Supplementary Document

Enhancing the prediction of disease-gene associations with multimodal deep learning

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1. Hyperparameters

Table S1. The AUC scores of dgMDL with different combinations of hyperparameters

	2	4	8	10
0.001	0.968	0.970	0.975	0.974
0.0005	0.966	**0.978**	0.974	0.973
0.0002	0.966	0.971	0.971	0.975
0.0001	0.970	0.970	0.976	0.974

2. Disease-gene associations

The data downloaded from OMIM are preprocessed using the approaches discussed in Section 2.6, and the resulted disease-gene associations are listed in the following table. Diseases removed in the third step are also listed in the table for future study. The classification results are indicated by the disease ID numbers, and disease terms with the same ID numbers are regarded as one disease. Braces, '{}', indicate the disease is multifactorial disorders or infections.

Table S2 Classified OMIM disease-gene associations

Disease ID	Disease name	Gene symbol
1	17-alpha-hydroxylase or 1720-lyase deficiency 202110	CYP17A1
1	1720-lyase deficiency isolated 202110	CYP17A1
2	2-aminoadipic 2-oxoadipic aciduria 204750	DHTKD1
3	2-methylbutyrylglycinuria 610006	ACADSB
4	3-M syndrome 1 273750	CUL7
4	3-M syndrome 2 612921	OBSL1
4	3-M syndrome 3 614205	CCDC8
5	3-Methylcrotonyl-CoA carboxylase 1 deficiency 210200	MCCC1
5	3-Methylcrotonyl-CoA carboxylase 2 deficiency 210210	MCCC2
6	3-hydroxyisobutryl-CoA hydrolase deficiency 250620	HIBCH
7	3-methylglutaconic aciduria type I 250950	AUH
7	3-methylglutaconic aciduria type III 258501	OPA3

7	3-methylglutaconic aciduria type IX 617698	TIMM50
7	3-methylglutaconic aciduria type V 610198	DNAJC19
7	3-methylglutaconic aciduria type VII with cataracts neurologic involvement and neutropenia 616271	CLPB
7	3-methylglutaconic aciduria type VIII 617248	HTRA2
7	3-methylglutaconic aciduria with deafness encephalopathy and Leigh-like syndrome 614739	SERAC1
8	3MC syndrome 1 257920	MASP1
8	3MC syndrome 2 265050	COLEC11
8	3MC syndrome 3 248340	COLEC10
9	46XX sex reversal 4 617480	NR5A1
9	46XX sex reversal 1 400045	SRY
9	46XY sex reversal 1 400044	SRY
9	46XY sex reversal 2 dosage-sensitive 300018	NR0B1
9	46XY sex reversal 3 612965	NR5A1
9	46XY sex reversal 6 613762	MAP3K1
9	46XY sex reversal 7 233420	DHH
9	46XY sex reversal 8 614279	AKR1C2
9	46XY sex reversal 9 616067	ZFPM2
9	{46XY sex reversal 8 modifier of} 614279	AKR1C4
9	46XY partial gonadal dysgenesis with minifascicular neuropathy 607080	DHH
10	5-oxoprolinase deficiency 260005	OPLAH
11	ABCD syndrome 600501	EDNRB
12	ACTH-independent macronodular adrenal hyperplasia 2 615954	ARMC5
12	ACTH-independent macronodular adrenal hyperplasia 219080	GNAS
13	ADULT syndrome 103285	TP63
14	AICA-ribosiduria due to ATIC deficiency 608688	ATIC
15	Abdominal obesity-metabolic syndrome 3 615812	DYRK1B
15	{Metabolic syndrome protection against} 605552	MTTP
16	Abetalipoproteinemia 200100	MTTP
17	Ablepharon-macrostomia syndrome 200110	TWIST2
18	Acatalasemia 614097	CAT
19	Achalasia-addisonianism-alacrimia syndrome 231550	AAAS
20	Acheiropody 200500	LMBR1
21	Achondrogenesis Ib 600972	SLC26A2
21	Achondrogenesis type IA 200600	TRIP11
21	Achondrogenesis type II or hypochondrogenesis 200610	COL2A1
21	Achondroplasia 100800	FGFR3
22	Achromatopsia 2 216900	CNGA3
22	Achromatopsia 3 262300	CNGB3
22	Achromatopsia 4 613856	GNAT2
22	Achromatopsia 6 610024	PDE6H
22	Achromatopsia 7 616517	ATF6
23	Acid-labile subunit deficiency of 615961	IGFALS

24	Acne inversa familial 1 142690	NCSTN
24	Acne inversa familial 2 613736	PSENEN
24	Acne inversa familial 3 613737	PSEN1
25	Acrocapitofemoral dysplasia 607778	IHH
26	Acrodermatitis enteropathica 201100	SLC39A4
27	Acrodysostosis 1 with or without hormone resistance 101800	PRKAR1A
27	Acrodysostosis 2 with or without hormone resistance 614613	PDE4D
28	Acrofacial dysostosis 1 Nager type 154400	SF3B4
28	Acrofacial dysostosis Cincinnati type 616462	POLR1A
29	Acrokeratosis verruciformis 101900	ATP2A2
30	Acromelic frontonasal dysostosis 603671	ZSWIM6
31	Acromesomelic dysplasia Demirhan type 609441	BMPR1B
31	Acromesomelic dysplasia Maroteaux type 602875	NPR2
31	Acromicric dysplasia 102370	FBN1
32	Acyl-CoA dehydrogenase medium chain deficiency of 201450	ACADM
32	Acyl-CoA dehydrogenase short-chain deficiency of 201470	ACADS
32	3-hydroxyacyl-CoA dehydrogenase deficiency 231530	HADH
33	Adams-Oliver syndrome 1 100300	ARHGAP31
33	Adams-Oliver syndrome 2 614219	DOCK6
33	Adams-Oliver syndrome 3 614814	RBPJ
33	Adams-Oliver syndrome 4 615297	EOGT
33	Adams-Oliver syndrome 5 616028	NOTCH1
33	Adams-Oliver syndrome 6 616589	DLL4
34	Adenine phosphoribosyltransferase deficiency 614723	APRT
35	Adenomas multiple colorectal 608456	MUTYH
35	Adenomas salivary gland pleomorphic somatic 181030	PLAG1
35	Adenomatous polyposis coli 175100	APC
35	Gardner syndrome 175100	APC
35	Brain tumor-polyposis syndrome 2 175100	APC
36	Adenosine deaminase deficiency partial 102700	ADA
36	Severe combined immunodeficiency due to ADA deficiency 102700	ADA
37	Adenosine triphosphate elevated of erythrocytes 102900	PKLR
38	Adenylosuccinase deficiency 103050	ADSL
39	Adermatoglyphia 136000	SMARCAD1
39	Basan syndrome 129200	SMARCAD1
40	Adiponectin deficiency 612556	ADIPOQ
41	Adrenal hyperplasia congenital due to 11-beta-hydroxylase deficiency 202010	CYP11B1
41	Adrenal hyperplasia congenital due to 21-hydroxylase deficiency 201910	CYP21A2
41	Hyperandrogenism nonclassic type due to 21-hydroxylase deficiency 201910	CYP21A2
41	Adrenal hyperplasia congenital due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency 201810	HSD3B2

41	Adrenal hypoplasia congenital 300200	NR0B1
41	Adrenal insufficiency congenital with 46XY sex reversal partial or complete 613743	CYP11A1
42	Adrenal cortical carcinoma 202300	TP53
42	Adrenocortical tumor somatic	PRKAR1A
43	Adrenocorticotropic hormone deficiency 201400	TBX19
44	Adrenoleukodystrophy 300100	ABCD1
44	Adrenomyeloneuropathy adult 300100	ABCD1
45	Adult i phenotype without cataract 110800	GCNT2
46	Advanced sleep phase syndrome familial 1 604348	PER2
46	Advanced sleep-phase syndrome familial 2 615224	CSNK1D
47	{Delayed sleep phase disorder susceptibility to} 614163	CRY1
48	Agammaglobulinemia 1 601495	IGHM
48	Agammaglobulinemia 2 613500	IGLL1
48	Agammaglobulinemia 3 613501	CD79A
48	Agammaglobulinemia 4 613502	BLNK
48	Agammaglobulinemia 6 612692	CD79B
48	Agammaglobulinemia 8 autosomal dominant 616941	TCF3
48	Agammaglobulinemia X-linked 1 300755	BTK
48	Agammaglobulinemia and isolated hormone deficiency 307200	BTK
49	Agenesis of the corpus callosum with peripheral neuropathy 218000	SLC12A6
50	Agnathia-otocephaly complex 202650	PRRX1
51	Aicardi-Goutieres syndrome 1 dominant and recessive 225750	TREX1
51	Aicardi-Goutieres syndrome 2 610181	RNASEH2B
51	Aicardi-Goutieres syndrome 3 610329	RNASEH2C
51	Aicardi-Goutieres syndrome 4 610333	RNASEH2A
51	Aicardi-Goutieres syndrome 5 612952	SAMHD1
51	Aicardi-Goutieres syndrome 6 615010	ADAR
51	Aicardi-Goutieres syndrome 7 615846	IFIH1
52	Al Kaissi syndrome 617694	CDK10
53	Al-Raqad syndrome 616459	DCPS
54	Alacrima achalasia and mental retardation syndrome 615510	GMPPA
55	Alagille syndrome 1 118450	JAG1
55	Alagille syndrome 2 610205	NOTCH2
56	Aland Island eye disease 300600	CACNA1F
57	Alazami syndrome 615071	LARP7
58	Alazami-Yuan syndrome 617126	TAF6
59	Albinism brown oculocutaneous 203200	OCA2
59	Albinism oculocutaneous type IA 203100	TYR
59	Albinism oculocutaneous type IB 606952	TYR
59	Albinism oculocutaneous type II 203200	OCA2
59	Albinism oculocutaneous type III 203290	TYRP1
59	Albinism oculocutaneous type IV 606574	SLC45A2
59	Albinism oculocutaneous type VI 113750	SLC24A5

59	Albinism oculocutaneous type VII 615179	LRMDA
59	{Albinism oculocutaneous type II modifier of} 203200	MC1R
60	Alcohol sensitivity acute 610251	ALDH2
60	{Hangover susceptibility to} 610251	ALDH2
61	Aldosteronism glucocorticoid-remediable 103900	CYP11B1
62	Alexander disease 203450	GFAP
63	Alkaptonuria 203500	HGD
64	Allan-Herndon-Dudley syndrome 300523	SLC16A2
65	Alopecia universalis 203655	HR
66	Alpha-2-plasmin inhibitor deficiency 262850	PLI
67	Alpha-fetoprotein deficiency 615969	AFP
68	Alpha-methylacetoacetic aciduria 203750	ACAT1
69	Alpha-methylacyl-CoA racemase deficiency 614307	AMACR
70	Alpha-thalassemia myelodysplasia syndrome somatic 300448	ATRX
70	Alpha-thalassemia or mental retardation syndrome 301040	ATRX
71	Alpha or beta T-cell lymphopenia with gamma or delta T-cell expansion severe cytomegalovirus infection and autoimmunity 609889	RAG1
72	Alport syndrome 301050	COL4A5
72	Alport syndrome autosomal dominant 104200	COL4A3
72	Alport syndrome autosomal recessive 203780	COL4A3
72	Alport syndrome autosomal recessive 203780	COL4A4
73	Alstrom syndrome 203800	ALMS1
74	Alternating hemiplegia of childhood 104290	ATP1A2
74	Alternating hemiplegia of childhood 2 614820	ATP1A3
75	Alveolar capillary dysplasia with misalignment of pulmonary veins 265380	FOXF1
76	Alveolar soft-part sarcoma 606243	ASPSCR1
77	Alzheimer disease 1 familial 104300	APP
77	Alzheimer disease type 3 607822	PSEN1
77	Alzheimer disease type 3 with spastic paraparesis and apraxia 607822	PSEN1
77	Alzheimer disease type 3 with spastic paraparesis and unusual plaques 607822	PSEN1
77	Alzheimer disease-2 104310	APOE
77	Alzheimer disease-4 606889	PSEN2
77	{Alzheimer disease 18 susceptibility to} 615590	ADAM10
77	{Alzheimer disease 9 susceptibility to} 608907	ABCA7
77	{Alzheimer disease late-onset susceptibility to} 104300	NOS3
77	{Alzheimer disease late-onset susceptibility to} 104300	PLAU
77	{Alzheimer disease susceptibility to} 104300	A2M
77	{Alzheimer disease susceptibility to} 104300	HFE
77	{Alzheimer disease susceptibility to} 104300	MPO
78	Amelogenesis imperfecta hypomaturation type IIA6 617217	GPR68
78	Amelogenesis imperfecta type 1E 301200	AMELX

78	Amelogenesis imperfecta type IA 104530	LAMB3
78	Amelogenesis imperfecta type IB 104500	ENAM
78	Amelogenesis imperfecta type IC 204650	ENAM
78	Amelogenesis imperfecta type IF 616270	AMBN
78	Amelogenesis imperfecta type IG (enamel-renal syndrome) 204690	FAM20A
78	Amelogenesis imperfecta type IH 616221	ITGB6
78	Amelogenesis imperfecta type IIA1 204700	KLK4
78	Amelogenesis imperfecta type IIA2 612529	MMP20
78	Amelogenesis imperfecta type IIA3 613211	WDR72
78	Amelogenesis imperfecta type IIA4 614832	ODAPH
78	Amelogenesis imperfecta type IIA5 615887	SLC24A4
78	Amelogenesis imperfecta type IIIA 130900	FAM83H
78	Amelogenesis imperfecta type IJ 617297	ACP4
78	Amelogenesis imperfecta type IV 104510	DLX3
79	Aminoacylase 1 deficiency 609924	ACY1
80	Amyloidosis 3 or more types 105200	APOA1
80	Amyloidosis Finnish type 105120	GSN
80	Amyloidosis familial visceral 105200	FGA
80	Amyloidosis hereditary transthyretin-related 105210	TTR
80	Amyloidosis primary localized cutaneous 1 105250	OSMR
80	Amyloidosis primary localized cutaneous 2 613955	IL31RA
80	Amyloidosis renal 105200	LYZ
81	Amyotrophic lateral sclerosis 1 105400	SOD1
81	Amyotrophic lateral sclerosis 10 with or without FTD 612069	TARDBP
81	Amyotrophic lateral sclerosis 11 612577	FIG4
81	Amyotrophic lateral sclerosis 12 613435	OPTN
81	Amyotrophic lateral sclerosis 14 with or without frontotemporal dementia 613954	VCP
81	Amyotrophic lateral sclerosis 15 with or without frontotemporal dementia 300857	UBQLN2
81	Amyotrophic lateral sclerosis 17 614696	CHMP2B
81	Amyotrophic lateral sclerosis 18 614808	PFN1
81	Amyotrophic lateral sclerosis 19 615515	ERBB4
81	Amyotrophic lateral sclerosis 2 juvenile 205100	ALS2
81	Amyotrophic lateral sclerosis 20 615426	HNRNPA1
81	Amyotrophic lateral sclerosis 21 606070	MATR3
81	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia 616208	TUBA4A
81	Amyotrophic lateral sclerosis 4 juvenile 602433	SETX
81	Amyotrophic lateral sclerosis 5 juvenile 602099	SPG11
81	Amyotrophic lateral sclerosis 6 with or without frontotemporal dementia 608030	FUS
81	Amyotrophic lateral sclerosis 8 608627	VAPB
81	Amyotrophic lateral sclerosis 9 611895	ANG

81	{Amyotrophic lateral sclerosis susceptibility to 13} 183090	ATXN2
81	{Amyotrophic lateral sclerosis susceptibility to} 105400	DCTN1
81	{Amyotrophic lateral sclerosis susceptibility to} 105400	PRPH
81	{Amyotrophic lateral sclerosis-parkinsonism or dementia complex susceptibility to} 105500	TRPM7
82	Amyotrophy hereditary neuralgic 162100	9-Sep
83	Analbuminemia 616000	ALB
84	Anauxetic dysplasia 1 607095	RMRP
84	Anauxetic dysplasia 2 617396	POP1
85	Andersen syndrome 170390	KCNJ2
86	Androgen insensitivity 300068	AR
86	Androgen insensitivity partial with or without breast cancer 312300	AR
87	Anemia X-linked with without neutropenia and or platelet abnormalities 300835	GATA1
87	Anemia hemolytic Rh-null regulator type 268150	RHAG
87	Anemia hemolytic due to UMPH1 deficiency 266120	NT5C3A
87	Anemia hypochromic microcytic with iron overload 1 206100	SLC11A2
87	Anemia sideroblastic 1 300751	ALAS2
87	Anemia sideroblastic 2 pyridoxine-refractory 205950	SLC25A38
87	Anemia sideroblastic 3 pyridoxine-refractory 616860	GLRX5
87	Anemia sideroblastic 4 182170	HSPA9
87	Anemia sideroblastic with ataxia 301310	ABCB7
87	Anemia neonatal hemolytic fatal and near-fatal	SPTB
88	Angelman syndrome 105830	UBE3A
89	Angioedema hereditary type III 610618	F12
89	Angioedema hereditary types I and II 106100	SERPING1
89	{Angioedema induced by ACE inhibitors susceptibility to} 300909	XPNPEP2
90	Angiopathy hereditary with nephropathy aneurysms and muscle cramps 611773	COL4A1
91	Anonychia congenita 206800	RSPO4
92	Anterior segment anomalies with or without cataract 602588	EYA1
92	Anterior segment dysgenesis 1 multiple subtypes 107250	PITX3
92	Anterior segment dysgenesis 2 multiple subtypes 610256	FOXE3
92	Anterior segment dysgenesis 3 multiple subtypes 601631	FOXC1
92	Anterior segment dysgenesis 4 137600	PITX2
92	Anterior segment dysgenesis 5 multiple subtypes 604229	PAX6
92	Anterior segment dysgenesis 6 multiple subtypes 617315	CYP1B1
92	Anterior segment dysgenesis 7 with sclerocornea 269400	PXDN
92	Anterior segment dysgenesis 8 617319	CPAMD8
93	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis 201750	POR
93	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis 207410	FGFR2
94	Aortic aneurysm familial thoracic 10 617168	LOX

