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

chrom	OMIM (gene)	gene	previous	OMIM (phen)	disease name (phenotype)	inheritance
						recessive
14	600225	GCH1		233910	Hyperphenylalaninemia, BH4-deficient, type B	Autosomal recessive
16	238330	GCSH		605899	?Glycine encephalopathy	Autosomal recessive
8	606598	GDAP1		608340	Charcot-Marie-Tooth disease, recessive intermediate, type A	Autosomal recessive
20	601146	GDF5		200700	Chondrodysplasia, Grebe type	Autosomal recessive
3	606639	GFM1		609060	Combined oxidative phosphorylation deficiency, type 1	Autosomal recessive
7	139191	GHRHR		612781	Growth hormone deficiency, isolated, type 1B	Autosomal recessive
X	304040	GJB1		302800	Charcot-Marie-Tooth neuropathy, X-linked dominant, type 1	X-linked
13	121011	GJB2		220290	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6	Autosomal recessive; Digenic inheritance (GJB6 gene)
13	604418	GJB6		612645; 220290	Deafness, autosomal recessive, type 1B; Deafness, digenic GJB2/GJB6	Autosomal recessive; Digenic inheritance (GJB2 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
6	606628	GNMT		606664	Glycine N-methyltransferase deficiency	Autosomal recessive
12	607840	GNPTAB		252500; 252600	Mucolipidosis 2 alpha/beta; Mucolipidosis 3 alpha/beta	Autosomal recessive
16	607838	GNPTG		252605	Mucolipidosis III gamma	Autosomal recessive
4	138850	GNRHR		146110	Hypogonadotropic hypogonadism, type 7, without anosmia	Autosomal recessive
12	607664	GNS		252940	Mucopolysaccharidosis, type 3D (Sanfilippo syndrome D)	Autosomal recessive
17	606672	GP1BA		231200	Bernard-Soulier syndrome, type A1	Autosomal recessive
22	138720	GP1BB		231200	Bernard-Soulier syndrome, type B	Autosomal recessive
3	173515	GP9		231200	Bernard-Soulier syndrome, type C	Autosomal recessive

chrom	OMIM (gene)	gene	previous	OMIM (phen)	disease name (phenotype)	inheritance
X	300808	GPR143		300500	Ocular albinism, type 1 (Nettleship-Falls type)	X-linked
9	604296	GRHPR		260000	Hyperoxaluria, primary, type 2	Autosomal recessive
20	601002	GSS		266130	Glutathione synthetase deficiency	Autosomal recessive
17	600179	GUCY2D		204000	Leber congenital amaurosis, type 1	Autosomal recessive
7	611499	GUSB		253220	Mucopolysaccharidosis, type 7	Autosomal recessive
4	601609	HADH		231530	3-hydroxyacyl-CoA dehydrogenase deficiency	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
X	300019	HCFC1		309541	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type)	X-linked
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
1	608374	HJV	HFE2	602390	Hemochromatosis, type 2A	Autosomal recessive
21	609018	HLCS		253270	Holocarboxylase synthetase deficiency	Autosomal recessive
1	613898	HMGCL		246450	HMG-CoA lyase deficiency	Autosomal recessive
22	141250	HMOX1		614034	Heme oxygenase-1 deficiency	Autosomal recessive
10	613597	HOGA1		613616	Hyperoxaluria, primary, type 3	Autosomal recessive
12	609695	HPD		276710	Tyrosinemia, type 3	Autosomal recessive
X	308000	HPRT1		300322	Lesch-Nyhan syndrome	X-linked
10	604982	HPS1		203300	Hermansky-Pudlak syndrome, type 1	Autosomal recessive
3	606118	HPS3		614072	Hermansky-Pudlak syndrome, type 3	Autosomal

