MULTIPLE CONGENITAL ANOMALIES GENE PANEL

Gene symbol	Depth (reads)	Coverage (avg %)	OMIM disease	Description
A4GALT	105	100	111400	NOR polyagglutination syndrome
AAAS	104	89	231550	Achalasia-addisonianism-alacrimia syndrome
AAGAB	121	95	148600	Keratoderma palmoplantar punctate type IA
AARS	101	84	613287	Charcot-Marie-Tooth disease axonal type 2N
AARS2	101	87	614096	Combined oxidative phosphorylation deficiency 8
AASS	104	95	238700	Hyperlysinemia
ABAT	82	86	613163	GABA-transaminase deficiency
ABCA1	105	84	604091	HDL deficiency type 2
ABCA12	112	94	242500	Ichthyosis autosomal recessive 4B (harlequin)
ABCA3	91	85	610921	Surfactant metabolism dysfunction pulmonary 3
ABCA4	100	85	604116	Cone-rod dystrophy 3
ABCB11	104	90	605479	Cholestasis benign recurrent intrahepatic 2
ABCB4	108	83	614972	Cholestasis intrahepatic of pregnancy 3
ABCB6	130	92	614497	Microphthalmia isolated with coloboma 7
ABCB7	99	90	301310	Anemia sideroblastic with ataxia
ABCC2	118	88	237500	Dubin-Johnson syndrome
ABCC6	79	82	614473	Arterial calcification generalized of infancy 2
ABCC8	92	86	125853	Diabetes mellitus noninsulin-dependent
ABCC9	115	96	614050	Atrial fibrillation familial 12
ABCD1	59	84	300100	Adrenoleukodystrophy
ABCD4	121	88	614857	Methylmalonic aciduria and homocystinuria cblJ type
ABCG5	132	86	210250	Sitosterolemia
ABCG8	99	85	611465	Gallbladder disease 4
ABHD12	72	98	612674	Polyneuropathy hearing loss ataxia retinitis pigmentosa and cataract
ABHD5	125	91	275630	Chanarin-Dorfman syndrome
ABL1	97	89	200	Leukemia Philadelphia chromosome-positive
ACAD8	101	80	611283	Isobutyryl-CoA dehydrogenase deficiency
ACAD9	105	89	611126	ACAD9 deficiency

ACADM	146	99	201450	Acyl-CoA dehydrogenase medium chain deficiency of
ACADS	106	95	201470	Acyl-CoA dehydrogenase short-chain deficiency of
ACADSB	89	95	610006	2-methylbutyrylglycinuria
ACADVL	100	89	201475	VLCAD deficiency
ACAN	134	46	165800	Osteochondritis dissecans short stature and early-onset osteoarthritis
ACAT1	101	96	203750	Alpha-methylacetoacetic aciduria
ACE	92	87	267430	Renal tubular dysgenesis
ACO2	76	67	614559	Infantile cerebellar-retinal degeneration
ACOX1	90	73	264470	Peroxisomal acyl-CoA oxidase deficiency
ACP5	96	95	200	Spondyloenchondrodysplasia with immune dysregulation
ACSF3	79	89	614265	Combined malonic and methylmalonic aciduria
ACSL4	98	95	300387	Mental retardation X-linked 63
ACSL6	95	92	200	Myelodysplastic syndrome
ACTA1	70	55	161800	Myopathy actin congenital with cores
ACTA2	107	70	611788	Aortic aneurysm familial thoracic 6
ACTB	101	17	243310	Baraitser-Winter syndrome 1
ACTC1	114	61	612794	Atrial septal defect 5
ACTG1	113	16	614583	Baraitser-Winter syndrome 2
ACTN1	99	79	615193	Bleeding disorder platelet-type 15
ACTN2	91	81	612158	Cardiomyopathy dilated 1AA
ACTN4	94	79	603278	Glomerulosclerosis focal segmental 1
ACVR1	115	84	135100	Fibrodysplasia ossificans progressiva
ACVR1B	112	87	200	Pancreatic cancer
ACVR2B	88	89	613751	Heterotaxy visceral 4 autosomal
ACVRL1	57	84	600376	Telangiectasia hereditary hemorrhagic type 2
ACY1	85	88	609924	Aminoacylase 1 deficiency
ADA	82	90	102700	Adenosine deaminase deficiency partial
ADAM17	125	89	614328	Inflammatory skin and bowel disease neonatal
ADAM9	114	95	612775	Cone-rod dystrophy 9
ADAMTS10	68	90	277600	Weill-Marchesani syndrome 1 recessive
ADAMTS13	67	72	274150	Thrombotic thrombocytopenic purpura familial
ADAMTS17	80	78	613195	Weill-Marchesani-like syndrome
ADAMTS18	108	85	608454	Knobloch syndrome 2

ADAMTS2	102	87	225410	Ehlers-Danlos syndrome type VIIC
ADAMTSL2	63	86	231050	Geleophysic dysplasia 1
ADAMTSL4	97	85	225100	Ectopia lentis isolated autosomal recessive
ADAR	136	91	615010	Aicardi-Goutieres syndrome 6
ADAT3	37	98	615286	Mental retardation autosomal recessive 36
ADCK3	97	87	200	-
ADCY5	80	89	606703	Dyskinesia familial with facial myokymia
ADIPOQ	163	93	612556	Adiponectin deficiency
ADK	98	94	614300	Hypermethioninemia due to adenosine kinase deficiency
ADRB2	138	100	200	Beta-2-adrenoreceptor agonist
ADSL	124	83	103050	Adenylosuccinase deficiency
AFF2	121	92	309548	Mental retardation X-linked FRAXE type
AFG3L2	93	83	614487	Ataxia spastic 5 autosomal recessive
AGA	121	94	208400	Aspartylglucosaminuria
AGK	121	84	614691	Cataract 38 autosomal recessive
AGL	134	97	232400	Glycogen storage disease IIIa
AGPAT2	53	98	608594	Lipodystrophy congenital generalized type 1
AGPS	101	98	600121	Rhizomelic chondrodysplasia punctata type 3
AGRN	76	91	254300	Myasthenia limb-girdle familial
AGT	125	91	267430	Renal tubular dysgenesis
AGTR1	147	100	145500	Hypertension essential
AGTR2	142	100	300852	Mental retardation X-linked 88
AGXT	88	95	259900	Hyperoxaluria primary type 1
AHCY	93	77	613752	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase
AHI1	117	97	608629	Joubert syndrome-3
AICDA	87	86	605258	Immunodeficiency with hyper-IgM type 2
AIFM1	106	80	300816	Combined oxidative phosphorylation deficiency 6
AIMP1	126	88	260600	Leukodystrophy hypomyelinating 3
AIP	92	88	219090	Pituitary adenoma ACTH-secreting
AIPL1	74	90	604393	Cone-rod dystrophy
AIRE	70	89	240300	Autoimmune polyendocrinopathy syndrome type I with or without reversible metaphyseal dysplasia
AK1	84	91	612631	Hemolytic anemia due to adenylate kinase deficiency
AK2	107	44	267500	Reticular dysgenesis

AKAP9	125	95	611820	Long QT syndrome-11
AKR1C2	167	59	614279	46XY sex reversal 8
AKR1D1	116	79	235555	Bile acid synthesis defect congenital 2
AKT1	104	91	114480	Breast cancer somatic
AKT2	104	85	125853	Diabetes mellitus type II
AKT3	99	95	603387	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome
ALAD	87	94	612740	Porphyria acute hepatic
ALAS2	84	77	300751	Anemia sideroblastic X-linked
ALB	101	90	200	-
ALDH18A1	103	82	219150	Cutis laxa autosomal recessive type IIIA
ALDH1A3	109	84	615113	Microphthalmia isolated 8
ALDH2	94	86	610251	Alcohol sensitivity acute
ALDH3A2	103	85	270200	Sjogren-Larsson syndrome
ALDH4A1	65	90	239510	Hyperprolinemia type II
ALDH5A1	71	88	271980	Succinic semialdehyde dehydrogenase deficiency
ALDH6A1	117	88	614105	Methylmalonate semialdehyde dehydrogenase deficiency
ALDH7A1	92	79	266100	Epilepsy pyridoxine-dependent
ALDOA	110	75	611881	Glycogen storage disease XII
ALDOB	115	79	229600	Fructose intolerance
ALG1	104	87	608540	Congenital disorder of glycosylation type Ik
ALG11	157	95	613661	Congenital disorder of glycosylation type Ip
ALG12	105	88	607143	Congenital disorder of glycosylation type Ig
ALG13	101	85	300884	Congenital disorder of glycosylation type Is
ALG2	109	97	607906	Congenital disorder of glycosylation type Ii
ALG3	117	86	601110	Congenital disorder of glycosylation type Id
ALG6	87	100	603147	Congenital disorder of glycosylation type Ic
ALG8	91	96	608104	Congenital disorder of glycosylation type Ih
ALG9	107	87	608776	Congenital disorder of glycosylation type Il
ALMS1	217	94	203800	Alstrom syndrome
ALOX12B	94	91	242100	Ichthyosis congenital autosomal recessive 2
ALOXE3	90	90	606545	Ichthyosis congenital autosomal recessive 3
ALPL	88	81	146300	Hypophosphatasia adult
ALS2	138	90	205100	Amyotrophic lateral sclerosis 2 juvenile

ALX1	180	94	613456	Frontonasal dysplasia 3
ALX3	77	83	136760	Frontonasal dysplasia 1
ALX4	77	93	613451	Frontonasal dysplasia 2
AMACR	85	91	614307	Alpha-methylacyl-CoA racemase deficiency
AMELX	114	88	301200	Amelogenesis imperfecta hypoplastic/hypomaturation type 1E
AMH	35	94	261550	Persistent Mullerian duct syndrome type I
AMHR2	98	79	261550	Persistent Mullerian duct syndrome type II
AMN	46	92	261100	Megaloblastic anemia-1 Norwegian type
AMPD1	119	92	200	-
AMT	135	94	605899	Glycine encephalopathy
ANG	173	96	611895	Amyotrophic lateral sclerosis 9
ANGPTL3	114	95	605019	Hypobetalipoproteinemia familial 2
ANK1	104	87	182900	Spherocytosis type 1
ANK2	113	88	600919	Cardiac arrhythmia ankyrin-B-related
ANKH	117	88	118600	Chondrocalcinosis 2
ANKK1	107	92	200	Dopamine receptor D2
ANKRD11	80	48	148050	KBG syndrome
ANKRD26	104	92	188000	Thrombocytopenia 2
ANO10	112	78	613728	Spinocerebellar ataxia autosomal recessive 10
ANO3	118	92	615034	Dystonia 24
ANO5	105	93	166260	Gnathodiaphyseal dysplasia
ANO6	105	89	262890	Scott syndrome
ANTXR2	120	96	228600	Hyaline fibromatosis syndrome
AP1S1	83	86	609313	MEDNIK syndrome
AP1S2	68	67	300630	Mental retardation X-linked syndromic Fried type
AP2S1	93	95	600740	Hypocalciuric hypercalcemia familial type III
AP3B1	108	93	608233	Hermansky-Pudlak syndrome 2
AP4B1	106	97	614066	Spastic paraplegia 47 autosomal recessive
AP4E1	127	92	613744	Spastic paraplegia 51 autosomal recessive
AP4M1	104	91	612936	Spastic paraplegia 50 autosomal recessive
AP4S1	75	84	614067	Spastic paraplegia 52 autosomal recessive
AP5Z1	74	86	613647	Spastic paraplegia 48 autosomal recessive
APC	152	99	200	Adenoma periampullary

APCDD1	127	91	605389	Hypotrichosis simplex
APOA1	80	89	105200	Amyloidosis 3 or more types
APOA2	112	81	200	-
APOA5	115	99	144650	Hyperchylomicronemia late-onset
APOB	179	94	144010	Hypercholesterolemia due to ligand-defective apo B
APOC2	168	100	207750	Hyperlipoproteinemia type Ib
APOC3	95	95	614028	Hyperalphalipoproteinemia 2
APOE	34	99	104310	Alzheimer disease-2
APP	105	78	104300	Alzheimer disease 1 familial
APRT	55	89	614723	Adenine phosphoribosyltransferase deficiency
APTX	143	96	208920	Ataxia early-onset with oculomotor apraxia and hypoalbuminemia
AQP2	74	84	125800	Diabetes insipidus nephrogenic
AR	81	41	300068	Androgen insensitivity
ARFGEF2	127	87	608097	Periventricular heterotopia with microcephaly
ARG1	60	100	207800	Argininemia
ARHGAP26	126	81	607785	Leukemia juvenile myelomonocytic
ARHGAP31	141	95	100300	Adams-Oliver syndrome 1
ARHGEF10	94	84	608236	Slowed nerve conduction velocity AD
ARHGEF12	127	93	601626	Leukemia acute myeloid
ARHGEF6	88	94	300436	Mental retardation X-linked 46
ARHGEF9	80	82	300607	Epileptic encephalopathy early infantile 8
ARID1A	112	91	614607	Mental retardation autosomal dominant 14
ARID1B	118	91	614562	Mental retardation autosomal dominant 12
ARL13B	113	94	612291	Joubert syndrome 8
ARL6	143	100	209900	Bardet-Biedl syndrome 3
ARNT	88	82	200	Leukemia
ARSA	83	94	250100	Metachromatic leukodystrophy
ARSB	91	85	253200	Mucopolysaccharidosis type VI (Maroteaux-Lamy)
ARSE	70	61	302950	Chondrodysplasia punctata X-linked recessive
ARX	37	94	308350	Epileptic encephalopathy early infantile 1
ASAH1	103	92	228000	Farber lipogranulomatosis
ASB10	78	88	603383	Glaucoma 1 open angle F
ASCC1	92	46	614266	Barrett esophagus/esophageal adenocarcinoma

ASCL1	120	100	209880	Central hypoventilation syndrome congenital
ASL	79	88	207900	Argininosuccinic aciduria
ASPA	118	96	271900	Canavan disease
ASPM	120	97	608716	Microcephaly 5 primary autosomal recessive
ASPSCR1	77	94	606243	Alveolar soft-part sarcoma
ASS1	68	37	215700	Citrullinemia
ASXL1	157	95	605039	Bohring-Opitz syndrome
ATCAY	88	94	601238	Ataxia cerebellar Cayman type
ATIC	116	87	608688	AICA-ribosiduria due to ATIC deficiency
ATL1	121	97	613708	Neuropathy hereditary sensory type ID
ATM	116	93	208900	Ataxia-telangiectasia
ATN1	129	60	125370	Dentatorubro-pallidoluysian atrophy
ATP13A2	81	88	606693	Parkinson disease 9
ATP1A2	106	82	104290	Alternating hemiplegia of childhood
ATP1A3	115	84	614820	Alternating hemiplegia of childhood 2
ATP2A1	115	89	601003	Brody myopathy
ATP2A2	119	92	101900	Acrokeratosis verruciformis
ATP2B3	88	83	302500	Spinocerebellar ataxia X-linked 1
ATP2C1	131	92	169600	Hailey-Hailey disease
ATP5E	148	100	614053	Mitochondrial complex V (ATP synthase) deficiency nuclear type 3
ATP6AP2	58	69	300423	Mental retardation X-linked with epilepsy
ATP6V0A2	110	93	219200	Cutis laxa autosomal recessive type IIA
ATP6V0A4	100	86	602722	Renal tubular acidosis distal autosomal recessive
ATP6V1B1	113	97	200	-
ATP7A	101	96	309400	Menkes disease
ATP7B	130	86	277900	Wilson disease
ATP8B1	122	87	243300	Cholestasis benign recurrent intrahepatic
ATPAF2	84	90	604273	Mitochondrial complex V (ATP synthase) deficiency nuclear type 1
ATR	118	94	614564	Cutaneous telangiectasia and cancer syndrome familial
ATRX	110	97	300448	Alpha-thalassemia myelodysplasia syndrome somatic
ATXN1	95	56	164400	Spinocerebellar ataxia 1
ATXN10	131	93	603516	Spinocerebellar ataxia 10
ATXN2	82	75	183090	Spinocerebellar ataxia 2

ATXN3	126	91	109150	Machado-Joseph disease
ATXN3	126	91	109150	Machado-Joseph disease
ATXN3	126	91	109150	Machado-Joseph disease
ATXN3	126	91	109150	Machado-Joseph disease
ATXN7	157	94	164500	Spinocerebellar ataxia 7
ATXN8OS	86	96	608768	Spinocerebellar ataxia 8
AUH	111	87	250950	3-methylglutaconic aciduria type I
AVP	25	94	125700	Diabetes insipidus neurohypophyseal
AVPR2	51	79	304800	Diabetes insipidus nephrogenic
AXIN1	115	91	607864	Caudal duplication anomaly
AXIN2	109	91	114500	Colorectal cancer somatic
B2M	171	95	241600	Hypoproteinemia hypercatabolic
B3GALNT2	99	79	615181	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies type A 11
B3GALT6	33	100	615349	Ehlers-Danlos syndrome progeroid type 2
B3GALTL	109	92	261540	Peters-plus syndrome
B3GAT3	87	61	245600	Multiple joint dislocations short stature craniofacial dysmorphism and congenital heart defects
B3GNT1	98	100	615287	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 13
B4GALNT1	89	86	609195	Spastic paraplegia 26 autosomal recessive
B4GALT1	86	95	607091	Congenital disorder of glycosylation type IId
B4GALT7	74	95	130070	Ehlers-Danlos syndrome progeroid type 1
B9D1	81	93	614209	Meckel syndrome 9
B9D2	53	96	614175	Meckel syndrome 10
BAAT	142	90	607748	Hypercholanemia familial
BAG3	161	97	613881	Cardiomyopathy dilated 1HH
BANF1	47	61	614008	Nestor-Guillermo progeria syndrome
BAP1	92	86	614327	Tumor predisposition syndrome
BAX	90	91	200	T-cell acute lymphoblastic leukemia
BBS1	126	89	209900	Bardet-Biedl syndrome 1
BBS10	128	100	209900	Bardet-Biedl syndrome 10
BBS12	163	100	209900	Bardet-Biedl syndrome 12
BBS2	130	91	209900	Bardet-Biedl syndrome 2
BBS4	106	87	209900	Bardet-Biedl syndrome 4
BBS5	114	96	209900	Bardet-Biedl syndrome 5

BBS7	121	98	209900	Bardet-Biedl syndrome 7
BBS9	117	95	209900	Bardet-Biedl syndrome 9
BCHE	166	98	200	Apnea
BCKDHA	109	92	248600	Maple syrup urine disease type Ia
BCKDHB	89	98	248600	Maple syrup urine disease type Ib
BCKDK	119	96	614923	Branched-chain ketoacid dehydrogenase kinase deficiency
BCL10	114	81	137245	Lymphoma MALT somatic
BCL2	129	89	2	Leukemia/lymphoma B-cell
BCL7A	91	86	200	B-cell non-Hodgkin lymphoma
BCMO1	129	84	115300	Hypercarotenemia and vitamin A deficiency autosomal dominant
BCOR	97	94	300166	Microphthalmia syndromic 2
BCR	105	89	613065	Leukemia acute lymphocytic
BCS1L	133	86	262000	Bjornstad syndrome
BDNF	216	100	209880	Central hypoventilation syndrome congenital
BEST1	110	94	153700	Best macular dystrophy
BFSP1	151	97	611391	Cataract 33
BFSP2	68	87	611597	Cataract 12 multiple types
BICD2	90	96	615290	Spinal muscular atrophy lower extremity-predominant 2 AD
BIN1	60	89	255200	Myopathy centronuclear autosomal recessive
BLK	110	88	613375	Maturity-onset diabetes of the young type 11
BLM	128	89	210900	Bloom syndrome
BLNK	102	91	613502	Agammaglobulinemia 4
BLOC1S3	19	90	614077	Hermansky-Pudlak syndrome 8
BLOC1S6	112	88	614171	Hermansky-pudlak syndrome 9
BLVRA	91	88	614156	Hyperbiliverdinemia
BMP1	97	86	614856	Osteogenesis imperfecta type XIII
BMP15	126	93	300510	Ovarian dysgenesis 2
BMP2	119	100	112600	Brachydactyly type A2
BMP4	115	94	607932	Microphthalmia syndromic 6
BMPER	141	91	608022	Diaphanospondylodysostosis
BMPR1A	80	40	174900	Juvenile polyposis syndrome infantile form
BMPR1B	118	90	112600	Brachydactyly type A2
BMPR2	163	97	178600	Pulmonary hypertension familial primary 1 with or without HHT

BOLA3	63	51	614299	Multiple mitochondrial dysfunctions syndrome 2
BPGM	145	100	222800	Erythrocytosis due to bisphosphoglycerate mutase deficiency
BRAF	82	81	211980	Adenocarcinoma of lung somatic
BRAT1	88	89	614498	Rigidity and multifocal seizure syndrome lethal neonatal
BRCA2	145	97	605724	Fanconi anemia complementation group D1
BRIP1	129	95	114480	Breast cancer early-onset
BRWD3	90	98	300659	Mental retardation X-linked 93
BSCL2	120	85	269700	Lipodystrophy congenital generalized type 2
BSND	109	89	602522	Bartter syndrome type 4a
BTD	146	94	253260	Biotinidase deficiency
BTK	86	90	307200	Agammaglobulinemia and isolated hormone deficiency
BUB1	124	88	200	Colorectal cancer with chromosomal instability
BUB1B	127	94	114500	Colorectal cancer somatic
C10orf11	113	77	615179	Albinism oculocutaneous type V
C10orf2	128	69	271245	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type)
C12orf57	71	78	218340	Temtamy syndrome
C12orf65	159	100	613559	Combined oxidative phosphorylation deficiency 7
C19orf12	90	90	614298	Neurodegeneration with brain iron accumulation 4
C1GALT1C1	118	100	300622	Tn polyagglutination syndrome somatic
C1QA	107	83	613652	C1q deficiency
C1QB	106	83	613652	C1q deficiency
C1QC	117	87	613652	C1q deficiency
C1QTNF5	80	69	605670	Retinal degeneration late-onset autosomal dominant
C1S	115	98	613783	C1s deficiency
C2	18	92	217000	C2 deficiency
C2orf71	110	99	613428	Retinitis pigmentosa 54
C3	99	93	613779	C3 deficiency
C4B	23	81	614379	C4B deficiency
C4orf26	152	100	614832	Amelogenesis imperfecta hypomaturation type IIA4
C5	106	90	609536	C5 deficiency
C5orf42	128	96	614615	Joubert syndrome 17
C6	123	89	612446	C6 deficiency
C7	96	89	610102	C7 deficiency