94	Aortic aneurysm familial thoracic 4 132900	MYH11
94	Aortic aneurysm familial thoracic 6 611788	ACTA2
94	Aortic aneurysm familial thoracic 7 613780	MYLK
94	Aortic aneurysm familial thoracic 8 615436	PRKG1
94	Aortic aneurysm familial thoracic 9 616166	MFAP5
94	{Aortic aneurysm familial thoracic 11 susceptibility to} 617349	FOXE3
95	Aortic valve disease 1 109730	NOTCH1
95	Aortic valve disease 2 614823	SMAD6
96	Apert syndrome 101200	FGFR2
97	Aplasia of lacrimal and salivary glands 180920	FGF10
98	Apolipoprotein C-III deficiency 614028	APOC3
98	ApoA-I and apoC-III deficiency combined	APOA1
98	Apolipoprotein A-II deficiency	APOA2
99	Apparent mineralocorticoid excess 218030	HSD11B2
100	Argininemia 207800	ARG1
100	Argininosuccinic aciduria 207900	ASL
101	Aromatase deficiency 613546	CYP19A1
101	Aromatase excess syndrome 139300	CYP19A1
102	Aromatic L-amino acid decarboxylase deficiency 608643	DDC
103	Arrhythmogenic right ventricular dysplasia 1 107970	TGFB3
103	Arrhythmogenic right ventricular dysplasia 10 610193	DSG2
103	Arrhythmogenic right ventricular dysplasia 11 610476	DSC2
103	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair 610476	DSC2
103	Arrhythmogenic right ventricular dysplasia 12 611528	JUP
103	Arrhythmogenic right ventricular dysplasia 2 600996	RYR2
103	Arrhythmogenic right ventricular dysplasia 5 604400	TMEM43
103	Arrhythmogenic right ventricular dysplasia 8 607450	DSP
103	Arrhythmogenic right ventricular dysplasia 9 609040	PKP2
103	Arrhythmogenic right ventricular dysplasia familial 13 615616	CTNNA3
104	Arterial calcification generalized of infancy 1 208000	ENPP1
104	Arterial calcification generalized of infancy 2 614473	ABCC6
105	Arterial tortuosity syndrome 208050	SLC2A10
106	Arthrogryposis distal type 1B 614335	MYBPC1
106	Arthrogryposis distal type 2A 193700	MYH3
106	Arthrogryposis distal type 2B 601680	MYH3
106	Arthrogryposis distal type 2B 601680	TNNT3
106	Arthrogryposis distal type 2B 601680	TPM2
106	Arthrogryposis distal type 3 114300	PIEZO2
106	Arthrogryposis distal type 5 108145	PIEZO2
106	Arthrogryposis distal type 5D 615065	ECEL1
106	Arthrogryposis distal type 8 178110	MYH3
106	Arthrogryposis distal with impaired proprioception and touch 617146	PIEZO2
106	Arthrogryposis lethal with anterior horn cell disease 611890	GLE1

106	Arthrogryposis multiplex congenita distal type 1 108120	TPM2
106	Arthrogryposis multiplex congenita distal type 2B 601680	TNNI2
106	Arthrogryposis multiplex congenita neurogenic with myelin defect 617468	LGI4
106	Arthrogryposis renal dysfunction and cholestasis 1 208085	VPS33B
106	Arthrogryposis renal dysfunction and cholestasis 2 613404	VIPAS39
107	Arts syndrome 301835	PRPS1
108	Asparagine synthetase deficiency 615574	ASNS
109	Aspartate aminotransferase serum level of QTL1 614419	GOT1
110	Aspartylglucosaminuria 208400	AGA
111	Asplenia isolated congenital 271400	RPSA
112	Asthma and nasal polyps 208550	TBX21
112	{Asthma aspirin-induced susceptibility to} 208550	PTGER2
112	{Asthma aspirin-induced susceptibility to} 208550	TBX21
112	{Asthma diminished response to antileukotriene treatment in} 600807	ALOX5
112	{Asthma nocturnal susceptibility to} 600807	ADRB2
112	{Asthma protection against} 600807	MUC7
112	{Asthma susceptibility 5} 611064	IRAK3
112	{Asthma susceptibility to 1} 607277	PTGDR
112	{Asthma susceptibility to 2} 608584	NPSR1
112	{Asthma susceptibility to} 600807	CCL11
112	{Asthma susceptibility to} 600807	HNMT
112	{Asthma susceptibility to} 600807	IL13
112	{Asthma susceptibility to} 600807	PLA2G7
112	{Asthma susceptibility to} 600807	SCGB3A2
112	{Asthma susceptibility to} 600807	TNF
112	{Asthma-related traits susceptibility to 7} 611960	CHI3L1
113	Ataxia cerebellar Cayman type 601238	ATCAY
113	Ataxia early-onset with oculomotor apraxia and hypoalbuminemia 208920	APTX
113	Ataxia posterior column with retinitis pigmentosa 609033	FLVCR1
113	Ataxia sensory 1 autosomal dominant 608984	RNF170
113	Ataxia with isolated vitamin E deficiency 277460	TTPA
113	Ataxia-oculomotor apraxia 3 615217	PIK3R5
113	Ataxia-oculomotor apraxia 4 616267	PNKP
113	Ataxia-pancytopenia syndrome 159550	SAMD9L
114	Ataxia-telangiectasia 208900	ATM
114	Ataxia-telangiectasia-like disorder 1 604391	MRE11A
115	De la Chapelle dysplasia 256050	SLC26A2
115	Atelosteogenesis II 256050	SLC26A2
115	Atelosteogenesis type I 108720	FLNB
115	Atelosteogenesis type III 108721	FLNB
116	Atransferrinemia 209300	TF
117	Atrial fibrillation familial 10 614022	SCN5A

117	Atrial fibrillation familial 11 614049	GJA5
117	Atrial fibrillation familial 12 614050	ABCC9
117	Atrial fibrillation familial 13 615377	SCN1B
117	Atrial fibrillation familial 14 615378	SCN2B
117	Atrial fibrillation familial 16 613120	SCN3B
117	Atrial fibrillation familial 17 611819	SCN4B
117	Atrial fibrillation familial 3 607554	KCNQ1
117	Atrial fibrillation familial 4 611493	KCNE2
117	Atrial fibrillation familial 6 612201	NPPA
117	Atrial fibrillation familial 7 612240	KCNA5
117	Atrial fibrillation familial 9 613980	KCNJ2
117	Atrial septal defect 2 607941	GATA4
117	Atrial septal defect 3 614089	MYH6
117	Atrial septal defect 4 611363	TBX20
117	Atrial septal defect 5 612794	ACTC1
117	Atrial septal defect 6 613087	TLL1
117	Atrial septal defect 7 with or without AV conduction defects 108900	NKX2-5
117	Atrial septal defect 8 614433	CITED2
117	Atrial septal defect 9 614475	GATA6
118	Atrial standstill 2 615745	NPPA
118	Atrial standstill digenic (GJA5 or SCN5A) 108770	GJA5
119	Atrichia with papular lesions 209500	HR
120	Atrioventricular septal defect 3 600309	GJA1
120	Atrioventricular septal defect 4 614430	GATA4
120	Atrioventricular septal defect 5 614474	GATA6
120	Atrioventricular septal defect partial with heterotaxy syndrome 606217	CRELD1
120	{Atrioventricular septal defect susceptibility to 2} 606217	CRELD1
121	Au-Kline syndrome 616580	HNRNPK
122	Auditory neuropathy and optic atrophy 617717	FDXR
122	Auditory neuropathy autosomal dominant 1 609129	DIAPH3
122	Auditory neuropathy autosomal recessive 1 601071	OTOF
123	Aural atresia congenital 607842	TSHZ1
124	Auriculocondylar syndrome 1 602483	GNAI3
124	Auriculocondylar syndrome 2 614669	PLCB4
124	Auriculocondylar syndrome 3 615706	EDN1
125	Autoimmune disease multisystem infantile-onset 1 615952	STAT3
125	Autoimmune disease multisystem infantile-onset 2 617006	ZAP70
125	Autoimmune disease multisystem with facial dysmorphism 613385	ITCH
125	{Autoimmune disease susceptibility to 1} 607836	FOXD3
125	{Autoimmune disease susceptibility to 6} 613551	SIAE
126	{Autoimmune interstitial lung joint and kidney disease} 616414	COPA
127	{Autoimmune thyroid disease susceptibility to 3} 608175	TG

127	{Autoimmune thyroid disease susceptibility to 3} 608175	ZFAT
128	Autoimmune lymphoproliferative syndrome type IA 601859	FAS
128	Autoimmune lymphoproliferative syndrome type IB 601859	FASLG
128	Autoimmune lymphoproliferative syndrome type II 603909	CASP10
128	Autoimmune lymphoproliferative syndrome type III 615559	PRKCD
128	Autoimmune lymphoproliferative syndrome type V 616100	CTLA4
128	{Autoimmune lymphoproliferative syndrome} 601859	FAS
128	Autoimmune polyendocrinopathy syndrome type I with or without reversible metaphyseal dysplasia 240300	AIRE
129	Autoinflammation antibody deficiency and immune dysregulation syndrome 614878	PLCG2
129	Autoinflammation lipodystrophy and dermatosis syndrome 256040	PSMB8
129	Autoinflammation panniculitis and dermatosis syndrome 617099	OTULIN
129	Autoinflammation with arthritis and dyskeratosis 617388	NLRP1
129	Autoinflammation with infantile enterocolitis 616050	NLRC4
129	Autoinflammatory syndrome familial Behcet-like 616744	TNFAIP3
130	Avascular necrosis of the femoral head 608805	COL2A1
131	Axenfeld-Rieger syndrome type 1 180500	PITX2
131	Axenfeld-Rieger syndrome type 3 602482	FOXC1
132	Ayme-Gripp syndrome 601088	MAF
133	B-cell expansion with NFKB and T-cell anergy 616452	CARD11
134	Bainbridge-Ropers syndrome 615485	ASXL3
135	Baller-Gerold syndrome 218600	RECQL4
136	Bamforth-Lazarus syndrome 241850	FOXE1
137	Band heterotopia 600348	EML1
138	Bannayan-Riley-Ruvalcaba syndrome 153480	PTEN
139	Baraitser-Winter syndrome 1 243310	ACTB
139	Baraitser-Winter syndrome 2 614583	ACTG1
140	Barber-Say syndrome 209885	TWIST2
141	Bardet-Biedl syndrome 1 209900	BBS1
141	Bardet-Biedl syndrome 10 615987	BBS10
141	Bardet-Biedl syndrome 12 615989	BBS12
141	Bardet-Biedl syndrome 13 615990	MKS1
141	Bardet-Biedl syndrome 16 615993	SDCCAG8
141	Bardet-Biedl syndrome 17 615994	LZTFL1
141	Bardet-Biedl syndrome 2 615981	BBS2
141	Bardet-Biedl syndrome 21 617406	C8orf37
141	Bardet-Biedl syndrome 3 600151	ARL6
141	Bardet-Biedl syndrome 4 615982	BBS4
141	Bardet-Biedl syndrome 5 615983	BBS5
141	Bardet-Biedl syndrome 6 605231	MKKS
141	Bardet-Biedl syndrome 7 615984	BBS7
141	Bardet-Biedl syndrome 8 615985	TTC8
141	Bardet-Biedl syndrome 9 615986	BBS9

141	{Bardet-Biedl syndrome 1 modifier of} 209900	ARL6
141	{Bardet-Biedl syndrome 1 modifier of} 209900	CCDC28B
141	{Bardet-Biedl syndrome 14 modifier of} 615991	TMEM67
142	Bare lymphocyte syndrome type I 604571	TAP1
142	Bare lymphocyte syndrome type I 604571	TAPBP
142	Bare lymphocyte syndrome type I due to TAP2 deficiency 604571	TAP2
142	Bare lymphocyte syndrome type II complementation group A 209920	CIITA
142	Bare lymphocyte syndrome type II complementation group C 209920	RFX5
142	Bare lymphocyte syndrome type II complementation group D 209920	RFXAP
142	Bare lymphocyte syndrome type II complementation group E 209920	RFX5
142	MHC class II deficiency complementation group B 209920	RFXANK
143	Barrett esophagus or esophageal adenocarcinoma 614266	ASCC1
143	Barrett esophagus or esophageal adenocarcinoma 614266	CTHRC1
143	Barrett esophagus or esophageal adenocarcinoma 614266	MSR1
144	Bart-Pumphrey syndrome 149200	GJB2
145	Barth syndrome 302060	TAZ
146	Bartter syndrome type 1 601678	SLC12A1
146	Bartter syndrome type 2 241200	KCNJ1
146	Bartter syndrome type 3 607364	CLCNKB
146	Bartter syndrome type 4a 602522	BSND
146	Bartter syndrome type 4b digenic 613090	CLCNKA
146	Bartter syndrome type 4b digenic 613090	CLCNKB
146	Bartter syndrome type 5 antenatal transient 300971	MAGED2
146	Sensorineural deafness with mild renal dysfunction 602522	BSND
147	Basal cell carcinoma somatic 605462	PTCH1
147	Basal cell carcinoma somatic 605462	PTCH2
147	Basal cell carcinoma somatic 605462	RASA1
147	Basal cell carcinoma somatic 605462	SMO
147	{Basal cell carcinoma 7} 614740	TP53
148	Basal cell nevus syndrome 109400	PTCH1
148	Basal cell nevus syndrome 109400	PTCH2
148	Basal cell nevus syndrome 109400	SUFU
149	Basal ganglia calcification idiopathic 1 213600	SLC20A2
149	Basal ganglia calcification idiopathic 4 615007	PDGFRB
149	Basal ganglia calcification idiopathic 5 615483	PDGFB
149	Basal ganglia calcification idiopathic 6 616413	XPR1
150	Basal laminar drusen 126700	CFH
151	Basel-Vanagait-Smirin-Yosef syndrome 616449	MED25
152	Beare-Stevenson cutis gyrata syndrome 123790	FGFR2
153	Beaulieu-Boycott-Innes syndrome 613680	THOC6
154	Becker muscular dystrophy 300376	DMD

155	Beckwith-Wiedemann syndrome 130650	CDKN1C
155	Beckwith-Wiedemann syndrome 130650	H19
155	Beckwith-Wiedemann syndrome 130650	H19-ICR
155	Beckwith-Wiedemann syndrome 130650	KCNQ1OT1
156	Behr syndrome 210000	OPA1
157	Bent bone dysplasia syndrome 614592	FGFR2
158	Bernard-Soulier syndrome type A1 (recessive) 231200	GP1BA
158	Bernard-Soulier syndrome type A2 (dominant) 153670	GP1BA
158	Bernard-Soulier syndrome type B 231200	GP1BB
158	Bernard-Soulier syndrome type C 231200	GP9
158	Giant platelet disorder isolated 231200	GP1BB
159	Bestrophinopathy autosomal recessive 611809	BEST1
160	Beta-ureidopropionase deficiency 613161	UPB1
161	Bethlem myopathy 1 158810	COL6A1
161	Bethlem myopathy 1 158810	COL6A2
161	Bethlem myopathy 1 158810	COL6A3
161	Bethlem myopathy 2 616471	COL12A1
162	Bietti crystalline corneoretinal dystrophy 210370	CYP4V2
163	Bifid nose with or without anorectal and renal anomalies 608980	FREM1
164	Bile acid malabsorption primary 613291	SLC10A2
165	Bile acid synthesis defect congenital 1 607765	HSD3B7
165	Bile acid synthesis defect congenital 2 235555	AKR1D1
165	Bile acid synthesis defect congenital 3 613812	CYP7B1
165	Bile acid synthesis defect congenital 4 214950	AMACR
165	Bile acid synthesis defect congenital 6 617308	ACOX2
166	Biotinidase deficiency 253260	BTD
167	Birk-Barel mental retardation dysmorphism syndrome 612292	KCNK9
168	Birt-Hogg-Dube syndrome 135150	FLCN
169	Bjornstad syndrome 262000	BCS1L
170	Bladder cancer somatic 109800	FGFR3
170	Bladder cancer somatic 109800	KRAS
170	Bladder cancer somatic 109800	RB1
170	{Bladder cancer somatic} 109800	HRAS
171	Blau syndrome 186580	NOD2
172	Bleeding disorder platelet-type 11 614201	GP6
172	Bleeding disorder platelet-type 15 615193	ACTN1
172	Bleeding disorder platelet-type 16 autosomal dominant 187800	ITGA2B
172	Bleeding disorder platelet-type 16 autosomal dominant 187800	ITGB3
172	Bleeding disorder platelet-type 17 187900	GFI1B
172	Bleeding disorder platelet-type 20 616913	SLFN14
172	Bleeding disorder platelet-type 21 617443	FLI1
172	Bleeding disorder platelet-type 8 609821	P2RY12
172	{Bleeding disorder platelet-type 13 susceptibility to} 614009	TBXA2R
173	Blepharocheilodontic syndrome 1 119580	CDH1
173	Blepharocheilodontic syndrome 2 617681	CTNND1