chrom	OMIM (gene)	gene	previous	OMIM (phen)	disease name (phenotype)	inheritance
						recessive
X	300256	HSD17B10		300438	HSD10 mitochondrial disease	X-linked
9	605573	HSD17B3		264300	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	Autosomal recessive
5	601860	HSD17B4		261515	D-bifunctional protein deficiency	Autosomal recessive
1	613890	HSD3B2		201810	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	Autosomal recessive
1	142461	HSPG2		224410	Dyssegmental dysplasia, Silverman-Handmaker type	Autosomal recessive
3	607071	HYAL1		601492	?Mucopolysaccharidosis, type 9	Autosomal recessive
11	610693	HYLS1		236680	Hydrolethalus syndrome	Autosomal recessive
20	604526	IDH3B		612572	Retinitis pigmentosa, type 46	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	300206	IL1RAPL1		300143	Mental retardation, X-linked, type 21/34	X-linked
X	308380	IL2RG		300400	Severe combined immunodeficiency, X-linked	X-linked
15	607036	IVD		243500	Isovaleric acidemia	Autosomal recessive
6	612025	IYD		274800	Thyroid dyshormonogenesis, type 4	Autosomal recessive
19	600173	JAK3		600802	Severe Combined Immunodeficiency, autosomal recessive, T-negative/B-positive type	Autosomal recessive
11	600937	KCNJ11		601820; 606176*	Hyperinsulinemic hypoglycemia, type 2 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive; Autosomal recessive*
X	314690	KDM5C		300534	Mental retardation, X-linked, syndromic, Claes-Jensen type	X-linked
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
22	603590	LARGE1	LARGE	613154; 608840	Muscular dystrophy-dystroglycanopathy, type 6A and 6B	Autosomal recessive

chrom	OMIM (gene)	gene	previous	OMIM (phen)	disease name (phenotype)	inheritance
6	611408	LCA5		604537	Leber congenital amaurosis, type 5	Autosomal recessive
2	152790	LHCGR		238320	Leydig cell hypoplasia	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
3	607365	LIPH		604379	Hypotrichosis, type 7 or woolly hair, autosomal recessive, type 2, with or without hypotrichosis	Autosomal recessive
6	612625	LMBRD1		277380	Methylmalonic aciduria and homocystinuria, cblF type	Autosomal recessive
18	613072	LOXHD1		613079	Deafness, autosomal recessive, type 77	Autosomal recessive
8	609708	LPL		238600	Lipoprotein lipase deficiency	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
4	609489	MANBA		248510	Mannosidosis, beta	Autosomal recessive
10	610550	MAT1A		250850	Methionine adenosyltransferase deficiency, autosomal recessive	Autosomal recessive
3	609010	MCCC1		210200	3-Methylcrotonyl-CoA carboxylase deficiency, type 1	Autosomal recessive
5	609014	MCCC2		210210	3-Methylcrotonyl-CoA carboxylase deficiency, type 2	Autosomal recessive
2	608419	MCEE		251120	Methylmalonyl-CoA epimerase deficiency	Autosomal recessive
19	605248	MCOLN1		252650	Mucolipidosis type 4	Autosomal recessive
X	300005	MECP2		300673; 312750	Encephalopathy, neonatal severe; Rett syndrome	X-linked
11	603810	MED17		613668	Microcephaly, postnatal progressive, with seizures and brain atrophy	Autosomal recessive
16	608107	MEFV		249100	Familial Mediterranean fever	Autosomal recessive
15	605195	MESP2		608681	Spondylocostal dysostosis, type 2, autosomal recessive	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
16	606761	MLYCD		248360	Malonyl-CoA decarboxylase deficiency	Autosomal

chrom	OMIM (gene)	gene	previous	OMIM (phen)	disease name (phenotype)	inheritance
						recessive
4	607481	MMAA		251100	Methylmalonic aciduria, vitamin B12-responsive	Autosomal recessive
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
1	607093	MTHFR		236250	Homocystinuria due to MTHFR deficiency	Autosomal recessive
X	300415	MTM1		310400	Myotubular myopathy, X-linked	X-linked
11	603557	MTMR2		601382	Charcot-Marie-Tooth disease, type 4B1	Autosomal recessive
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
12	251170	MVK		610377	Mevalonic aciduria	Autosomal recessive
17	602666	MYO15A		600316	Deafness, autosomal recessive, type 3	Autosomal recessive
11	276903	MYO7A		276900; 600060	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2	Autosomal recessive
2	615787	NADK2		616034	2,4-dienoyl-CoA reductase deficiency	Autosomal recessive
17	609701	NAGLU		252920	Mucopolysaccharidosis, type 3B (Sanfilippo B)	Autosomal recessive
17	608300	NAGS		237310	N-acetylglutamate synthase deficiency	Autosomal recessive
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
X	300658	NDP		310600	Norrie disease	X-linked