C8A	94	88	613790	C8 deficiency type I
C8B	112	82	613789	C8 deficiency type II
C8orf37	94	86	614500	Cone-rod dystrophy 16
C9	117	100	613825	C9 deficiency with dermatomyositis
C9orf72	83	95	105550	Amyotrophic lateral sclerosis and/or frontotemporal dementia
CA12	88	90	143860	Hyperchlorhidrosis isolated
CA2	157	87	259730	Osteopetrosis autosomal recessive 3 with renal tubular acidosis
CA4	79	91	600852	Retinitis pigmentosa 17
CA8	75	99	613227	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3
CABP2	74	74	614899	Deafness autosomal recessive 93
CABP4	71	87	610427	Night blindness congenital stationary (incomplete) 2B autosomal recessive
CACNA1A	89	88	108500	Episodic ataxia type 2
CACNA1C	111	83	611875	Brugada syndrome 3
CACNA1C	111	83	611875	Brugada syndrome 3
CACNA1C	111	83	611875	Brugada syndrome 3
CACNA1D	126	87	614896	Sinoatrial node dysfunction and deafness
CACNA1F	69	90	300600	Aland Island eye disease
CACNA1S	93	92	170400	Hypokalemic periodic paralysis type 1
CACNA2D4	91	91	610478	Retinal cone dystrophy 4
CACNB2	132	92	611876	Brugada syndrome 4
CACNB4	100	92	613855	Episodic ataxia type 5
CACNG2	89	100	614256	Mental retardation autosomal dominant 10
CALM1	152	70	614916	Ventricular tachycardia catecholaminergic polymorphic 4
CALR3	107	87	613875	Cardiomyopathy familial hypertrophic 19
CAMTA1	133	91	614756	Cerebellar ataxia nonprogressive with mental retardation
CANT1	90	92	251450	Desbuquois dysplasia
CAPN3	128	93	253600	Muscular dystrophy limb-girdle type 2A
CARD11	105	88	615206	Persistent polyclonal B-cell lymphocytosis 606445 (3) Immunodeficiency primary autosomal recessive CARD11-related
CARD14	79	64	173200	Pityriasis rubra pilaris
CARD9	59	97	212050	Candidiasis familial 2 autosomal recessive
CASC5	151	99	604321	Microcephaly 4 primary autosomal recessive
CASK	77	93	300422	FG syndrome 4

CASP10	98	97	603909	Autoimmune lymphoproliferative syndrome type II
CASP8	141	91	114550	Hepatocellular carcinoma somatic
CASQ2	95	94	611938	Ventricular tachycardia catecholaminergic polymorphic 2
CASR	125	96	239200	Hyperparathyroidism neonatal
CAT	99	88	614097	Acatalasemia
CATSPER1	99	96	612997	Spermatogenic failure 7
CAV1	121	100	612526	Lipodystrophy congenital generalized type 3
CAV3	132	100	192600	Cardiomyopathy familial hypertrophic
CBL	140	91	613563	Noonan syndrome-like disorder with or without juvenile meylomonocytic leukemia
CBS	77	86	236200	Homocystinuria B6-responsive and nonresponsive types
CBX2	139	94	613080	46XY sex reversal 5
CC2D1A	99	90	608443	Mental retardation autosomal recessive 3
CC2D2A	101	90	216360	COACH syndrome
CCBE1	97	91	235510	Hennekam lymphangiectasia-lymphedema syndrome
CCDC103	115	88	614679	Ciliary dyskinesia primary 17
CCDC11	181	92	614779	Heterotaxy visceral 6 autosomal recessive
CCDC114	74	90	615067	Ciliary dyskinesia primary 20
CCDC39	100	90	613807	Ciliary dyskinesia primary 14
CCDC40	100	86	613808	Ciliary dyskinesia primary 15
CCDC50	131	92	607453	Deafness autosomal dominant 44
CCDC78	84	95	614807	Myopathy centronuclear 4
CCDC8	121	48	614205	Three M syndrome 3
CCDC88C	98	88	236600	Hydrocephalus nonsyndromic autosomal recessive
CCT5	119	60	256840	Neuropathy hereditary sensory with spastic paraplegia
CD151	98	85	609057	Nephropathy with pretibial epidermolysis bullosa and deafness
CD19	83	93	613493	Immunodeficiency common variable 3
CD247	107	83	610163	Immunodeficiency due to defect in CD3-zeta
CD27	89	85	615122	Lymphoproliferative syndrome 2
CD2AP	114	96	607832	Glomerulosclerosis focal segmental 3
CD320	80	93	613646	Methylmalonic aciduria due to transcobalamin receptor defect
CD36	119	95	608404	Platelet glycoprotein IV deficiency
CD3D	107	100	608971	Severe combined immunodeficiency T cell-negative B-cell/natural killer-cell positive
CD3E	124	87	200	-

CD3G	106	94	200	-
CD4	89	89	613949	OKT4 epitope deficiency
CD40	110	87	606843	Immunodeficiency with hyper-IgM type 3
CD40LG	89	95	308230	Immunodeficiency, X-linked, with hyper-IgM
CD59	133	93	612300	CD59 deficiency
CD79A	63	93	613501	Agammaglobulinemia 3
CD79B	128	99	612692	Agammaglobulinemia 6
CD81	70	85	613496	Immunodeficiency common variable 6
CD8A	76	94	608957	CD8 deficiency familial
CD96	120	97	211750	C syndrome
CDAN1	114	84	224120	Anemia congenital dyserythropoietic type I
CDC6	120	92	613805	Meier-Gorlin syndrome 5
CDC73	141	93	145000	Hyperparathyroidism familial primary
CDH1	124	83	608089	Endometrial carcinoma somatic
CDH15	75	89	612580	Mental retardation autosomal dominant 3
CDH23	92	88	601386	Deafness autosomal recessive 12
CDH3	104	86	225280	Ectodermal dysplasia ectrodactyly and macular dystrophy
CDHR1	108	86	613660	Cone-rod dystrophy 15
CDK5RAP2	121	88	604804	Microcephaly 3 primary autosomal recessive
CDKL5	118	92	105830	Angelman syndrome-like
CDKN1B	94	44	610755	Multiple endocrine neoplasia type IV
CDKN1C	34	37	130650	Beckwith-Wiedemann syndrome
CDKN2A	75	66	155755	Melanoma and neural system tumor syndrome
CDON	132	83	614226	Holoprosencephaly 11
CDSN	16	95	146520	Hypotrichosis simplex of scalp 1
CDT1	51	85	613804	Meier-Gorlin syndrome 4
CEACAM16	91	86	614614	Deafness autosomal dominant 4B
CEBPA	24	100	601626	Leukemia acute myeloid
CEBPE	90	97	245480	Specific granule deficiency
CEL	84	46	609812	Maturity-onset diabetes of the young type VIII
CENPJ	131	96	608393	Microcephaly 6 primary autosomal recessive
CEP135	115	93	614673	Microcephaly 8 primary autosomal recessive
CEP152	138	93	614852	Microcephaly 9 primary autosomal recessive

CEP164	89	87	614845	Nephronophthisis 15
CEP290	95	92	209900	Bardet-Biedl syndrome 14
CEP41	94	92	614464	Joubert syndrome 15
CEP57	99	72	614114	Mosaic variegated aneuploidy syndrome 2
CEP63	113	88	614728	Seckel syndrome 6
CERKL	127	93	608380	Retinitis pigmentosa 26
CERS3	88	93	615023	Ichthyosis congenital autosomal recessive 9
CES1	115	47	1	-
CETP	102	94	143470	Hyperalphalipoproteinemia
CFD	53	92	613912	Complement factor D deficiency
CFH	112	70	609814	Complement factor H deficiency
CFHR5	109	80	614809	Nephropathy due to CFHR5 deficiency
CFI	141	92	610984	Complement factor I deficiency
CFL2	113	100	610687	Nemaline myopathy 7 autosomal recessive
CFP	77	82	312060	Properdin deficiency, X-linked
CFTR	136	94	277180	Congenital bilateral absence of vas deferens
CHAT	83	83	254210	Myasthenic syndrome congenital associated with episodic apnea
CHD7	129	92	214800	CHARGE syndrome
CHEK2	86	86	609265	Li-Fraumeni syndrome
CHKB	89	90	602541	Muscular dystrophy congenital megaconial type
CHM	79	94	303100	Choroideremia
CHMP1A	98	84	614961	Pontocerebellar hypoplasia type 8
CHMP2B	129	97	614696	Amyotrophic lateral sclerosis 17
CHMP4B	115	78	605387	Cataract 31 multiple types
CHN1	123	92	604356	Duane retraction syndrome 2
CHRDL1	106	82	200	Megalocornea 1
CHRM3	174	100	100100	Eagle-Barrett syndrome
CHRNA1	111	82	253290	Multiple pterygium syndrome lethal type
CHRNA2	127	53	610353	Epilepsy nocturnal frontal lobe type 4
CHRNA4	105	53	600513	Epilepsy nocturnal frontal lobe 1
CHRNB1	113	84	608931	Myasthenic syndrome congenital associated with acetylcholine receptor deficiency
CHRNB2	142	94	605375	Epilepsy nocturnal frontal lobe 3
CHRND	106	84	253290	Multiple pterygium syndrome lethal type

CHRNE	139	100	608931	Myasthenic syndrome congenital associated with acetylcholine receptor deficiency
CHRNG	98	82	265000	Escobar syndrome
CHST14	107	100	601776	Ehlers-Danlos syndrome musculocontractural type
CHST3	71	100	143095	Spondyloepiphyseal dysplasia with congenital joint dislocations
CHST6	109	48	217800	Macular corneal dystrophy
CHSY1	154	100	605282	Temtamy preaxial brachydactyly syndrome
CHUK	100	89	613630	Cocoon syndrome
CIB2	76	79	609439	Deafness autosomal recessive 48
CIITA	95	91	209920	Bare lymphocyte syndrome, type II, complementation group A
CIITA	95	91	180300	{Rheumatoid arthritis, susceptibility to}
CIRH1A	105	89	604901	Cirrhosis North American Indian childhood type
CISD2	220	100	604928	Wolfram syndrome 2
CITED2	104	94	614433	Atrial septal defect 8
CLCF1	46	85	610313	Cold-induced sweating syndrome 1
CLCN1	96	90	160800	Myotonia congenita dominant
CLCN5	123	88	300009	Dent disease
CLCN7	80	88	166600	Osteopetrosis autosomal dominant 2
CLCNKA	109	63	613090	Bartter syndrome type 4b digenic
CLCNKB	95	64	607364	Bartter syndrome type 3
CLDN1	99	92	607626	Ichthyosis leukocyte vacuoles alopecia and sclerosing cholangitis
CLDN14	61	100	614035	Deafness autosomal recessive 29
CLDN16	146	92	248250	Hypomagnesemia 3 renal
CLDN19	70	89	248190	Hypomagnesemia 5 renal with ocular involvement
CLEC7A	106	99	613108	Candidiasis familial 4 autosomal recessive
CLIC2	59	87	300886	Mental retardation X-linked syndromic 32
CLMP	112	84	615237	Congenital short bowel syndrome
CLN3	89	87	204200	Ceroid lipofuscinosis neuronal 3
CLN5	124	98	256731	Ceroid lipofuscinosis neuronal 5
CLN6	64	88	601780	Ceroid lipofuscinosis neuronal 6
CLN8	131	96	600143	Ceroid lipofuscinosis neuronal 8
CLPP	84	94	614129	Perrault syndrome 3
CLRN1	170	98	614180	Retinitis pigmentosa 61
CNGA1	128	98	613756	Retinitis pigmentosa 49

CNGA3	144	97	216900	Achromatopsia-2
CNGB1	96	87	613767	Retinitis pigmentosa 45
CNGB3	111	85	262300	Achromatopsia-3
CNNM2	131	95	613882	Hypomagnesemia 6 renal
CNNM4	162	94	217080	Jalili syndrome
CNTN1	104	94	612540	Myopathy congenital Compton-North
CNTNAP2	115	86	610042	Cortical dysplasia-focal epilepsy syndrome
COA5	120	64	220110	Mitochondrial complex IV deficiency
COCH	139	93	601369	Deafness autosomal dominant 9
COG1	125	90	611209	Congenital disorder of glycosylation type IIg
COG4	96	84	613489	Congenital disorder of glycosylation type IIj
COG5	105	95	613612	Congenital disorder of glycosylation type IIi
COG6	91	96	615328	Shaheen syndrome
COG7	99	76	608779	Congenital disorder of glycosylation type IIe
COG8	119	79	611182	Congenital disorder of glycosylation type IIh
COL10A1	98	96	156500	Metaphyseal chondrodysplasia Schmid type
COL11A1	95	96	228520	Fibrochondrogenesis
COL11A2	15	85	601868	Deafness autosomal dominant 13
COL17A1	103	86	226650	Epidermolysis bullosa junctional non-Herlitz type
COL18A1	77	90	267750	Knobloch syndrome type 1
COL1A1	106	91	114000	Caffey disease
COL1A2	110	82	225320	Ehlers-Danlos syndrome cardiac valvular form
COL2A1	95	86	200610	Achondrogenesis type II or hypochondrogenesis
COL3A1	83	86	130020	Ehlers-Danlos syndrome type III
COL4A1	109	77	611773	Angiopathy hereditary with nephropathy aneurysms and muscle
COL4A2	92	84	614483	Porencephaly 2
COL4A3	82	83	104200	Alport syndrome autosomal dominant
COL4A4	100	83	203780	Alport syndrome autosomal recessive
COL4A5	64	87	301050	Alport syndrome
COL5A1	109	87	130000	Ehlers-Danlos syndrome type I
COL5A2	90	90	130000	Ehlers-Danlos syndrome type I
COL6A1	78	93	158810	Bethlem myopathy
COL6A2	80	95	158810	Bethlem myopathy

COL6A3	130	94	158810	Bethlem myopathy
COL7A1	104	93	226600	EBD inversa
COL8A2	63	95	609140	Corneal dystrophy polymorphous posterior 2
COL9A1	110	91	614135	Epiphyseal dysplasia multiple 6
COL9A2	76	86	600204	Epiphyseal dysplasia multiple 2
COL9A3	80	87	600969	Epiphyseal dysplasia multiple 3
COLEC11	109	96	265050	3MC syndrome 2
COLQ	85	87	603034	Endplate acetylcholinesterase deficiency
COMP	86	89	132400	Epiphyseal dysplasia multiple 1
COQ2	79	93	607426	Coenzyme Q10 deficiency primary 1
COQ6	147	90	614650	Coenzyme Q10 deficiency primary 6
COQ9	113	83	614654	Coenzyme Q10 deficiency primary 5
CORIN	125	92	614595	Preeclampsia/eclampsia 5
COX10	147	83	200	Encephalopathy progressive mitochondrial
COX14	165	68	220110	Mitochondrial complex IV deficiency
COX15	84	88	615119	Cardioencephalomyopathy fatal infantile due to cytochrome c oxidase deficiency 2
COX4I2	63	84	612714	Exocrine pancreatic insufficiency dyserythropoietic anemia and calvarial hyperostosis
COX6B1	99	67	220110	Cytochrome c oxidase deficiency
COX7B	47	78	300887	Aplasia cutis congenita reticulolinear with mmicrocephaly facial dysmorphism and other congenital anomalies
CP	97	72	604290	Cerebellar ataxia
CPA6	123	92	614417	Epilepsy familial temporal lobe 5
CPN1	93	87	212070	Carboxypeptidase N deficiency
CPOX	84	93	121300	Coproporphyria
CPS1	107	93	237300	Carbamoylphosphate synthetase I deficiency
CPT1A	111	81	255120	CPT deficiency hepatic type IA
CPT2	116	96	600649	CPT deficiency hepatic type II
CR2	127	92	614699	Immunodeficiency common variable 7
CRADD	162	90	614499	Mental retardation autosomal recessive 34
CRB1	171	94	613835	Leber congenital amaurosis 8
CRBN	135	98	607417	Mental retardation autosomal recessive 2
CREB1	96	91	612160	Histiocytoma angiomatoid fibrous somatic
CREBBP	90	92	180849	Rubinstein-Taybi syndrome
CRELD1	96	92	606217	Atrioventricular septal defect partial with heterotaxy syndrome

CRLF1	64	86	272430	Cold-induced sweating syndrome
CRTAP	109	88	610682	Osteogenesis imperfecta type VII
CRTC1	67	89	200	Mucoepidermoid salivary gland carcinoma
CRX	149	91	120970	Cone-rod retinal dystrophy-2
CRYAA	114	93	604219	Cataract 9 multiple types
CRYAB	125	98	615184	Cardiomyopathy dilated 1II
CRYBA1	120	89	600881	Cataract 10 multiple types
CRYBA4	67	87	610425	Cataract 23
CRYBB1	64	84	611544	Cataract 17 multiple types
CRYBB2	110	83	601547	Cataract 3 multiple types
CRYBB3	108	85	609741	Cataract 22 autosomal recessive
CRYGB	111	89	200	Cataract 39 multiple types
CRYGC	102	88	604307	Cataract 2 multiple types
CRYGD	71	58	115700	Cataract 4 multiple types
CRYGS	103	90	116100	Cataract 20 multiple types
CRYM	72	87	200	Deafness
CSF1R	84	85	221820	Leukoencephalopathy diffuse hereditary with spheroids
CSF2RB	101	88	614370	Surfactant metabolism dysfunction pulmonary 5
CSF3R	96	90	162830	Neutrophilia hereditary
CSNK1D	107	84	615224	Advanced sleep-phase syndrome familial 2
CSRP3	154	76	607482	Cardiomyopathy dilated 1M
CST3	44	97	105150	Cerebral amyloid angiopathy
CSTA	122	97	607936	Exfoliative ichthyosis autosomal recessive ichthyosis bullosa of Siemens-like
CSTB	146	96	254800	Epilepsy progressive myoclonic 1A (Unverricht and Lundborg)
CTC1	118	86	612199	Cerebroretinal microangiopathy with calcifications and cysts
CTDP1	79	88	604168	Congenital cataracts facial dysmorphism and neuropathy
CTH	120	95	219500	Cystathioninuria
CTHRC1	85	100	614266	Barrett esophagus/esophageal adenocarcinoma
CTNNB1	140	95	114500	Colorectal cancer somatic
CTNS	116	93	219800	Cystinosis atypical nephropathic
CTSA	99	90	256540	Galactosialidosis
CTSC	99	96	245010	Haim-Munk syndrome
CTSD	84	97	610127	Ceroid lipofuscinosis neuronal 10

CTSK	128	85	265800	Pycnodysostosis
CUBN	100	78	261100	Megaloblastic anemia-1 Finnish type
CUL3	111	96	614496	Pseudohypoaldosteronism type IIE
CUL4B	89	95	300354	Mental retardation X-linked syndromic 15 (Cabezas type)
CUL7	106	89	273750	3-M syndrome 1
CXCR4	198	100	200	Myelokathexis
CYB5A	80	72	250790	Methemoglobinemia type IV
CYB5R3	66	91	250800	Methemoglobinemia type I
CYBA	43	85	233690	Chronic granulomatous disease autosomal due to deficiency of CYBA
CYBB	80	85	300645	Atypical mycobacteriosis familial X-linked 2
CYCS	105	22	612004	Thrombocytopenia 4
CYLD	129	96	605041	Brooke-Spiegler syndrome
CYP11A1	86	94	200	-
CYP11B1	124	64	202010	Adrenal hyperplasia congenital due to 11-beta-hydroxylase deficiency
CYP11B2	112	53	200	-
CYP17A1	112	87	202110	1720-lyase deficiency isolated
CYP19A1	140	92	613546	Aromatase deficiency
CYP1B1	108	98	231300	Glaucoma 3A primary open angle congenital juvenile or adult onset
CYP21A2	19	82	201910	Adrenal hyperplasia congenital due to 21-hydroxylase deficiency
CYP24A1	99	90	143880	Hypercalcemia infantile
CYP26B1	67	92	614416	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies
CYP26C1	61	90	614974	Focal facial dermal dysplasia 4
CYP27A1	126	86	213700	Cerebrotendinous xanthomatosis
CYP27B1	119	85	264700	Vitamin D-dependent rickets type I
CYP2A6	50	40	122700	Coumarin resistance
CYP2B6	133	55	200	Efavirenz poor metabolism of
CYP2C8	133	85	200	Rhabdomyolysis
CYP2C9	149	45	200	-
CYP2R1	108	93	600081	Rickets due to defect in vitamin D 25-hydroxylation
CYP2U1	109	94	615030	Spastic paraplegia 56 autosomal recessive
CYP4F22	101	85	604777	Ichthyosis congenital autosomal recessive 5
CYP4V2	113	89	210370	Bietti crystalline corneoretinal dystrophy
CYP7B1	98	88	613812	Bile acid synthesis defect congenital 3

D2HGDH	58	84	600721	D-2-hydroxyglutaric aciduria
DAG1	127	100	613818	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 9
DARS	112	99	615281	Hypomyelination with brainstem and spinal cord involvement and leg spasticity
DARS2	112	97	611105	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation
DBH	99	89	223360	Dopamine beta-hydroxylase deficiency
DBT	100	93	248600	Maple syrup urine disease type II
DCAF17	96	97	241080	Woodhouse-Sakati syndrome
DCC	130	85	114500	Colorectal cancer somatic
DCLRE1C	117	98	603554	Omenn syndrome
DCN	108	85	610048	Corneal dystrophy congenital stromal
DCTN1	133	90	607641	Neuropathy distal hereditary motor type VIIB
DCX	111	94	300067	Lissencephaly X-linked
DDB2	102	85	278740	Xeroderma pigmentosum group E DDB-negative subtype
DDC	102	93	608643	Aromatic L-amino acid decarboxylase deficiency
DDHD1	117	99	609340	Spastic paraplegia 28 autosomal recessive
DDHD2	112	91	615033	Spastic paraplegia 54 autosomal recessive
DDOST	113	89	614507	Congenital disorder of glycosylation type Ir
DDR2	138	83	271665	Spondylometaepiphyseal dysplasia short limb-hand type
DDX11	89	82	613398	Warsaw breakage syndrome
DEPDC5	118	87	604364	Epilepsy familial focal with variable foci
DES	94	94	604765	Cardiomyopathy dilated 1I
DFNA5	98	81	600994	Deafness autosomal dominant 5
DFNB31	79	87	611383	Usher syndrome, type 2D
DGKE	109	91	615008	Nephrotic syndrome type 7
DGUOK	102	90	251880	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type)
DHCR24	95	79	602398	Desmosterolosis
DHCR7	118	88	270400	Smith-Lemli-Opitz syndrome
DHDDS	97	83	613861	Retinitis pigmentosa 59
DHFR	53	34	613839	Megaloblastic anemia due to dihydrofolate reductase deficiency
DHH	76	94	607080	46XY partial gonadal dysgenesis with minifascicular neuropathy
DHODH	107	81	263750	Miller syndrome
DHTKD1	114	86	204750	2-aminoadipic 2-oxoadipic aciduria
DIAPH1	95	92	124900	Deafness autosomal dominant 1