174	Blepharophimosis epicanthus inversus and ptosis type 1 110100	FOXL2
174	Blepharophimosis epicanthus inversus and ptosis type 2 110100	FOXL2
175	Blood group--Lutheran inhibitor 111150	KLF1
176	Bloom syndrome 210900	RECQL3
177	Blue cone monochromacy 303700	OPN1LW
177	Blue cone monochromacy 303700	OPN1MW
178	Bohring-Opitz syndrome 605039	ASXL1
179	Bone marrow failure syndrome 1 614675	SRP72
179	Bone marrow failure syndrome 2 615715	ERCC6L2
179	Bone marrow failure syndrome 3 617052	DNAJC21
180	Bone mineral density QTL18 osteoporosis 300910	PLS3
180	{Bone mineral density QTL 12 osteoporosis} 612560	UGT2B17
180	{Bone mineral density low susceptibility to} 615311	LGR4
180	{Bone mineral density variation QTL osteoporosis} 166710	COL1A1
180	{Osteoporosis postmenopausal susceptibility} 166710	CALCR
180	{Osteoporosis postmenopausal} 166710	COL1A2
180	{Osteoporosis susceptibility to} 166710	PDLIM4
180	{Osteoporosis} 166710	LRP5
180	{Osteoporosis early-onset susceptibility to autosomal dominant} 615221	WNT1
181	Boomerang dysplasia 112310	FLNB
182	Borjeson-Forssman-Lehmann syndrome 301900	PHF6
183	Bosch-Boonstra-Schaaf optic atrophy syndrome 615722	NR2F1
184	Bosley-Salih-Alorainy syndrome 601536	HOXA1
184	Athabaskan brainstem dysgenesis syndrome 601536	HOXA1
185	Bosma arhinia microphthalmia syndrome 603457	SMCHD1
186	Bothnia retinal dystrophy 607475	RLBP1
187	Boucher-Neuhauser syndrome 215470	PNPLA6
188	Bowen-Conradi syndrome 211180	EMG1
189	Brachycephaly trichomegaly and developmental delay 617412	RPS23
190	Brachydactyly type A1 112500	IHH
190	Brachydactyly type A1 C 615072	GDF5
190	Brachydactyly type A1 D 616849	BMPR1B
190	Brachydactyly type A2 112600	BMP2
190	Brachydactyly type A2 112600	BMPR1B
190	Brachydactyly type A2 112600	GDF5
190	Brachydactyly type B1 113000	ROR2
190	Brachydactyly type B2 611377	NOG
190	Brachydactyly type C 113100	GDF5
190	Brachydactyly type D 113200	HOXD13
190	Brachydactyly type E 113300	HOXD13
190	Brachydactyly type E2 613382	PTHLH
191	Brachyolmia 4 with mild epiphyseal and metaphyseal changes 612847	PAPSS2
191	Brachyolmia type 3 113500	TRPV4

192	Bradyopsia 608415	RGS9
192	Bradyopsia 608415	RGS9BP
193	Brain malformations with or without urinary tract defects 613735	NFIA
194	Brain small vessel disease with or without ocular anomalies 607595	COL4A1
195	Branched-chain ketoacid dehydrogenase kinase deficiency 614923	BCKDK
196	Branchiooculofacial syndrome 113620	TFAP2A
197	Branchiootic syndrome 1 602588	EYA1
197	Branchiootic syndrome 3 608389	SIX1
197	Branchiootorenal syndrome 1 with or without cataracts 113650	EYA1
197	Branchiootorenal syndrome 2 610896	SIX5
198	Breast cancer 114480	TP53
198	Breast cancer early-onset 114480	BRIP1
198	Breast cancer somatic 114480	AKT1
198	Breast cancer somatic 114480	KRAS
198	Breast cancer somatic 114480	PIK3CA
198	Breast cancer somatic 114480	PPM1D
198	Breast cancer somatic 114480	RB1CC1
198	Breast cancer somatic 114480	SLC22A1
198	Breast cancer somatic 114480	TSG101
198	{Breast cancer invasive ductal} 114480	RAD54L
198	{Breast cancer lobular} 114480	CDH1
198	{Breast cancer male susceptibility to} 114480	BRCA2
198	{Breast cancer protection against} 114480	CASP8
198	{Breast cancer susceptibility to} 114480	ATM
198	{Breast cancer susceptibility to} 114480	BARD1
198	{Breast cancer susceptibility to} 114480	CHEK2
198	{Breast cancer susceptibility to} 114480	HMMR
198	{Breast cancer susceptibility to} 114480	PALB2
198	{Breast cancer susceptibility to} 114480	PHB
198	{Breast cancer susceptibility to} 114480	RAD51
198	{Breast cancer susceptibility to} 114480	XRCC3
198	{Breast-ovarian cancer familial 1} 604370	BRCA1
198	{Breast-ovarian cancer familial 2} 612555	BRCA2
198	{Breast-ovarian cancer familial susceptibility to 3} 613399	RAD51C
198	{Breast-ovarian cancer familial susceptibility to 4} 614291	RAD51D
198	{Breast and colorectal cancer, susceptibility to}	CHEK2
199	Brittle cornea syndrome 1 229200	ZNF469
199	Brittle cornea syndrome 2 614170	PRDM5
200	Brody myopathy 601003	ATP2A1
201	Bronchiectasis with or without elevated sweat chloride 1 211400	SCNN1B
201	Bronchiectasis with or without elevated sweat chloride 2 613021	SCNN1A
201	Bronchiectasis with or without elevated sweat chloride 3 613071	SCNN1G

201	{Bronchiectasis with or without elevated sweat chloride 1 modifier of} 211400	CFTR
202	Brooke-Spiegler syndrome 605041	CYLD
203	Brown-Vialetto-Van Laere syndrome 1 211530	SLC52A3
203	Brown-Vialetto-Van Laere syndrome 2 614707	SLC52A2
204	Bruck syndrome 1 259450	FKBP10
204	Bruck syndrome 2 609220	PLOD2
205	Brugada syndrome 1 601144	SCN5A
205	Brugada syndrome 2 611777	GPD1L
205	Brugada syndrome 3 611875	CACNA1C
205	Brugada syndrome 4 611876	CACNB2
205	Brugada syndrome 5 612838	SCN1B
205	Brugada syndrome 6 613119	KCNE3
205	Brugada syndrome 7 613120	SCN3B
205	Brugada syndrome 8 613123	HCN4
205	Brugada syndrome 9 616399	KCND3
206	Brunner syndrome 300615	MAOA
206	{Antisocial behavior} 300615	MAOA
207	Burn-McKeown syndrome 608572	TXNL4A
208	C syndrome 211750	CD96
209	C1q deficiency 613652	C1QA
209	C1q deficiency 613652	C1QB
209	C1q deficiency 613652	C1QC
209	C1s deficiency 613783	C1S
209	C2 deficiency 217000	C2
209	C3 deficiency 613779	C3
209	C4B deficiency 614379	C4B
209	C4a deficiency 614380	C4A
209	C5 deficiency 609536	C5
209	C6 deficiency 612446	C6
209	C7 deficiency 610102	C7
209	C8 deficiency type I 613790	C8A
209	C8 deficiency type II 613789	C8B
209	C9 deficiency 613825	C9
209	Combined C6/C7 deficiency	C6
210	CAPOS syndrome 601338	ATP1A3
211	CARASIL syndrome 600142	HTRA1
212	CATSHL syndrome 610474	FGFR3
213	CD8 deficiency familial 608957	CD8A
214	CHARGE syndrome 214800	CHD7
215	CHILD syndrome 308050	NSDHL
216	CHIME syndrome 280000	PIGL
217	CHOPS syndrome 616368	AFF4
218	CINCA syndrome 607115	NLRP3
219	CK syndrome 300831	NSDHL

220	CLOVE syndrome somatic 612918	PIK3CA
221	COACH syndrome 216360	CC2D2A
221	COACH syndrome 216360	RPGRIP1L
221	COACH syndrome 216360	TMEM67
222	CODAS syndrome 600373	LONP1
223	COMMAD syndrome 617306	MITF
224	COPD rate of decline of lung function in 606963	MMP1
224	{Pulmonary disease chronic obstructive susceptibility to} 606963	HMOX1
225	CPT II deficiency infantile 600649	CPT2
225	CPT II deficiency lethal neonatal 608836	CPT2
225	CPT II deficiency myopathic stress-induced 255110	CPT2
225	CPT deficiency hepatic type IA 255120	CPT1A
226	Caffey disease 114000	COL1A1
227	Calcification of joints and arteries 211800	NT5E
228	Campomelic dysplasia 114290	SOX9
228	Campomelic dysplasia with autosomal sex reversal 114290	SOX9
228	Acampomelic campomelic dysplasia 114290	SOX9
229	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome 208250	PRG4
230	Camurati-Engelmann disease 131300	TGFB1
231	Canavan disease 271900	ASPA
232	Candidiasis familial 2 autosomal recessive 212050	CARD9
232	Candidiasis familial 4 autosomal recessive 613108	CLEC7A
232	Candidiasis familial 9 616445	IL17RC
233	Capillary malformation-arteriovenous malformation 608354	RASA1
233	Capillary malformations congenital 1 somatic mosaic 163000	GNAQ
234	Carbamoylphosphate synthetase I deficiency 237300	CPS1
235	Carboxypeptidase N deficiency 212070	CPN1
236	Carcinoid tumors intestinal 114900	SDHD
237	Cardiac conduction defect nonspecific 612838	SCN1B
237	{Cardiac conduction defect susceptibility to} 115080	AKAP10
238	Cardiac valvular defect developmental 212093	PLD1
238	Cardiac valvular dysplasia X-linked 314400	FLNA
239	Cardioencephalomyopathy fatal infantile due to cytochrome c oxidase deficiency 1 604377	SCO2
239	Cardioencephalomyopathy fatal infantile due to cytochrome c oxidase deficiency 2 615119	COX15
239	Cardioencephalomyopathy fatal infantile due to cytochrome c oxidase deficiency 4 616501	COA6
240	Cardiofaciocutaneous syndrome 115150	BRAF
240	Cardiofaciocutaneous syndrome 2 615278	KRAS
240	Cardiofaciocutaneous syndrome 3 615279	MAP2K1
240	Cardiofaciocutaneous syndrome 4 615280	MAP2K2
241	Cardiomyopathy dilated 1A 115200	LMNA
241	Cardiomyopathy dilated 1AA with or without LVNC 612158	ACTN2

241	Cardiomyopathy dilated 1BB 612877	DSG2
241	Cardiomyopathy dilated 1C with or without LVNC 601493	LDB3
241	Cardiomyopathy dilated 1CC 613122	NEXN
241	Cardiomyopathy dilated 1D 601494	TNNT2
241	Cardiomyopathy dilated 1DD 613172	RBM20
241	Cardiomyopathy dilated 1E 601154	SCN5A
241	Cardiomyopathy dilated 1EE 613252	MYH6
241	Cardiomyopathy dilated 1FF 613286	TNNI3
241	Cardiomyopathy dilated 1G 604145	TTN
241	Cardiomyopathy dilated 1GG 613642	SDHA
241	Cardiomyopathy dilated 1HH 613881	BAG3
241	Cardiomyopathy dilated 1I 604765	DES
241	Cardiomyopathy dilated 1II 615184	CRYAB
241	Cardiomyopathy dilated 1J 605362	EYA4
241	Cardiomyopathy dilated 1JJ 615235	LAMA4
241	Cardiomyopathy dilated 1KK 615248	MYPN
241	Cardiomyopathy dilated 1L 606685	SGCD
241	Cardiomyopathy dilated 1LL 615373	PRDM16
241	Cardiomyopathy dilated 1MM 615396	MYBPC3
241	Cardiomyopathy dilated 1NN 615916	RAF1
241	Cardiomyopathy dilated 1O 608569	ABCC9
241	Cardiomyopathy dilated 1P 609909	PLN
241	Cardiomyopathy dilated 1R 613424	ACTC1
241	Cardiomyopathy dilated 1S 613426	MYH7
241	Cardiomyopathy dilated 1U 613694	PSEN1
241	Cardiomyopathy dilated 1V 613697	PSEN2
241	Cardiomyopathy dilated 1W 611407	VCL
241	Cardiomyopathy dilated 1X 611615	FKTN
241	Cardiomyopathy dilated 1Y 611878	TPM1
241	Cardiomyopathy dilated 1Z 611879	TNNC1
241	Cardiomyopathy dilated 3B 302045	DMD
241	Cardiomyopathy dilated with woolly hair and keratoderma 605676	DSP
241	Cardiomyopathy familial hypertrophic 192600	CAV3
241	Cardiomyopathy familial hypertrophic 26	FLNC
241	Cardiomyopathy familial hypertrophic 9 613765	TTN
241	Cardiomyopathy familial restrictive 1 115210	TNNI3
241	Cardiomyopathy familial restrictive 3 612422	TNNT2
241	Cardiomyopathy familial restrictive 4 615248	MYPN
241	Cardiomyopathy familial restrictive 5 617047	FLNC
241	Cardiomyopathy hypertrophic 1 192600	MYH7
241	Cardiomyopathy hypertrophic 1 digenic 192600	MYLK2
241	Cardiomyopathy hypertrophic 10 608758	MYL2
241	Cardiomyopathy hypertrophic 11 612098	ACTC1
241	Cardiomyopathy hypertrophic 12 612124	CSRP3

241	Cardiomyopathy hypertrophic 13 613243	TNNC1
241	Cardiomyopathy hypertrophic 14 613251	MYH6
241	Cardiomyopathy hypertrophic 15 613255	VCL
241	Cardiomyopathy hypertrophic 16 613838	MYOZ2
241	Cardiomyopathy hypertrophic 17 613873	JPH2
241	Cardiomyopathy hypertrophic 18 613874	PLN
241	Cardiomyopathy hypertrophic 2 115195	TNNT2
241	Cardiomyopathy hypertrophic 20 613876	NEXN
241	Cardiomyopathy hypertrophic 22 615248	MYPN
241	Cardiomyopathy hypertrophic 23 with or without LVNC 612158	ACTN2
241	Cardiomyopathy hypertrophic 24 601493	LDB3
241	Cardiomyopathy hypertrophic 25 607487	TCAP
241	Cardiomyopathy hypertrophic 3 115196	TPM1
241	Cardiomyopathy hypertrophic 4 115197	MYBPC3
241	Cardiomyopathy hypertrophic 6 600858	PRKAG2
241	Cardiomyopathy hypertrophic 7 613690	TNNI3
241	Cardiomyopathy hypertrophic 8 608751	MYL3
242	Cardiospondylocarpofacial syndrome 157800	MAP3K7
243	Carey-Fineman-Ziter syndrome 254940	MYMK
244	Carney complex type 1 160980	PRKAR1A
244	Carney complex variant 608837	MYH8
245	Carnitine deficiency systemic primary 212140	SLC22A5
246	Carnitine-acylcarnitine translocase deficiency 212138	SLC25A20
247	Carotid intimal medial thickness 1 609338	PPARG
248	Carpal tunnel syndrome familial 115430	TTR
249	Carpenter syndrome 2 614976	MEGF8
249	Carpenter syndrome 201000	RAB23
250	Cartilage-hair hypoplasia 250250	RMRP
251	Cataract 1 multiple types 116200	GJA8
251	Cataract 10 multiple types 600881	CRYBA1
251	Cataract 11 multiple types 610623	PITX3
251	Cataract 11 syndromic 610623	PITX3
251	Cataract 12 multiple types 611597	BFSP2
251	Cataract 13 with adult i phenotype 116700	GCNT2
251	Cataract 14 multiple types 601885	GJA3
251	Cataract 15 multiple types 615274	MIP
251	Cataract 16 multiple types 613763	CRYAB
251	Cataract 17 multiple types 611544	CRYBB1
251	Cataract 18 autosomal recessive 610019	FYCO1
251	Cataract 19 multiple types 615277	LIM2
251	Cataract 2 multiple types 604307	CRYGC
251	Cataract 20 multiple types 116100	CRYGS
251	Cataract 21 multiple types 610202	MAF
251	Cataract 22 609741	CRYBB3
251	Cataract 23 610425	CRYBA4

251	Cataract 3 multiple types 601547	CRYBB2
251	Cataract 30 pulverulent 116300	VIM
251	Cataract 31 multiple types 605387	CHMP4B
251	Cataract 33 multiple types 611391	BFSP1
251	Cataract 34 multiple types 612968	FOXE3
251	Cataract 36 613887	TDRD7
251	Cataract 38 autosomal recessive 614691	AGK
251	Cataract 39 multiple types autosomal dominant 615188	CRYGB
251	Cataract 4 multiple types 115700	CRYGD
251	Cataract 40 X-linked 302200	NHS
251	Cataract 44 616509	LSS
251	Cataract 46 juvenile-onset 212500	LEMD2
251	Cataract 47 juvenile with microcornea 612018	SLC16A12
251	Cataract 5 multiple types 116800	HSF4
251	Cataract 6 multiple types 116600	EPHA2
251	Cataract 9 multiple types 604219	CRYAA
251	Cataract with late-onset corneal dystrophy 106210	PAX6
251	Aniridia 106210	PAX6
252	Catel-Manzke syndrome 616145	TGDS
253	Caudal regression syndrome 600145	VANGL1
254	Cavitary optic disc anomalies 611543	MMP19
255	Cenani-Lenz syndactyly syndrome 212780	LRP4
256	Central core disease 117000	RYR1
256	Neuromuscular disease congenital with uniform type 1 fiber 117000	RYR1
257	Central hypoventilation syndrome 209880	GDNF
257	Central hypoventilation syndrome congenital 209880	ASCL1
257	Central hypoventilation syndrome congenital 209880	BDNF
257	Central hypoventilation syndrome congenital 209880	EDN3
257	Central hypoventilation syndrome congenital 209880	RET
257	Central hypoventilation syndrome congenital with or without Hirschsprung disease 209880	PHOX2B
257	Haddad syndrome 209880	ASCL1
258	Centronuclear myopathy 1 160150	DNM2
258	Centronuclear myopathy 2 255200	BIN1
258	Centronuclear myopathy 3 614408	MYF6
258	Centronuclear myopathy 5 615959	SPEG
258	Centronuclear myopathy 6 with fiber-type disproportion 617760	ZAK
258	{Centronuclear myopathy autosomal modifier of} 160150	MTMR14
259	Cerebellar ataxia 604290	CP
259	Hemosiderosis systemic due to aceruloplasminemia 604290	CP
259	Cerebellar ataxia and hypogonadotropic hypogonadism 212840	RNF216
259	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3 613227	CA8