chrom	OMIM (gene)	gene	previous	OMIM (phen)	disease name (phenotype)	inheritance
8	605262	NDRG1		601455	Charcot-Marie-Tooth disease, type 4D	Autosomal recessive
5	609653	NDUFAF2		618233	Mitochondrial complex I deficiency, nuclear type 10	Autosomal recessive
20	612360	NDUFAF5		618238	Mitochondrial complex I deficiency, nuclear type 16	Autosomal recessive
5	602694	NDUFS4		252010	Mitochondrial complex I deficiency, nuclear type 1	Autosomal recessive
5	603848	NDUFS6		618232	Mitochondrial complex I deficiency, nuclear type 9	Autosomal recessive
19	601825	NDUFS7		618224	Mitochondrial complex I deficiency, nuclear type 3	Autosomal recessive
11	161015	NDUFV1		618225	Mitochondrial complex I deficiency, nuclear type 4	Autosomal recessive
2	161650	NEB		256030	Nemaline myopathy type 2	Autosomal recessive
6	608272	NEU1		256550	Sialidosis, type 1 and type 2	Autosomal recessive
5	606470	NHP2		613987	Dyskeratosis congenita, autosomal recessive type 2	Autosomal recessive
19	609661	NLRP7		231090	Hydatidiform mole, recurrent, type 1	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
2	607100	NPHP1		609583	Joubert syndrome type 4	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
19	606580	OPA3		258501	3-methylglutaconic aciduria, type 3	Autosomal recessive
X	300127	OPHN1		300486	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	X-linked
6	607649	OSTM1		259720	Osteopetrosis, autosomal recessive	Autosomal

chrom	OMIM (gene)	gene	previous	OMIM (phen)	disease name (phenotype)	inheritance
					type 5	recessive
X	300461	OTC		311250	Ornithine transcarbamylase deficiency	X-linked
2	603681	OTOF		601071	Deafness, autosomal recessive, type 9	Autosomal recessive
1	610339	P3H1	LEPRE1	610915	Osteogenesis imperfecta, type 8	Autosomal recessive
12	612349	PAH		261600	Phenylketonuria	Autosomal recessive
X	300142	PAK3		300558	Mental retardation, X-linked, type 30	X-linked
20	606157	PANK2		234200	Neurodegeneration with brain iron accumulation type 1	Autosomal recessive
11	608786	PC		266150	Pyruvate carboxylase deficiency	Autosomal recessive
10	126090	PCBD1		264070	Hyperphenylalaninemia, BH4-deficient, type D	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
5	180071	PDE6A		613810	Retinitis pigmentosa type 43	Autosomal recessive
X	300502	PDHA1		312170	Pyruvate dehydrogenase E1-alpha deficiency	X-linked
3	179060	PDHB		614111	Pyruvate dehydrogenase E1-beta deficiency	Autosomal recessive
7	602136	PEX1		234580	Heimler syndrome type 1	Autosomal recessive
1	602859	PEX10		614870; 614871	Peroxisome biogenesis disorder, type 6A (Zellweger syndrome); Peroxisome biogenesis disorder, type 6B	Autosomal recessive
17	601758	PEX12		614859	Peroxisome biogenesis disorder type 3A (Zellweger)	Autosomal recessive
8	170993	PEX2		614866	Peroxisome biogenesis disorder type 5A (Zellweger)	Autosomal recessive
22	608666	PEX26		614872	Peroxisome biogenesis disorder type 7A (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
12	610681	PFKM		232800	Glycogen storage disease, type 7	Autosomal recessive
X	311800	PGK1		300653	Phosphoglycerate kinase 1 deficiency	X-linked
X	300560	PHF8		300263	Mental retardation syndrome, X-linked, Siderius type	X-linked