DIAPH2	87	95	300511	Premature ovarian failure
DIAPH3	105	95	609129	Auditory neuropathy autosomal dominant 1
DICER1	128	93	138800	Goiter multinodular 1 with or without Sertoli-Leydig cell tumors
DIP2B	110	87	136630	Mental retardation FRA12A type
DIS3L2	152	90	267000	Perlman syndrome
DKC1	84	94	305000	Dyskeratosis congenita X-linked
DLAT	116	89	245348	Pyruvate dehydrogenase E2 deficiency
DLC1	147	94	200	Colorectal cancer
DLD	141	94	246900	Dihydrolipoamide dehydrogenase deficiency
DLG3	64	87	300850	Mental retardation X-linked 90
DLL3	71	85	277300	Spondylocostal dysostosis autosomal recessive 1
DLX3	67	91	104510	Amelogenesis imperfecta hypomaturation-hypoplastic type with taurodontism
DLX5	103	92	220600	Split-hand/foot malformation 1 with sensorineural hearing loss
DMD	91	91	300376	Becker muscular dystrophy
DMGDH	124	88	605850	Dimethylglycine dehydrogenase deficiency
DMP1	117	99	241520	Hypophosphatemic rickets AR
DMPK	100	89	160900	Myotonic dystrophy 1
DNA2	114	74	615156	Progressive external ophthalmoplegia with mitochondrial DNA deletions autosomal dominant 6
DNAAF3	73	87	606763	Ciliary dyskinesia primary 2
DNAH11	122	87	611884	Ciliary dyskinesia primary 7 with or without situs inversus
DNAH5	106	88	608644	Ciliary dyskinesia primary 3 with or without situs inversus
DNAI1	140	90	244400	Ciliary dyskinesia primary 1 with or without situs inversus
DNAI2	112	92	612444	Ciliary dyskinesia primary 9 with or without situs inversus
DNAJB2	120	90	614881	Spinal muscular atrophy distal autosomal recessive 5
DNAJB6	70	48	603511	Muscular dystrophy limb-girdle type 1E
DNAJC19	101	83	610198	3-methylglutaconic aciduria type V
DNAJC5	90	76	162350	Ceroid lipofuscinosis neuronal 4 Parry type
DNAL1	99	81	614017	Ciliary dyskinesia primary 16
DNASE1L3	108	90	614420	Systemic lupus erythematosus 16
DNM1L	92	98	614388	Encephalopahty lethal due to defective mitochondrial peroxisomal fission
DNM2	85	89	606482	Charcot-Marie-Tooth disease axonal type 2M
DNMT1	105	89	604121	Cerebellar ataxia deafness and narcolepsy autosomal dominant
DNMT3B	93	82	242860	Immunodeficiency-centromeric instability-facial anomalies syndrome 1

DOCK6	91	88	614219	Adams-Oliver syndrome 2
DOCK8	97	84	243700	Hyper-IgE recurrent infection syndrome autosomal recessive
DOK7	59	85	208150	Fetal akinesia deformation sequence
DOLK	142	100	200	-
DPAGT1	116	89	608093	Congenital disorder of glycosylation type Ij
DPM1	145	100	608799	Congenital disorder of glycosylation type Ie
DPM2	72	90	615042	Congenital disorder of glycosylation type Iu
DPM3	87	100	612937	Congenital disorder of glycosylation type Io
DPP6	102	90	612956	Ventricular fibrillation paroxysmal familial 2
DPY19L2	116	54	613958	Spermatogenic failure 9
DPYD	122	88	274270	5-fluorouracil toxicity
DPYS	90	78	222748	Dihydropyrimidinuria
DRC1	85	92	615294	Ciliary dyskinesia primary 21
DRD2	106	88	159900	Dystonia myoclonic
DRD4	33	93	200	Autonomic nervous system dysfunction
DRD5	65	9	200	Dystonia
DSC2	111	53	610476	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair
DSC3	97	95	613102	Hypotrichosis and recurrent skin vesicles
DSG1	134	93	148700	Keratosis palmoplantaris striata I
DSG2	135	94	610193	Arrhythmogenic right ventricular dysplasia 10
DSG4	129	92	607903	Hypotrichosis localized autosomal recessive
DSP	144	93	607450	Arrhythmogenic right ventricular dysplasia 8
DSPP	227	26	605594	Deafness autosomal dominant 36 with dentinogenesis
DST	133	93	614653	Neuropathy hereditary sensory and autonomic type VI
DTNA	91	32	604169	Left ventricular noncompaction 1 with or without congenital heart defects
DTNBP1	119	82	614076	Hermansky-Pudlak syndrome 7
DUOX2	108	78	607200	Thryoid dyshormonogenesis 6
DUOXA2	88	92	274900	Thyroid dyshormonogenesis 5
DUSP6	155	90	615269	Hypogonadotropic hypogonadism 19 with or without anosmia
DYM	98	94	223800	Dyggve-Melchior-Clausen disease
DYNC1H1	133	88	614228	Charcot-Marie-Tooth disease axonal type 20
DYNC2H1	111	96	613091	Asphyxiating thoracic dystrophy 3
DYRK1A	182	43	614104	Mental retardation autosomal dominant 7

DYSF	105	88	254130	Miyoshi muscular dystrophy 1
EARS2	91	93	614924	Combined oxidative phosphorylation deficiency 12
EBP	71	78	302960	Chondrodysplasia punctata X-linked dominant
ECE1	100	89	613870	Hirschsprung disease cardiac defects and autonomic dysfunction
ECEL1	50	66	615065	Arthrogryposis distal type 5D
ECM1	128	88	247100	Urbach-Wiethe disease
EDAR	85	88	129490	Ectodermal dysplasia 10A hypohidrotic/hair/nail type autosomal dominant
EDARADD	99	97	614940	Ectodermal dysplasia 11A hypohidrotic/hair/tooth type
EDN3	93	84	209880	Central hypoventilation syndrome congenital
EDNRA	125	91	157300	Migraine resistance to
EDNRB	148	95	600501	ABCD syndrome
EFEMP1	123	95	126600	Doyne honeycomb degeneration of retina
EFEMP2	101	91	614437	Cutis laxa autosomal recessive type IB
EFNB1	88	93	304110	Craniofrontonasal dysplasia
EFTUD2	108	87	610536	Mandibulofacial dysostosis Guion-Almeida type
EGF	124	88	611718	Hypomagnesemia 4 renal
EGFR	109	86	211980	Adenocarcinoma of lung response to tyrosine kinase inhibitor in
EGLN1	63	95	609820	Erythrocytosis familial 3
EGR2	92	91	607678	Charcot-Marie-Tooth disease type 1D
EHMT1	105	85	610253	Kleefstra syndrome
EIF2AK3	117	94	226980	Wolcott-Rallison syndrome
EIF2B1	139	82	603896	Leukoencephalopathy with vanishing white matter
EIF2B2	109	86	603896	Leukoencephalopathy with vanishing white matter
EIF2B3	109	94	603896	Leukoencephalopathy with vanishing white matter
EIF2B4	130	86	603896	Leukoencephaly with vanishing white matter
EIF2B5	112	94	603896	Leukoencephalopathy with vanishing white matter
EIF4G1	116	92	614251	Parkinson disease 18
ELANE	124	83	162800	Neutropenia cyclic
ELN	79	87	123700	Cutis laxa AD
ELOVL4	111	92	614457	Ichthyosis spastic quadriplegia and mental retardation
EMD	127	100	310300	Emery-Dreifuss muscular dystrophy 1 X-linked
EMG1	118	82	211180	Bowen-Conradi syndrome
EMX2	125	100	269160	Schizencephaly

ENAM	144	99	104500	Amelogenesis imperfecta type IB
ENG	73	84	187300	Telangiectasia hereditary hemorrhagic type 1
ENO3	116	88	612932	Glycogen storage disease XIII
ENPP1	112	93	208000	Arterial calcification generalized of infancy 1
EOGT	97	95	615297	Adams-Oliver syndrome 4
EP300	141	93	114500	Colorectal cancer somatic
EPAS1	105	83	611783	Erythrocytosis familial 4
EPB41	143	92	611804	Elliptocytosis-1
EPB41L1	93	86	614257	Mental retardation autosomal dominant 11
EPB42	108	86	612690	Spherocytosis hereditary type 5
EPCAM	110	90	613244	Colorectal cancer hereditary nonpolyposis type 8
EPG5	97	87	242840	Vici syndrome
EPHA2	95	89	116600	Cataract 6 multiple types
EPHB2	109	92	603688	Prostate cancer progression and metastasis of
EPHX1	92	90	607748	Hypercholanemia familial
EPM2A	76	86	254780	Epilepsy progressive myoclonic 2A (Lafora)
EPX	105	83	261500	Eosinophil peroxidase deficiency
ERBB2	99	90	211980	Adenocarcinoma of lung somatic
ERBB3	137	87	607598	Lethal congenital contractural syndrome 2
ERCC1	74	95	610758	Cerebrooculofacioskeletal syndrome 4
ERCC2	88	89	610756	Cerebrooculofacioskeletal syndrome 2
ERCC3	134	93	601675	Trichothiodystrophy
ERCC4	162	93	615272	Fanconi anemia complementation group Q
ERCC5	130	94	278780	Xeroderma pigmentosum group G
ERCC6	157	92	214150	Cerebrooculofacioskeletal syndrome 1
ERCC8	84	94	216400	Cockayne syndrome type A
ERF	112	92	600775	Craniosynostosis 4
ERLIN2	125	94	611225	Spastic paraplegia 18 autosomal recessive
ESCO2	91	97	268300	Roberts syndrome
ESPN	47	48	609006	Deafness autosomal recessive 36
ESR1	132	87	200	-
ESRRB	64	88	608565	Deafness autosomal recessive 35
ETFA	109	97	231680	Glutaric acidemia IIA

ETFB	101	86	231680	Glutaric acidemia IIB
ETFDH	123	96	231680	Glutaric acidemia IIC
ETHE1	65	93	602473	Ethylmalonic encephalopathy
ETV6	120	95	601626	Leukemia acute myeloid somatic
EVC	93	82	225500	Ellis-van Creveld syndrome
EWSR1	93	56	612219	Ewing sarcoma
EXOSC3	141	65	614678	Pontocerebellar hypoplasia type 1B
EXPH5	156	98	615028	Epidermolysis bullosa nonspecific autosomal recessive
EXT1	111	90	215300	Chondrosarcoma
EXT2	127	79	133701	Exostoses multiple type 2
EYA1	125	82	113650	Anterior segment anomalies with or without cataract
EYA4	126	93	605362	Cardiomyopathy dilated 1J
EYS	128	95	602772	Retinitis pigmentosa 25
EZH2	93	91	277590	Weaver syndrome
F10	94	94	227600	Factor X deficiency
F11	112	91	612416	Factor XI deficiency autosomal dominant
F12	107	88	610618	Angioedema hereditary type III
F13A1	110	85	613225	Factor XIIIA deficiency
F13B	100	86	613235	Factor XIIIB deficiency
F2	88	85	613679	Dysprothrombinemia
F5	169	59	227400	Factor V deficiency
F7	92	90	227500	Factor VII deficiency
F8	108	92	306700	Hemophilia A
F9	127	98	306900	Hemophilia B
FA2H	61	88	612319	Spastic paraplegia 35 autosomal recessive
FADD	98	94	613759	Infections recurrent with encephalopathy hepatic dysfunction and cardiovasuclar malformations
FAH	119	88	276700	Tyrosinemia type I
FAM111A	201	98	602361	Gracile bone dysplasia
FAM126A	122	96	610532	Leukodystrophy hypomyelinating 5
FAM134B	89	97	613115	Neuropathy hereditary sensory and autonomic type IIB
FAM161A	136	96	606068	Retinitis pigmentosa 28
FAM20A	74	76	614253	Amelogenesis imperfecta and gingival fibromatosis syndrome
FAM20C	79	90	259775	Raine syndrome

FAM58A	47	30	300707	STAR syndrome
FAM83H	66	99	130900	Amelogenesis imperfecta type 3
FAN1	118	56	614817	Interstitial nephritis karyomegalic
FANCA	98	89	227650	Fanconi anemia complementation group A
FANCB	112	96	200	-
FANCC	87	91	227645	Fanconi anemia complementation group C
FANCD2	119	84	227646	Fanconi anemia complementation group D2
FANCE	89	91	600901	Fanconi anemia complementation group E
FANCF	143	100	603467	Fanconi anemia complementation group F
FANCG	130	86	200	-
FANCI	127	92	609053	Fanconi anemia complementation group I
FANCL	94	92	200	-
FANCM	108	94	614087	Fanconi anemia complementation group M
FARS2	100	93	614946	Combined oxidative phosphorylation deficiency 14
FAS	185	88	601859	Autoimmune lymphoproliferative syndrome, type IA
FASLG	91	94	211980	{Lung cancer, susceptibility to}
FASLG	91	94	601859	Autoimmune lymphoproliferative syndrome, type IB
FASTKD2	128	87	220110	Mitochondrial complex IV deficiency
FBLN1	106	85	608180	Synpolydactyly 3/34 associated with metacarpal and metatarsal synostoses
FBLN5	95	81	614434	Cutis laxa autosomal dominant 2
FBN1	110	88	102370	Acromicric dysplasia
FBN2	108	89	121050	Contractural arachnodactyly congenital
FBP1	88	90	229700	Fructose-16-bidphosphatase deficiency
FBXO7	151	92	260300	Parkinson disease 15 autosomal recessive
FCGR3B	146	29	200	Neutropenia
FCN3	101	88	613860	Immunodeficiency due to ficolin 3 deficiency
FECH	109	84	177000	Protoporphyria erythropoietic autosomal recessive
FERMT3	89	94	612840	Leukocyte adhesion deficiency, type III
FGA	179	95	202400	Afibrinogenemia congenital
FGB	129	96	202400	Afibrinogenemia congenital
FGD1	77	91	305400	Aarskog-Scott syndrome
FGD4	132	96	609311	Charcot-Marie-Tooth disease type 4H
FGF10	119	100	180920	Aplasia of lacrimal and salivary glands

FGF14	132	85	609307	Spinocerebellar ataxia 27
FGF17	103	88	615270	Hypogonadotropic hypogonadism 20 with or without anosmia
FGF23	70	88	193100	Hypophosphatemic rickets autosomal dominant
FGF3	69	100	610706	Deafness congenital with inner ear agenesis microtia and microdontia
FGF8	98	86	612702	Hypogonadotropic hypogonadism 6 with or without anosmia
FGF9	142	100	612961	Multiple synostoses syndrome 3
FGFR1	130	83	147950	Hypogonadotropic hypogonadism 2 with or without anosmia
FGFR2	135	78	207410	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis
FGFR3	57	93	100800	Achondroplasia
FGG	120	92	200	Dysfibrinogenemia
FH	88	80	606812	Fumarase deficiency
FHL1	81	32	300696	Emery-Dreifuss muscular dystrophy 6 X-linked
FIG4	124	93	612577	Amyotrophic lateral sclerosis 11
FIGLA	96	81	612310	Premature ovarian failure 6
FKBP10	83	87	610968	Osteogenesis imperfecta type XI
FKBP14	120	100	614557	Ehlers-Danlos syndrome with progressive kyphoscoliosis myopathy and hearing loss
FKRP	67	98	613153	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 5
FKTN	112	91	611615	Cardiomyopathy dilated 1X
FLCN	107	91	135150	Birt-Hogg-Dube syndrome
FLG	141	2	146700	Ichthyosis vulgaris
FLNA	83	89	314400	Cardiac valvular dysplasia X-linked
FLNB	99	77	108720	Atelosteogenesis type I
FLNC	89	92	614065	Myopathy distal 4
FLRT3	222	100	615271	Hypogonadotropic hypogonadism 21 with anosmia
FLT3	117	88	200	Leukemia
FLT4	90	91	602089	Hemangioma capillary infantile somatic
FLVCR1	95	96	609033	Ataxia posterior column with retinitis pigmentosa
FLVCR2	159	88	225790	Proliferative vasculopathy and hydraencephaly-hydrocephaly syndrome
FMO3	127	84	602079	Trimethylaminuria
FMR1	83	94	300624	Fragile X syndrome
FN1	108	80	601894	Glomerulopathy with fibronectin deposits 2
FOLR1	99	81	613068	Neurodegeneration due to cerebral folate transport deficiency
FOXC1	34	46	602482	Axenfeld-Rieger syndrome type 3

FOXC2	78	48	153400	Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus
FOXE1	32	100	241850	Bamforth-Lazarus syndrome
FOXE3	9	46	107250	Anterior segment mesenchymal dysgenesis
FOXF1	96	95	265380	Alveolar capillary dysplasia with misalignment of pulmonary veins
FOXG1	97	100	613454	Rett syndrome congenital variant
FOXI1	91	95	600791	Enlarged vestibular aqueduct
FOXL2	58	100	110100	Blepharophimosis epicanthus inversus and ptosis type 1
FOXN1	116	92	601705	T-cell immunodeficiency congenital alopecia and nail dystrophy
FOXP1	112	82	613670	Mental retardation with language impairment and autistic features
FOXP2	122	30	602081	Speech-language disorder-1
FOXP3	69	86	304790	Immunodysregulation polyendocrinopathy and enteropathy X-linked
FOXRED1	110	83	256000	Leigh syndrome due to mitochondrial complex I deficiency
FRAS1	116	82	219000	Fraser syndrome
FREM1	121	87	608980	Bifid nose with or without anorectal and renal anomalies
FREM2	141	93	219000	Fraser syndrome
FRMD7	106	93	310700	Nystagmus 1 congenital X-linked
FSCN2	84	94	607921	Retinitis pigmentosa 30
FSHB	93	94	229070	Follicle-stimulating hormone deficiency isolated
FSHR	107	91	233300	Ovarian dysgenesis 1
FTCD	49	84	229100	Glutamate formiminotransferase deficiency
FTH1	106	23	200	Iron overload
FTL	130	48	600886	Hyperferritinemia-cataract syndrome
FTO	124	87	612938	Growth retardation developmental delay coarse facies and early death
FTSJ1	84	86	309549	Mental retardation X-linked 9
FUCA1	81	88	230000	Fucosidosis
FUS	95	78	608030	Amyotrophic lateral sclerosis 6 autosomal recessive with or without frontotemporal dementia
FUT6	119	26	613852	Fucosyltransferase 6 deficiency
FUZ	73	89	182940	Neural tube defects
FXN	106	74	229300	Friedreich ataxia with retained reflexes
FXYD2	44	93	154020	Hypomagnesemia-2 renal
FYCO1	103	91	610019	Cataract 18 autosomal recessive
FZD4	144	100	133780	Exudative vitreoretinopathy
FZD6	149	98	614157	Nail disorder nonsyndromic congenital 10 (claw-shaped nails)

G6PC	137	91	232200	Glycogen storage disease Ia
G6PC3	123	86	612541	Dursun syndrome
G6PD	76	94	134700	Favism
GAA	98	91	232300	Glycogen storage disease II
GABRB3	116	91	200	-
GABRG2	129	90	611277	Epilepsy generalized with febrile seizures plus type 3
GAD1	112	87	603513	Cerebral palsy spastic quadriplegic 1
GALC	100	96	245200	Krabbe disease
GALE	107	97	230350	Galactose epimerase deficiency
GALK1	84	90	230200	Galactokinase deficiency with cataracts
GALNS	63	92	253000	Mucopolysaccharidosis IVA
GALNT3	108	96	211900	Tumoral calcinosis hyperphosphatemic familial
GALT	128	90	230400	Galactosemia
GAMT	77	92	612736	Cerebral creatine deficiency syndrome 2
GAN	146	91	256850	Giant axonal neuropathy-1
GARS	118	87	601472	Charcot-Marie-Tooth disease type 2D
GATA1	98	91	300835	Anemia X-linked with/without neutropenia and/or platelet abnormalities
GATA2	98	92	614172	Dendritic cell monocyte B lymphocyte and natural killer lymphocyte deficiency
GATA3	133	94	146255	Hypoparathyroidism sensorineural deafness and renal dysplasia
GATA4	59	86	607941	Atrial septal defect 2
GATA6	63	90	614475	Atrial septal defect 9
GATAD1	90	86	614672	Cardiomyopathy dilated 2B
GATAD2B	132	88	615074	Mental retardation autosomal dominant 18
GATM	91	87	612718	Cerebral creatine deficiency syndrome 3
GBA	145	76	608013	Gaucher disease perinatal lethal
GBA2	142	96	614409	Spastic paraplegia 46 autosomal recessive
GBE1	109	96	232500	Glycogen storage disease IV
GCDH	89	86	231670	Glutaricaciduria type I
GCH1	85	98	128230	Dystonia DOPA-responsive with or without hyperphenylalaninemia
GCK	77	87	125851	Diabetes mellitus gestational
GCLC	124	93	230450	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency
GCNT2	190	99	110800	Adult i phenotype without cataract
GCSH	50	31	605899	Glycine encephalopathy

GDAP1	109	92	607831	Charcot-Marie-Tooth disease axonal type 2K
GDF1	27	90	217095	Double-outlet right ventricle
GDF3	130	94	613702	Klippel-Feil syndrome 3 autosomal dominant
GDF5	96	100	201250	Acromesomelic dysplasia Hunter-Thompson type
GDF6	119	100	118100	Klippel-Feil syndrome 1 autosomal dominant
GDI1	99	87	300849	Mental retardation X-linked 41
GDNF	191	93	209880	Central hypoventilation syndrome
GFAP	89	89	203450	Alexander disease
GFER	58	83	613076	Myopathy mitochondrial progressive with congenital cataract hearing loss and developmental delay
GFI1	67	86	607847	Neutropenia nonimmune chronic idiopathic of adults
GFM1	119	90	609060	Combined oxidative phosphorylation deficiency 1
GFPT1	109	96	610542	Myasthenia congenital with tubular aggregates 1
GGCX	107	87	610842	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency
GH1	143	48	262400	Growth hormone deficiency isolated type IA
GHR	144	98	200	-
GHRHR	103	89	612781	Growth hormone deficiency isolated type IB
GHSR	115	95	604271	Short stature
GIF	126	79	261000	Intrinsic factor deficiency
GIGYF2	117	91	607688	Parkinson disease 11
GIPC3	93	90	601869	Deafness autosomal recessive 15
GJA1	80	13	600309	Atrioventricular septal defect 3
GJA3	82	100	601885	Cataract 14 multiple types
GJA5	125	99	614049	Atrial fibrillation familial 11
GJA8	114	100	116200	Cataract 1 multiple types
GJB1	117	100	302800	Charcot-Marie-Tooth neuropathy X-linked dominant 1
GJB2	161	100	149200	Bart-Pumphrey syndrome
GJB3	135	100	612644	Deafness autosomal dominant 2B
GJB4	128	100	133200	Erythrokeratodermia variabilis with erythema gyratum repens
GJB6	177	100	612643	Deafness autosomal dominant 3B
GJC2	34	99	608804	Leukodystrophy hypomyelinating 2
GK	59	29	307030	Glycerol kinase deficiency
GLA	82	80	301500	Fabry disease
GLB1	88	84	230500	GM1-gangliosidosis type I