259	Cerebellar ataxia deafness and narcolepsy autosomal dominant 604121	DNMT1
259	Cerebellar ataxia mental retardation and dysequilibrium syndrome 2 610185	WDR81
259	Cerebellar ataxia nonprogressive with mental retardation 614756	CAMTA1
259	Cerebellar atrophy visual impairment and psychomotor retardation 616875	EMC1
259	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1 224050	VLDLR
260	Cerebellofaciodental syndrome 616202	BRF1
261	Cerebral amyloid angiopathy 105150	CST3
261	Cerebral amyloid angiopathy Dutch Italian Iowa Flemish Arctic variants 605714	APP
261	Cerebral amyloid angiopathy PRNP-related 137440	PRNP
261	Gerstmann-Straussler disease 137440	PRNP
262	Cerebral arteriopathy autosomal dominant with subcortical infarcts and leukoencephalopathy type 2 616779	HTRA1
262	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1 125310	NOTCH3
263	Cerebral cavernous malformations 3 603285	PDCD10
263	Cerebral cavernous malformations-1 116860	KRIT1
263	Cerebral cavernous malformations-2 603284	CCM2
263	Cavernous malformations of CNS and retina 116860	KRIT1
263	Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations 116860	KRIT1
264	Cerebral creatine deficiency syndrome 1 300352	SLC6A8
264	Cerebral creatine deficiency syndrome 2 612736	GAMT
264	Cerebral creatine deficiency syndrome 3 612718	GATM
265	Cerebral dysgenesis neuropathy ichthyosis and palmoplantar keratoderma syndrome 609528	SNAP29
266	Cerebral palsy spastic quadriplegic 2 612900	KANK1
266	Cerebral palsy spastic quadriplegic 3 617008	ADD3
267	{Cerebral infarction susceptibility to} 601367	PRKCH
267	{Ischemic stroke susceptibility to} 601367	NOS3
267	{Stroke ischemic susceptibility to} 601367	F2
267	{Stroke ischemic susceptibility to} 601367	F5
267	{Stroke susceptibility to} 601367	ALOX5AP
267	{Stroke susceptibility to 1} 606799	PDE4D
267	{Stroke susceptibility to} 601367	ALOX5AP
268	Cerebrocostomandibular syndrome 117650	SNRPB
269	Cerebrooculofacioskeletal syndrome 1 214150	ERCC6
269	Cerebrooculofacioskeletal syndrome 3 616570	ERCC5
269	Cerebrooculofacioskeletal syndrome 4 610758	ERCC1
270	Cerebroretinal microangiopathy with calcifications and cysts 2 617341	STN1

270	Cerebroretinal microangiopathy with calcifications and cysts 612199	CTC1
271	Cerebrotendinous xanthomatosis 213700	CYP27A1
272	Ceroid lipofuscinosis neuronal 1 256730	PPT1
272	Ceroid lipofuscinosis neuronal 10 610127	CTSD
272	Ceroid lipofuscinosis neuronal 11 614706	GRN
272	Ceroid lipofuscinosis neuronal 13 Kufs type 615362	CTSF
272	Ceroid lipofuscinosis neuronal 2 204500	TPP1
272	Ceroid lipofuscinosis neuronal 3 204200	CLN3
272	Ceroid lipofuscinosis neuronal 4 Parry type 162350	DNAJC5
272	Ceroid lipofuscinosis neuronal 5 256731	CLN5
272	Ceroid lipofuscinosis neuronal 6 601780	CLN6
272	Ceroid lipofuscinosis neuronal 7 610951	MFSD8
272	Ceroid lipofuscinosis neuronal 8 600143	CLN8
272	Ceroid lipofuscinosis neuronal 8 Northern epilepsy variant 610003	CLN8
272	Ceroid lipofuscinosis neuronal Kufs type adult onset 204300	CLN6
273	Cervical cancer somatic 603956	FGFR3
274	Chanarin-Dorfman syndrome 275630	ABHD5
275	Char syndrome 169100	TFAP2B
276	Charcot-Marie-Tooth disease X-linked recessive 5 311070	PRPS1
276	Charcot-Marie-Tooth disease axonal type 20 614228	DYNC1H1
276	Charcot-Marie-Tooth disease axonal type 2A2A 609260	MFN2
276	Charcot-Marie-Tooth disease axonal type 2A2B 617087	MFN2
276	Charcot-Marie-Tooth disease axonal type 2CC 616924	NEFH
276	Charcot-Marie-Tooth disease axonal type 2F 606595	HSPB1
276	Charcot-Marie-Tooth disease axonal type 2K 607831	GDAP1
276	Charcot-Marie-Tooth disease axonal type 2L 608673	HSPB8
276	Charcot-Marie-Tooth disease axonal type 2M 606482	DNM2
276	Charcot-Marie-Tooth disease axonal type 2N 613287	AARS
276	Charcot-Marie-Tooth disease axonal type 2P 614436	LRSAM1
276	Charcot-Marie-Tooth disease axonal type 2S 616155	IGHMBP2
276	Charcot-Marie-Tooth disease axonal type 2T 617017	MME
276	Charcot-Marie-Tooth disease axonal type 2U 616280	MARS
276	Charcot-Marie-Tooth disease axonal type 2W 616625	HARS
276	Charcot-Marie-Tooth disease axonal type 2X 616668	SPG11
276	Charcot-Marie-Tooth disease axonal type 2Z 616688	MORC2
276	Charcot-Marie-Tooth disease axonal with vocal cord paresis 607706	GDAP1
276	Charcot-Marie-Tooth disease dominant intermediate B 606482	DNM2
276	Charcot-Marie-Tooth disease dominant intermediate C 608323	YARS
276	Charcot-Marie-Tooth disease dominant intermediate D 607791	MPZ
276	Charcot-Marie-Tooth disease dominant intermediate E 614455	INF2
276	Charcot-Marie-Tooth disease dominant intermediate F 615185	GNB4
276	Charcot-Marie-Tooth disease foot deformity of 192950	HOXD10

276	Charcot-Marie-Tooth disease recessive intermediate A 608340	GDAP1
276	Charcot-Marie-Tooth disease recessive intermediate C 615376	PLEKHG5
276	Charcot-Marie-Tooth disease recessive intermediate D 616039	COX6A1
276	Charcot-Marie-Tooth disease type 1A 118220	PMP22
276	Charcot-Marie-Tooth disease type 1B 118200	MPZ
276	Charcot-Marie-Tooth disease type 1C 601098	LITAF
276	Charcot-Marie-Tooth disease type 1D 607678	EGR2
276	Charcot-Marie-Tooth disease type 1E 118300	PMP22
276	Charcot-Marie-Tooth disease type 1F 607734	NEFL
276	Charcot-Marie-Tooth disease type 2B 600882	RAB7A
276	Charcot-Marie-Tooth disease type 2B1 605588	LMNA
276	Charcot-Marie-Tooth disease type 2D 601472	GARS
276	Charcot-Marie-Tooth disease type 2E 607684	NEFL
276	Charcot-Marie-Tooth disease type 2I 607677	MPZ
276	Charcot-Marie-Tooth disease type 2J 607736	MPZ
276	Charcot-Marie-Tooth disease type 2R 615490	TRIM2
276	Charcot-Marie-Tooth disease type 2Y 616687	VCP
276	Charcot-Marie-Tooth disease type 4A 214400	GDAP1
276	Charcot-Marie-Tooth disease type 4B1 601382	MTMR2
276	Charcot-Marie-Tooth disease type 4B2 604563	SBF2
276	Charcot-Marie-Tooth disease type 4B3 615284	SBF1
276	Charcot-Marie-Tooth disease type 4C 601596	SH3TC2
276	Charcot-Marie-Tooth disease type 4D 601455	NDRG1
276	Charcot-Marie-Tooth disease type 4F 614895	PRX
276	Charcot-Marie-Tooth disease type 4H 609311	FGD4
276	Charcot-Marie-Tooth disease type 4J 611228	FIG4
276	Charcot-Marie-Tooth disease type 4K 616684	SURF1
276	Charcot-Marie-Tooth neuropathy X-linked dominant 1 302800	GJB1
276	Vertical talus congenital 192950	HOXD10
277	Chediak-Higashi syndrome 214500	LYST
278	Cherubism 118400	SH3BP2
279	Chilblain lupus 610448	TREX1
280	Chitayat syndrome 617180	ERF
281	Choanal atresia and lymphedema 613611	PTPN14
282	Cholestasis benign recurrent intrahepatic 2 605479	ABCB11
282	Cholestasis benign recurrent intrahepatic 243300	ATP8B1
282	Cholestasis intrahepatic of pregnancy 1 147480	ATP8B1
282	Cholestasis intrahepatic of pregnancy 3 614972	ABCB4
282	Cholestasis progressive familial intrahepatic 1 211600	ATP8B1
282	Cholestasis progressive familial intrahepatic 2 601847	ABCB11
282	Cholestasis progressive familial intrahepatic 3 602347	ABCB4
282	Cholestasis progressive familial intrahepatic 4 615878	TJP2
282	Cholestasis progressive familial intrahepatic 5 617049	NR1H4
283	Chondrocalcinosis 2 118600	ANKH
284	Chondrodysplasia Blomstrand type 215045	PTH1R

285	Chondrodysplasia Grebe type 200700	GDF5
286	Chondrodysplasia punctata X-linked dominant 302960	EBP
286	Chondrodysplasia punctata X-linked recessive 302950	ARSE
287	Chondrodysplasia with joint dislocations GPAPP type 614078	IMPAD1
288	Chondrosarcoma 215300	EXT1
288	Chondrosarcoma extraskeletal myxoid 612237	NR4A3
289	Chorea hereditary benign 118700	NKX2-1
290	Choreoacanthocytosis 200150	VPS13A
291	Choreoathetosis hypothyroidism and neonatal respiratory distress 610978	NKX2-1
292	Choroid plexus papilloma 260500	TP53
293	Choroidal dystrophy central areolar 2 613105	PRPH2
294	Choroideremia 303100	CHM
295	Chronic atrial and intestinal dysrhythmia 616201	SGOL1
296	Chronic granulomatous disease X-linked 306400	CYBB
296	Chronic granulomatous disease autosomal due to deficiency of CYBA 233690	CYBA
296	Chronic granulomatous disease due to deficiency of NCF-1 233700	NCF1
296	Chronic granulomatous disease due to deficiency of NCF-2 233710	NCF2
297	Chudley-McCullough syndrome 604213	GPSM2
298	Chylomicron retention disease 246700	SAR1B
299	Ciliary dyskinesia primary 1 with or without situs inversus 244400	DNAI1
299	Ciliary dyskinesia primary 10 612518	DNAAF2
299	Ciliary dyskinesia primary 11 612649	RSPH4A
299	Ciliary dyskinesia primary 12 612650	RSPH9
299	Ciliary dyskinesia primary 13 613193	DNAAF1
299	Ciliary dyskinesia primary 14 613807	CCDC39
299	Ciliary dyskinesia primary 15 613808	CCDC40
299	Ciliary dyskinesia primary 16 614017	DNAL1
299	Ciliary dyskinesia primary 17 614679	CCDC103
299	Ciliary dyskinesia primary 18 614874	DNAAF5
299	Ciliary dyskinesia primary 19 614935	LRRC6
299	Ciliary dyskinesia primary 2 606763	DNAAF3
299	Ciliary dyskinesia primary 20 615067	CCDC114
299	Ciliary dyskinesia primary 21 615294	DRC1
299	Ciliary dyskinesia primary 22 615444	ZMYND10
299	Ciliary dyskinesia primary 23 615451	ARMC4
299	Ciliary dyskinesia primary 24 615481	RSPH1
299	Ciliary dyskinesia primary 25 615482	DNAAF4
299	Ciliary dyskinesia primary 26 615500	C21orf59
299	Ciliary dyskinesia primary 27 615504	CCDC65
299	Ciliary dyskinesia primary 28 615505	SPAG1
299	Ciliary dyskinesia primary 29 615872	CCNO

299	Ciliary dyskinesia primary 3 with or without situs inversus 608644	DNAH5
299	Ciliary dyskinesia primary 30 616037	CCDC151
299	Ciliary dyskinesia primary 32 616481	RSPH3
299	Ciliary dyskinesia primary 33 616726	GAS8
299	Ciliary dyskinesia primary 34 617091	DNAJB13
299	Ciliary dyskinesia primary 35 617092	TTC25
299	Ciliary dyskinesia primary 36 X-linked 300991	PIH1D3
299	Ciliary dyskinesia primary 5 608647	HYDIN
299	Ciliary dyskinesia primary 6 610852	NME8
299	Ciliary dyskinesia primary 7 with or without situs inversus 611884	DNAH11
299	Ciliary dyskinesia primary 9 with or without situs inversus 612444	DNAI2
300	Cirrhosis cryptogenic 215600	KRT18
300	Cirrhosis cryptogenic 215600	KRT8
300	{Cirrhosis noncryptogenic susceptibility to} 215600	KRT18
300	{Cirrhosis noncryptogenic susceptibility to} 215600	KRT8
301	Citrullinemia 215700	ASS1
301	Citrullinemia adult-onset type II 603471	SLC25A13
301	Citrullinemia type II neonatal-onset 605814	SLC25A13
302	Cleft lip or palate-ectodermal dysplasia syndrome 225060	NECTIN1
302	Cleft palate cardiac defects and mental retardation 600987	MEIS2
302	Cleft palate psychomotor retardation and distinctive facial features 616728	KDM1A
302	Cleft palate with ankyloglossia 303400	TBX22
303	Cleidocranial dysplasia 119600	RUNX2
303	Cleidocranial dysplasia forme fruste dental anomalies only 119600	RUNX2
303	Cleidocranial dysplasia forme fruste with brachydactyly 119600	RUNX2
304	Clopidogrel impaired responsiveness to 609535	CYP2C19
304	Proguanil poor metabolizer 609535	CYP2C19
305	Clubfoot congenital with or without deficiency of long bones and or mirror-image polydactyly 119800	PITX1
306	Cockayne syndrome type A 216400	ERCC8
306	Cockayne syndrome type B 133540	ERCC6
307	Cocoon syndrome 613630	CHUK
308	Coenzyme Q10 deficiency primary 1 607426	COQ2
308	Coenzyme Q10 deficiency primary 2 614651	PDSS1
308	Coenzyme Q10 deficiency primary 3 614652	PDSS2
308	Coenzyme Q10 deficiency primary 4 612016	ADCK3
308	Coenzyme Q10 deficiency primary 5 614654	COQ9
308	Coenzyme Q10 deficiency primary 6 614650	COQ6
308	Coenzyme Q10 deficiency primary 7 616276	COQ4
309	Coffin-Lowry syndrome 303600	RPS6KA3
310	Coffin-Siris syndrome 1 135900	ARID1B
310	Coffin-Siris syndrome 2 614607	ARID1A
310	Coffin-Siris syndrome 3 614608	SMARCB1

310	Coffin-Siris syndrome 4 614609	SMARCA4
310	Coffin-Siris syndrome 5 616938	SMARCE1
311	Cohen syndrome 216550	VPS13B
312	Cohen-Gibson syndrome 617561	EED
313	Cold-induced sweating syndrome 1 272430	CRLF1
313	Cold-induced sweating syndrome 2 610313	CLCF1
313	Cold-induced sweating syndrome 3 617055	KLHL7
314	Cole disease 615522	ENPP1
315	Cole-Carpenter syndrome 1 112240	P4HB
315	Cole-Carpenter syndrome 2 616294	SEC24D
316	Coloboma ocular 120433	YAP1
316	Coloboma ocular with or without hearing impairment cleft lip palate and or mental retardation 120433	YAP1
317	Colorblindness deutan 303800	OPN1MW
317	Colorblindness protan 303900	OPN1LW
317	Colorblindness tritan 190900	OPN1SW
318	Colorectal adenomatous polyposis autosomal recessive with pilomatricomas 132600	MUTYH
318	Pilomatricoma somatic 132600	CTNNB1
318	Colorectal cancer 114500	TP53
318	Colorectal cancer hereditary nonpolyposis type 1 120435	MSH2
318	Colorectal cancer hereditary nonpolyposis type 2 609310	MLH1
318	Colorectal cancer hereditary nonpolyposis type 4 614337	PMS2
318	Colorectal cancer hereditary nonpolyposis type 5 614350	MSH6
318	Colorectal cancer hereditary nonpolyposis type 6 614331	TGFBR2
318	Colorectal cancer hereditary nonpolyposis type 7 614385	MLH3
318	Colorectal cancer hereditary nonpolyposis type 8 613244	EPCAM
318	Colorectal cancer somatic 114500	AKT1
318	Colorectal cancer somatic 114500	APC
318	Colorectal cancer somatic 114500	AXIN2
318	Colorectal cancer somatic 114500	BAX
318	Colorectal cancer somatic 114500	BUB1B
318	Colorectal cancer somatic 114500	CTNNB1
318	Colorectal cancer somatic 114500	DCC
318	Colorectal cancer somatic 114500	DLC1
318	Colorectal cancer somatic 114500	EP300
318	Colorectal cancer somatic 114500	FGFR3
318	Colorectal cancer somatic 114500	FLCN
318	Colorectal cancer somatic 114500	MCC
318	Colorectal cancer somatic 114500	MLH3
318	Colorectal cancer somatic 114500	NRAS
318	Colorectal cancer somatic 114500	PDGFRL
318	Colorectal cancer somatic 114500	PIK3CA
318	Colon cancer advanced somatic 114500	SRC
318	Colon cancer somatic 114500	PTPN12