chrom	OMIM (gene)	gene	previous	OMIM (phen)	disease name (phenotype)	inheritance
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
X	300401	PLP1		312080	Pelizaeus-Merzbacher disease	X-linked
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
6	610060	POLR1C		616494; 248390	Leukodystrophy, hypomyelinating, type 11; Treacher Collins syndrome 3	Autosomal recessive
1	606822	POMGNT1		253280; 613151; 613157	Muscular dystrophy-dystroglycanopathy, 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-dystroglycanopathy, 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-dystroglycanopathy, type 2A (Walker-Warburg syndrome); Type 2B; Type 2C (limb-girdle muscular dystrophy, type 14 [LGMD R14])	Autosomal recessive
7	124015	POR		201750	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	Autosomal recessive
3	173110	POU1F1		613038*	Pituitary hormone deficiency, combined, type 1	Autosomal recessive*
X	300039	POU3F4		304400	Deafness, X-linked, type 2	X-linked
4	611065	PPM1K		615135	?Maple syrup urine disease, mild variant	Autosomal recessive
1	600722	PPT1		256730	Ceroid lipofuscinosis, neuronal, type 1	Autosomal recessive
X	300463	PQBP1		309500	Renpenning syndrome	X-linked
1	176763	PRDX1		277400	Methylmalonic aciduria and homocystinuria, cblC type, digenic	Autosomal recessive, digenic inheritance (MMACHC gene)
10	170280	PRF1		603553	Hemophagocytic lymphohistiocytosis, familial, type 2	Autosomal recessive
22	606810	PRODH		239500	Hyperprolinemia, type 1	Autosomal recessive

chrom	OMIM (gene)	gene	previous	OMIM (phen)	disease name (phenotype)	inheritance
5	601538	PROP1		262600	Pituitary hormone deficiency, combined, type 2	Autosomal recessive
X	311850	PRPS1		300661; 304500; 311070; 301835	PRPS1-related disoders	X-linked
10	176801	PSAP		611721	Combined SAP deficiency	Autosomal recessive
11	612719	PTS		261640	Hyperphenylalaninemia, BH4-deficient, type A	Autosomal recessive
12	608109	PUS1		600462	Myopathy, lactic acidosis, and sideroblastic anemia, type 1	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
6	611524	RARS2		611523	Pontocerebellar hypoplasia, type 6	Autosomal recessive
18	601881	RAX		611038	Isolated microphthalmia, type 3	Autosomal recessive
14	608830	RDH12		612712	Leber congenital amaurosis, type 13	Autosomal recessive
11	610330	RNASEH2C		610329	Aicardi-Goutieres syndrome, type 3	Autosomal recessive
X	300757	RP2		312600	Retinitis pigmentosa, type 2, X-linked	X-linked
1	180069	RPE65		204100; 613794	RPE65-related Leber congenital amaurosis/early-onset severe retinal dystrophy	Autosomal recessive
X	312610	RPGR		300029; 304020	Retinitis pigmentosa, type 3, X-linked; Cone-rod dystrophy, X-linked, 1	X-linked
16	610937	RPGRIP1L		611560; 611561; 619113	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome	Autosomal recessive
X	300839	RS1		312700	Retinoschisis	X-linked
20	608833	RTEL1		615190*	Dyskeratosis congenita, autosomal recessive type 5	Autosomal recessive*
13	604490	SACS		270550	Spastic ataxia, Charlevoix-Saguenay, type	Autosomal recessive
2	181031	SAG		258100	Oguchi disease, type 1	Autosomal recessive
20	606754	SAMHD1		612952	Aicardi-Goutieres syndrome, type 5	Autosomal recessive
7	607444	SBDS		260400	Shwachman-Diamond syndrome	Autosomal