GLDC	74	67	605899	Glycine encephalopathy
GLE1	112	81	611890	Arthrogryposis lethal with anterior horn cell disease
GLI2	111	89	610829	Holoprosencephaly-9
GLI3	113	90	175700	Greig cephalopolysyndactyly syndrome
GLIS2	95	93	611498	Nephronophthisis 7
GLIS3	104	88	610199	Diabetes mellitus neonatal with congenital hypothyroidism
GLRA1	116	85	149400	Hyperekplexia hereditary 1 autosomal dominant or recessive
GLRB	123	89	614619	Hyperekplexia 2 autosomal recessive
GLRX5	37	76	205950	Anemia sideroblastic pyridoxine-refractory autosomal recessive
GLUD1	130	67	606762	Hyperinsulinism-hyperammonemia syndrome
GLUL	66	35	610015	Glutamine deficiency congenital
GLYCTK	93	91	220120	D-glyceric aciduria
GM2A	107	93	272750	GM2-gangliosidosis AB variant
GMPPB	106	100	615350	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 14
GMPS	128	87	601626	Leukemia acute myelogenous
GNAI2	69	88	192605	Ventricular tachycardia, idiopathic
GNAI3	104	87	602483	Auriculocondylar syndrome 1
GNAL	82	99	615073	Dystonia 25
GNAQ	87	60	163000	Capillary malformations congenital 1 somatic mosaic
GNAS	132	82	102200	Acromegaly
GNAT1	83	88	610444	Night blindness congenital stationary autosomal dominant 3
GNAT2	129	83	613856	Achromatopsia-4
GNB4	131	94	615185	Charcot-Marie-Tooth disease dominant intermediate F
GNE	112	84	600737	Inclusion body myopathy autosomal recessive
GNMT	77	95	606664	Glycine N-methyltransferase deficiency
GNPAT	139	83	222765	Chondrodysplasia punctata rhizomelic type 2
GNPTAB	138	93	252500	Mucolipidosis II alpha/beta
GNPTG	69	100	252605	Mucolipidosis III gamma
GNRH1	62	69	614841	Hypogonadotropic hypogonadism 12 with or without anosmia
GNRHR	162	97	228300	Fertile eunuch syndrome
GNS	86	85	252940	Mucopolysaccharidosis type IIID
GOLGA5	132	93	188550	Thyroid carcinoma papillary
GORAB	149	91	231070	Geroderma osteodysplasticum

GOSR2	115	82	614018	Epilepsy progressive myoclonic 6
GOT1	110	85	614419	Aspartate aminotransferase serum level of QTL1
GP1BA	144	47	231200	Bernard-Soulier syndrome type A1 (recessive)
GP1BB	9	88	231200	Bernard-Soulier syndrome type B
GP6	85	55	614201	Bleeding disorder platelet-type 11
GP9	44	89	231200	Bernard-Soulier syndrome type C
GPC3	85	94	312870	Simpson-Golabi-Behmel syndrome type 1
GPC6	121	84	258315	Omodysplasia 1
GPD1	77	88	614480	Hypertriglyceridemia transient infantile
GPD1L	111	85	611777	Brugada syndrome 2
GPHN	121	95	252150	Molybdenum cofactor deficiency type C
GPI	103	92	613470	Hemolytic anemia nonspherocytic due to glucose phosphate isomerase deficiency
GPR143	50	88	300814	Nystagmus 6 congenital X-linked
GPR179	150	98	614565	Night blindness congenital stationary (complete) 1E autosomal recessive
GPR56	92	95	606854	Polymicrogyria bilateral frontoparietal
GPR98	120	93	604352	Febrile seizures familial 4
GPSM2	144	92	604213	Chudley-McCullough syndrome
GRHL2	111	90	608641	Deafness autosomal dominant 28
GRHPR	99	86	260000	Hyperoxaluria primary type II
GRIA3	97	80	300699	Mental retardation X-linked 94
GRIK2	125	60	611092	Mental retardation autosomal recessive 6
GRIN1	66	93	614254	Mental retardation autosomal dominant 8
GRIN2A	144	96	613971	Epilepsy with neurodevelopmental defects
GRIN2B	147	92	613970	Mental retardation autosomal dominant 6
GRK1	96	89	613411	Oguchi disease-2
GRM1	148	55	614831	Spinocerebellar ataxia autosomal recessive 13
GRM6	79	91	257270	Night blindness congenital stationary (complete) 1B autosomal recessive
GRN	104	92	607485	Aphasia primary progressive
GRXCR1	194	93	613285	Deafness autosomal recessive 25
GSN	78	87	105120	Amyloidosis Finnish type
GSS	104	77	266130	Glutathione synthetase deficiency
GTF2H5	101	37	601675	Trichothiodystrophy complementation group A
GUCA1A	80	81	602093	Cone dystrophy-3

GUCA1B	118	100	613827	Retinitis pigmentosa 48
GUCY2C	106	93	614616	Diarrhea 6
GUCY2D	77	90	200	Cone-rod dystrophy 6
GUSB	79	70	253220	Mucopolysaccharidosis VII
GYG1	72	39	613507	Glycogen storage disease XV
GYS1	74	86	611556	Glycogen storage disease 0 muscle
GYS2	93	96	240600	Glycogen storage disease type 0
H19	91	98	130650	Beckwith-Wiedemann syndrome
H6PD	128	98	604931	Cortisone reductase deficiency 1
HADH	91	94	200	-
HADHA	110	68	609016	Fatty liver acute of pregnancy
HADHB	90	76	609015	Trifunctional protein deficiency
HAMP	128	81	613313	Hemochromatosis type 2B
HARS	136	91	614504	Usher syndrome type 3B
HARS2	166	84	614926	Perrault syndrome 2
HAX1	157	100	610738	Neutropenia severe congenital 3 autosomal recessive
HBA1	136	100	200	Erythremias
HBA2	160	100	200	-
HBB	187	87	141749	Delta-beta thalassemia
HBD	223	80	200	Thalassemia due to Hb Lepore
HBG1	57	36	141749	Fetal hemoglobin quantitative trait locus 1
HBG2	64	36	613977	Cyanosis transient neonatal
HCCS	100	88	309801	Microphthalmia syndromic 7
HCFC1	56	90	309541	Mental retardation X-linked 3
HCN4	62	88	613123	Brugada syndrome 8
HCRT	48	81	161400	Narcolepsy 1
HDAC4	77	82	600430	Brachydactyly-mental retardation syndrome
HDAC8	91	90	300882	Cornelia de Lange syndrome 5
HEATR2	79	83	614874	Ciliary dyskinesia primary 18
HEPACAM	79	80	613925	Megalencephalic leukoencephalopathy with subcortical cysts 2A
HES7	36	98	613686	Spondylocostal dysostosis 4 autosomal recessive
HESX1	86	87	182230	Growth hormone deficiency with pituitary anomalies
HEXA	101	88	272800	GM2-gangliosidosis several forms

HEXB	106	97	268800	Sandhoff disease infantile juvenile and adult forms
HFE	127	85	200	Hemochromatosis
HGD	117	86	203500	Alkaptonuria
HGF	106	97	608265	Deafness autosomal recessive 39
HGSNAT	112	91	252930	Mucopolysaccharidosis type IIIC (Sanfilippo C)
HIBCH	75	77	250620	3-hydroxyisobutryl-CoA hydrolase deficiency
HINT1	63	60	137200	Neuromyotonia and axonal neuropathy autosomal recessive
HK1	117	87	235700	Hemolytic anemia due to hexokinase deficiency
HLCS	143	89	253270	Holocarboxylase synthetase deficiency
HMBS	122	81	176000	Porphyria acute intermittent
HMGCL	123	83	246450	HMG-CoA lyase deficiency
HMGCS2	129	85	605911	HMG-CoA synthase-2 deficiency
HMOX1	66	86	614034	Heme oxygenase-1 deficiency
HMX1	19	96	612109	Oculoauricular syndrome
HNF1A	82	91	612520	Diabetes mellitus insulin-dependent 20
HNF1B	85	81	125853	Diabetes mellitus noninsulin-dependent
HNF4A	92	77	125850	MODY type I
HOGA1	65	96	613616	Hyperoxaluria primary type III
HOXA1	118	94	601536	Athabaskan brainstem dysgenesis syndrome
HOXA11	112	94	605432	Radioulnar synostosis with amegakaryocytic thrombocytopenia
HOXA13	50	99	176305	Guttmacher syndrome
HOXA2	137	96	612290	Microtia hearing impairment and cleft palate
HOXB1	87	100	614744	Facial paresis hereditary congenital 3
HOXC13	80	92	614931	Ectodermal dysplasia 9 hair/nail type
HOXD10	148	99	192950	Charcot-Marie-Tooth disease foot deformity of
HOXD13	91	96	113200	Brachydactyly type D
HPD	95	97	140350	Hawkinsinuria
HPGD	81	88	259100	Cranioosteoarthropathy
HPRT1	75	80	300323	HPRT-related gout
HPS1	85	82	203300	Hermansky-Pudlak syndrome 1
HPS3	136	91	614072	Hermansky-Pudlak syndrome 3
HPS4	118	90	614073	Hermansky-Pudlak syndrome 4
HPS5	112	92	614074	Hermansky-Pudlak syndrome 5

HPS6	85	100	614075	Hermansky-Pudlak syndrome 6
HPSE2	89	89	236730	Urofacial syndrome 1
HR	77	92	203655	Alopecia universalis
HRAS	80	92	218040	Congenital myopathy with excess of muscle spindles
HRG	149	94	613116	Thrombophilia due to HRG deficiency
HSD11B1	105	87	614662	Cortisone reductase deficiency 2
HSD11B2	105	89	218030	Apparent mineralocorticoid excess
HSD17B10	87	89	300438	17-beta-hydroxysteroid dehydrogenase X deficiency
HSD17B3	102	92	264300	Pseudohermaphroditism male with gynecomastia
HSD17B4	103	89	261515	D-bifunctional protein deficiency
HSD3B2	98	21	201810	3-beta-hydroxysteroid dehydrogenase type II deficiency
HSD3B7	50	77	607765	Bile acid synthesis defect congenital 1
HSF4	93	93	116800	Cataract 5 multiple types
HSPB1	51	68	606595	Charcot-Marie-Tooth disease axonal type 2F
HSPB3	184	100	613376	Neuronopathy distal hereditary motor type IIC
HSPB8	97	88	608673	Charcot-Marie-Tooth disease axonal type 2L
HSPD1	43	20	612233	Leukodystrophy hypomyelinating 4
HSPG2	77	87	224410	Dyssegmental dysplasia Silverman-Handmaker type
HTR1A	106	100	614674	Periodic fever menstrual cycle dependent
HTRA1	75	89	600142	CARASIL syndrome
HTRA2	116	96	610297	Parkinson disease 13
HTT	114	84	143100	Huntington disease
HUWE1	84	85	300706	Mental retardation X-linked syndromic Turner type
HYAL1	85	96	601492	Mucopolysaccharidosis type IX
HYDIN	121	83	608647	Ciliary dyskinesia primary 5
HYLS1	164	100	236680	Hydrolethalus syndrome
ICK	104	94	612651	Endocrine-cerebroosteodysplasia
ICOS	133	100	607594	Immunodeficiency common variable 1
IDH2	109	94	613657	D-2-hydrosyglutaric aciduria 2
IDH3B	141	92	612572	Retinitis pigmentosa 46
IDS	92	89	309900	Mucopolysaccharidosis II
IDUA	83	88	607014	Mucopolysaccharidosis Ih
IER3IP1	92	49	614231	Microcephaly epilepsy and diabetes syndrome

IFITM5	88	95	610967	Osteogenesis imperfecta type V
IFNGR1	139	97	209950	BCG infection generalized familial
IFT122	108	81	218330	Cranioectodermal dysplasia 1
IFT140	97	86	266920	Mainzer-Saldino syndrome
IFT43	116	79	614099	Cranioectodermal dysplasia 3
IFT80	84	81	611263	Asphyxiating thoracic dystrophy 2
IGBP1	93	40	300472	Corpus callosum agenesis of with mental retardation ocular coloboma and micrognathia
IGF1	84	73	608747	Growth retardation with deafness and mental retardation due to IGF1 deficiency
IGF1R	112	89	270450	Insulin-like growth factor I resistance to
IGF2R	110	83	200	Hepatocellular carcinoma
IGFALS	61	96	200	Acid-labile subunit
IGFBP7	52	93	614224	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis
IGHMBP2	80	85	604320	Neuronopathy distal hereditary motor type VI
IGLL1	47	68	613500	Agammaglobulinemia 2
IGSF1	98	85	300888	Hypothyroidism central and testicular enlargement
IHH	96	95	607778	Acrocapitofemoral dysplasia
IKBKAP	119	90	223900	Dysautonomia familial
IKBKG	84	79	300291	Ectodermal dysplasia hypohidrotic with immune deficiency
IKZF1	116	92	200	Leukemia
IL10RA	105	89	613148	Inflammatory bowel disease 28 early onset autosomal recessive
IL10RB	114	88	612567	Inflammatory bowel disease 25, early onset, autosomal recessive
IL10RB	114	88	610424	{Hepatitis B virus, susceptibility to}
IL11RA	106	84	614188	Craniosynostosis and dental anomalies
IL17F	101	86	613956	Candidiasis familial 6 autosomal dominant
IL17RA	91	94	613953	Candidiasis familial 5 autosomal recessive
IL17RD	119	89	615267	Hypogonadotropic hypogonadism 18 with or without anosmia
IL1RAPL1	121	94	300143	Mental retardation X-linked 21/34
IL1RN	100	91	612852	Interleukin 1 receptor antagonist deficiency
IL21R	115	91	615207	Immunodeficiency primary autosomal recessive IL21R-related
IL2RA	107	89	606367	Interleukin-2 receptor alpha chain deficiency of
IL2RG	80	79	312863	Combined immunodeficiency X-linked moderate
IL31RA	137	89	613955	Amyloidosis primary localized cutaneous 2
IL36RN	83	79	614204	Psoriasis generalized pustular

IL7R	118	93	608971	Severe combined immunodeficiency T-cell negative B-cell/natural killer cell-positive type
ILDR1	62	92	609646	Deafness autosomal recessive 42
IMPAD1	97	99	614078	Chondrodysplasia with joint dislocations GRAPP type
IMPDH1	54	35	613837	Leber congenital amaurosis 11
IMPG2	136	93	613581	Maculopathy IMPG2-related
INF2	70	92	614455	Charcot-Marie-Tooth disease dominant intermediate E
ING1	119	98	275355	Squamous cell carcinoma head and neck somatic
INPP5E	67	95	213300	Joubert syndrome 1
INPPL1	90	90	258480	Opsismodysplasia
INS	54	85	125852	Diabetes mellitus insulin-dependent 2
INSL3	48	58	219050	Cryptorchidism
INSR	130	88	610549	Diabetes mellitus insulin-resistant with acanthosis nigricans
INVS	128	90	602088	Nephronophthisis 2 infantile
IQCB1	94	73	609254	Senior-Loken syndrome 5
IQSEC2	59	92	309530	Mental retardation X-linked 1
IRAK4	105	95	610799	Invasive pneumococcal disease recurrent isolated 1
IRF1	118	96	613659	Gastric cancer somatic
IRF4	120	91	254500	Multiple myeloma
IRF6	109	95	608864	Orofacial cleft 6
IRF8	80	88	614893	CD11C+/CD1C+ dendritic cell deficiency dominant
IRGM	180	100	612278	Inflammatory bowel disease 19
IRX5	53	70	611174	Hamamy syndrome
ISCU	87	75	255125	Myopathy with lactic acidosis hereditary
ISPD	100	89	614643	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 7
ITCH	116	95	613385	Autoimmune disease syndromic multisystem
ITGA2B	79	86	187800	Bleeding disorder platelet-type 16 autosomal dominant
ITGA3	106	54	614748	Interstitial lung disease nephrotic syndrome and epidermolysis bullosa congenital
ITGA6	112	57	226730	Epidermolysis bullosa junctional with pyloric stenosis
ITGA7	92	89	613204	Muscular dystrophy congenital due to ITGA7 deficiency
ITGB2	85	89	116920	Leukocyte adhesion deficiency
ITGB3	110	86	187800	Bleeding disorder platelet-type 16 autosomal dominant
ITGB4	86	89	131800	Epidermolysis bullosa of hands and feet
ITK	113	87	613011	Lymphoproliferative syndrome 1

ITM2B	97	92	176500	Dementia familial British
ITPR1	117	81	606658	Spinocerebellar ataxia 15
IVD	94	82	243500	Isovaleric acidemia
IYD	93	89	274800	Thyroid dyshormonogenesis 4
JAG1	125	89	118450	Alagille syndrome
JAK2	112	95	133100	Erythrocytosis somatic
JAK3	97	85	600802	SCID autosomal recessive T-negative/B-positive type
JAM3	76	89	613730	Hemorrhagic destruction of the brain subependymal calcification and cataracts
JPH2	58	95	613873	Cardiomyopathy familial hypertrophic 17
JPH3	107	96	606438	Huntington disease-like 2
JUP	78	86	611528	Arrhythmogenic right ventricular dysplasia 12
KAL1	87	81	308700	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1)
KANK1	152	97	612900	Cerebral palsy spastic quadriplegic 2
KANSL1	81	86	610443	Koolen-De Vries syndrome
KARS	132	86	613641	Charcot-Marie-Tooth disease recessive intermediate B
KAT6B	153	63	606170	Genitopatellar syndrome
KBTBD13	44	99	609273	Nemaline myopathy 6 autosomal dominant
KCNA1	124	48	160120	Episodic ataxia/myokymia syndrome
KCNA5	106	31	612240	Atrial fibrillation familial 7
KCNC3	56	48	605259	Spinocerebellar ataxia 13
KCNE1	177	100	612347	Jervell and Lange-Nielsen syndrome 2
KCNE2	151	100	611493	Atrial fibrillation familial 4
KCNE3	112	100	613119	Brugada syndrome 6
KCNH2	65	86	613688	Long QT syndrome-2
KCNJ1	175	100	241200	Bartter syndrome type 2
KCNJ10	158	94	600791	Enlarged vestibular aqueduct digenic
KCNJ11	107	100	606176	Diabetes mellitus permanent neonatal with neurologic features
KCNJ2	136	98	170390	Andersen syndrome
KCNJ5	176	97	613677	Hyperaldosteronism familial type III
KCNK3	76	96	615344	Pulmonary hypertension primary 4
KCNK9	119	97	612292	Birk-Barel mental retardation dysmorphism syndrome
KCNMA1	95	88	609446	Generalized epilepsy and paroxysmal dyskinesia
KCNQ1	64	92	607554	Atrial fibrillation familial 3

KCNQ2	73	96	613720	Epileptic encephalopathy early infantile 7
KCNQ3	99	94	121201	Seizures benign neonatal type 2
KCNQ4	93	91	600101	Deafness autosomal dominant 2A
KCNT1	71	94	615005	Epilepsy nocturnal frontal lobe 5
KCNV2	75	95	610356	Retinal cone dystrophy 3B
KCTD1	180	81	181270	Scalp-ear-nipple syndrome
KCTD7	109	87	611726	Epilepsy progressive myoclonic 3 with or without intracellular inclusions
KDM5C	98	86	300534	Mental retardation X-linked syndromic Claes-Jensen type
KDM6A	95	84	300867	Kabuki syndrome 2
KDR	116	86	602089	Hemangioma capillary infantile somatic
KERA	153	100	217300	Cornea plana congenita recessive
KHDC3L	103	96	614293	Hydatidiform mole recurrent 2
KIAA0196	108	89	603563	Spastic paraplegia 8 autosomal dominant
KIAA1279	120	97	609460	Goldberg-Shprintzen megacolon syndrome
KIF11	104	93	152950	Microcephaly with or without chorioretinopathy lymphedema or mental retardation
KIF1A	68	91	614255	Mental retardation autosomal dominant 9
KIF1B	126	87	118210	Charcot-Marie-Tooth disease type 2A1
KIF21A	111	92	135700	Fibrosis of extraocular muscles congenital 1
KIF22	113	90	603546	Spondyloepimetaphyseal dysplasia with joint laxity type 2
KIF5A	113	82	604187	Spastic paraplegia 10 autosomal dominant
KIF7	67	89	200990	Acrocallosal syndrome
KIRREL3	78	84	612581	Mental retardation autosomal dominant 4
KISS1	43	86	614842	Hypogonadotropic hypogonadism 13 with or without anosmia
KISS1R	48	90	614837	Hypogonadotropic hypogonadism 8 with or without anosmia
KIT	119	90	606764	Gastrointestinal stromal tumor familial
KITLG	59	19	145250	Hyperpigmentation familial progressive 2
KL	145	94	211900	Tumoral calcinosis hyperphosphatemic
KLF1	47	90	613673	Anemia dyserythropoietic congenital type IV
KLF11	175	96	610508	Maturity-onset diabetes of the young type VII
KLF6	133	93	613659	Gastric cancer somatic
KLHDC8B	54	87	236000	Hodgkin lymphoma
KLHL10	144	97	615081	Spermatogenic failure 11
KLHL3	103	86	614495	Pseudohypoaldosteronism type IID

KLHL40	92	99	615348	Nemaline myopathy 8 autosomal recessive
KLHL7	123	94	612943	Retinitis pigmentosa 42
KLK4	147	97	204700	Amelogenesis imperfecta type IIA1
KLKB1	153	83	612423	Fletcher factor deficiency
KLLN	101	93	615107	Cowden syndrome 4
KMT2A	158	97	200	-
KMT2D	111	96	147920	Kabuki syndrome 1
KRAS	68	74	109800	Bladder cancer somatic
KRT1	107	93	113800	Epidermolytic hyperkeratosis
KRT10	111	92	113800	Epidermolytic hyperkeratosis
KRT12	116	91	122100	Meesmann corneal dystrophy
KRT13	101	82	193900	White sponge nevus
KRT14	61	47	125595	Dermatopathia pigmentosa reticularis
KRT16	35	20	167200	Pachyonychia congenita Jadassohn-Lewandowsky type
KRT17	44	37	167210	Pachyonychia congenita Jackson-Lawler type
KRT18	62	18	200	Cirrhosis
KRT2	142	88	146800	Ichthyosis bullosa of Siemens
KRT3	82	86	122100	Meesmann corneal dystrophy
KRT4	87	85	193900	White sponge nevus
KRT5	107	67	179850	Dowling-Degos disease 1
KRT6A	61	8	167200	Pachyonychia congenita Jadassohn-Lewandowsky type
KRT6B	74	5	167210	Pachyonychia congenita Jackson-Lawler type
KRT74	103	69	613981	Hypotrichosis simplex of the scalp 2
KRT8	53	24	200	Cirrhosis
KRT81	47	39	158000	Monilethrix
KRT83	68	33	158000	Monilethrix
KRT85	51	52	602032	Ectodermal dysplasia 4 hair/nail type
KRT86	64	28	158000	Monilethrix
KRT9	134	94	144200	Epidermolytic palmoplantar keratoderma
L1CAM	95	93	304100	Corpus callosum partial agenesis of
L2HGDH	81	97	236792	L-2-hydroxyglutaric aciduria
LAMA2	112	86	607855	Muscular dystrophy congenital merosin-deficient
LAMA3	106	87	226650	Epidermolysis bullosa generalized atrophic benign