318	Colon cancer somatic 114500	PTPRJ
318	Colon cancer somatic 114500	RAD54B
318	{Colon cancer susceptibility to} 114500	AURKA
318	{Colonic adenoma recurrence reduced risk of} 114500	ODC1
318	{Colorectal cancer susceptibility to 10} 612591	POLD1
318	{Colorectal cancer susceptibility to 12} 615083	POLE
318	{Colorectal cancer susceptibility to 1} 608812	GALNT12
318	{Colorectal cancer susceptibility to 3} 612229	SMAD7
318	{Colorectal cancer susceptibility to} 114500	CCND1
318	{Colorectal cancer susceptibility to} 114500	TLR2
318	{Colorectal cancer} 114500	PLA2G2A
318	Colonic Adenocarcinoma somatic	RAD54L
318	Colorectal cancer with chromosomal instability somatic	BUB1
318	Colorectal cancer somatic	BRAF
318	{Breast and colorectal cancer, susceptibility to}	CHEK2
318	{Breast cancer, poor survival after chemotherapy for}	NQO1
319	Combined D-2- and L-2-hydroxyglutaric aciduria 615182	SLC25A1
320	Combined SAP deficiency 611721	PSAP
321	Combined cellular and humoral immune defects with granulomas 233650	RAG1
321	Combined cellular and humoral immune defects with granulomas 233650	RAG2
322	Combined factor V and VIII deficiency 227300	LMAN1
323	Combined hyperlipidemia familial 144250	LPL
323	{Hyperlipidemia familial combined susceptibility to} 602491	USF1
324	Combined immunodeficiency X-linked moderate 312863	IL2RG
324	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia 617780	MTHFD1
325	Combined malonic and methylmalonic aciduria 614265	ACSF3
326	Combined oxidative phosphorylation deficiency 1 609060	GFM1
326	Combined oxidative phosphorylation deficiency 10 614702	MTO1
326	Combined oxidative phosphorylation deficiency 11 614922	RMND1
326	Combined oxidative phosphorylation deficiency 12 614924	EARS2
326	Combined oxidative phosphorylation deficiency 13 614932	PNPT1
326	Combined oxidative phosphorylation deficiency 14 614946	FARS2
326	Combined oxidative phosphorylation deficiency 15 614947	MTFMT
326	Combined oxidative phosphorylation deficiency 17 615440	ELAC2
326	Combined oxidative phosphorylation deficiency 18 615578	SFXN4
326	Combined oxidative phosphorylation deficiency 2 610498	MRPS16
326	Combined oxidative phosphorylation deficiency 20 615917	VARS2
326	Combined oxidative phosphorylation deficiency 23 616198	GTPBP3
326	Combined oxidative phosphorylation deficiency 24 616239	NARS2
326	Combined oxidative phosphorylation deficiency 26 616539	TRMT5
326	Combined oxidative phosphorylation deficiency 27 616672	CARS2
326	Combined oxidative phosphorylation deficiency 28 616794	SLC25A26

326	Combined oxidative phosphorylation deficiency 3 610505	TSFM
326	Combined oxidative phosphorylation deficiency 30 616974	TRMT10C
326	Combined oxidative phosphorylation deficiency 31 617228	MIPEP
326	Combined oxidative phosphorylation deficiency 32 617664	MRPS34
326	Combined oxidative phosphorylation deficiency 33 617713	C1QBP
326	Combined oxidative phosphorylation deficiency 4 610678	TUFM
326	Combined oxidative phosphorylation deficiency 5 611719	MRPS22
326	Combined oxidative phosphorylation deficiency 6 300816	AIFM1
326	Combined oxidative phosphorylation deficiency 7 613559	C12orf65
326	Combined oxidative phosphorylation deficiency 8 614096	AARS2
326	Combined oxidative phosphorylation deficiency 9 614582	MRPL3
327	Complement component 4 partial deficiency of 120790	SERPING1
328	Complement factor D deficiency 613912	CFD
328	Complement factor H deficiency 609814	CFH
328	Complement factor I deficiency 610984	CFI
329	Complement hyperactivation angiopathic thrombosis and protein-losing enteropathy 226300	CD55
330	Cone dystrophy 4 613093	PDE6C
330	Cone dystrophy-3 602093	GUCA1A
330	Cone-rod dystrophy 10 610283	SEMA4A
330	Cone-rod dystrophy 11 610381	RAX2
330	Cone-rod dystrophy 12 612657	PROM1
330	Cone-rod dystrophy 13 608194	RPGRIP1
330	Cone-rod dystrophy 14 602093	GUCA1A
330	Cone-rod dystrophy 15 613660	CDHR1
330	Cone-rod dystrophy 16 614500	C8orf37
330	Cone-rod dystrophy 18 615374	RAB28
330	Cone-rod dystrophy 19 615860	TTLL5
330	Cone-rod dystrophy 20 615973	POC1B
330	Cone-rod dystrophy 21 616502	DRAM2
330	Cone-rod dystrophy 3 604116	ABCA4
330	Cone-rod dystrophy 5 600977	PITPNM3
330	Cone-rod dystrophy 6 601777	GUCY2D
330	Cone-rod dystrophy 604393	AIPL1
330	Cone-rod dystrophy 7 603649	RIMS1
330	Cone-rod dystrophy 9 612775	ADAM9
330	Cone-rod dystrophy X-linked 1 304020	RPGR
330	Cone-rod dystrophy X-linked 3 300476	CACNA1F
330	Cone-rod dystrophy and hearing loss 617236	CEP78
330	Cone-rod retinal dystrophy-2 120970	CRX
330	Cone-rod synaptic disorder congenital nonprogressive 610427	CABP4
331	Congenital anomalies of kidney and urinary tract 2 143400	TBX18
331	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss abnormal ears or developmental delay 617641	PBX1

331	{Congenital anomalies of kidney and urinary tract susceptibility to} 610805	DSTYK
332	Congenital bilateral absence of vas deferens 277180	CFTR
333	Congenital cataracts facial dysmorphism and neuropathy 604168	CTDP1
333	Congenital cataracts hearing loss and neurodegeneration 614482	SLC33A1
334	Congenital contractures of the limbs and face hypotonia and developmental delay 616266	NALCN
335	Congenital disorder of deglycosylation 615273	NGLY1
335	Congenital disorder of glycosylation type IIa 212066	MGAT2
335	Congenital disorder of glycosylation type IIb 606056	MOGS
335	Congenital disorder of glycosylation type IIc 266265	SLC35C1
335	Congenital disorder of glycosylation type IId 607091	B4GALT1
335	Congenital disorder of glycosylation type IIe 608779	COG7
335	Congenital disorder of glycosylation type IIf 603585	SLC35A1
335	Congenital disorder of glycosylation type IIg 611209	COG1
335	Congenital disorder of glycosylation type IIh 611182	COG8
335	Congenital disorder of glycosylation type IIi 613612	COG5
335	Congenital disorder of glycosylation type IIj 613489	COG4
335	Congenital disorder of glycosylation type IIk 614727	TMEM165
335	Congenital disorder of glycosylation type IIl 614576	COG6
335	Congenital disorder of glycosylation type IIm 300896	SLC35A2
335	Congenital disorder of glycosylation type IIn 616721	SLC39A8
335	Congenital disorder of glycosylation type IIo 616828	CCDC115
335	Congenital disorder of glycosylation type IIp 616829	TMEM199
335	Congenital disorder of glycosylation type Ia 212065	PMM2
335	Congenital disorder of glycosylation type Ib 602579	MPI
335	Congenital disorder of glycosylation type Ic 603147	ALG6
335	Congenital disorder of glycosylation type Id 601110	ALG3
335	Congenital disorder of glycosylation type Ie 608799	DPM1
335	Congenital disorder of glycosylation type If 609180	MPDU1
335	Congenital disorder of glycosylation type Ig 607143	ALG12
335	Congenital disorder of glycosylation type Ih 608104	ALG8
335	Congenital disorder of glycosylation type Ij 608093	DPAGT1
335	Congenital disorder of glycosylation type Ik 608540	ALG1
335	Congenital disorder of glycosylation type Il 608776	ALG9
335	Congenital disorder of glycosylation type Im 610768	DOLK
335	Congenital disorder of glycosylation type In 612015	RFT1
335	Congenital disorder of glycosylation type Io 612937	DPM3
335	Congenital disorder of glycosylation type Ip 613661	ALG11
335	Congenital disorder of glycosylation type Iq 612379	SRD5A3
335	Congenital disorder of glycosylation type It 614921	PGM1
335	Congenital disorder of glycosylation type Iu 615042	DPM2
335	Congenital disorder of glycosylation type Iy 300934	SSR4
336	Congenital heart defects and ectodermal dysplasia 617364	PRKD1

336	Congenital heart defects and skeletal malformations syndrome 617602	ABL1
336	Congenital heart defects dysmorphic facial features and intellectual developmental disorder 617360	CDK13
336	Congenital heart defects multiple types 4 615779	NR2F2
336	Congenital heart defects nonsyndromic 1 X-linked 306955	ZIC3
336	Congenital heart defects nonsyndromic 2 614980	TAB2
337	Congenital myopathy with excess of muscle spindles 218040	HRAS
337	Costello syndrome 218040	HRAS
338	Congenital short bowel syndrome 300048	FLNA
338	Intestinal pseudoobstruction neuronal 300048	FLNA
338	Congenital short bowel syndrome 615237	CLMP
339	Conotruncal anomaly face syndrome 217095	TBX1
339	Conotruncal heart malformations 217095	NKX2-6
339	Conotruncal heart malformations variable 217095	NKX2-5
339	Persistent truncus arteriosus 217095	GATA6
339	Persistent truncus arteriosus 217095	NKX2-6
339	Double-outlet right ventricle 217095	GDF1
340	Contractural arachnodactyly congenital 121050	FBN2
341	Convulsions familial infantile with paroxysmal choreoathetosis 602066	PRRT2
342	Cornea plana 2 autosomal recessive 217300	KERA
343	Corneal dystrophy Avellino type 607541	TGFBI
343	Corneal dystrophy Fuchs endothelial 1 136800	COL8A2
343	Corneal dystrophy Fuchs endothelial 3 613267	TCF4
343	Corneal dystrophy Fuchs endothelial 4 613268	SLC4A11
343	Corneal dystrophy Fuchs endothelial 6 613270	ZEB1
343	Corneal dystrophy Fuchs endothelial 8 615523	AGBL1
343	Corneal dystrophy Groenouw type I 121900	TGFBI
343	Corneal dystrophy Reis-Bucklers type 608470	TGFBI
343	Corneal dystrophy Schnyder type 121800	UBIAD1
343	Corneal dystrophy Thiel-Behnke type 602082	TGFBI
343	Corneal dystrophy congenital stromal 610048	DCN
343	Corneal dystrophy epithelial basement membrane 121820	TGFBI
343	Corneal dystrophy gelatinous drop-like 204870	TACSTD2
343	Corneal dystrophy lattice type I 122200	TGFBI
343	Corneal dystrophy lattice type IIIA 608471	TGFBI
343	Corneal dystrophy posterior polymorphous 1 122000	OVOL2
343	Corneal dystrophy posterior polymorphous 2 609140	COL8A2
343	Corneal dystrophy posterior polymorphous 3 609141	ZEB1
343	Corneal endothelial dystrophy and perceptive deafness 217400	SLC4A11
343	Corneal endothelial dystrophy autosomal recessive 217700	SLC4A11
343	Corneal fleck dystrophy 121850	PIKFYVE
343	Corneal clouding autosomal recessive	APOA1
344	Cornelia de Lange syndrome 1 122470	NIPBL

344	Cornelia de Lange syndrome 2 300590	SMC1A
344	Cornelia de Lange syndrome 3 610759	SMC3
344	Cornelia de Lange syndrome 4 614701	RAD21
344	Cornelia de Lange syndrome 5 300882	HDAC8
345	Corpus callosum agenesis of with mental retardation ocular coloboma and micrognathia 300472	IGBP1
345	Corpus callosum partial agenesis of 304100	L1CAM
346	Cortical dysplasia complex with other brain malformations 1 614039	TUBB3
346	Cortical dysplasia complex with other brain malformations 2 615282	KIF5C
346	Cortical dysplasia complex with other brain malformations 3 615411	KIF2A
346	Cortical dysplasia complex with other brain malformations 4 615412	TUBG1
346	Cortical dysplasia complex with other brain malformations 5 615763	TUBB2A
346	Cortical dysplasia complex with other brain malformations 6 615771	TUBB
346	Cortical dysplasia complex with other brain malformations 7 610031	TUBB2B
346	Cortical dysplasia complex with other brain malformations 8 613180	TUBA8
346	Cortical dysplasia-focal epilepsy syndrome 610042	CNTNAP2
346	Cortical malformations occipital 614115	LAMC3
347	Corticosteroid-binding globulin deficiency 611489	CBG
348	Cortisone reductase deficiency 1 604931	H6PD
348	Cortisone reductase deficiency 2 614662	HSD11B1
349	Cousin syndrome 260660	TBX15
350	Cowchock syndrome 310490	AIFM1
351	Lhermitte-Duclos syndrome 158350	PTEN
351	Cowden syndrome 1 158350	PTEN
351	Cowden syndrome 2 612359	SDHB
351	Cowden syndrome 3 615106	SDHD
351	Cowden syndrome 4 615107	KLLN
351	Cowden syndrome 5 615108	PIK3CA
351	Cowden syndrome 6 615109	AKT1
351	Cowden syndrome 7 616858	SEC23B
352	Craniodiaphyseal dysplasia autosomal dominant 122860	SOST
352	Cranioectodermal dysplasia 1 218330	IFT122
352	Cranioectodermal dysplasia 2 613610	WDR35
352	Cranioectodermal dysplasia 3 614099	IFT43
353	Craniofacial dysmorphism skeletal anomalies and mental retardation syndrome 213980	TMCO1
354	Craniofacial-deafness-hand syndrome 122880	PAX3

355	Craniofrontonasal dysplasia 304110	EFNB1
356	Craniolenticulosutural dysplasia 607812	SEC23A
357	Craniometaphyseal dysplasia 123000	ANKH
357	Craniometaphyseal dysplasia autosomal recessive 218400	GJA1
358	Craniosynostosis 1 123100	TWIST1
358	Craniosynostosis 2 604757	MSX2
358	Craniosynostosis 3 615314	TCF12
358	Craniosynostosis 4 600775	ERF
358	Craniosynostosis 6 616602	ZIC1
358	Craniosynostosis and dental anomalies 614188	IL11RA
358	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies 614416	CYP26B1
358	{Craniosynostosis 5 susceptibility to} 615529	ALX4
358	{Craniosynostosis 7 susceptibility to} 617439	SMAD6
358	Craniosynostosis nonspecific	FGFR2
359	Creatine phosphokinase elevated serum 123320	CAV3
360	Creutzfeldt-Jakob disease 123400	PRNP
360	{Creutzfeldt-Jakob disease variant resistance to} 123400	HLA-DQB1
361	Crigler-Najjar syndrome type I 218800	UGT1A1
361	Crigler-Najjar syndrome type II 606785	UGT1A1
362	Crouzon syndrome 123500	FGFR2
362	Crouzon syndrome with acanthosis nigricans 612247	FGFR3
363	Cryohydrocytosis 185020	SLC4A1
364	Cryptorchidism 219050	INSL3
365	Culler-Jones syndrome 615849	GLI2
366	Currarino syndrome 176450	MNX1
367	Curry-Jones syndrome somatic mosaic 601707	SMO
368	Cushing syndrome ACTH-independent adrenal somatic 615830	PRKACA
369	Cutis laxa autosomal dominant 123700	ELN
369	Cutis laxa autosomal dominant 2 614434	FBLN5
369	Cutis laxa autosomal dominant 3 616603	ALDH18A1
369	Cutis laxa autosomal recessive type IA 219100	FBLN5
369	Cutis laxa autosomal recessive type IB 614437	EFEMP2
369	Cutis laxa autosomal recessive type IC 613177	LTBP4
369	Cutis laxa autosomal recessive type IIA 219200	ATP6V0A2
369	Cutis laxa autosomal recessive type IIB 612940	PYCR1
369	Cutis laxa autosomal recessive type IIC 617402	ATP6V1E1
369	Cutis laxa autosomal recessive type IID 617403	ATP6V1A
369	Cutis laxa autosomal recessive type IIIA 219150	ALDH18A1
369	Cutis laxa autosomal recessive type IIIB 614438	PYCR1
370	Cyanosis transient neonatal 613977	HBG2
371	Cylindromatosis familial 132700	CYLD
372	Cystathioninuria 219500	CTH
373	Cystic fibrosis 219700	CFTR
373	{Cystic fibrosis lung disease modifier of} 219700	TGFB1

373	{Pseudomonas aeruginosa susceptibility to chronic infection by in cystic fibrosis} 219700	FCGR2A
374	Cystinosis atypical nephropathic 219800	CTNS
374	Cystinosis late-onset juvenile or adolescent nephropathic 219900	CTNS
374	Cystinosis nephropathic 219800	CTNS
374	Cystinosis ocular nonnephropathic 219750	CTNS
374	Cystinuria 220100	SLC3A1
374	Cystinuria 220100	SLC7A9
375	Czech dysplasia 609162	COL2A1
376	D-2-hydroxyglutaric aciduria 2 613657	IDH2
376	D-2-hydroxyglutaric aciduria 600721	D2HGDH
377	D-bifunctional protein deficiency 261515	HSD17B4
378	D-glyceric aciduria 220120	GLYCTK
379	DOOR syndrome 220500	TBC1D24
380	Danon disease 300257	LAMP2
381	Darier disease 124200	ATP2A2
382	De Sanctis-Cacchione syndrome 278800	ERCC6
383	Deafness autosomal recessive 86 614617	TBC1D24
383	Deafness X-linked 1 304500	PRPS1
383	Deafness X-linked 2 304400	POU3F4
383	Deafness X-linked 4 300066	SMPX
383	Deafness X-linked 5 300614	AIFM1
383	Deafness and myopia 221200	SLITRK6
383	Deafness autosomal dominant 1 124900	DIAPH1
383	Deafness autosomal dominant 10 601316	EYA4
383	Deafness autosomal dominant 11 601317	MYO7A
383	Deafness autosomal dominant 13 601868	COL11A2
383	Deafness autosomal dominant 15 602459	POU4F3
383	Deafness autosomal dominant 17 603622	MYH9
383	Deafness autosomal dominant 20 or 26 604717	ACTG1
383	Deafness autosomal dominant 22 606346	MYO6
383	Deafness autosomal dominant 22 with hypertrophic cardiomyopathy 606346	MYO6
383	Deafness autosomal dominant 23 605192	SIX1
383	Deafness autosomal dominant 25 605583	SLC17A8
383	Deafness autosomal dominant 28 608641	GRHL2
383	Deafness autosomal dominant 2A 600101	KCNQ4
383	Deafness autosomal dominant 2B 612644	GJB3
383	Deafness autosomal dominant 34 with or without inflammation 617772	NLRP3
383	Deafness autosomal dominant 36 606705	TMC1
383	Deafness autosomal dominant 39 with dentinogenesis 605594	DSPP
383	Deafness autosomal dominant 3A 601544	GJB2
383	Deafness autosomal dominant 3B 612643	GJB6
383	Deafness autosomal dominant 40 616357	CRYM