chrom	OMIM (gene)	gene	previous	OMIM (phen)	disease name (phenotype)	inheritance
						recessive
4	613009	SEPSECS		613811	Pontocerebellar hypoplasia, type 2D	Autosomal recessive
14	107400	SERPINA1		613490	Alpha-1 antitrypsin deficiency	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
5	601411	SGCD		601287	Limb-girdle muscular dystrophy, type 6 (LGMD R6)	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
5	608206	SH3TC2		601596	Charcot-Marie-Tooth disease, type 4C	Autosomal recessive
16	600968	SLC12A3		263800	Gitelman syndrome	Autosomal recessive
15	604878	SLC12A6		218000	Agenesis of the corpus callosum with peripheral neuropathy	Autosomal recessive
X	300095	SLC16A2		300523	Allan-Herndon-Dudley syndrome	X-linked
6	604322	SLC17A5		604369	Salla disease	Autosomal recessive
1	603941	SLC19A2		249270	Thiamine-responsive megaloblastic anemia syndrome	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
3	613698	SLC25A20		212138	Carnitine-acylcarnitine translocase deficiency	Autosomal recessive
5	606718	SLC26A2		600972	Achondrogenesis, type 1B (diastrophic dysplasia)	Autosomal recessive
7	126650	SLC26A3		214700	Diarrhea 1, secretory chloride, congenital	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

chrom	OMIM (gene)	gene	previous	OMIM (phen)	disease name (phenotype)	inheritance
8	607059	SLC39A4		201100	Acrodermatitis enteropathica	Autosomal recessive
2	104614	SLC3A1		220100*	Cystinuria	Autosomal recessive*
5	606202	SLC45A2		606574	Albinism, oculocutaneous, type 4	Autosomal recessive
17	611672	SLC46A1		229050	Folate malabsorption, hereditary	Autosomal recessive
20	610206	SLC4A11		217700	Corneal endothelial dystrophy, autosomal recessive	Autosomal recessive
19	601843	SLC5A5		274400	Thyroid dyshormonogenesis, type 1	Autosomal recessive
5	608893	SLC6A19		234500	Hartnup disorder	Autosomal recessive
X	300036	SLC6A8		300352	Cerebral creatine deficiency syndrome, type 1	X-linked
14	603593	SLC7A7		222700	Lysinuric protein intolerance	Autosomal recessive
19	604144	SLC7A9		220100*	Cystinuria	Autosomal recessive*
2	606622	SMARCAL1		242900	Schimke immunoosseous dysplasia	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
15	610844	SPG11		602099	Amyotrophic lateral sclerosis, type 5, juvenile	Autosomal recessive
16	602783	SPG7		607259	Spastic paraplegia, type 7, autosomal recessive	Autosomal recessive
2	607306	SRD5A2		264600	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency (pseudovaginal perineoscrotal hypospadias)	Autosomal recessive
2	604402	ST3GAL5		609056	Salt and pepper developmental regression syndrome	Autosomal recessive
8	600617	STAR		201710	Lipoid adrenal hyperplasia	Autosomal recessive
3	607939	SUMF1		272200	Multiple sulfatase deficiency	Autosomal recessive
9	185620	SURF1		616684; 256000	Charcot-Marie-Tooth disease, type 4K; Leigh syndrome, due to COX IV deficiency	Autosomal recessive
X	313440	SYN1		300491	Epilepsy, X-linked, with variable learning disabilities and behavior disorders	X-linked
16	613018	TAT		276600	Tyrosinemia, type 2	Autosomal recessive
11	604592	TCIRG1		259700	Osteopetrosis, autosomal recessive, type 1	Autosomal recessive
14	615000	TECPR2		615031	Spastic paraplegia, type 49, autosomal recessive	Autosomal recessive
7	604720	TFR2		604250	Hemochromatosis, type 3	Autosomal recessive