LAMA4	110	85	615235	Cardiomyopathy dilated 1JJ
LAMB1	131	82	615191	Lissencephaly 5
LAMB2	108	94	614199	Nephrotic syndrome type 5 with or without ocular abnormalities
LAMB3	81	77	226700	Epidermolysis bullosa junctional Herlitz type
LAMC2	117	82	226700	Epidermolysis bullosa junctional Herlitz type
LAMC3	99	85	614115	Cortical malformations occipital
LAMP2	90	87	300257	Danon disease
LAMTOR2	90	84	610798	Immunodeficiency due to defect in MAPBP-interacting protein
LARGE	116	82	613154	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 6
LARP7	107	80	615071	Alazami syndrome
LARS2	121	87	615300	Perrault syndrome 4
LBR	98	87	215140	HEM skeletal dysplasia
LCA5	157	98	604537	Leber congenital amaurosis 5
LCAT	98	91	136120	Fish-eye disease
LCT	137	92	223000	Lactase deficiency congenital
LDB3	88	87	601493	Cardiomyopathy dilated 1C
LDHA	78	22	612933	Glycogen storage disease XI
LDHB	102	66	614128	Lactate dehydrogenase-B deficiency
LDLR	122	89	143890	Hypercholesterolemia familial
LDLRAP1	88	87	603813	Hypercholesterolemia familial autosomal recessive
LEF1	108	55	200	Sebaceous tumors
LEMD3	106	98	166700	Buschke-Ollendorff syndrome
LEP	116	88	614962	Obesity morbid due to leptin deficiency
LEPR	133	92	614963	Obesity morbid due to leptin receptor deficiency
LEPRE1	110	91	610915	Osteogenesis imperfecta type VIII
LEPREL1	89	86	614292	Myopia high with cataract and vitreoretinal degeneration
LFNG	83	81	609813	Spondylocostal dysostosis autosomal recessive 3
LGI1	151	95	600512	Epilepsy familial temporal lobe 1
LHB	32	77	200	Hypogonadism
LHCGR	157	96	176410	Leydig cell adenoma somatic with precocious puberty
LHFPL5	168	96	610265	Deafness autosomal recessive 67
LHX3	55	91	221750	Pituitary hormone deficiency combined 3
LHX4	90	93	262700	Pituitary hormone deficiency combined 4

LIAS	116	94	614462	Pyruvate dehydrogenase lipoic acid synthetase deficiency
LIFR	113	90	601559	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome
LIG1	90	83	200	-
LIG4	186	100	606593	LIG4 syndrome
LIM2	52	91	615277	Cataract 19
LIPA	108	93	278000	Cholesteryl ester storage disease
LIPC	95	86	614025	Hepatic lipase deficiency
LIPH	130	86	604379	Hypotrichosis localized autosomal recessive 2
LIPN	117	92	613943	Ichthyosis congenital autosomal recessive 8
LITAF	78	28	601098	Charcot-Marie-Tooth disease type 1C
LMAN1	116	98	227300	Combined factor V and VIII deficiency
LMBR1	96	96	200500	Acheiropody
LMBRD1	103	98	277380	Methylmalonic aciduria and homocystinuria cblF type
LMF1	85	56	246650	Lipase deficiency combined
LMNA	80	84	115200	Cardiomyopathy dilated 1A
LMNB1	84	92	169500	Leukodystrophy adult-onset autosomal dominant
LMX1B	92	88	161200	Nail-patella syndrome
LOR	35	100	604117	Vohwinkel syndrome with ichthyosis
LOXHD1	116	88	613079	Deafness autosomal recessive 77
LPIN1	112	87	268200	Myoglobinuria acute recurrent autosomal recessive
LPIN2	89	85	609628	Majeed syndrome
LPL	123	80	144250	Combined hyperlipidemia familial
LPP	141	84	601626	Leukemia acute myeloid
LRAT	182	100	613341	Leber congenital amaurosis 14
LRBA	110	95	614700	Immunodeficiency common variable 8 with autoimmunity
LRIG2	129	90	615112	Urofacial syndrome 2
LRIT3	161	97	615058	Night blindness congenital stationary (complete) 1F autosomal recessive
LRP2	124	87	222448	Donnai-Barrow syndrome
LRP4	105	86	212780	Cenani-Lenz syndactyly syndrome
LRP5	88	84	601813	Exudative vitreoretinopathy 4
LRPPRC	98	90	220111	Leigh syndrome French-Canadian type
LRRC6	125	95	614935	Ciliary dyskinesia primary 19
LRRC8A	127	98	613506	Agammaglobulinemia 5

LRRK2	117	94	607060	Parkinson disease 8
LRSAM1	88	86	614436	Charcot-Marie-Toothe disease axonal type 2P
LRTOMT	84	90	611451	Deafness autosomal recessive 63
LTBP2	78	83	613086	Glaucoma 3 primary congenital D
LTBP3	64	92	613097	Tooth agenesis selective 6
LTBP4	100	81	613177	Cutis laxa autosomal recessive type IC
LYST	124	94	214500	Chediak-Higashi syndrome
LYZ	118	93	105200	Amyloidosis renal
LZTS1	104	99	133239	Esophageal squamous cell carcinoma
MAD1L1	87	77	200	Lymphoma
MAF	61	100	610202	Cataract pulverulent or cerulean with or without microcornea
MAFB	74	100	166300	Multicentric carpotarsal osteolysis syndrome
MAGT1	86	94	300853	Immunodeficiency X-linked with magnesium defect Epstein-Barr virus infection and neoplasia
MAK	90	84	614181	REtinitis pigmentosa 62
MAL	88	83	200	-
MALT1	102	95	200	MALT lymphoma
MAML2	96	48	200	Mucoepidermoid salivary gland carcinoma
MAMLD1	102	99	300758	Hypospadias 2 X-linked
MAN1B1	97	85	614202	Mental retardation autosomal recessive 15
MAN2B1	89	85	248500	Mannosidosis alpha- types I and II
MANBA	98	92	248510	Mannosidosis beta
MAOA	81	90	300615	Brunner syndrome
MAP2K1	104	73	615279	Cardiofaciocutaneous syndrome 3
MAP2K2	99	73	615280	Cardiofaciocutaneous syndrome 4
MAP3K1	127	94	613762	46XY sex reversal 6
MAP3K8	138	88	211980	Lung cancer somatic
MAPK10	112	87	606369	Epileptic encephalopathy Lennox-Gastaut type
MAPT	58	91	600274	Dementia frontotemporal with or without parkinsonism
MARVELD2	157	91	610153	Deafness autosomal recessive 49
MASP1	93	86	257920	3MC syndrome 1
MASP2	114	92	613791	MASP2 deficiency
MASTL	128	99	188000	Thrombocytopenia-2
MAT1A	105	87	250850	Hypermethioninemia persistent autosomal dominant due to methionine adenosyltransferase I/III deficiency

MATN3	97	96	607078	Epiphyseal dysplasia multiple 5
MATR3	148	51	606070	Myopathy distal 2
MBD5	161	97	156200	Mental retardation autosomal dominant 1
MBTPS2	101	93	308205	IFAP syndrome with or without BRESHECK syndrome
MC2R	126	98	202200	Glucocorticoid deficiency due to ACTH unresponsiveness
MC4R	183	100	601665	Obesity autosomal dominant
MCC	110	79	200	Colorectal cancer
MCCC1	117	91	210200	3-Methylcrotonyl-CoA carboxylase 1 deficiency
MCCC2	118	91	210210	3-Methylcrotonyl-CoA carboxylase 2 deficiency
MCEE	84	97	251120	Methylmalonyl-CoA epimerase deficiency
MCFD2	105	59	613625	Factor V and factor VIII combined deficiency of
MCM4	121	80	609981	Natural killer cell and glucocorticoid deficiency with DNA repair defect
MCM6	111	89	223100	Lactase persistance/nonpersistance
MCOLN1	97	90	252650	Mucolipidosis IV
MCPH1	121	97	251200	Microcephaly 1 primary autosomal recessive
MECP2	107	100	105830	Angelman syndrome
MED12	115	88	309520	Lujan-Fryns syndrome
MED13L	129	86	608808	Transposition of the great arteries dextro-looped 1
MED17	142	89	613668	Microcephaly postnatal progressive with seizures and brain atrophy
MED23	118	94	614249	Mental retardation autosomal recessive 18
MED25	121	86	605589	Charcot-Marie-Tooth disease type 2B2
MEF2C	119	86	613443	Mental retardation stereotypic movements epilepsy and/or cerebral malformations
MEFV	108	96	134610	Familial Mediterranean fever AD
MEGF10	114	81	614399	Myopathy areflexia respiratory distress and dysphagia early-onset
MEGF8	89	89	614976	Carpenter syndrome 2
MEN1	105	85	200	Adrenal adenoma
MEOX1	57	81	214300	Klippel-Feil syndrome 2
MERTK	120	90	613862	Retinitis pigmentosa 38
MESP2	60	92	608681	Spondylocostal dysostosis autosomal recessive 2
MET	132	96	114550	Hepatocellular carcinoma childhood type
MFN2	112	88	609260	Charcot-Marie-Tooth disease type 2A2
MFSD8	115	93	610951	Ceroid lipofuscinosis neuronal 7
MGAT2	207	100	212066	Congenital disorder of glycosylation type IIa

MGME1	169	97	615084	Mitochondrial DNA depletion syndrome 11
MGP	102	83	245150	Keutel syndrome
MIB1	106	94	615092	Left ventricular noncompaction 7
MID1	115	91	300000	Opitz GBBB syndrome type I
MINPP1	111	98	188470	Thyroid carcinoma follicular
MIP	76	92	615274	Cataract 15 multiple types
MIR17HG	?	?	614326	Feingold syndrome 2
MIR184	44	91	614303	EDICT syndrome
MIR96	70	100	613074	Deafness autosomal dominant 50
MITF	135	87	103500	Tietz albinism-deafness syndrome
MKKS	147	97	209900	Bardet-Biedl syndrome 6
MKL1	65	92	200	Megakaryoblastic leukemia
MKRN3	107	100	615346	Precocious puberty central 2
MKS1	126	86	209900	Bardet-Biedl syndrome 13
MLC1	94	92	604004	Megalencephalic leukoencephalopathy with subcortical cysts
MLH1	111	88	609310	Colorectal cancer hereditary nonpolyposis type 2
MLH3	163	94	614385	Colon cancer hereditary nonpolyposis type 7
MLLT11	122	100	200	Leukemia
MLPH	95	78	609227	Griscelli syndrome type 3
MLYCD	90	86	248360	Malonyl-CoA decarboxylase deficiency
MMAA	172	98	251100	Methylmalonic aciduria vitamin B12-responsive
MMAB	90	92	251110	Methylmalonic aciduria vitamin B12-responsive due to defect in synthesis of adenosylcobalamin cblB complementation type
MMACHC	166	89	277400	Methylmalonic aciduria and homocystinuria cblC type
MMADHC	79	80	200	-
MMP1	123	93	606963	COPD rate of decline of lung function in
MMP13	156	93	602111	Metaphyseal anadysplasia 1
MMP2	99	86	259600	Torg-Winchester syndrome
MMP20	109	87	612529	Amelogenesis imperfecta type IIA2
MMP9	104	84	613073	Metaphyseal anadysplasia 2
MN1	72	50	607174	Meningioma
MNX1	39	100	176450	Currarino syndrome
MOCS1	125	19	252150	Molybdenum cofactor deficiency type A

MOCS2	85	70	252150	Molybdenum cofactor deficiency type B
MOG	15	89	614250	Narcolepsy 7
MOGS	129	96	200	-
MPC1	75	79	200	-
MPDU1	134	96	609180	Congenital disorder of glycosylation type If
MPDZ	114	90	615219	Hydrocephalus nonsyndromic autosomal recessive 2
MPI	110	82	602579	Congenital disorder of glycosylation type Ib
MPL	127	88	254450	Myelofibrosis with myeloid metaplasia somatic
MPLKIP	76	100	234050	Trichothiodystrophy nonphotosensitive 1
MPO	86	84	254600	Myeloperoxidase deficiency
MPV17	152	87	256810	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)
MPZ	100	88	607791	Charcot-Marie-Tooth disease dominant intermediate D
MR1	95	75	118800	Paroxysmal nonkinesigenic dyskinesia
MRAP	86	92	607398	Glucocorticoid deficiency 2
MRE11A	99	79	604391	Ataxia-telangiectasia-like disorder
MRPL3	97	69	614582	Combined oxidative phosphorylation deficiency 9
MRPS16	132	89	610498	Combined oxidative phosphorylation deficiency 2
MRPS22	104	77	611719	Combined oxidative phosphorylation deficiency 5
MS4A1	161	94	613495	Immunodeficiency common variable 5
MSH2	101	91	120435	Colorectal cancer hereditary nonpolyposis type 1
MSH3	112	93	200	-
MSH6	161	98	614350	Colorectal cancer hereditary nonpolyposis type 5
MSR1	130	94	614266	Barrett esophagus/esophageal adenocarcinoma
MSRB3	117	75	613718	Deafness autosomal recessive 74
MSTN	162	95	200	-
MSX1	47	99	189500	Ectodermal dysplasia 3 Witkop type
MSX2	59	70	604757	Craniosynostosis type 2
MTAP	97	80	112250	Diaphyseal medullary stenosis with malignant fibrous histiocytoma
MTFMT	103	92	614947	Combined oxidative phosphorylation deficiency 15
MTHFR	98	91	236250	Homocystinuria due to MTHFR deficiency
MTM1	89	89	310400	Myotubular myopathy X-linked
MTMR2	107	84	601382	Charcot-Marie-Tooth disease type 4B1
MTO1	129	89	614702	Combined oxidative phosphorylation deficiency 10

MTPAP	113	94	613672	Ataxia spastic 4
MTR	119	86	250940	Homocystinuria-megaloblastic anemia cblG complementation type
MTRR	113	93	236270	Homocystinuria-megaloblastic anemia cbl E type
MTTP	111	90	545000	Merff syndrome
MUC1	109	99	174000	Medullary cystic kidney disease 1
MUC1	109	99	174000	Medullary cystic kidney disease 1
MUC1	109	99	174000	Medullary cystic kidney disease 1
MUSK	132	87	608931	Myasthenic syndrome congenital associated with acetylcholine receptor deficiency
MUT	125	94	251000	Methylmalonic aciduria mut(0) type
MUTYH	114	97	608456	Adenomas multiple colorectal
MVK	101	78	260920	Hyper-IgD syndrome
MXI1	96	93	200	Neurofibrosarcoma
MYBPC1	106	87	614335	Arthrogryposis distal type 1B
MYBPC3	86	85	115197	Cardiomyopathy familial hypertrophic 4
MYC	174	94	113970	Burkitt lymphoma
MYCN	78	98	164280	Feingold syndrome
MYD88	148	90	153600	Macroglobulinemia Waldenstrom somatic
MYF6	135	100	614408	Myopathy centronuclear 3
MYH11	133	74	132900	Aortic aneurysm familial thoracic 4
MYH14	75	79	600652	Deafness autosomal dominant 4A
MYH2	135	52	605637	Inclusion body myopathy-3
MYH3	144	62	193700	Arthrogryposis distal type 2A
MYH6	122	60	614089	Atrial septal defect 3
MYH7	117	61	613426	Cardiomyopathy dilated 1S
MYH8	142	60	608837	Carney complex variant
MYH9	100	85	603622	Deafness autosomal dominant 17
MYL2	105	99	608758	Cardiomyopathy familial hypertrophic 10
MYL3	90	78	608751	Cardiomyopathy familial hypertrophic 8
MYLK	111	68	613780	Aortic aneurysm familial thoracic 7
MYLK2	87	96	192600	Cardiomyopathy hypertrophic midventricular digenic
MYO15A	95	92	600316	Deafness autosomal recessive 3
MYO1A	123	84	607841	Deafness autosomal dominant 48
MYO1E	108	86	614131	Glomerulosclerosis focal segmental 6

MYO3A	108	92	607101	Deafness autosomal recessive 30
MYO5A	101	86	214450	Griscelli syndrome type 1
MYO5B	101	78	251850	Microvillus inclusion disease
MYO6	105	94	606346	Deafness autosomal dominant 22
MYO7A	82	87	601317	Deafness autosomal dominant 11
MYOC	194	95	137750	Glaucoma 1A primary open angle
MYOT	136	94	159000	Muscular dystrophy limb-girdle type 1A
MYOZ2	100	95	613838	Cardiomyopathy familial hypertrophic 16
MYPN	134	88	615248	Cardiomyopathy dilated 1KK
NAA10	73	95	300855	N-terminal acetyltransferase deficiency
NAGA	93	84	609242	Kanzaki disease
NAGLU	62	92	252920	Mucopolysaccharidosis type IIIB (Sanfilippo B)
NAGS	53	88	237310	N-acetylglutamate synthase deficiency
NAT8L	23	92	614063	N-acetylaspartate deficiency
NBAS	109	94	614800	Short stature optic nerve atrophy and Pelger-Huet anomaly
NBEAL2	102	96	139090	Gray platelet syndrome
NBEAP1	341	72	200	Lymphoma
NBN	127	94	613065	Leukemia, acute lymphoblastic
NBN	127	94	251260	Nijmegen breakage syndrome
NBN	127	94	609135	Aplastic anemia
NCF2	119	89	233710	Chronic granulomatous disease due to deficiency of NCF-2
NCF4	98	84	613960	Granulomatous disease chronic autosomal recessive cytochrome b-positive type III
NCOA4	108	30	188550	Thyroid carcinoma papillary
NCSTN	105	75	142690	Acne inversa familial 1
NDE1	92	60	614019	Lissencephaly 4 (with microcephaly)
NDN	36	100	176270	Prader-Willi syndrome
NDP	112	86	305390	Exudative vitreoretinopathy X-linked
NDRG1	90	94	601455	Charcot-Marie-Tooth disease type 4D
NDUFA1	128	100	252010	Mitochondrial complex I deficiency
NDUFA10	101	90	256000	Leigh syndrome
NDUFA11	59	90	252010	Mitochondrial complex I deficiency
NDUFA12	104	92	256000	Leigh syndrome due to mitochondrial complex 1 deficiency
NDUFA2	105	100	256000	Leigh syndrome due to mitochondrial complex I deficiency

NDUFA9	107	80	256000	Leigh syndrome due to mitochondrial complex I deficiency
NDUFAF1	122	87	252010	Mitochondrial complex I deficiency
NDUFAF2	57	37	256000	Leigh syndrome
NDUFAF3	96	94	252010	Mitochondrial complex I deficiency
NDUFAF4	71	42	252010	Mitochondrial complex I deficiency
NDUFAF5	120	93	252010	Mitochondrial complex 1 deficiency
NDUFAF6	84	99	256000	Leigh syndrome due to mitochondrial complex I deficiency
NDUFB3	5	26	252010	Mitochondrial complex I deficiency
NDUFS1	87	94	252010	Mitochondrial complex I deficiency
NDUFS2	161	83	252010	Mitochondrial complex I deficiency
NDUFS3	180	89	256000	Leigh syndrome due to mitochondrial complex I deficiency
NDUFS4	120	94	256000	Leigh syndrome
NDUFS6	109	91	252010	Complex I mitochondrial respiratory chain deficiency of
NDUFS7	103	91	256000	Leigh syndrome
NDUFS8	93	86	256000	Leigh syndrome due to mitochondrial complex I deficiency
NDUFV1	69	85	252010	Mitochondrial complex I deficiency
NDUFV2	126	64	252010	Mitochondrial complex I deficiency
NEB	122	85	256030	Nemaline myopathy 2 autosomal recessive
NEFL	127	95	607734	Charcot-Marie-Tooth disease type 1F
NEK1	110	96	263520	Short rib-polydactyly syndrome type IIA
NEK8	118	89	613824	Nephronophthisis 9
NEU1	16	87	256550	Sialidosis type I
NEUROD1	124	96	606394	Maturity-onset diabetes of the young 6
NEUROG3	108	95	610370	Diarrhea 4 malabsorptive congenital
NEXN	130	92	613122	Cardiomyopathy dilated 1CC
NF1	109	85	607785	Leukemia juvenile myelomonocytic
NF2	87	94	607174	Meningioma NF2-related somatic
NFIX	115	24	602535	Marshall-Smith syndrome
NFKBIA	115	88	612132	Ectodermal dysplasia anhidrotic with T-cell immunodeficiency
NFU1	89	80	605711	Multiple mitochondrial dysfunctions syndrome 1
NHEJ1	90	80	611291	Severe combined immunodeficiency with microcephaly growth retardation and sensitivity to ionizing radiation
NHLRC1	111	100	254780	Epilepsy progressive myoclonic 2B (Lafora)
NHP2	74	37	613987	Dyskeratosis congenita, autosomal recessive 2

NHS	109	94	302200	Cataract 40 X-linked
NIN	146	90	614851	Seckel syndrome 7
NIPA1	140	93	600363	Spastic paraplegia 6 autosomal dominant
NIPAL4	129	91	612281	Ichthyosis congenital autosomal recessive 6
NIPBL	125	97	122470	Cornelia de Lange syndrome 1
NKX2-1	88	93	118700	Chorea hereditary benign
NKX2-5	82	100	271400	Asplenia isolated congenital
NKX2-6	84	92	217095	Persistent truncus arteriosus
NKX3-2	50	99	613330	Spondylo-megaepiphyseal-metaphyseal dysplasia
NLGN4X	94	24	200	-
NLRP12	110	92	611762	Familial cold autoinflammatory syndrome 2
NLRP3	128	96	607115	CINCA syndrome
NME1	157	79	256700	Neuroblastoma
NME8	103	90	610852	Ciliary dyskinesia primary 6
NMNAT1	144	52	608553	Leber congenital amaurosis 9
NNT	107	91	614736	Glucocorticoid deficiency 4
NOBOX	97	91	611548	Premature ovarian failure 5
NOD2	95	94	186580	Blau syndrome
NODAL	120	99	270100	Heterotaxy visceral 5
NOG	96	100	611377	Brachydactyly type B2
NOL3	132	100	614937	Myoclonus familial cortical
NOP10	141	100	224230	Dyskeratosis congenita, autosomal recessive 1
NOP56	128	87	614153	Spinocerebellar ataxia 36
NOTCH1	69	93	109730	Aortic valve disease
NOTCH2	113	86	610205	Alagille syndrome 2
NOTCH3	70	90	125310	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy
NPC1	110	84	257220	Niemann-Pick disease type C1
NPC2	76	86	607625	Niemann-pick disease type C2
NPHP1	117	92	609583	Joubert syndrome 4
NPHP3	112	95	267010	Meckel syndrome 7
NPHP4	94	82	606966	Nephronophthisis 4
NPHS1	89	87	256300	Nephrotic syndrome type 1
NPHS2	133	92		