383	Deafness autosomal dominant 41 608224	P2RX2
383	Deafness autosomal dominant 4A 600652	MYH14
383	Deafness autosomal dominant 4B 614614	CEACAM16
383	Deafness autosomal dominant 5 600994	GSDME
383	Deafness autosomal dominant 50 613074	MIR96
383	Deafness autosomal dominant 56 615629	TNC
383	Deafness autosomal dominant 6 or 14 or 38 600965	WFS1
383	Deafness autosomal dominant 64 614152	DIABLO
383	Deafness autosomal dominant 65 616044	TBC1D24
383	Deafness autosomal dominant 67 616340	OSBPL2
383	Deafness autosomal dominant 69 unilateral or asymmetric 616697	KITLG
383	Deafness autosomal dominant 73 617663	PTPRR
383	Deafness autosomal dominant 8 or 12 601543	TECTA
383	Deafness autosomal dominant 9 601369	COCH
383	Deafness autosomal recessive 105 616958	CDC14A
383	Deafness autosomal recessive 106 617637	EPS8L2
383	Deafness autosomal recessive 107 617639	WBP2
383	Deafness autosomal recessive 12 601386	CDH23
383	Deafness autosomal recessive 15 601869	GIPC3
383	Deafness autosomal recessive 16 603720	STRC
383	Deafness autosomal recessive 18A 602092	USH1C
383	Deafness autosomal recessive 18B 614945	OTOG
383	Deafness autosomal recessive 1A 220290	GJB2
383	Deafness autosomal recessive 1B 612645	GJB6
383	Deafness autosomal recessive 2 600060	MYO7A
383	Deafness autosomal recessive 21 603629	TECTA
383	Deafness autosomal recessive 22 607039	OTOA
383	Deafness autosomal recessive 23 609533	PCDH15
383	Deafness autosomal recessive 24 611022	RDX
383	Deafness autosomal recessive 25 613285	GRXCR1
383	Deafness autosomal recessive 28 609823	TRIOBP
383	Deafness autosomal recessive 29 614035	CLDN14
383	Deafness autosomal recessive 3 600316	MYO15A
383	Deafness autosomal recessive 30 607101	MYO3A
383	Deafness autosomal recessive 31 607084	WHRN
383	Deafness autosomal recessive 35 608565	ESRRB
383	Deafness autosomal recessive 36 609006	ESPN
383	Deafness autosomal recessive 37 607821	MYO6
383	Deafness autosomal recessive 39 608265	HGF
383	Deafness autosomal recessive 4 with enlarged vestibular aqueduct 600791	SLC26A4
383	Deafness autosomal recessive 42 609646	ILDR1
383	Deafness autosomal recessive 48 609439	CIB2
383	Deafness autosomal recessive 49 610153	MARVELD2

383	Deafness autosomal recessive 53 609706	COL11A2
383	Deafness autosomal recessive 59 610220	PJVK
383	Deafness autosomal recessive 6 600971	TMIE
383	Deafness autosomal recessive 63 611451	LRTOMT
383	Deafness autosomal recessive 67 610265	LHFPL5
383	Deafness autosomal recessive 68 610419	S1PR2
383	Deafness autosomal recessive 7 600974	TMC1
383	Deafness autosomal recessive 70 614934	PNPT1
383	Deafness autosomal recessive 74 613718	MSRB3
383	Deafness autosomal recessive 76 615540	SYNE4
383	Deafness autosomal recessive 77 613079	LOXHD1
383	Deafness autosomal recessive 79 613307	TPRN
383	Deafness autosomal recessive 8 or 10 601072	TMPRSS3
383	Deafness autosomal recessive 84A 613391	PTPRR
383	Deafness autosomal recessive 84B 614944	OTOGL
383	Deafness autosomal recessive 89 613916	KARS
383	Deafness autosomal recessive 9 601071	OTOF
383	Deafness autosomal recessive 93 614899	CABP2
383	Deafness autosomal recessive 98 614861	TSPEAR
383	{Deafness autosomal recessive 12 modifier of} 601386	ATP2B2
383	Deafness congenital with inner ear agenesis microtia and microdontia 610706	FGF3
383	Deafness congenital with onychodystrophy autosomal dominant 124480	ATP6V1B2
383	Deafness digenic GJB2 or GJB3 220290	GJB3
383	Deafness digenic GJB2 or GJB6 220290	GJB6
383	Deafness dystonia and cerebral hypomyelination 300475	BCAP31
383	{Deafness mitochondrial modifier of} 580000	TRMU
383	Deafness autosomal dominant with peripheral neuropathy	GJB3
383	Deafness autosomal recessive	GJB3
383	Deafness neurosensory without vestibular involvement autosomal dominant	ESPN
384	Dehydrated hereditary stomatocytosis 2 616689	KCNN4
384	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and or perinatal edema 194380	PIEZO1
385	Dejerine-Sottas disease 145900	EGR2
385	Dejerine-Sottas disease 145900	MPZ
385	Dejerine-Sottas disease 145900	PMP22
385	Dejerine-Sottas disease 145900	PRX
386	Dementia Lewy body 127750	SNCA
386	Dementia Lewy body 127750	SNCB
386	{Lewy body dementia susceptibility to} 127750	GBA
386	Dementia familial British 176500	ITM2B
386	Dementia familial Danish 117300	ITM2B
386	Dementia familial nonspecific 600795	CHMP2B

386	Dementia frontotemporal 600274	PSEN1
386	Dementia frontotemporal with or without parkinsonism 600274	MAPT
386	{Dementia, vascular, susceptibility to}	TNF
387	Dent disease 2 300555	OCRL
387	Dent disease 300009	CLCN5
388	Dental anomalies and short stature 601216	LTBP3
389	Dentatorubro-pallidoluysian atrophy 125370	ATN1
390	Dentin dysplasia type I with microdontia and misshapen teeth 125400	SMOC2
390	Dentin dysplasia type II 125420	DSPP
391	Dentinogenesis imperfecta Shields type II 125490	DSPP
391	Dentinogenesis imperfecta Shields type III 125500	DSPP
392	Denys-Drash syndrome 194080	WT1
393	Dermatofibrosarcoma protuberans 607907	PDGFB
394	Dermatopathia pigmentosa reticularis 125595	KRT14
395	Desanto-Shinawi syndrome 616708	WAC
396	Desbuquois dysplasia 1 251450	CANT1
396	Desbuquois dysplasia 2 615777	XYLT1
397	Desmoid disease hereditary 135290	APC
398	Desmosterolosis 602398	DHCR24
399	Developmental delay with short stature dysmorphic features and sparse hair 616901	DPH1
400	DiGeorge syndrome 188400	TBX1
401	Diabetes insipidus nephrogenic 125800	AQP2
401	Diabetes insipidus nephrogenic 304800	AVPR2
401	Diabetes insipidus neurohypophyseal 125700	AVP
402	Diabetes mellitus insulin-dependent 2 125852	INS
402	Diabetes mellitus insulin-dependent 20 612520	HNF1A
402	Diabetes mellitus insulin-resistant with acanthosis nigricans 610549	INSR
402	Diabetes mellitus neonatal with congenital hypothyroidism 610199	GLIS3
402	Diabetes mellitus noninsulin-dependent 125853	ABCC8
402	Diabetes mellitus noninsulin-dependent 125853	HNF1B
402	Diabetes mellitus noninsulin-dependent late onset 125853	GCK
402	Diabetes mellitus permanent neonatal 606176	ABCC8
402	Diabetes mellitus permanent neonatal 606176	GCK
402	Diabetes mellitus permanent neonatal 606176	INS
402	Diabetes mellitus transient neonatal 1 601410	ZFP57
402	Diabetes mellitus transient neonatal 2 610374	ABCC8
402	Diabetes mellitus transient neonatal 3 610582	KCNJ11
402	Diabetes mellitus type 2 125853	PAX4
402	Diabetes mellitus type II 125853	AKT2
402	Diabetes permanent neonatal with or without neurologic features 606176	KCNJ11

402	{Diabetes mellitus insulin-dependent 12} 601388	CTLA4
402	{Diabetes mellitus insulin-dependent 22} 612522	CCR5
402	{Diabetes mellitus insulin-dependent 5} 600320	SUMO4
402	{Diabetes mellitus insulin-dependent susceptibility to 10} 601942	IL2RA
402	{Diabetes mellitus insulin-dependent} 222100	HNF1A
402	{Diabetes mellitus ketosis-prone susceptibility to} 612227	PAX4
402	{Diabetes mellitus non-insulin-dependent susceptibility to} 125853	ENPP1
402	{Diabetes mellitus noninsulin-dependent 1} 601283	CAPN10
402	{Diabetes mellitus noninsulin-dependent 2} 125853	HNF1A
402	{Diabetes mellitus noninsulin-dependent 5} 616087	TBC1D4
402	{Diabetes mellitus noninsulin-dependent association with} 125853	WFS1
402	{Diabetes mellitus noninsulin-dependent susceptibility to} 125853	CDKAL1
402	{Diabetes mellitus noninsulin-dependent susceptibility to} 125853	HMGA1
402	{Diabetes mellitus noninsulin-dependent susceptibility to} 125853	IGF2BP2
402	{Diabetes mellitus noninsulin-dependent susceptibility to} 125853	RETN
402	{Diabetes mellitus noninsulin-dependent susceptibility to} 125853	SLC30A8
402	{Diabetes mellitus noninsulin-dependent} 125853	GCGR
402	{Diabetes mellitus noninsulin-dependent} 125853	HNF4A
402	{Diabetes mellitus noninsulin-dependent} 125853	IRS1
402	{Diabetes mellitus noninsulin-dependent} 125853	IRS2
402	{Diabetes mellitus noninsulin-dependent} 125853	LIPC
402	{Diabetes mellitus noninsulin-dependent} 125853	MAPK8IP1
402	{Diabetes mellitus noninsulin-dependent} 125853	NEUROD1
402	{Diabetes mellitus noninsulin-dependent} 125853	SLC2A2
402	{Diabetes mellitus type 1 susceptibility to} 222100	OAS1
402	{Diabetes mellitus type 2 susceptibility to} 125853	KCNJ11
402	{Diabetes mellitus type 2 susceptibility to} 125853	MTNR1B
402	{Diabetes mellitus type 2 susceptibility to} 125853	TCF7L2
402	{Diabetes mellitus type I susceptibility to} 222100	FOXP3
402	{Diabetes mellitus type II susceptibility to} 125853	PDX1
402	{Diabetes susceptibility to} 222100 125853	IL6
402	{Diabetes type 1 susceptibility to} 222100	PTPN22
402	{Diabetes type 2 susceptibility to} 125853	GPD2
402	{Diabetes type 2} 125853	PPARG
402	Maturity-onset diabetes of the young 6 606394	NEUROD1
402	Maturity-onset diabetes of the young type 10 613370	INS
402	Maturity-onset diabetes of the young type 11 613375	BLK
402	Maturity-onset diabetes of the young type 13 616329	KCNJ11

402	Maturity-onset diabetes of the young type IX 612225	PAX4
402	Maturity-onset diabetes of the young type VII 610508	KLF11
402	Maturity-onset diabetes of the young type VIII 609812	CEL
402	{Maturity-onset diabetes of the young type 14} 616511	APPL1
403	Diamond-Blackfan anemia 15 with mandibulofacial dysostosis 606164	RPS28
403	Diamond-Blackfan anemia 1 105650	RPS19
403	Diamond-Blackfan anemia 10 613309	RPS26
403	Diamond-Blackfan anemia 13 615909	RPS29
403	Diamond-Blackfan anemia 4 612527	RPS17
403	Diamond-Blackfan anemia 5 612528	RPL35A
403	Diamond-Blackfan anemia 6 612561	RPL5
403	Diamond-Blackfan anemia 7 612562	RPL11
403	Diamond-Blackfan anemia 8 612563	RPS7
403	Diamond-Blackfan anemia 9 613308	RPS10
403	Diamond-blackfan anemia 3 610629	RPS24
404	Diaphanospondylodysostosis 608022	BMPER
405	Diaphragmatic hernia 3 610187	ZFPM2
406	Diaphyseal medullary stenosis with malignant fibrous histiocytoma 112250	MTAP
407	Diarrhea 1 secretory chloride congenital 214700	SLC26A3
407	Diarrhea 3 secretory sodium congenital syndromic 270420	SPINT2
407	Diarrhea 4 malabsorptive congenital 610370	NEUROG3
407	Diarrhea 5 with tufting enteropathy congenital 613217	EPCAM
407	Diarrhea 6 614616	GUCY2C
407	Diarrhea 8 secretory sodium congenital 616868	SLC9A3
408	Dias-Logan syndrome 617101	BCL11A
409	Diastrophic dysplasia 222600	SLC26A2
409	Diastrophic dysplasia broad bone-platyspondylic variant 222600	SLC26A2
410	Dicarboxylic aminoaciduria 222730	SLC1A1
411	Digital arthropathy-brachydactyly familial 606835	TRPV4
412	Digital clubbing isolated congenital 119900	HPGD
413	Dihydrolipoamide dehydrogenase deficiency 246900	DLD
414	Dihydropyrimidine dehydrogenase deficiency 274270	DPYD
414	5-fluorouracil toxicity 274270	DPYD
415	Dihydropyrimidinuria 222748	DPYS
416	Dilated cardiomyopathy with woolly hair keratoderma and tooth agenesis 615821	DSP
417	Dimethylglycine dehydrogenase deficiency 605850	DMGDH
418	Disordered steroidogenesis due to cytochrome P450 oxidoreductase 613571	POR
419	Donnai-Barrow syndrome 222448	LRP2
420	Dopamine beta-hydroxylase deficiency 223360	DBH
421	Dowling-Degos disease 1 179850	KRT5
421	Dowling-Degos disease 2 615327	POFUT1

421	Dowling-Degos disease 4 615696	POGLUT1
422	Doyne honeycomb degeneration of retina 126600	EFEMP1
423	Du Pan syndrome 228900	GDF5
424	Duane retraction syndrome 2 604356	CHN1
424	Duane retraction syndrome 3 617041	MAFB
425	Duane-radial ray syndrome 607323	SALL4
426	Dubin-Johnson syndrome 237500	ABCC2
427	Duchenne muscular dystrophy 310200	DMD
428	Dyggve-Melchior-Clausen disease 223800	DYM
429	Dysautonomia familial 223900	IKBKAP
430	Dyschromatosis symmetrica hereditaria 127400	ADAR
430	Dyschromatosis universalis hereditaria 3 615402	ABCB6
431	Dyserythropoietic anemia congenital type II 224100	SEC23B
431	Dyserythropoietic anemia congenital type IV 613673	KLF1
431	Dyserythropoietic anemia congenital type Ia 224120	CDAN1
431	Dyserythropoietic anemia congenital type Ib 615631	C15orf41
432	Dysfibrinogenemia congenital 616004	FGA
432	Dysfibrinogenemia congenital 616004	FGB
432	Dysfibrinogenemia congenital 616004	FGG
432	Hypodysfibrinogenemia 616004	FGG
432	Hypodysfibrinogenemia congenital 616004	FGA
432	Hypofibrinogenemia congenital 202400	FGB
432	Hypofibrinogenemia congenital 202400	FGG
432	Afibrinogenemia congenital 202400	FGA
432	Afibrinogenemia congenital 202400	FGB
432	Afibrinogenemia congenital 202400	FGG
433	Dyskeratosis congenita X-linked 305000	DKC1
433	Dyskeratosis congenita autosomal dominant 1 127550	TERC
433	Dyskeratosis congenita autosomal dominant 3 613990	TINF2
433	Dyskeratosis congenita autosomal dominant 4 615190	RTEL1
433	Dyskeratosis congenita autosomal recessive 1 224230	NOP10
433	Dyskeratosis congenita autosomal recessive 2 613987	NHP2
433	Dyskeratosis congenita autosomal recessive 3 613988	WRAP53
433	Dyskeratosis congenita autosomal recessive 5 615190	RTEL1
433	Dyskeratosis congenita autosomal recessive 6 616353	PARN
433	{Dyskeratosis congenita autosomal dominant 2} 613989	TERT
433	{Dyskeratosis congenita autosomal recessive 4} 613989	TERT
434	Dyskinesia familial with facial myokymia 606703	ADCY5
435	Dyskinesia limb and orofacial infantile-onset 616921	PDE10A
436	Dyssegmental dysplasia Silverman-Handmaker type 224410	HSPG2
437	Dystonia 16 612067	PRKRA
437	Dystonia 2 torsion autosomal recessive 224500	HPCA
437	Dystonia 24 615034	ANO3
437	Dystonia 25 615073	GNAL
437	Dystonia 26 myoclonic 616398	KCTD17

437	Dystonia 27 616411	COL6A3
437	Dystonia 28 childhood-onset 617284	KMT2B
437	Dystonia 4 torsion autosomal dominant 128101	TUBB4A
437	Dystonia 6 torsion 602629	THAP1
437	Dystonia 9 601042	SLC2A1
437	Dystonia DOPA-responsive with or without hyperphenylalaninemia 128230	GCH1
437	Dystonia childhood-onset with optic atrophy and basal ganglia abnormalities 617282	MECR
437	Dystonia dopa-responsive due to sepiapterin reductase deficiency 612716	SPR
437	Dystonia-1 torsion 128100	TOR1A
437	Dystonia-11 myoclonic 159900	SGCE
437	Dystonia-12 128235	ATP1A3
437	Dystonia-Parkinsonism X-linked 314250	TAF1
437	Dystonia primary cervical	DRD5
437	{Dystonia-1, modifier of}	TOR1A
438	EDICT syndrome 614303	MIR184
439	Ectodermal dysplasia 1 hypohidrotic X-linked 305100	EDA
439	Ectodermal dysplasia 10A hypohidrotic or hair or nail type autosomal dominant 129490	EDAR
439	Ectodermal dysplasia 10B hypohidrotic or hair or tooth type autosomal recessive 224900	EDAR
439	Ectodermal dysplasia 11A hypohidrotic or hair or tooth type autosomal dominant 614940	EDARADD
439	Ectodermal dysplasia 11B hypohidrotic or hair or tooth type autosomal recessive 614941	EDARADD
439	Ectodermal dysplasia 13 hair or tooth type 617392	KREMEN1
439	Ectodermal dysplasia 2 Clouston type 129500	GJB6
439	Ectodermal dysplasia 3 Witkop type 189500	MSX1
439	Ectodermal dysplasia 4 hair or nail type 602032	KRT85
439	Ectodermal dysplasia 9 hair or nail type 614931	HOXC13
439	Ectodermal dysplasia anhidrotic lymphedema and immunodeficiency 300301	IKBKG
439	Ectodermal dysplasia anhidrotic with T-cell immunodeficiency 612132	NFKBIA
439	Ectodermal dysplasia ectrodactyly and macular dystrophy 225280	CDH3
439	Ectodermal dysplasia hypohidrotic with immune deficiency 300291	IKBKG
439	Ectodermal dysplasia-syndactyly syndrome 1 613573	NECTIN4
439	Ectodermal dysplasia or short stature syndrome 616029	GRHL2
439	Ectodermal dysplasia or skin fragility syndrome 604536	PKP1
440	Ectopia lentis et pupillae 225200	ADAMTSL4
440	Ectopia lentis familial 129600	FBN1
440	Ectopia lentis isolated autosomal recessive 225100	ADAMTSL4