chrom	OMIM (gene)	gene	previous	OMIM (phen)	disease name (phenotype)	inheritance
8	188450	TG		274700	Thyroid dyshormonogenesis, type 3	Autosomal recessive
14	190195	TGM1		242300	Ichthyosis, congenital, autosomal recessive, type 1	Autosomal recessive
11	191290	TH		605407	Segawa syndrome, recessive	Autosomal recessive
X	300395	THOC2		300957	Mental retardation, X-linked 12	X-linked
11	613277	TMEM216		608091; 603194	Joubert syndrome, type 2; Meckel syndrome, type 2	Autosomal recessive
8	609884	TMEM67		610688; 607361; 216360	Joubert syndrome, type 6; Meckel syndrome, type 3; COACH syndrome	Autosomal recessive
21	605511	TMPRSS3		601072	Deafness, autosomal recessive, type 8/10	Autosomal recessive
2	606765	TPO		274500	Thyroid dyshormonogenesis, type 2A	Autosomal recessive
11	607998	TPP1		204500; 609270	Ceroid lipofuscinosis, neuronal, type 2; Spinocerebellar ataxia, autosomal recessive, type 7	Autosomal recessive
6	603283	TRDN		615441	Ventricular tachycardia, catecholaminergic polymorphic, type 5, with or without muscle weakness	Autosomal recessive
3	606609	TREX1		225750	Aicardi-Goutieres syndrome, type 1	Autosomal recessive
9	602290	TRIM32		254110	Limb-girdle muscular dystrophy, type 8 (LGMD R8)	Autosomal recessive
17	605073	TRIM37		253250	Mulibrey nanism	Autosomal recessive
22	610230	TRMU		613070	Liver failure, transient infantile	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
1	188540	TSHB		275100	Hypothyroidism, congenital, nongoitrous, type 4	Autosomal recessive
14	603372	TSHR		275200	Hypothyroidism, congenital, nongoitrous, type 1	Autosomal recessive
5	614589	TTC37		222470	Trichohepatoenteric syndrome, type 1 (diarrhea, syndromic)	Autosomal recessive
8	600415	TTPA		277460	Ataxia with isolated vitamin E deficiency	Autosomal recessive
22	131222	TYMP		603041	Mitochondrial DNA depletion syndrome, type 1 (MNGIE type)	Autosomal recessive
11	606933	TYR		203100; 606952	Oculocutaneous albinism (OCA) type 1A; OCA type 1B	Autosomal recessive
9	115501	TYRP1		203290	Albinism, oculocutaneous, type 3	Autosomal recessive
15	601623	UBE3A		105830	Angelman syndrome	Autosomal dominant
2	191740	UGT1A1		606785; 218800	Crigler-Najjar syndrome, type 1; Crigler-Najjar syndrome, type 2	Autosomal recessive
17	608897	UNC13D		608898	Hemophagocytic lymphohistiocytosis,	Autosomal

chrom	OMIM (gene)	gene	previous	OMIM (phen)	disease name (phenotype)	inheritance
					familial, type 3	recessive
X	300298	UPF3B		300676	Mental retardation, X-linked, syndromic, type 14	X-linked
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
9	605978	VPS13A		200150	Choreoacanthocytosis	Autosomal recessive
8	607817	VPS13B		216550	Cohen syndrome	Autosomal recessive
1	610035	VPS45		615285	Neutropenia, severe congenital, type 5	Autosomal recessive
17	615850	VPS53		615851	Pontocerebellar hypoplasia, type 2E	Autosomal recessive
14	602168	VRK1		607596	Pontocerebellar hypoplasia, type 1A	Autosomal recessive
14	142993	VSX2		610092; 610093	Microphthalmia with coloboma 3; Isolated microphthalmia 2	Autosomal recessive
X	300392	WAS		301000; 313900	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked	X-linked
9	607928	WHRN	DFNB31	611383; 607084	Usher syndrome, type 2D; Deafness, autosomal recessive, type 31	Autosomal recessive
2	606268	WNT10A		257980	Odontoonychodermal dysplasia	Autosomal recessive
8	604611	WRN		277700	Werner syndrome	Autosomal recessive
9	611153	XPA		278700	Xeroderma pigmentosum, group A	Autosomal recessive
3	613208	XPC		278720	Xeroderma pigmentosum, group C	Autosomal recessive
X	300646	ZDHHC9		300799	Mental retardation, X-linked syndromic, Raymond type	X-linked
14	612012	ZFYVE26		270700	Spastic paraplegia, type 15, autosomal recessive	Autosomal recessive
X	314990	ZNF711		300803	Mental retardation, X-linked, type 97	X-linked

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