NPM1	72	30	601626	Leukemia acute myeloid
NPPA	163	100	612201	Atrial fibrillation familial 6
NPR2	153	89	602875	Acromesomelic dysplasia Maroteaux type
NR0B2	66	89	601665	Obesity mild early-onset
NR2E3	85	91	268100	Enhanced S-cone syndrome
NR3C1	106	60	200	-
NR3C2	136	95	605115	Hypertension early-onset autosomal dominant with exacerbation in pregnancy
NR5A1	76	96	612965	46XY sex reversal 3
NRAS	128	94	614470	Autoimmune lymphoproliferative syndrome type IV
NRL	48	95	200	Retinal degeneration autosomal recessive
NRXN1	130	92	614325	Pitt-Hopkins-like syndrome 2
NSD1	138	92	130650	Beckwith-Wiedemann syndrome
NSDHL	77	91	308050	CHILD syndrome
NSMF	83	91	614838	Hypogonadotropic hypogonadism 9 with or without anosmia
NSUN2	130	87	611091	Mental retardation autosomal recessive 5
NT5C3A	85	66	200	-
NT5E	115	93	211800	Calcification of joints and arteries
NTF4	41	83	613100	Glaucoma 1 open angle 1O
NTRK1	77	87	256800	Insensitivity to pain congenital with anhidrosis
NTRK2	106	87	613886	Obesity hyperphagia and developmental delay
NUBPL	86	99	252010	Mitochondrial complex I deficiency
NUMA1	100	94	200	Leukemia acute promyelocytic
NUP214	138	90	601626	Leukemia acute myeloid
NUP62	84	97	271930	Striatonigral degeneration infantile
NYX	49	97	310500	Night blindness congenital stationary (complete) 1A X-linked
OAT	58	58	258870	Gyrate atrophy of choroid and retina with or without ornithinemia
OBSL1	82	91	612921	3-M syndrome 2
OCA2	110	86	203200	Albinism brown oculocutaneous
OCLN	144	89	251290	Band-like calcification with simplified gyration and polymicrogyria
OCRL	107	89	300555	Dent disease 2
OFD1	61	54	300804	Joubert syndrome 10
OGG1	95	91	144700	Renal cell carcinoma clear cell somatic
OPA1	121	99	165500	Optic atrophy 1

OPA3	85	87	258501	3-methylglutaconic aciduria type III
OPHN1	95	84	300486	Mental retardation X-linked with cerebellar hypoplasia and distinctive facial appearance
OPLAH	92	95	260005	5-oxoprolinase deficiency
OPN1LW	?	?	303700	Blue cone monochromacy
OPN1SW	108	81	190900	Colorblindness tritan
OPTN	119	83	613435	Amyotrophic lateral sclerosis 12
ORAI1	82	92	612782	Immune dysfunction with T-cell inactivation due to calcium entry defect 1
ORC1	120	84	224690	Meier-Gorlin syndrome 1
ORC4	98	93	613800	Meier-Gorlin syndrome 2
ORC6	94	89	613803	Meier-Gorlin syndrome 3
OSMR	148	90	105250	Amyloidosis primary localized cutaneous 1
OSTM1	111	96	259720	Osteopetrosis autosomal recessive 5
OTC	85	93	311250	Ornithine transcarbamylase deficiency
OTOA	119	83	607039	Deafness autosomal recessive 22
OTOF	95	80	601071	Auditory neuropathy autosomal recessive 1
OTOG	96	88	614945	Deafness autosomal recessive 18B
OTOGL	115	90	614944	Deafness autosomal recessive 84B
OTX2	153	97	610125	Microphthalmia syndromic 5
OXCT1	107	97	245050	Succinyl CoA:3-oxoacid CoA transferase deficiency
P2RX1	79	89	200	Bleeding disorder due to P2RX1 defect
P2RY12	184	100	609821	Bleeding disorder platelet-type 8
PABPN1	50	45	164300	Oculopharyngeal muscular dystrophy
PACS1	112	92	615009	Mental retardation autosomal dominant 17
PAFAH1B1	87	63	607432	Lissencephaly 1
PAH	93	91	261600	Phenylketonuria
PAK3	85	91	300558	Mental retardation X-linked 30/47
PALB2	142	95	610832	Fanconi anemia complementation group N
PANK2	152	93	607236	HARP syndrome
PAPSS2	91	86	612847	Brachyolmia 4 with mild epiphyseal and metaphyseal changes
PAX2	106	87	120330	Papillorenal syndrome
PAX3	109	89	122880	Craniofacial-deafness-hand syndrome
PAX4	84	81	612227	Diabetes mellitus ketosis-prone
PAX6	102	82	106210	Aniridia

PAX7	93	81	268220	Rhabdomyosarcoma 2 alveolar
PAX8	73	83	218700	Hypothyroidism congenital due to thyroid dysgenesis or hypoplasia
PAX9	185	100	604625	Tooth agenesis selective 3
PC	98	91	266150	Pyruvate carboxylase deficiency
PCBD1	80	85	200	-
PCCA	96	90	606054	Propionicacidemia
PCCB	116	84	606054	Propionicacidemia
PCDH15	119	94	609533	Deafness autosomal recessive 23
PCDH19	108	96	300088	Epileptic encephalopathy early infantile 9
PCM1	131	92	188550	Thyroid carcinoma papillary
PCNT	106	81	210720	Microcephalic osteodysplastic primordial dwarfism type II
PCSK1	115	86	600955	Obesity with impaired prohormone processing
PCSK9	82	80	603776	Hypercholesterolemia familial 3
PDCD10	80	75	603285	Cerebral cavernous malformations 3
PDE11A	104	91	610475	Pigmented nodular adrenocortical disease primary 2
PDE4D	124	89	614613	Acrodysostosis 2 with or without hormone resistance
PDE6A	109	90	613810	Retinitis pigmentosa 43
PDE6B	96	96	163500	Night blindness congenital stationary autosomal dominant 2
PDE6C	116	97	613093	Cone dystrophy 4
PDE6G	72	91	613582	Retinitis pigmentosa 57
PDE6H	49	84	610024	Achromatopsia 6
PDE8B	101	90	614190	Pigmented nodular adrenocortical disease primary 3
PDGFB	71	79	607807	Dermatofibrosarcoma protuberans
PDGFRA	135	88	606764	Gastrointestinal stromal tumor somatic
PDGFRB	83	86	615007	Basal ganglia calcification idiopathic 4
PDGFRL	131	89	114500	Colorectal cancer somatic
PDHA1	108	86	308930	Leigh syndrome X-linked
PDHB	105	95	614111	Pyruvate dehydrogenase E1-beta deficiency
PDP1	160	92	608782	Pyruvate dehydrogenase phosphatase deficiency
PDSS1	109	86	614651	Coenzyme Q10 deficiency primary 2
PDSS2	94	92	614652	Coenzyme Q10 deficiency primary 3
PDX1	37	100	245349	Lacticacidemia due to PDX1 deficiency
PDYN	153	92	610245	Spinocerebellar ataxia 23

PDZD7	85	86	605472	Usher syndrome type IIC GPR98/PDZD7 digenic
PEPD	78	89	170100	Prolidase deficiency
PER2	95	84	604348	Advanced sleep phase syndrome familial 1
PEX1	122	93	214100	Peroxisome biogenesis disorder 1A (Zellweger)
PEX10	76	90	614870	Peroxisome biogenesis disorder 6A (Zellweger)
PEX11B	184	88	614920	Peroxisome biogenesis disorder 14B
PEX12	131	90	614859	Peroxisome biogenesis disorder 3A (Zellweger)
PEX13	131	95	614883	Peroxisome biogenesis disorder 11A (Zellweger)
PEX14	98	85	614887	Peroxisome biogenesis disorder 13A (Zellweger)
PEX16	101	90	614876	Peroxisome biogenesis disorder 8A (Zellweger)
PEX19	112	86	614886	Peroxisome biogenesis disorder 12A (Zellweger)
PEX2	148	87	614867	Peroxisome biogenesis disorder 5B
PEX2	148	87	614866	Peroxisome biogenesis disorder 5A (Zellweger)
PEX26	118	91	614872	Peroxisome biogenesis disorder 7A (Zellweger)
PEX3	124	100	614882	Peroxisome biogenesis disorder 10A (Zellweger)
PEX5	98	74	214110	Peroxisome biogenesis disorder 2A (Zellweger)
PEX6	113	88	614862	Peroxisome biogenesis disorder 4A (Zellweger)
PEX7	112	91	614879	Peroxisome biogenesis disorder 9B
PFKM	130	80	232800	Glycogen storage disease VII
PFN1	74	50	614808	Amyotrophic lateral sclerosis 18
PGAM2	77	100	261670	Glycogen storage disease X
PGAP2	116	97	614207	Hyperphosphatasia with mental retardation syndrome 3
PGK1	82	43	300653	Phosphoglycerate kinase 1 deficiency
PGM1	109	76	614921	Congenital disorder of glycosylation type It
PHEX	104	94	307800	Hypophosphatemic rickets X-linked dominant
PHF6	87	94	301900	Borjeson-Forssman-Lehmann syndrome
PHF8	93	83	300263	Mental retardation syndrome X-linked Siderius type
PHGDH	103	86	601815	Phosphoglycerate dehydrogenase deficiency
PHKA1	80	90	300559	Muscle glycogenosis
PHKA2	84	87	306000	Glycogen storage disease type IXa1
PHKB	117	92	261750	Phosphorylase kinase deficiency of liver and muscle autosomal recessive
PHKG2	125	100	200	-
PHOX2A	33	83	602078	Fibrosis of extraocular muscles congenital 2

PHOX2B	60	96	200	-
PHYH	90	93	266500	Refsum disease
PICALM	92	95	601626	Leukemia acute myeloid
PIEZO1	78	94	194380	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema
PIGA	110	89	300868	Multiple congenital anomalies-hypotonia-seizures syndrome 2
PIGL	96	85	280000	CHIME syndrome
PIGM	128	100	610293	Glycosylphosphatidylinositol deficiency
PIGN	101	97	614080	Multiple congenital anomalies-hypotonia-seizures syndrome 1
PIGO	114	93	614749	Hyperphosphatasia with mental retardation syndrome 2
PIGV	191	97	239300	Hyperphosphatasia with mental retardation syndrome 1
PIK3CA	133	93	114480	Breast cancer somatic
PIK3R1	162	93	615214	Agammaglobulinemia 7 autosomal recessive
PIK3R2	84	89	603387	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome
PIK3R5	82	91	615217	Ataxia-oculomotor apraxia 3
PIKFYVE	144	96	121850	Corneal fleck dystrophy
PINK1	89	83	605909	Parkinson disease 6 early onset
PIP5K1C	61	90	611369	Lethal congenital contractural syndrome 3
PITPNM3	74	86	600977	Cone-rod dystrophy 5
PITX1	66	94	119800	Clubfoot congenital with or without deficiency of long bones and/or mirror-image polydactyly
PITX2	126	100	180500	Axenfeld-Rieger syndrome type 1
PITX3	42	88	107250	Anterior segment mesenchymal dysgenesis
PKD1	59	91	173900	Polycystic kidney disease adult type I
PKD2	98	93	613095	Polycystic kidney disease 2
PKHD1	114	90	200	-
PKLR	113	86	102900	Adenosine triphosphate elevated of erythrocytes
PKP1	89	90	604536	Ectodermal dysplasia/skin fragility syndrome
PKP2	99	54	609040	Arrhythmogenic right ventricular dysplasia 9
PLA2G4A	131	95	200	Phospholipase A2 group IV A
PLA2G5	123	74	228980	Fleck retina familial benign
PLA2G6	79	85	256600	Infantile neuroaxonal dystrophy 1
PLA2G7	114	92	614278	Platelet-activating factor acetylhydrolase deficiency
PLAG1	184	97	181030	Adenomas salivary gland pleomorphic
PLAU	102	83	601709	Quebec platelet disorder

PLCB1	117	93	613722	Epileptic encephalopathy early infantile 12
PLCB4	98	90	614669	Auriculocondylar syndrome 2
PLCD1	98	93	151600	Nail disorder nonsyndromic congenital 3 (leukonychia)
PLCE1	132	86	610725	Nephrotic syndrome type 3
PLCG2	125	83	614878	Autoinflammation antibody deficiency and immune dysregulation syndrome
PLEC	101	50	200	-
PLEKHG5	91	95	611067	Spinal muscular atrophy distal autosomal recessive 4
PLEKHM1	59	33	611497	Osteopetrosis autosomal recessive 6
PLG	116	75	217090	Conjunctivitis ligneous
PLIN1	56	87	613877	Lipodystrophy familial partial type 4
PLN	136	100	609909	Cardiomyopathy dilated 1P
PLOD1	84	88	225400	Ehlers-Danlos syndrome type VI
PLOD2	124	96	609220	Bruck syndrome 2
PLOD3	82	89	612394	Lysyl hydroxylase 3 deficiency
PLP1	79	79	312080	Pelizaeus-Merzbacher disease
PML	92	94	200	Leukemia acute promyelocytic
PMM2	118	89	212065	Congenital disorder of glycosylation type Ia
PMP22	124	81	118220	Charcot-Marie-Tooth disease type 1A
PMS2	134	36	614337	Colorectal cancer hereditary nonpolyposis type 4
PNKP	77	92	613402	Epileptic encephalopathy early infantile 10
PNP	138	97	613179	Immunodeficiency due to purine nucleoside phosphorylase deficiency
PNPLA1	140	92	615024	Ichthyosis congenital autosomal recessive 10
PNPLA2	69	91	610717	Neutral lipid storage disease with myopathy
PNPLA6	90	88	612020	Spastic paraplegia 39 autosomal recessive
PNPO	71	86	610090	Pyridoxamine 5-phosphate oxidase deficiency
PNPT1	96	84	614932	Combined oxidative phosphorylation deficiency 13
POC1A	112	89	614813	Short stature onychodysplasia facial dysmorphism and hypotrichosis
POFUT1	121	91	615327	Dowling-Degos disease 2
POLG	89	85	203700	Mitochondrial DNA depletion syndrome 4A (Alpers type)
POLG2	130	93	610131	Progressive external ophthalmoplegia with mitochondrial DNA deletions autosomal dominant 4
POLH	145	91	278750	Xeroderma pigmentosum variant type
POLR1C	139	89	248390	Treacher Collins syndrome 3
POLR1D	112	100	613717	Treacher Collins syndrome 2

POLR3A	107	83	607694	Leukodystrophy hypomyelinating 7 with or without oligodontia and/or hypogonadotropic hypogonadism
POLR3B	112	89	614381	Leukodystrophy hypomyelinating 8 with or without oligodontia and/or hypogonadotropic hypogonadism
POMC	61	89	609734	Obesity adrenal insufficiency and red hair due to POMC deficiency
POMGNT1	106	90	253280	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 3
POMGNT2	114	100	200	-
POMGNT2	114	100	200	-
POMP	154	93	601952	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma
POMT1	116	87	236670	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 1
POMT2	84	90	613150	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 2
POR	96	97	201750	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis
PORCN	88	84	305600	Focal dermal hypoplasia
POU1F1	126	88	613038	Pituitary hormone deficiency combined 1
POU3F4	123	100	304400	Deafness X-linked 2
POU4F3	153	100	602459	Deafness autosomal dominant 15
PPARG	122	88	609338	Carotid intimal medial thickness 1
PPIB	90	97	259440	Osteogenesis imperfecta type IX
PPM1D	151	97	114480	Breast cancer
PPM1K	98	97	615135	Maple syrup urine disease mild variant
PPOX	107	93	176200	Porphyria variegata
PPP1R3A	200	99	604367	Insulin resistance severe digenic
PPP2R1B	114	95	211980	Lung cancer
PPP2R2B	114	86	604326	Spinocerebellar ataxia 12
PPT1	77	91	200	Ceroid lipofuscinosis neuronal 1
PQBP1	105	80	309500	Renpenning syndrome
PRCC	106	93	605074	Renal cell carcinoma papillary
PRCD	115	44	610599	Retinitis pigmentosa 36
PRDM5	114	93	614170	Brittle cornea syndrome 2
PRF1	82	100	603553	Hemophagocytic lymphohistiocytosis familial 2
PRG4	114	25	208250	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome
PRICKLE1	124	89	612437	Epilepsy progressive myoclonic 1B
PRICKLE2	119	91	613832	Epilepsy progressive myoclonic 5
PRKAG2	92	90	600858	Cardiomyopathy familial hypertrophic 6
PRKAR1A	117	78	101800	Acrodysostosis 1 with or without hormone resistance

PRKCA	120	86	200	Pituitary tumor
PRKCG	117	86	605361	Spinocerebellar ataxia 14
PRKCSH	87	89	174050	Polycystic liver disease
PRKRA	103	97	612067	Dystonia 16
PRNP	104	100	123400	Creutzfeldt-Jakob disease
PROC	81	90	176860	Thrombophilia due to protein C deficiency autosomal dominant
PRODH	63	81	239500	Hyperprolinemia type I
PROK2	93	100	610628	Hypogonadotropic hypogonadism 4 with or without anosmia
PROKR2	183	95	244200	Hypogonadotropic hypogonadism 3 with or without anosmia
PROM1	90	91	612657	Cone-rod dystrophy 12
PROP1	82	86	262600	Pituitary hormone deficiency combined 2
PROS1	65	65	612336	Thrombophilia due to protein S deficiency autosomal dominant
PRPF3	116	78	601414	Retinitis pigmentosa 18
PRPF31	89	72	600138	Retinitis pigmentosa 11
PRPF6	96	82	613983	Retinitis pigmentosa 60
PRPF8	141	89	600059	Retinitis pigmentosa 13
PRPH2	152	93	613105	Choriodal dystrophy central areolar 2
PRPS1	95	64	301835	Arts syndrome
PRRT2	73	97	602066	Convulsions familial infantile with paroxysmal choreoathetosis
PRRX1	67	17	202650	Agnathia-otocephaly complex
PRSS1	179	65	167800	Pancreatitis hereditary
PRSS12	113	87	249500	Mental retardation autosomal recessive 1
PRSS56	74	79	613517	Microphthalmia isolated 6
PRX	87	77	614895	Charcot-Marie-Tooth disease type 4F
PSAP	105	84	611721	Combined SAP deficiency
PSAT1	83	40	610992	Phosphoserine aminotransferase deficiency
PSEN1	104	90	613737	Acne inversa familial 3
PSEN2	90	90	606889	Alzheimer disease-4
PSENEN	145	77	613736	Acne inversa familial 2
PSMB8	12	80	256040	Autoinflammation lipodystrophy and dermatosis syndrome
PSMC3IP	162	88	614324	Ovarian dysgenesis 3
PSTPIP1	58	94	604416	Pyogenic sterile arthritis pyoderma gangrenosum and acne
PTCH1	86	48	605462	Basal cell carcinoma somatic

PTCH2	77	92	605462	Basal cell carcinoma somatic
PTEN	130	65	153480	Bannayan-Riley-Ruvalcaba syndrome
PTF1A	33	100	609069	Diabetes mellitus permanent neonatal with cerebellar agenesis
PTGIS	74	76	145500	Hypertension essential
PTH	142	100	146200	Hypoparathyroidism autosomal dominant
PTH1R	86	95	200	-
PTHLH	184	61	613382	Brachydactyly type E2
PTPN11	90	25	151100	LEOPARD syndrome 1
PTPN12	117	98	200	Colon cancer
PTPN14	121	93	613611	Choanal atresia and lymphedema
PTPRC	110	89	608971	Severe combined immunodeficiency T cell-negative B-cell/natural killer-cell positive
PTPRJ	126	87	114500	Colon cancer somatic
PTPRO	109	90	614196	Nephrotic syndrome type 6
PTPRQ	118	94	613391	Deafness autosomal recessive 84A
PTRF	135	97	613327	Lipodystrophy congenital generalized type 4
PTS	116	93	261640	Hyperphenylalaninemia BH4-deficient A
PUS1	60	95	200	Mitochondrial myopathy and sideroblastic anemia 1
PVRL1	63	93	200	-
PVRL4	106	84	613573	Ectodermal dysplasia-syndactyly syndrome 1
PYCR1	78	86	612940	Cutis laxa autosomal recessive type IIB
PYGL	109	84	232700	Glycogen storage disease VI
PYGM	100	86	232600	McArdle disease
QDPR	89	82	261630	Hyperphenylalaninemia BH4-deficient C
RAB18	109	90	614222	Warburg micro syndrome 3
RAB23	123	95	201000	Carpenter syndrome
RAB27A	137	91	607624	Griscelli syndrome type 2
RAB33B	136	94	615222	Smith-McCort dysplasia 2
RAB39B	116	96	300271	Mental retardation X-linked 72
RAB3GAP1	121	95	600118	Warburg micro syndrome 1
RAB3GAP2	111	93	212720	Martsolf syndrome
RAB40AL	71	9	300519	Mental retardation X-linked syndromic Martin-Probst type
RAC2	53	85	608203	Neutrophil immunodeficiency syndrome
RAD21	96	79	614701	Cornelia de Lange syndrome 4

RAD50	109	91	613078	Nijmegen breakage syndrome-like disorder
RAD51C	93	100	613390	Fanconi anemia complementation group O
RAD54B	118	93	200	Colon adenocarcinoma
RAD54L	113	87	200	Adenocarcinoma colonic
RAF1	115	82	611554	LEOPARD syndrome 2
RAG1	159	100	609889	A/b T-cell lymphopenia with g/d T-cell expansion severe cytomegalovirus infection and autoimmunity
RAG2	201	100	233650	Combined cellular and humoral immune defects with granulomas
RAI1	134	100	182290	Smith-Magenis syndrome
RAP1GDS1	84	90	200	Lymphocytic leukemia
RAPSN	95	83	208150	Fetal akinesia deformation sequence
RARS2	96	96	611523	Pontocerebellar hypoplasia type 6
RASA1	99	97	605462	Basal cell carcinoma somatic
RAX	80	92	611038	Microphthalmia isolated 3
RAX2	46	98	610381	Cone-rod dystrophy 11
RB1	111	95	109800	Bladder cancer somatic
RB1CC1	122	93	114480	Breast cancer somatic
RBBP8	114	98	251255	Jawad syndrome
RBM10	75	91	311900	TARP syndrome
RBM20	113	92	613172	Cardiomyopathy dilated 1DD
RBM28	110	85	612079	Alopecia neurologic defects and endocrinopathy syndrome
RBM8A	106	64	274000	Thrombocytopenia-absent radius syndrome
RBP4	105	88	615147	Retinol dystrophy iris coloboma and comedogenic acne syndrome
RBPJ	86	51	614814	Adams-Oliver syndrome 3
RD3	43	96	610612	Leber congenital amaurosis 12
RDH12	83	78	612712	Leber congenital amaurosis 13
RDH5	115	83	136880	Fundus albipunctatus
RDX	62	57	611022	Deafness autosomal recessive 24
RECQL4	88	97	218600	Baller-Gerold syndrome
REEP1	105	87	614751	Neuronopathy distal hereditary motor type VB
RELN	119	87	257320	Lissencephaly 2 (Norman-Roberts type)
REN	97	88	613092	Hyperuricemic nephropathy familial juvenile 2
RET	97	85	209880	Central hypoventilation syndrome congenital
RFT1	91	84	612015	Congenital disorder of glycosylation type In