441	Ectrodactyly ectodermal dysplasia and cleft lip or palate syndrome 3 604292	TP63
442	Efavirenz poor metabolism of 614546	CYP2B6
442	{Efavirenz central nervous system toxicity susceptibility to} 614546	CYP2B6
443	Ehlers-Danlos syndrome cardiac valvular form 225320	COL1A2
443	Ehlers-Danlos syndrome classic 130000	COL1A1
443	Ehlers-Danlos syndrome classic type 130000	COL5A1
443	Ehlers-Danlos syndrome classic type 130000	COL5A2
443	Ehlers-Danlos syndrome due to tenascin X deficiency 606408	TNXB
443	Ehlers-Danlos syndrome musculocontractural type 1 601776	CHST14
443	Ehlers-Danlos syndrome musculocontractural type 2 615539	DSE
443	Ehlers-Danlos syndrome periodontal type 1 130080	C1R
443	Ehlers-Danlos syndrome periodontal type 2 617174	C1S
443	Ehlers-Danlos syndrome progeroid type 2 615349	B3GALT6
443	Ehlers-Danlos syndrome type IV 130050	COL3A1
443	Ehlers-Danlos syndrome type VI 225400	PLOD1
443	Ehlers-Danlos syndrome type VIIA 130060	COL1A1
443	Ehlers-Danlos syndrome type VIIB 130060	COL1A2
443	Ehlers-Danlos syndrome type VIIC 225410	ADAMTS2
443	Ehlers-Danlos syndrome with progressive kyphoscoliosis myopathy and hearing loss 614557	FKBP14
443	Ehlers-Danlos syndrome with short stature and limb anomalies 130070	B4GALT7
444	Eiken syndrome 600002	PTH1R
445	Elliptocytosis-1 611804	EPB41
445	Elliptocytosis-2 130600	SPTA1
445	Elliptocytosis-3	SPTB
446	Ellis-van Creveld syndrome 225500	EVC
446	Ellis-van Creveld syndrome 225500	EVC2
447	Emberger syndrome 614038	GATA2
448	Emery-Dreifuss muscular dystrophy 1 X-linked 310300	EMD
448	Emery-Dreifuss muscular dystrophy 2 AD 181350	LMNA
448	Emery-Dreifuss muscular dystrophy 3 AR 616516	LMNA
448	Emery-Dreifuss muscular dystrophy 4 autosomal dominant 612998	SYNE1
448	Emery-Dreifuss muscular dystrophy 5 autosomal dominant 612999	SYNE2
448	Emery-Dreifuss muscular dystrophy 6 X-linked 300696	FHL1
448	Emery-Dreifuss muscular dystrophy 7 AD 614302	TMEM43
448	Myopathy X-linked with postural muscle atrophy 300696	FHL1
449	Emphysema due to AAT deficiency 613490	SERPINA1
449	Emphysema-cirrhosis due to AAT deficiency 613490	SERPINA1
449	Hemorrhagic diathesis due to antithrombin Pittsburgh 613490	SERPINA1
450	Encephalocraniocutaneous lipomatosis 613001	FGFR1

450	Lipoma	LPP
450	Lipoma somatic	MEN1
451	Encephalopathy due to defective mitochondrial and peroxisomal fission 2 617086	MFF
451	Encephalopathy familial with neuroserpin inclusion bodies 604218	SERPINI1
451	Encephalopathy lethal due to defective mitochondrial peroxisomal fission 1 614388	DNM1L
451	Encephalopathy neonatal severe 300673	MECP2
451	Encephalopathy neonatal severe with lactic acidosis and brain abnormalities 617668	LIPT2
451	Encephalopathy progressive early-onset with brain atrophy and spasticity 617669	TRAPPC12
451	Encephalopathy progressive early-onset with brain atrophy and thin corpus callosum 617193	TBCD
451	Encephalopathy progressive early-onset with brain edema and or leukoencephalopathy 617186	NAXE
451	Encephalopathy progressive with amyotrophy and optic atrophy 617207	TBCE
451	Encephalopathy progressive with or without lipodystrophy 615924	BSCL2
452	Endocrine-cerebroosteodysplasia 612651	ICK
453	Endometrial cancer familial 608089	MSH6
453	Endometrial carcinoma somatic 608089	CDH1
453	Endometrial carcinoma somatic 608089	MSH3
453	Endometrial carcinoma somatic 608089	PTEN
453	{Endometrial cancer susceptibility to} 608089	MLH3
454	Enhanced S-cone syndrome 268100	NR2E3
455	Enlarged vestibular aqueduct 600791	FOXI1
455	Enlarged vestibular aqueduct digenic 600791	KCNJ10
456	Enterokinase deficiency 226200	PRSS7
457	Epidermodysplasia verruciformis 226400	TMC6
457	Epidermodysplasia verruciformis 226400	TMC8
458	Epidermolysis bullosa dystrophica AD 131750	COL7A1
458	Epidermolysis bullosa dystrophica AR 226600	COL7A1
458	EBD Bart type 132000	COL7A1
458	EBD inversa 226600	COL7A1
458	EBD localisata variant	COL7A1
458	{Epidermolysis bullosa dystrophica autosomal recessive modifier of} 226600	MMP1
458	Epidermolysis bullosa generalized atrophic benign 226650	LAMA3
458	Epidermolysis bullosa junctional Herlitz type 226700	LAMA3
458	Epidermolysis bullosa junctional Herlitz type 226700	LAMB3
458	Epidermolysis bullosa junctional Herlitz type 226700	LAMC2
458	Epidermolysis bullosa junctional localisata variant 226650	COL17A1

458	Epidermolysis bullosa junctional non-Herlitz type 226650	COL17A1
458	Epidermolysis bullosa junctional non-Herlitz type 226650	ITGB4
458	Epidermolysis bullosa junctional non-Herlitz type 226650	LAMB3
458	Epidermolysis bullosa junctional non-Herlitz type 226650	LAMC2
458	Epidermolysis bullosa junctional with pyloric atresia 226730	ITGB4
458	Epidermolysis bullosa junctional with pyloric stenosis 226730	ITGA6
458	Epidermolysis bullosa lethal acantholytic 609638	DSP
458	Epidermolysis bullosa nonspecific autosomal recessive 615028	EXPH5
458	Epidermolysis bullosa of hands and feet 131800	ITGB4
458	Epidermolysis bullosa pretibial 131850	COL7A1
458	Epidermolysis bullosa pruriginosa 604129	COL7A1
458	Epidermolysis bullosa simplex Dowling-Meara type 131760	KRT14
458	Epidermolysis bullosa simplex Dowling-Meara type 131760	KRT5
458	Epidermolysis bullosa simplex Koebner type 131900	KRT14
458	Epidermolysis bullosa simplex Koebner type 131900	KRT5
458	Epidermolysis bullosa simplex Ogna type 131950	PLEC
458	Epidermolysis bullosa simplex Weber-Cockayne type 131800	KRT14
458	Epidermolysis bullosa simplex Weber-Cockayne type 131800	KRT5
458	Epidermolysis bullosa simplex autosomal recessive 2 615425	DST
458	Epidermolysis bullosa simplex generalized with scarring and hair loss 617294	KLHL24
458	Epidermolysis bullosa simplex recessive 1 601001	KRT14
458	Epidermolysis bullosa simplex recessive 1 601001	KRT5
458	Epidermolysis bullosa simplex with muscular dystrophy 226670	PLEC
458	Epidermolysis bullosa simplex with pyloric atresia 612138	PLEC
458	Epidermolysis bullosa simplex-MCR 609352	KRT5
458	Epidermolysis bullosa simplex-MP 131960	KRT5
459	Epidermolytic hyperkeratosis 113800	KRT1
459	Epidermolytic hyperkeratosis 113800	KRT10
460	Epilepsy X-linked with variable learning disabilities and behavior disorders 300491	SYN1
460	Epilepsy early-onset vitamin B6-dependent 617290	PROSC
460	Epilepsy familial focal with variable foci 1 604364	DEPDC5
460	Epilepsy familial focal with variable foci 2 617116	NPRL2
460	Epilepsy familial focal with variable foci 3 617118	NPRL3
460	Epilepsy familial temporal lobe 1 600512	LGI1
460	Epilepsy familial temporal lobe 5 614417	CPA6
460	Epilepsy focal with speech disorder and with or without mental retardation 245570	GRIN2A
460	Epilepsy generalized with febrile seizures plus type 1 604233	SCN1B
460	Epilepsy generalized with febrile seizures plus type 2 604403	SCN1A
460	Epilepsy generalized with febrile seizures plus type 3 611277	GABRG2
460	Epilepsy generalized with febrile seizures plus type 7 613863	SCN9A
460	Epilepsy hearing loss and mental retardation syndrome 616577	SPATA5
460	Epilepsy myoclonic familial adult 2 607876	ADRA2B

460	Epilepsy nocturnal frontal lobe 1 600513	CHRNA4
460	Epilepsy nocturnal frontal lobe 3 605375	CHRNB2
460	Epilepsy nocturnal frontal lobe 5 615005	KCNT1
460	Epilepsy nocturnal frontal lobe type 4 610353	CHRNA2
460	Epilepsy progressive myoclonic 1A (Unverricht and Lundborg) 254800	CSTB
460	Epilepsy progressive myoclonic 1B 612437	PRICKLE1
460	Epilepsy progressive myoclonic 2A (Lafora) 254780	EPM2A
460	Epilepsy progressive myoclonic 2B (Lafora) 254780	NHLRC1
460	Epilepsy progressive myoclonic 3 with or without intracellular inclusions 611726	KCTD7
460	Epilepsy progressive myoclonic 4 with or without renal failure 254900	SCARB2
460	Epilepsy progressive myoclonic 6 614018	GOSR2
460	Epilepsy progressive myoclonic 7 616187	KCNC1
460	Epilepsy pyridoxine-dependent 266100	ALDH7A1
460	Epileptic encephalopathy childhood-onset 615369	CHD2
460	Epileptic encephalopathy early infantile 1 308350	ARX
460	Epileptic encephalopathy early infantile 11 613721	SCN2A
460	Epileptic encephalopathy early infantile 12 613722	PLCB1
460	Epileptic encephalopathy early infantile 13 614558	SCN8A
460	Epileptic encephalopathy early infantile 14 614959	KCNT1
460	Epileptic encephalopathy early infantile 16 615338	TBC1D24
460	Epileptic encephalopathy early infantile 17 615473	GNAO1
460	Epileptic encephalopathy early infantile 18 615476	SZT2
460	Epileptic encephalopathy early infantile 19 615744	GABRA1
460	Epileptic encephalopathy early infantile 2 300672	CDKL5
460	Epileptic encephalopathy early infantile 23 615859	DOCK7
460	Epileptic encephalopathy early infantile 24 615871	HCN1
460	Epileptic encephalopathy early infantile 25 615905	SLC13A5
460	Epileptic encephalopathy early infantile 26 616056	KCNB1
460	Epileptic encephalopathy early infantile 27 616139	GRIN2B
460	Epileptic encephalopathy early infantile 28 616211	WWOX
460	Epileptic encephalopathy early infantile 29 616339	AARS
460	Epileptic encephalopathy early infantile 3 609304	SLC25A22
460	Epileptic encephalopathy early infantile 30 616341	SIK1
460	Epileptic encephalopathy early infantile 31 616346	DNM1
460	Epileptic encephalopathy early infantile 32 616366	KCNA2
460	Epileptic encephalopathy early infantile 33 616409	EEF1A2
460	Epileptic encephalopathy early infantile 34 616645	SLC12A5
460	Epileptic encephalopathy early infantile 35 616647	ITPA
460	Epileptic encephalopathy early infantile 36 300884	ALG13
460	Epileptic encephalopathy early infantile 37 616981	FRRS1L
460	Epileptic encephalopathy early infantile 38 617020	ARV1
460	Epileptic encephalopathy early infantile 39 612949	SLC25A12

460	Epileptic encephalopathy early infantile 4 612164	STXBP1
460	Epileptic encephalopathy early infantile 41 617105	SLC1A2
460	Epileptic encephalopathy early infantile 42 617106	CACNA1A
460	Epileptic encephalopathy early infantile 43 617113	GABRB3
460	Epileptic encephalopathy early infantile 44 617132	UBA5
460	Epileptic encephalopathy early infantile 45 617153	GABRB1
460	Epileptic encephalopathy early infantile 46 617162	GRIN2D
460	Epileptic encephalopathy early infantile 47 617166	FGF12
460	Epileptic encephalopathy early infantile 48 617276	AP3B2
460	Epileptic encephalopathy early infantile 49 617281	DENND5A
460	Epileptic encephalopathy early infantile 5 613477	SPTAN1
460	Epileptic encephalopathy early infantile 50 616457	CAD
460	Epileptic encephalopathy early infantile 51 617339	MDH2
460	Epileptic encephalopathy early infantile 52 617350	SCN1B
460	Epileptic encephalopathy early infantile 53 617389	SYNJ1
460	Epileptic encephalopathy early infantile 54 617391	HNRNPU
460	Epileptic encephalopathy early infantile 56 617665	YWHAG
460	Epileptic encephalopathy early infantile 6 (Dravet syndrome) 607208	SCN1A
460	{Dravet syndrome modifier of} 607208	SCN9A
460	Epileptic encephalopathy early infantile 7 613720	KCNQ2
460	Epileptic encephalopathy early infantile 8 300607	ARHGEF9
460	Epileptic encephalopathy early infantile 9 300088	PCDH19
460	Epileptic encephalopathy infantile or early childhood 617711	PPP3CA
460	{Epilepsy childhood absence susceptibility to 2} 607681	GABRG2
460	{Epilepsy childhood absence susceptibility to 4} 611136	GABRA1
460	{Epilepsy childhood absence susceptibility to 5} 612269	GABRB3
460	{Epilepsy childhood absence susceptibility to 6} 611942	CACNA1H
460	{Epilepsy familial temporal lobe 7} 616436	RELN
460	{Epilepsy generalized with febrile seizures plus type 5 susceptibility to} 613060	GABRD
460	{Epilepsy idiopathic generalized 10} 613060	GABRD
460	{Epilepsy idiopathic generalized susceptibility to 11} 607628	CLCN2
460	{Epilepsy idiopathic generalized susceptibility to 12} 614847	SLC2A1
460	{Epilepsy idiopathic generalized susceptibility to 14} 616685	SLC12A5
460	{Epilepsy idiopathic generalized susceptibility to 6} 611942	CACNA1H
460	{Epilepsy idiopathic generalized susceptibility to 8} 612899	CASR
460	{Epilepsy idiopathic generalized susceptibility to 9} 607682	CACNB4
460	{Epilepsy juvenile absence susceptibility to 1} 607631	EFHC1
460	{Epilepsy juvenile absence susceptibility to 2} 607628	CLCN2
460	{Epilepsy juvenile myoclonic susceptibility to 5} 611136	GABRA1
460	{Epilepsy juvenile myoclonic susceptibility to 6} 607682	CACNB4
460	{Epilepsy juvenile myoclonic susceptibility to 8} 607628	CLCN2
460	{Epilepsy juvenile myoclonic susceptibility to} 613060	GABRD
461	Epiphyseal chondrodysplasia Miura type 615923	NPR2

461	Epiphyseal dysplasia multiple 1 132400	COMP
461	Epiphyseal dysplasia multiple 2 600204	COL9A2
461	Epiphyseal dysplasia multiple 3 with or without myopathy 600969	COL9A3
461	Epiphyseal dysplasia multiple 4 226900	SLC26A2
461	Epiphyseal dysplasia multiple 5 607078	MATN3
461	Epiphyseal dysplasia multiple 7 617719	CANT1
461	Epiphyseal dysplasia multiple with myopia and deafness 132450	COL2A1
462	Episodic ataxia type 2 108500	CACNA1A
462	Episodic ataxia type 5 613855	CACNB4
462	Episodic ataxia type 6 612656	SLC1A3
462	Episodic ataxia or myokymia syndrome 160120	KCNA1
462	Episodic kinesigenic dyskinesia 1 128200	PRRT2
463	Episodic pain syndrome familial 2 615551	SCN10A
463	Episodic pain syndrome familial 3 615552	SCN11A
464	Epithelial recurrent erosion dystrophy 122400	COL17A1
465	Epstein syndrome 153650	MYH9
466	Erythrocyte lactate transporter defect 245340	SLC16A1
467	Erythrocytosis due to bisphosphoglycerate mutase deficiency 222800	BPGM
467	Erythrocytosis familial 2 263400	VHL
467	Erythrocytosis familial 3 609820	EGLN1
467	Erythrocytosis familial 4 611783	EPAS1
467	Erythrocytosis somatic 133100	JAK2
467	Erythrocytosis somatic 133100	SH2B3
467	Erythrocytosis	HBA2
468	Erythroderma congenital with palmoplantar keratoderma hypotrichosis and hyper IgE 615508	DSG1
469	Erythrokeratodermia variabilis et progressiva 1 133200	GJB3
469	Erythrokeratodermia variabilis et progressiva 2 617524	GJB4
469	Erythrokeratodermia variabilis et progressiva 3 617525	GJA1
469	Erythrokeratodermia variabilis et progressiva 4 617526	KDSR
469	Erythrokeratodermia variabilis et progressiva 5 617756	KRT83
470	Escobar syndrome 265000	CHRNG
471	Esophageal cancer somatic 133239	TGFBR2
471	Esophageal carcinoma somatic 133239	DCC
471	Esophageal carcinoma somatic 133239	RNF6
471	Esophageal squamous cell carcinoma 133239	LZTS1
471	Esophageal squamous cell carcinoma somatic 133239	WWOX
471	{Esophageal cancer, alcohol-related, susceptibility to}	ALDH2
472	Essential tremor hereditary 4 614782	FUS
472	Essential tremor hereditary 5 616736	TENM4
472	{Essential tremor hereditary 1} 190300	DRD3
473	Estrogen resistance 615363	ESR1
474	Ethylmalonic encephalopathy 602473	ETHE1
475	Even-plus syndrome 616854	HSPA9