RFX5	128	93	209920	Bare lymphocyte syndrome type II complementation group C
RFX6	139	90	601346	Martinez-Frias syndrome
RFXANK	83	51	209920	MHC class II deficiency complementation group B
RFXAP	81	100	209920	Bare lymphocyte syndrome type II complementation group D
RGR	95	81	613769	Retinitis pigmentosa 44
RGS9	121	88	608415	Bradyopsia
RGS9BP	34	99	608415	Bradyopsia
RHAG	94	86	268150	Anemia hemolytic Rh-null regulator type
RHBDF2	69	85	148500	Tylosis with esophageal cancer
RHCE	143	79	200	Rh-null disease
RHO	114	88	610445	Night blindness congenital stationary autosomal dominant 1
RIMS1	103	93	603649	Cone-rod dystrophy 7
RIN2	110	90	613075	Macrocephaly alopecia cutis laxa and scoliosis
RIPK4	103	90	263650	Popliteal pterygium syndrome 2 lethal type
RIT1	122	93	615355	Noonan syndrome 8
RLBP1	104	75	607475	Bothnia retinal dystrophy
RMND1	95	100	614922	Combined oxidative phosphorylation deficiency 11
RNASEH2A	103	87	610333	Aicardi-Goutieres syndrome 4
RNASEH2B	94	95	610181	Aicardi-Goutieres syndrome 2
RNASEH2C	134	100	610329	Aicardi-Goutieres syndrome 3
RNASEL	163	96	601518	Prostate cancer 1
RNASET2	112	87	612951	Leukoencephalopathy cystic without megalencephaly
RNF135	61	90	614192	Macrocephaly macrosomia facial dysmorphism syndrome
RNF139	154	100	144700	Renal cell carcinoma
RNF168	192	95	611943	RIDDLE syndrome
RNF170	130	94	608984	Ataxia sensory 1 autosomal dominant
RNF212	114	99	612042	Recombination rate QTL 1
RNF216	109	80	212840	Cerebellar ataxia and hypogonadotropic hypogonadism
RNF6	178	51	133239	Esophageal carcinoma somatic
ROBO2	119	83	610878	Vesicoureteral reflux 2
ROBO3	95	82	607313	Gaze palsy horizontal with progressive scoliosis
ROGDI	83	96	226750	Kohlschutter-Tonz syndrome
ROM1	89	100	608133	Retinitis pigmentosa 7 digenic

ROR2	99	92	113000	Brachydactyly type B1
RP1	192	98	180100	Retinitis pigmentosa 1
RP1L1	151	25	613587	Occult macular dystrophy
RP2	93	99	312600	Retinitis pigmentosa 2
RP9	56	77	180104	Retinitis pigmentosa 9
RPE65	129	88	204100	Leber congenital amaurosis 2
RPGR	100	89	304020	Cone-rod dystrophy X-linked 1
RPGRIP1	133	85	608194	Cone-rod dystrophy 13
RPGRIP1L	103	95	216360	COACH syndrome
RPIA	83	85	608611	Ribose 5-phosphate isomerase deficiency
RPL11	104	61	612562	Diamond-Blackfan anemia 7
RPL26	50	22	614900	Diamond-Blackfan anemia 11
RPL35A	78	21	612528	Diamond-Blackfan anemia 5
RPL5	94	30	612561	Diamond-Blackfan anemia 6
RPS10	89	29	613308	Diamond-Blackfan anemia 9
RPS14	81	33	153550	Macrocytic anemia refractory due to 5q deletion somatic
RPS19	44	44	105650	Diamond-Blackfan anemia 1
RPS24	116	58	610629	Diamond-blackfan anemia 3
RPS26	53	26	613309	Diamond-Blackfan anemia 10
RPS6KA3	76	98	303600	Coffin-Lowry syndrome
RPS7	49	22	612563	Diamond-Blackfan anemia 8
RRAS2	115	74	200	Ovarian carcinoma
RRM2B	112	98	612075	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)
RS1	64	88	312700	Retinoschisis
RSPH4A	156	96	612649	Ciliary dyskinesia primary 11
RSPH9	86	92	612650	Ciliary dyskinesia primary 12
RSPO1	60	67	610644	Palmoplantar hyperkeratosis and true hermaphroditism
RSPO4	66	88	206800	Anonychia congenita
RTEL1	80	84	615190	Dyskeratosis congenita autosomal recessive 5
RTN2	74	89	604805	Spastic paraplegia 12 autosomal dominant
RTTN	94	93	614833	Polymicrogyria with seizures
RUNX1	73	83	601626	Leukemia acute myeloid
RUNX2	113	76	119600	Cleidocranial dysplasia

RYR1	86	89	117000	Central core disease
RYR2	121	92	600996	Arrhythmogenic right ventricular dysplasia 2
SACS	165	100	270550	Spastic ataxia Charlevoix-Saguenay type
SAG	120	89	258100	Oguchi disease-1
SALL1	135	52	107480	Townes-Brocks branchiootorenal-like syndrome
SALL4	104	37	607323	Duane-radial ray syndrome
SAMD9	204	100	610455	Tumoral calcinosis familial normophosphatemic
SAMHD1	127	89	612952	Aicardi-Goutieres syndrome 5
SAR1B	114	96	246700	Chylomicron retention disease
SARS2	75	92	613845	Hyperuricemia pulmonary hypertension renal failure and alkalosis
SART3	108	86	175900	Porokeratosis disseminated superficial actinic 1
SAT1	99	92	308800	Keratosis follicularis spinulosa decalvans
SATB2	124	86	119540	Cleft palate and mental retardation
SBDS	84	70	260400	Shwachman-Bodian-Diamond syndrome
SBF2	108	89	604563	Charcot-Marie-Tooth disease type 4B2
SC5D	145	90	200	-
SCARB2	85	87	254900	Epilepsy progressive myoclonic 4 with or without renal failure
SCARF2	38	94	600920	Van den Ende-Gupta syndrome
SCN1A	116	54	607208	Dravet syndrome
SCN1B	91	90	612838	Brugada syndrome 5
SCN2A	149	56	613721	Epileptic encephalopathy early infantile 11
SCN3B	96	81	613120	Brugada syndrome 7
SCN4A	127	92	170500	Hyperkalemic periodic paralysis type 2
SCN4B	78	88	611819	Long QT syndrome-10
SCN5A	106	74	614022	Atrial fibrillation familial 10
SCN8A	149	89	614306	Cognitive impairment with or without cerebellar ataxia
SCN9A	133	64	613863	Epilepsy generalized with febrile seizures plus type 7
SCNN1A	89	93	613021	Bronchiectasis with or without elevated sweat chloride 2
SCNN1B	90	91	211400	Bronchiectasis with or without elevated sweat chloride 1
SCNN1G	135	97	613071	Bronchiectasis with or without elevated sweat chloride 3
SCO1	106	81	200	-
SCO2	88	100	604377	Cardioencephalomyopathy fatal infantile due to cytochrome c oxidase deficiency 1
SCP2	102	90	613724	Leukoencephalopathy with dystonia and motor neuropathy

SDCCAG8	97	89	613615	Senior-Loken syndrome 7
SDHA	39	18	613642	Cardiomyopathy dilated 1GG
SDHAF1	57	86	200	Mitochondrial complex II deficiency
SDHAF2	134	83	200	Paragangliomas 2
SDHB	97	87	612359	Cowden syndrome 2
SDHC	83	88	606764	Gastrointestinal stromal tumor
SDHD	71	4	114900	Carcinoid tumors intestinal
SEC23A	114	99	607812	Craniolenticulosutural dysplasia
SEC23B	132	87	224100	Anemia dyserythropoietic congenital type II
SEC63	97	85	174050	Polycystic liver disease
SECISBP2	107	85	609698	Thyroid hormone metabolism abnormal
SEMA3E	111	95	214800	CHARGE syndrome
SEMA4A	100	90	610283	Cone-rod dystrophy 10
sep-09	108	95	162100	Amyotrophy hereditary neuralgic
sep-12	77	93	614822	Spermatogenic failure 10
SEPN1	80	90	602771	Muscular dystrophy rigid spine 1
SEPSECS	104	93	613811	Pontocerebellar hypoplasia type 2D
SERAC1	95	95	614739	3-methylglutaconic aciduria with deafness encephalopathy and Leigh-like syndrome
SERPINA1	134	93	613490	Emphysema due to AAT deficiency
SERPINA3	175	84	200	Cerebrovascular disease, occlusive
SERPINB6	130	98	613453	Deafness autosomal recessive 91
SERPINC1	137	90	613118	Thrombophilia due to antithrombin III deficiency
SERPINF1	118	83	613982	Osteogenesis imperfecta type VI
SERPING1	144	93	120790	Complement component 4, partial deficiency of
SERPING1	144	93	106100	Angioedema, hereditary, types I and II
SERPINH1	118	97	613848	Osteogenesis imperfecta type X
SERPINI1	83	89	604218	Encephalopathy familial with neuroserpin inclusion bodies
SETBP1	164	97	269150	Schinzel-Giedion midface retraction syndrome
SETX	153	96	602433	Amyotrophic lateral sclerosis 4 juvenile
SF3B1	111	95	614286	Myelodysplastic syndrome somatic
SF3B4	104	46	154400	Acrofacial dysostosis 1 Nager type
SFTPA2	127	31	178500	Pulmonary fibrosis idiopathic
SFTPB	62	87	265120	Surfactant metabolism dysfunction pulmonary 1

SFTPC	83	82	610913	Surfactant metabolism dysfunction pulmonary 2
SGCA	100	78	608099	Muscular dystrophy limb-girdle type 2D
SGCB	136	97	604286	Muscular dystrophy limb-girdle type 2E
SGCD	108	87	606685	Cardiomyopathy dilated 1L
SGCE	92	92	159900	Dystonia-11 myoclonic
SGCG	84	99	253700	Muscular dystrophy limb-girdle type 2C
SGSH	80	93	252900	Mucopolysaccharidisis type IIIA (Sanfilippo A)
SH2B3	90	93	133100	Erythrocytosis somatic
SH2D1A	74	87	308240	Lymphoproliferative syndrome X-linked
SH3BP2	94	96	118400	Cherubism
SH3PXD2B	114	96	249420	Frank-ter Haar syndrome
SH3TC2	109	91	601596	Charcot-Marie-Tooth disease type 4C
SHANK3	71	95	606232	Phelan-McDermid syndrome
SHH	81	100	142945	Holoprosencephaly-3
SHOC2	127	96	607721	Noonan-like syndrome with loose anagen hair
SHROOM4	102	79	300434	Stocco dos Santos X-linked mental retardation syndrome
SI	104	97	222900	Sucrase-isomaltase deficiency congenital
SIGMAR1	89	92	614373	Amyotrophic lateral sclerosis 16 juvenile
SIL1	111	82	248800	Marinesco-Sjogren syndrome
SIM1	121	90	601665	Obesity severe
SIX1	80	94	608389	Brachiootic syndrome 3
SIX3	101	96	157170	Holoprosencephaly-2
SIX5	52	93	610896	Branchiootorenal syndrome 2
SIX6	129	93	212550	Microphthalmia with cataract 2
SKI	66	89	182212	Shprintzen-Goldberg syndrome
SKIV2L	19	91	614602	Trichohepatoenteric syndrome 2
SLC10A2	140	93	613291	Bile acid malabsorption primary
SLC12A1	144	92	601678	Bartter syndrome type 1
SLC12A3	88	95	263800	Gitelman syndrome
SLC12A6	105	82	218000	Agenesis of the corpus callosum with peripheral neuropathy
SLC16A1	145	94	245340	Erythrocyte lactate transporter defect
SLC16A12	132	93	612018	Cataract juvenile with microcornea and glucosuria
SLC16A2	89	90	300523	Allan-Herndon-Dudley syndrome

SLC17A5	107	94	604369	Salla disease
SLC17A8	131	92	605583	Deafness autosomal dominant 25
SLC19A2	97	96	249270	Thiamine-responsive megaloblastic anemia syndrome
SLC19A3	123	91	607483	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)
SLC1A3	119	88	612656	Episodic ataxia type 6
SLC20A2	96	84	614540	Basal ganglia cancification idiopathic 3
SLC22A12	97	83	220150	Hypouricemia renal
SLC22A5	128	81	212140	Carnitine deficiency systemic primary
SLC24A1	148	80	613830	Night blindness congenital stationary (complete) 1D autosomal recessive
SLC25A1	66	89	615182	Combined D-2- and L-2-hydroxyglutaric aciduria
SLC25A12	120	95	612949	Hypomyelination global cerebral
SLC25A13	101	91	603471	Citrullinemia adult-onset type II
SLC25A15	146	88	238970	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome
SLC25A19	94	83	607196	Microcephaly Amish type
SLC25A20	95	83	212138	Carnitine-acylcarnitine translocase deficiency
SLC25A22	64	94	609304	Epileptic encephalopathy early infantile 3
SLC25A3	119	77	610773	Mitochondrial phosphate carrier deficiency
SLC25A38	93	65	205950	Anemia sideroblastic pyridoxine-refractory autosomal recessive
SLC25A4	102	94	192600	Cardiomyopathy familial hypertrophic
SLC26A2	148	99	600972	Achondrogenesis Ib
SLC26A3	131	87	214700	Chloride diarrhea congenital Finnish type
SLC26A4	108	89	600791	Deafness autosomal recessive 4 with enlarged vestibular aqueduct
SLC26A5	119	80	613865	Deafness autosomal recessive 61
SLC26A8	120	88	606766	Spermatogenic failure 3
SLC27A4	72	87	608649	Ichthyosis prematurity syndrome
SLC29A3	137	67	602782	Histiocytosis-lymphadenopathy plus syndrome
SLC2A1	90	94	601042	Dystonia 9
SLC2A10	90	94	208050	Arterial tortuosity syndrome
SLC2A2	139	96	227810	Fanconi-Bickel syndrome
SLC2A9	90	81	612076	Hypouricemia renal 2
SLC30A10	125	97	613280	Hypermanganesemia with dystonia polycythemia and cirrhosis
SLC30A2	89	84	608118	Zinc deficiency transient neonatal
SLC33A1	93	96	614482	Congenital cataracts hearing loss and neurodegeneration

SLC34A1	85	89	613388	Fanconi renotubular syndrome 2
SLC34A2	151	86	265100	Pulmonary alveolar microlithiasis
SLC34A3	63	92	241530	Hypophosphatemic rickets with hypercalciuria
SLC35A1	106	97	603585	Congenital disorder of glycosylation type IIf
SLC35A2	80	91	300896	Congenital disorder of glycosylation type IIm
SLC35C1	83	100	266265	Congenital disorder of glycosylation type IIc
SLC35D1	111	98	269250	Schneckenbecken dysplasia
SLC36A2	111	78	138500	Hyperglycinuria
SLC37A4	94	91	232220	Glycogen storage disease Ib
SLC39A13	102	89	612350	Spondylocheirodysplasia Ehlers-Danlos syndrome-like
SLC39A4	73	90	201100	Acrodermatitis enteropathica
SLC3A1	132	92	220100	Cystinuria
SLC40A1	132	96	606069	Hemochromatosis type 4
SLC45A2	128	83	606574	Oculocutaneous albinism type IV
SLC46A1	94	91	229050	Folate malabsorption hereditary
SLC4A1	84	93	200	-
SLC4A11	89	98	613268	Corneal dystrophy Fuchs endothelial 4
SLC4A4	74	41	604278	Renal tubular acidosis proximal with ocular abnormalities
SLC52A1	125	100	615026	Riboflavin deficiency
SLC52A2	105	92	614707	Brown-Vialetto-Van Laere syndrome 2
SLC52A3	69	97	211530	Brown-Vialetto-Van Laere syndrome 1
SLC5A1	109	89	606824	Glucose/galactose malabsorption
SLC5A2	69	90	233100	Renal glucosuria
SLC5A5	63	89	274400	Thyroid dyshormonogenesis 1
SLC5A7	112	95	158580	Neuronopathy distal hereditary motor type VIIA
SLC6A19	96	87	234500	Hartnup disorder
SLC6A2	103	90	604715	Orthostatic intolerance
SLC6A20	94	84	138500	Hyperglycinuria
SLC6A3	78	94	613135	Parkinsonism-dystonia infantile
SLC6A5	112	90	614618	Hyperekplexia 3
SLC6A8	15	27	300352	Cerebral creatine deficiency syndrome 1
SLC7A7	106	89	222700	Lysinuric protein intolerance
SLC7A9	83	92	220100	Cystinuria

SLC9A3R1	100	85	612287	Nephrolithiasis/osteoporosis hypophosphatemic 2
SLC9A6	95	91	300243	Mental retardation X-linked syndromic Christianson type
SLCO1B1	107	80	237450	Hyperbilirubinemia Rotor type digenic
SLCO1B3	97	77	237450	Hyperbilirubinemia Rotor type digenic
SLCO2A1	87	80	614441	Hypertrophic osteoarthropathy primary autosomal recessive 2
SLITRK1	139	100	137580	Tourette syndrome
SLURP1	34	78	248300	Meleda disease
SLX4	131	93	613951	Fanconi anemia complementation group P
SMAD3	85	88	613795	Loeys-Dietz syndrome type 3
SMAD4	123	93	200	-
SMAD6	72	97	614823	Aortic valve disease 2
SMARCA2	101	85	601358	Nicolaides-Baraitser syndrome
SMARCA4	88	86	614609	Mental retardation autosomal dominant 16
SMARCAD1	117	94	136000	Adermatoglyphia
SMARCAL1	137	89	242900	Schimke immunoosseous dysplasia
SMARCB1	152	84	614608	Mental retardation autosomal dominant 15
SMC1A	106	93	200	-
SMC3	117	83	200	-
SMCHD1	104	96	158901	Fascioscapulohumeral muscular dystrophy 2 digenic
SMOC1	98	82	206920	Microphthalmia with limb anomalies
SMOC2	89	82	125400	Dentin dysplasia type I with microdontia and misshapen teeth
SMPD1	95	94	257200	Niemann-Pick disease type A
SMPX	100	82	300066	Deafness X-linked 4
SMS	52	23	309583	Mental retardation X-linked Snyder-Robinson type
SNAI2	76	100	172800	Piebaldism
SNAP29	109	93	609528	Cerebral dysgenesis neuropathy ichthyosis and palmoplantar keratoderma syndrome
SNCA	144	96	127750	Dementia Lewy body
SNCB	67	97	127750	Dementia Lewy body
SNIP1	143	88	614501	Psychomotor retardation epilepsy and craniofacial dysmorphism
SNRNP200	129	85	610359	Retinitis pigmentosa 33
SNRPE	66	70	615059	Hypotrichosis 11
SNRPN	85	74	176270	Prader-Willi syndrome
SNX10	101	92	615085	Osteopetrosis autosomal recessive 8

SOBP	111	92	613671	Mental retardation anterior maxillary protrusion and strabismus
SOD1	119	87	105400	Amyotrophic lateral sclerosis 1
SOS1	112	94	135300	Fibromatosis gingival
SOST	110	90	122860	Craniodiaphyseal dysplasia autosomal dominant
SOX10	55	89	609136	PCWH syndrome
SOX17	70	97	613674	Vesicoureteral reflux 3
SOX18	12	90	607823	Hypotrichosis-lymphedema-telangiectasia syndrome
SOX2	96	47	206900	Microphthalmia syndromic 3
SOX3	42	48	300123	Mental retardation X-linked with isolated growth hormone deficiency
SOX9	116	84	114290	Acampomelic campomelic dysplasia
SP110	95	82	235550	Hepatic venoocclusive disease with immunodeficiency
SP7	75	100	613849	Osteogenesis imperfecta type XII
SPAST	87	99	182601	Spastic paraplegia 4 autosomal dominant
SPATA16	128	94	102530	Spermatogenic failure 6
SPATA7	137	98	604232	Leber congenital amaurosis 3
SPECC1L	151	94	600251	Facial clefting oblique 1
SPG11	111	90	604360	Spastic paraplegia 11 autosomal recessive
SPG20	127	95	275900	Troyer syndrome
SPINK1	89	100	167800	Pancreatitis hereditary
SPINK5	113	89	147050	Atopy
SPINT2	43	93	270420	Diarrhea 3 secretory sodium congenital syndromic
SPR	69	90	612716	Dystonia dopa-responsive due to sepiapterin reductase deficiency
SPRED1	135	95	611431	Legius syndrome
SPRY4	86	100	615266	Hypogonadotropic hypogonadism 17 with or without anosmia
SPTA1	117	84	130600	Elliptocytosis-2
SPTAN1	116	82	613477	Epileptic encephalopathy early infantile 5
SPTB	111	88	200	Anemia neonatal hemolytic
SPTBN2	99	89	600224	Spinocerebellar ataxia 5
SPTLC1	96	91	162400	Neuropathy hereditary sensory and autonomic type IA
SPTLC2	104	94	613640	Neuropathy hereditary sensory and autonomic type IC
SQSTM1	77	81	602080	Paget disease of bone
SRC	75	85	200	Colon cancer
SRCAP	143	92	136140	Floating-Harbor syndrome

SRD5A2	71	96	264600	Pseudovaginal perineoscrotal hypospadias
SRD5A3	107	84	612379	Congenital disorder of glycosylation type Iq
SRP72	105	74	614675	Bone marrow failure familial
SRPX2	87	77	300643	Rolandic epilepsy mental retardation and speech dyspraxia
SRY	34	100	400045	46XX sex reversal 1
SSTR5	105	97	102200	Somatostatin analog resistance to
ST14	77	89	610765	Ichthyosis with hypotrichosis
ST3GAL3	134	97	615006	Epileptic encephalopathy early infantile 15
ST3GAL3	134	97	615006	Epileptic encephalopathy early infantile 15
ST3GAL3	134	97	615006	Epileptic encephalopathy early infantile 15
ST3GAL5	107	86	200	-
STAR	120	81	201710	Lipoid adrenal hyperplasia
STAT1	103	87	614162	Candidiasis familial 7
STAT3	103	87	147060	Hyper-IgE recurrent infection syndrome
STAT5B	93	79	245590	Growth hormone insensitivity with immunodeficiency
STIL	146	98	612703	Microcephaly 7 primary autosomal recessive
STIM1	85	79	612783	Immune dysfunction with T-cell inactivation due to calcium entry defect 2
STK11	71	89	200	Melanoma malignant
STK4	111	83	614868	T-cell immunodeficiency recurrent infections autoimmunity and cardiac malformations
STOX1	153	92	609404	Preeclampsia/eclampsia 4
STRA6	79	86	601186	Microphthalmia isolated with coloboma 8
STRADA	95	90	611087	Polyhydramnios megalencephaly and symptomatic epilepsy
STRC	132	38	603720	Deafness autosomal recessive 16
STS	119	87	308100	Ichthyosis X-linked
STX11	133	100	603552	Hemophagocytic lymphohistiocytosis familial 4
STX16	104	91	603233	Pseudohypoparathyroidism type IB
STXBP1	109	86	612164	Epileptic encephalopathy early infantile 4
STXBP2	81	96	613101	Hemophagocytic lymphohistiocytosis familial 5
SUCLA2	93	57	612073	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with methylmalonic aciduria)
SUCLG1	120	86	245400	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)
SUFU	93	89	155255	Medulloblastoma desmoplastic
SUMF1	88	90	272200	Multiple sulfatase deficiency
SUMO1	40	38	613705	Orofacial cleft 10

SUOX	174	99	272300	Sulfite oxidase deficiency
SURF1	90	79	256000	Leigh syndrome due to COX deficiency
SYCP3	120	97	270960	Spermatogenic failure 4
SYN1	55	88	300491	Epilepsy X-linked with variable learning disabilities and behavior disorders
SYNE1	120	87	612998	Emery-Dreifuss muscular dystrophy 4 autosomal dominant
SYNE2	114	90	612999	Emery-Dreifuss muscular dystrophy 5 autosomal dominant
SYNGAP1	56	88	612621	Mental retardation autosomal dominant 5
SYP	68	98	300802	Mental retardation X-linked 96
SYT14	136	80	614229	Spinocerebellar ataxia autosomal recessive 11
TAB2	190	98	614980	Congenital heart defects nonsyndromic 2
TAC3	101	85	614839	Hypogonadotropic hypogonadism 10 with or without anosmia
TACR3	160	96	614840	Hypogonadotropic hypogonadism 11 with or without anosmia
TACSTD2	155	100	204870	Corneal dystrophy gelatinous drop-like
TAF1	128	71	314250	Dystonia-Parkinsonism X-linked
TAL1	50	90	200	Leukemia-1
TAL2	166	100	200	Leukemia-2
TALDO1	101	86	606003	Transaldolase deficiency
TAP1	16	86	604571	Bare lymphocyte syndrome type I
TAP2	13	90	604571	Bare lymphocyte syndrome type I due to TAP2 deficiency
TAPBP	21	88	604571	Bare lymphocyte syndrome type I
TARDBP	89	39	612069	Amyotrophic lateral sclerosis 10 with or without FTD
TAT	122	85	276600	Tyrosinemia type II
TAZ	68	97	302060	Barth syndrome
TBC1D24	99	99	615338	Epileptic encephalopathy early infantile 16
TBCE	131	91	241410	Hypoparathyroidism-retardation-dysmorphism syndrome
TBP	115	62	607136	Spinocerebellar ataxia 17
TBX1	80	86	217095	Conotruncal anomaly face syndrome
TBX15	99	91	260660	Cousin syndrome
TBX19	143	88	201400	Adrenocorticotropic hormone deficiency
TBX20	68	71	611363	Atrial septal defect 4
TBX21	106	91	208550	Asthma and nasal polyps
TBX22	125	94	303400	Cleft palate with ankyloglossia
TBX3	79	91	181450	Ulnar-mammary syndrome

TBX4	116	92	147891	Small patella syndrome
TBX5	93	84	142900	Holt-Oram syndrome
TBXAS1	123	86	231095	Ghosal hematodiaphyseal syndrome
TCAP	57	81	607487	Cardiomyopathy dilated 1N
TCF12	129	49	615314	Craniosynostosis 3
TCF4	117	81	610954	Pitt-Hopkins syndrome
TCIRG1	76	91	259700	Osteopetrosis autosomal recessive 1
TCN2	112	85	275350	Transcobalamin II deficiency
TCOF1	112	91	154500	Treacher Collins syndrome 1
TCTN1	112	90	200	-
TCTN2	106	84	613885	Meckel syndrome 8
TCTN3	124	92	614815	Joubert syndrome 18
TDGF1	97	39	200	-
TDP1	127	90	607250	Spinocerebellar ataxia autosomal recessive with axonal neuropathy
TDRD7	130	92	613887	Cataract 36
TEAD1	97	86	108985	Sveinsson choreoretinal atrophy
TECPR2	116	86	615031	Spastic paraplegia 49 autosomal recessive
TECR	77	77	614020	Mental retardation autosomal recessive 14
TECTA	125	90	601543	Deafness autosomal dominant 8/12
TEK	122	86	600195	Venous malformations multiple cutaneous and mucosal
TERC	64	100	127550	Dyskeratosis congenita autosomal dominant 1
TET2	150	97	614286	Myelodysplastic syndrome somatic
TF	119	84	209300	Atransferrinemia
TFAP2A	85	89	113620	Branchiooculofacial syndrome
TFAP2B	116	88	169100	Char syndrome
TFE3	62	88	300854	Renal cell carcinoma papillary 1
TFG	117	81	604484	Hereditary motor and sensory neuropathy proximal type
TFR2	75	93	604250	Hemochromatosis type 3
TG	117	83	274700	Thyroid dyshormonogenesis 3
TGFB1	63	85	131300	Camurati-Engelmann disease
TGFB2	133	86	614816	Loeys-Dietz syndrome type 4
TGFB3	102	84	107970	Arrhythmogenic right ventricular dysplasia 1
TGFBI	114	88	607541	Corneal dystrophy Avellino type

TGFBR1	123	91	609192	Loeys-Dietz syndrome type 1A
TGFBR2	101	87	614331	Colorectal cancer hereditary nonpolyposis type 6
TGIF1	205	97	200	-
TGM1	108	89	242300	Ichthyosis congenital autosomal recessive 1
TGM5	106	89	609796	Peeling skin syndrome acral type
TGM6	62	87	613908	Spinocerebellar ataxia 35
TH	86	91	605407	Segawa syndrome recessive
THAP1	112	94	602629	Dystonia 6 torsion
THBD	71	100	614486	Thrombophilia due to thrombomodulin defect
THPO	92	97	187950	Thrombocythemia 1
THRA	110	94	614450	Hypothyroidism congenital nongoitrous 6
THRB	129	82	188570	Thyroid hormone resistance
TIA1	105	94	604454	Welander distal myopathy
TIMM8A	56	60	200	Deafness X-linked 1
TIMP3	133	89	136900	Sorsby fundus dystrophy
TINF2	206	92	613990	Dyskeratosis congenita autosomal dominant 3
TJP2	97	84	607748	Hypercholanemia familial
TK2	110	85	609560	Mitochondrial DNA depletion syndrome 2 (myopathic type)
TLL1	125	91	613087	Atrial septal defect 6
TLR4	153	90	200	Endotoxin hyporesponsiveness
TMC1	117	91	606705	Deafness autosomal dominant 36
TMC6	68	90	226400	Epidermodysplasia verruciformis
TMC8	92	87	226400	Epidermodysplasia verruciformis
TMCO1	82	75	614132	Craniofacial dysmorphism skeletal anomalies and mental retardation syndrome
TMEM126A	79	87	612989	Optic atrophy-7
TMEM138	95	93	614465	Joubert syndrome 16
TMEM165	83	91	614727	Congenital disorder of glycosylation type IIk
TMEM216	77	88	608091	Joubert syndrome 2
TMEM231	87	66	614970	Joubert syndrome 20
TMEM237	99	91	614424	Joubert syndrome 14
TMEM38B	102	96	615066	Osteogenesis imperfecta type XIV
TMEM43	87	92	604400	Arrhythmogenic right ventricular dysplasia 5
TMEM5	151	88	615041	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 10

TMEM67	108	94	216360	COACH syndrome
TMEM70	174	44	614052	Mitochondrial complex V (ATP synthase) deficiency nuclear type 2
TMIE	65	91	600971	Deafness autosomal recessive 6
TMLHE	68	81	300872	Epsilon-trimethyllysine hydroxylase deficiency
TMPO	112	70	613740	Cardiomyopathy dilated 1T
TMPRSS3	88	47	601072	Deafness autosomal recessive 8/10
TMPRSS6	75	83	206200	Iron-refractory iron deficiency anemia
TNFRSF10B	116	91	275355	Squamous cell carcinoma head and neck
TNFRSF11A	99	94	174810	Osteolysis familial expansile
TNFRSF11B	173	95	239000	Paget disease juvenile
TNFRSF13B	70	86	240500	Immunodeficiency common variable 2
TNFRSF13C	67	80	613494	Immunodeficiency common variable 4
TNFRSF1A	72	89	142680	Periodic fever familial
TNFSF11	156	99	259710	Osteopetrosis autosomal recessive 2
TNNC1	110	100	611879	Cardiomyopathy dilated 1Z
TNNI2	55	100	601680	Arthrogryposis multiplex congenita distal type 2B
TNNI3	105	82	613286	Cardiomyopathy dilated 1FF
TNNT1	90	96	605355	Nemaline myopathy 5 Amish type
TNNT2	104	92	601494	Cardiomyopathy dilated 1D
TNNT3	86	83	601680	Arthyrgryposis distal type 2B
TNXB	15	64	130020	Ehlers-Danlos syndrome autosomal dominant hypermobility type
TOP1	117	85	200	DNA topoisomerase I
TOP2A	117	95	200	DNA topoisomerase II resistance to inhibition of
TOPORS	167	100	609923	Retinitis pigmentosa 31
TP53	81	95	202300	Adrenal cortical carcinoma
TP63	141	81	103285	ADULT syndrome
TPI1	63	39	200	-
TPK1	84	100	614458	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type)
TPM1	133	91	611878	Cardiomyopathy dilated 1Y
TPM2	102	86	108120	Arthrogryposis multiplex congenita distal type 1
TPM3	97	60	609284	CAP myopathy 1
TPMT	91	82	610460	6-mercaptopurine sensitivity
TPO	94	84	274500	Thyroid dyshormonogenesis 2A

TPP1	137	95	204500	Ceroid lipofuscinosis neuronal 2
TPRN	35	100	613307	Deafness autosomal recessive 79
TRAPPC11	124	91	615356	Muscular dystrophy limb-girdle type 2S
TRAPPC2	71	60	313400	Spondyloepiphyseal dysplasia tarda
TRAPPC9	86	85	613192	Mental retardation autosomal recessive 13
TREM2	104	90	221770	Nasu-Hakola disease
TREX1	114	70	225750	Aicardi-Goutieres syndrome 1 dominant and recessive
TRHR	142	92	200	Thyrotropin-releasing hormone resistance
TRIM24	97	95	188550	Thyroid carcinoma papillary
TRIM32	110	100	209900	Bardet-Biedl syndrome 11
TRIM33	97	95	188550	Thyroid carcinoma papillary
TRIM37	103	95	253250	Mulibrey nanism
TRIOBP	125	52	609823	Deafness autosomal recessive 28
TRIP11	123	90	200600	Achondrogenesis type IA
TRMU	82	89	613070	Liver failure transient infantile
TRPA1	81	84	615040	Episodic pain syndrome familial
TRPC6	97	72	603965	Glomerulosclerosis focal segmental 2
TRPM1	142	88	613216	Night blindness congenital stationary (complete) 1C autosomal recessive
TRPM4	96	87	604559	Progressive familial heart block type IB
TRPM6	127	93	602014	Hypomagnesemia 1 intestinal
TRPS1	143	98	190350	Trichorhinophalangeal syndrome type I
TRPV3	110	90	614594	Olmsted syndrome
TRPV4	88	93	113500	Brachyolmia type 3
TSC1	106	87	607341	Focal cortical dysplasia Taylor balloon cell type
TSC2	83	92	606690	Lymphangioleiomyomatosis somatic
TSEN2	126	88	612389	Pontocerebellar hypoplasia type 2B
TSEN34	64	94	612390	Pontocerebellar hypoplasia type 2C
TSEN54	101	97	277470	Pontocerebellar hypoplasia type 2A
TSFM	116	76	610505	Combined oxidative phosphorylation deficiency 3
TSG101	106	88	114480	Breast cancer somatic
TSHB	172	100	275100	Hypothryoidism congenital nongoitrous 4
TSHR	175	91	603373	Hyperthyroidism familial gestational
TSHZ1	118	100	607842	Aural atresia congenital

TSPAN12	127	100	613310	Exudative vitreoretinopathy 5
TSPAN7	77	87	300210	Mental retardation X-linked 58
TSPEAR	119	93	614861	Deafness autosomal recessive 98
TSPYL1	198	100	608800	Sudden infant death with dysgenesis of the testes syndrome
TTBK2	131	96	604432	Spinocerebellar ataxia 11
TTC19	63	83	615157	Mitochondrial complex III deficiency nuclear type 2
TTC21B	113	96	613819	Asphyxiating thoracic dystrophy 4
TTC37	115	97	222470	Trichohepatoenteric syndrome 1
TTC7A	78	89	243150	Intestinal atresia multiple
TTC8	107	91	209900	Bardet-Biedl syndrome 8
TTN	159	97	604145	Cardiomyopathy dilated 1G
TTPA	97	97	277460	Ataxia with isolated vitamin E deficiency
TTR	110	74	105210	Amyloidosis hereditary transthyretin-related
TUBA1A	58	9	611603	Lissencephaly 3
TUBA8	116	50	613180	Polymicrogyria with optic nerve hypoplasia
TUBB1	128	96	613112	Macrothrombocytopenia autosomal dominant TUBB1-related
TUBB2B	129	13	610031	Polymicrogyria symmetric or asymmetric
TUBB3	46	20	614039	Cortical dysplasia complex with other brain malformations
TUBB4A	125	25	612438	Leukodystrophy hypomyelinating 6
TUBGCP6	105	73	251270	Microcephaly and chorioretinopathy with or without mental retardation
TUFM	128	86	610678	Combined oxidative phosphorylation deficiency 4
TULP1	92	87	613843	Leber congenital amaurosis 15
TUSC3	73	33	611093	Mental retardation autosomal recessive 7
TWIST1	61	100	123100	Craniosynostosis type 1
TWIST2	55	100	227260	Focal facial dermal dysplasia 3 Setleis type
TYK2	85	89	611521	Tyrosine kinase 2 deficiency
TYMP	78	95	603041	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
TYR	196	100	203100	Albinism oculocutaneous type IA
TYROBP	64	85	221770	Nasu-Hakola disease
TYRP1	114	89	203290	Albinism oculocutaneous type III
UBA1	107	94	301830	Spinal muscular atrophy X-linked 2 infantile
UBE2A	74	93	300860	Mental retardation X-linked syndromic Nascimento-type
UBE3A	100	31	105830	Angelman syndrome

UBE3B	121	83	615057	Blepharophimosis-ptosis-intellectual disability syndrome
UBIAD1	95	100	121800	Corneal dystrophy Schnyder type
UBQLN2	119	100	300857	Amyotrophic lateral sclerosis 15 with or without frontotemporal dementia
UBR1	105	97	243800	Johanson-Blizzard syndrome
UGT1A1	147	97	218800	Crigler-Najjar syndrome type I
UMOD	89	84	609886	Glomerulocystic kidney disease with hyperuricemia and isosthenuria
UMPS	118	92	258900	Orotic aciduria
UNC119	106	92	200	-
UNC13D	60	88	608898	Hemophagocytic lymphohistiocytosis familial 3
UNC93B1	77	88	610551	Herpes simplex encephalitis susceptibility to 1
UNG	100	80	608106	Immunodeficiency with hyper IgM type 5
UPB1	124	81	613161	Beta-ureidopropionase deficiency
UPF3B	87	86	300676	Mental retardation X-linked syndromic 14
UPK3A	70	93	191830	Renal adysplasia
UQCRB	112	89	615158	Mitochondrial complex III deficiency nuclear type 3
UQCRC2	102	81	615160	Mitochondrial complex III deficiency nuclear type 5
UQCRQ	48	85	615159	Mitochondrial complex III deficiency nuclear type 4
UROC1	76	85	276880	Urocanase deficiency
UROD	104	82	176100	Porphyria cutanea tarda
UROS	86	92	263700	Porphyria congenital erythropoietic
USB1	53	83	604173	Poikiloderma with neutropenia
USH1C	89	90	602092	Deafness autosomal recessive 18A
USH1G	103	93	606943	Usher syndrome, type 1G
USH2A	121	91	613809	Retinitis pigmentosa 39
USP9Y	30	80	415000	Spermatogenic failure Y-linked 2
UVSSA	77	90	614640	UV-sensitive syndrome 3
VANGL1	159	90	600145	Caudal regression syndrome
VAPB	170	83	608627	Amyotrophic lateral sclerosis 8
VAX1	106	91	614402	Microphthalmia syndromic 11
VCAN	128	92	143200	Wagner syndrome 1
VCL	108	83	611407	Cardiomyopathy dilated 1W
VCP	131	86	613954	Amyotrophic lateral sclerosis 14 with or without frontotemporal dementia
VDR	95	90	277440	Rickets vitamin D-resistant type IIA

VHL	101	95	263400	Erythrocytosis familial 2
VIM	118	90	116300	Cataract 30 pulverulent
VIPAS39	103	76	613404	Arthrogryposis renal dysfunction and cholestasis 2
VKORC1	122	75	607473	Vitamin K-dependent clotting factors combined deficiency of 2
VLDLR	130	88	224050	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1
VPS13A	110	95	200150	Choreoacanthocytosis
VPS13B	117	92	216550	Cohen syndrome
VPS33B	121	89	208085	Arthrogryposis renal dysfunction and cholestasis 1
VPS35	94	76	614203	Parkinson disease 17
VPS37A	86	76	614898	Spastic paraplegia 53 autosomal recessive
VRK1	140	97	607596	Pontocerebellar hypoplasia type 1A
VSX1	70	69	122000	Corneal dystrophy hereditary polymorphous posterior
VWF	80	67	193400	von Willebrand disease type 1
WAS	56	85	300299	Neutropenia severe congenital X-linked
WDPCP	90	93	209900	Bardet-Biedl syndrome 15
WDR11	94	88	614858	Hypogonadotropic hypogonadism 14 with or without anosmia
WDR19	122	94	614376	Asphyxiating thoracic dystrophy 5
WDR35	108	96	613610	Cranioectodermal dysplasia 2
WDR36	114	96	609887	Glaucoma 1 open angle G
WDR45	75	68	300894	Neurodegeneration with brain iron acculation 5
WDR62	103	89	604317	Microcephaly 2 primary autosomal recessive with or without cortical malformations
WDR72	76	44	613211	Amelogenesis imperfecta hypomaturation type IIA3
WDR81	95	94	610185	Cerebellar ataxia mental retardation and dysequilibrium syndrome 2
WFS1	141	92	600965	Deafness autosomal dominant 6/14/38
WHSC1L1	126	71	601626	Leukemia acute myeloid
WIPF1	105	85	614493	Wiskott-Aldrich syndrome 2
WISP3	140	93	208230	Arthropathy progressive pseudorheumatoid of childhood
WNK1	151	94	201300	Neuropathy hereditary sensory and autonomic type II
WNK4	111	93	614491	Pseudohypoaldosteronism type IIB
WNT1	138	93	615220	Osteogenesis imperfecta type XV
WNT10A	64	90	257980	Odontoonychodermal dysplasia
WNT10B	102	91	225300	Split-hand/foot malformation 6
WNT3	132	90	273395	Tetra-amelia autosomal recessive

WNT4	130	95	158330	Mullerian aplasia and hyperandrogenism
WNT5A	98	97	180700	Robinow syndrome autosomal dominant
WNT7A	113	92	228930	Fuhrmann syndrome
WRAP53	129	89	613988	Dyskeratosis congenita autosomal recessive 3
WRN	133	96	200	-
WT1	64	91	194080	Denys-Drash syndrome
WWOX	111	86	133239	Esophageal squamous cell carcinoma
XDH	103	86	278300	Xanthinuria type I
XIAP	120	86	300635	Lymphoproliferative syndrome X-linked 2
XK	107	95	300842	McLeod syndrome with or without chronic granulomatous disease
XPA	79	89	278700	Xeroderma pigmentosum group A
XPC	121	91	278720	Xeroderma pigmentosum group C
XPNPEP3	132	87	613159	Nephronophthisis-like nephropathy 1
YARS	116	81	608323	Charcot-Marie-Tooth disease dominant intermediate C
YARS2	110	94	613561	Myopathy lactic acidosis and sideroblastic anemia 2
ZAP70	76	93	269840	Selective T-cell defect
ZBTB16	119	93	200	Leukemia acute promyelocytic
ZBTB24	155	98	614069	Immunodeficiency-centromeric instability-facial anomalies syndrome-2
ZC4H2	87	78	314580	Wieacker-Wolf syndrome
ZDHHC15	88	91	300577	Mental retardation X-linked 91
ZDHHC9	86	83	300799	Mental retardation X-linked syndromic Raymond type
ZEB1	162	95	613270	Corneal dystrophy Fuchs endothelial 6
ZEB2	167	92	235730	Mowat-Wilson syndrome
ZFP57	17	86	601410	Diabetes mellitus transient neonatal 1
ZFPM2	203	99	610187	Diaphragmatic hernia 3
ZFYVE26	101	82	270700	Spastic paraplegia 15 autosomal recessive
ZFYVE27	97	77	610244	Spastic paraplegia 33 autosomal dominant
ZIC2	40	95	609637	Holoprosencephaly-5
ZIC3	90	97	306955	Congenital heart defects nonsyndromic 1 X-linked
ZMPSTE24	135	96	608612	Mandibuloacral dysplasia with type B lipodystrophy
ZNF335	78	92	615095	Microcephaly 10 primary autosomal recessive
ZNF41	107	51	300848	Mental retardation X-linked 89
ZNF423	123	97	614844	Joubert syndrome 19

ZNF469	90	100	229200	Brittle cornea syndrome
ZNF513	93	96	613617	Retinitis pigmentosa 58
ZNF592	128	86	606937	Spinocerebellar ataxia autosomal recessive 5
ZNF644	111	86	614167	Myopia 21 autosomal dominant
ZNF674	106	38	300851	Mental retardation X-linked 92
ZNF711	107	98	300803	Mental retardation X-linked 97
ZNF750	127	99	610227	Seborrhea-like dermatitis with psoriasiform elements
ZNF81	84	99	300498	Mental retardation X-linked 45

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
Depth describes the average number of reads seen across 50 exomes
Coverage describes the average coverage of a gene across 50 exomes in percentiles
OMIM release used for OMIM disease identifiers and descriptions : 15 october 2013

Ad 1. OMIM identifier 200 signifies a gene without a current OMIM association

Ad 2. OMIM phenotype descriptions between {} signify risk factors