476	Ewing sarcoma 612219	EWSR1
476	Neuroepithelioma 612219	EWSR1
477	Exocrine pancreatic insufficiency dyserythropoietic anemia and calvarial hyperostosis 612714	COX4I2
478	Exostoses multiple type 1 133700	EXT1
478	Exostoses multiple type 2 133701	EXT2
479	Exudative vitreoretinopathy 1 133780	FZD4
479	Retinopathy of prematurity 133780	FZD4
479	Exudative vitreoretinopathy 2 X-linked 305390	NDP
479	Exudative vitreoretinopathy 4 601813	LRP5
479	Exudative vitreoretinopathy 5 613310	TSPAN12
479	Exudative vitreoretinopathy 7 617572	CTNNB1
479	Vitreoretinopathy with phalangeal epiphyseal dysplasia	COL2A1
479	Vitreoretinopathy neovascular inflammatory 193235	CAPN5
480	FG syndrome 2 300321	FLNA
480	FG syndrome 4 300422	CASK
481	FILS syndrome 615139	POLE
482	Fabry disease 301500	GLA
482	Fabry disease cardiac variant 301500	GLA
483	Facial paresis hereditary congenital 3 614744	HOXB1
484	Factor V and factor VIII combined deficiency of 613625	MCFD2
484	Factor V deficiency 227400	F5
484	Factor VII deficiency 227500	F7
484	Factor X deficiency 227600	F10
484	Factor XI deficiency autosomal dominant 612416	F11
484	Factor XI deficiency autosomal recessive 612416	F11
484	Factor XII deficiency 234000	F12
484	Factor XIIIA deficiency 613225	F13A1
484	Factor XIIIB deficiency 613235	F13B
485	Failure of tooth eruption primary 125350	PTH1R
486	Familial Mediterranean fever AD 134610	MEFV
486	Familial Mediterranean fever AR 249100	MEFV
487	Familial adenomatous polyposis 3 616415	NTHL1
487	Familial adenomatous polyposis 4 617100	MSH3
488	Familial cold autoinflammatory syndrome 2 611762	NLRP12
488	Familial cold autoinflammatory syndrome 3 614468	PLCG2
488	Familial cold-induced inflammatory syndrome 1 120100	NLRP3
489	Fanconi anemia complementation group A 227650	FANCA
489	Fanconi anemia complementation group B 300514	FANCB
489	Fanconi anemia complementation group C 227645	FANCC
489	Fanconi anemia complementation group D1 605724	BRCA2
489	Fanconi anemia complementation group D2 227646	FANCD2
489	Fanconi anemia complementation group E 600901	FANCE
489	Fanconi anemia complementation group F 603467	FANCF
489	Fanconi anemia complementation group G 614082	FANCG

489	Fanconi anemia complementation group I 609053	FANCI
489	Fanconi anemia complementation group J 609054	BRIP1
489	Fanconi anemia complementation group L 614083	FANCL
489	Fanconi anemia complementation group N 610832	PALB2
489	Fanconi anemia complementation group O 613390	RAD51C
489	Fanconi anemia complementation group P 613951	SLX4
489	Fanconi anemia complementation group Q 615272	ERCC4
489	Fanconi anemia complementation group T 616435	UBE2T
490	Fanconi renotubular syndrome 4 with maturity-onset diabetes of the young 616026	HNF4A
490	Fanconi-Bickel syndrome 227810	SLC2A2
491	Farber lipogranulomatosis 228000	ASAH1
492	Fascioscapulohumeral muscular dystrophy 2 digenic 158901	SMCHD1
493	Fatty liver acute of pregnancy 609016	HADHA
493	LCHAD deficiency 609016	HADHA
494	Favism 134700	G6PD
495	Febrile seizures familial 11 614418	CPA6
495	Febrile seizures familial 3A 604403	SCN1A
495	Febrile seizures familial 3B 613863	SCN9A
495	Febrile seizures familial 8 611277	GABRG2
496	Fechtner syndrome 153640	MYH9
497	Feingold syndrome 1 164280	MYCN
497	Feingold syndrome 2 614326	MIR17HG
498	Fetal akinesia deformation sequence 208150	MUSK
498	Fetal akinesia deformation sequence 208150	RAPSN
499	Fetal hemoglobin quantitative trait locus 1 141749	HBG1
499	Fetal hemoglobin quantitative trait locus 1 141749	HBG2
499	Hereditary persistence of fetal hemoglobin 141749	HBB
500	Fibrochondrogenesis 1 228520	COL11A1
500	Fibrochondrogenesis 2 614524	COL11A2
501	Fibrodysplasia ossificans progressiva 135100	ACVR1
502	Fibromatosis gingival 5 617626	REST
503	Fibrosis of extraocular muscles congenital 1 135700	KIF21A
503	Fibrosis of extraocular muscles congenital 2 602078	PHOX2A
503	Fibrosis of extraocular muscles congenital 3A 600638	TUBB3
503	Fibrosis of extraocular muscles congenital 3B 135700	KIF21A
503	Fibrosis of extraocular muscles congenital 5 616219	COL25A1
504	Filippi syndrome 272440	CKAP2L
505	Fish-eye disease 136120	LCAT
506	Fletcher factor (prekallikrein) deficiency 612423	KLKB1
507	Floating-Harbor syndrome 136140	SRCAP
508	Focal cortical dysplasia type II somatic 607341	MTOR
508	Focal cortical dysplasia type II somatic 607341	TSC1
509	Focal dermal hypoplasia 305600	PORCN
510	Focal facial dermal dysplasia 3 Setleis type 227260	TWIST2

510	Focal facial dermal dysplasia 4 614974	CYP26C1
511	Focal segmental glomerulosclerosis 8 616032	ANLN
511	Focal segmental glomerulosclerosis 9 616220	CRB2
512	Folate malabsorption hereditary 229050	SLC46A1
513	Foveal hypoplasia 1 136520	PAX6
513	Foveal hypoplasia 2 with or without optic nerve misrouting and or anterior segment dysgenesis 609218	SLC38A8
514	Fragile X syndrome 300624	FMR1
514	Fragile X tremor or ataxia syndrome 300623	FMR1
515	Frank-ter Haar syndrome 249420	SH3PXD2B
516	Fraser syndrome 1 219000	FRAS1
516	Fraser syndrome 2 617666	FREM2
516	Fraser syndrome 3 617667	GRIP1
517	Frasier syndrome 136680	WT1
518	Friedreich ataxia 229300	FXN
518	Friedreich ataxia with retained reflexes 229300	FXN
519	Frontometaphyseal dysplasia 1 305620	FLNA
519	Frontometaphyseal dysplasia 2 617137	MAP3K7
520	Frontonasal dysplasia 1 136760	ALX3
520	Frontonasal dysplasia 2 613451	ALX4
521	Frontotemporal dementia and or amyotrophic lateral sclerosis 1 105550	C9orf72
521	Frontotemporal dementia and or amyotrophic lateral sclerosis 2 615911	CHCHD10
521	Frontotemporal dementia and or amyotrophic lateral sclerosis 3 616437	SQSTM1
521	Frontotemporal dementia and or amyotrophic lateral sclerosis 4 616439	TBK1
522	Frontotemporal lobar degeneration TARDBP-related 612069	TARDBP
522	Frontotemporal lobar degeneration with ubiquitin-positive inclusions 607485	GRN
522	Aphasia primary progressive 607485	GRN
523	Fructose intolerance hereditary 229600	ALDOB
523	Fructose-16-bisphosphatase deficiency 229700	FBP1
524	Fucosidosis 230000	FUCA1
525	Fucosyltransferase 6 deficiency 613852	FUT6
526	Fuhrmann syndrome 228930	WNT7A
527	Fumarase deficiency 606812	FH
528	Fundus albipunctatus 136880	RDH5
528	Fundus albipunctatus 136880	RLBP1
528	Fundus flavimaculatus 248200	ABCA4
529	GABA-transaminase deficiency 613163	ABAT
530	GAPO syndrome 230740	ANTXR1
531	GLUT1 deficiency syndrome 1 infantile onset severe 606777	SLC2A1
531	GLUT1 deficiency syndrome 2 childhood onset 612126	SLC2A1

532	GM1-gangliosidosis type I 230500	GLB1
532	GM1-gangliosidosis type II 230600	GLB1
532	GM1-gangliosidosis type III 230650	GLB1
532	GM2-gangliosidosis AB variant 272750	GM2A
532	GM2-gangliosidosis several forms 272800	HEXA
532	Tay-Sachs disease 272800	HEXA
533	GRACILE syndrome 603358	BCS1L
534	Gabriele-de Vries syndrome 617557	YY1
535	Galactokinase deficiency with cataracts 230200	GALK1
536	Galactose epimerase deficiency 230350	GALE
537	Galactosemia 230400	GALT
538	Galactosialidosis 256540	CTSA
539	Gallbladder disease 1 600803	ABCB4
539	{Gallbladder disease 4} 611465	ABCG8
540	Galloway-Mowat syndrome 1 251300	WDR73
540	Galloway-Mowat syndrome 2 X-linked 301006	LAGE3
540	Galloway-Mowat syndrome 3 617729	OSGEP
540	Galloway-Mowat syndrome 4 617730	TP53RK
540	Galloway-Mowat syndrome 5 617731	TPRKB
541	Gastric cancer familial diffuse with or without cleft lip and or palate 137215	CDH1
541	Gastric cancer somatic 137215	KRAS
541	Gastric cancer somatic 613659	APC
541	Gastric cancer somatic 613659	CASP10
541	Gastric cancer somatic 613659	ERBB2
541	Gastric cancer somatic 613659	FGFR2
541	Gastric cancer somatic 613659	IRF1
541	Gastric cancer somatic 613659	KLF6
541	Gastric cancer somatic 613659	MUTYH
541	Gastric cancer somatic 613659	PIK3CA
541	{Gastric cancer risk after H. pylori infection} 137215	IL1B
541	{Gastric cancer risk after H. pylori infection} 137215	IL1RN
542	Gastrointestinal defects and immunodeficiency syndrome 243150	TTC7A
543	Gastrointestinal stromal tumor 606764	SDHB
543	Gastrointestinal stromal tumor 606764	SDHC
543	Gastrointestinal stromal tumor familial 606764	KIT
543	Gastrointestinal stromal tumor somatic 606764	PDGFRA
544	Gaucher disease atypical 610539	PSAP
544	Gaucher disease perinatal lethal 608013	GBA
544	Gaucher disease type I 230800	GBA
544	Gaucher disease type II 230900	GBA
544	Gaucher disease type III 231000	GBA
544	Gaucher disease type IIIC 231005	GBA
545	Gaze palsy familial horizontal with progressive scoliosis 1 607313	ROBO3
545	Gaze palsy familial horizontal with progressive scoliosis 2 617542	DCC

546	Geleophysic dysplasia 1 231050	ADAMTSL2
546	Geleophysic dysplasia 2 614185	FBN1
547	Generalized epilepsy with febrile seizures plus type 9 616172	STX1B
548	Genitopatellar syndrome 606170	KAT6B
549	Geroderma osteodysplasticum 231070	GORAB
550	Ghosal hematodiaphyseal syndrome 231095	TBXAS1
551	Giant axonal neuropathy-1 256850	GAN
552	Gillespie syndrome 206700	ITPR1
553	Gillessen-Kaesbach-Nishimura syndrome 263210	ALG9
554	Gitelman syndrome 263800	SLC12A3
555	Glanzmann thrombasthenia 273800	ITGA2B
555	Glanzmann thrombasthenia 273800	ITGB3
556	Glass syndrome 612313	SATB2
557	Glaucoma 1 open angle 1O 613100	NTF4
557	Glaucoma 1 open angle E 137760	OPTN
557	Glaucoma 1 open angle F 603383	ASB10
557	Glaucoma 1 open angle G 609887	WDR36
557	Glaucoma 1A primary open angle 137750	MYOC
557	Glaucoma 3 primary congenital D 613086	LTBP2
557	Glaucoma 3 primary congenital E 617272	TEK
557	Glaucoma 3A primary open angle congenital juvenile or adult onset 231300	CYP1B1
558	Glioblastoma somatic 137800	ERBB2
558	{Glioblastoma 3} 613029	BRCA2
558	{Glioma susceptibility 1} 137800	TP53
558	{Glioma susceptibility to somatic} 137800	IDH1
558	{Glioma susceptibility 2} 613028	PTEN
558	{Glioma susceptibility 9} 616568	POT1
559	Global developmental delay absent or hypoplastic corpus callosum and dysmorphic facies 617260	ZNF148
560	Glomerulocystic kidney disease with hyperuricemia and isosthenuria 609886	UMOD
561	Glomerulopathy with fibronectin deposits 2 601894	FN1
562	Glomerulosclerosis focal segmental 1 603278	ACTN4
562	Glomerulosclerosis focal segmental 2 603965	TRPC6
562	Glomerulosclerosis focal segmental 3 607832	CD2AP
562	Glomerulosclerosis focal segmental 5 613237	INF2
562	Glomerulosclerosis focal segmental 6 614131	MYO1E
562	Glomerulosclerosis focal segmental 7 616002	PAX2
562	{Glomerulosclerosis focal segmental 4 susceptibility to} 612551	APOL1
562	{End-stage renal disease nondiabetic susceptibility to} 612551	APOL1
563	Glomuvenous malformations 138000	GLML
564	Glucocorticoid deficiency 2 607398	MRAP
564	Glucocorticoid deficiency 4 with or without mineralocorticoid deficiency 614736	NNT

564	Glucocorticoid deficiency due to ACTH unresponsiveness 202200	MC2R
564	Glucocorticoid resistance 615962	NR3C1
565	Glucose or galactose malabsorption 606824	SLC5A1
566	Glutamate formiminotransferase deficiency 229100	FTCD
567	Glutamine deficiency congenital 610015	GLUL
568	Glutaric acidemia IIA 231680	ETFA
568	Glutaric acidemia IIB 231680	ETFB
568	Glutaric acidemia IIC 231680	ETFDH
568	Glutaric aciduria III 231690	SUGCT
568	Glutaric aciduria type I 231670	GCDH
569	Glutathione synthetase deficiency 266130	GSS
570	Glycerol kinase deficiency 307030	GK
571	Glycine N-methyltransferase deficiency 606664	GNMT
571	Glycine encephalopathy 605899	AMT
571	Glycine encephalopathy 605899	GCSH
571	Glycine encephalopathy 605899	GLDC
571	Glycine encephalopathy with normal serum glycine 617301	SLC6A9
572	Glycogen storage disease 0 liver 240600	GYS2
572	Glycogen storage disease 0 muscle 611556	GYS1
572	Glycogen storage disease II 232300	GAA
572	Glycogen storage disease IIIa 232400	AGL
572	Glycogen storage disease IIIb 232400	AGL
572	Glycogen storage disease IV 232500	GBE1
572	Glycogen storage disease IXc 613027	PHKG2
572	Glycogen storage disease Ia 232200	G6PC
572	Glycogen storage disease Ib 232220	SLC37A4
572	Glycogen storage disease Ic 232240	SLC37A4
572	Glycogen storage disease VI 232700	PYGL
572	Glycogen storage disease VII 232800	PFKM
572	Glycogen storage disease X 261670	PGAM2
572	Glycogen storage disease XI 612933	LDHA
572	Glycogen storage disease XII 611881	ALDOA
572	Glycogen storage disease of heart lethal congenital 261740	PRKAG2
572	Glycogen storage disease type IXa1 306000	PHKA2
572	Glycogen storage disease type IXa2 306000	PHKA2
573	Glycosylphosphatidylinositol deficiency 610293	PIGM
574	Gnathodiaphyseal dysplasia 166260	ANO5
575	Goiter multinodular 1 with or without Sertoli-Leydig cell tumors 138800	DICER1
576	Goldberg-Shprintzen megacolon syndrome 609460	KIAA1279
577	Gracile bone dysplasia 602361	FAM111A
578	Grange syndrome 602531	YY1AP1
579	Gray platelet syndrome 139090	NBEAL2
580	Greenberg skeletal dysplasia 215140	LBR
581	Greig cephalopolysyndactyly syndrome 175700	GLI3

582	Griscelli syndrome type 1 214450	MYO5A
582	Griscelli syndrome type 2 607624	RAB27A
582	Griscelli syndrome type 3 609227	MLPH
583	Growth hormone deficiency isolated partial 615925	GHSR
583	Growth hormone deficiency isolated type IA 262400	GH1
583	Growth hormone deficiency isolated type IB 612781	GH1
583	Growth hormone deficiency isolated type IB 612781	GHRHR
583	Growth hormone deficiency isolated type II 173100	GH1
583	Growth hormone deficiency with pituitary anomalies 182230	HESX1
583	Septooptic dysplasia 182230	HESX1
583	Growth hormone insensitivity partial 604271	GHR
583	Growth hormone insensitivity with immunodeficiency 245590	STAT5B
583	Increased responsiveness to growth hormone	GHR
584	Growth retardation developmental delay facial dysmorphism 612938	FTO
584	Growth retardation intellectual developmental disorder hypotonia and hepatopathy 617093	IARS
584	Growth retardation with deafness and mental retardation due to IGF1 deficiency 608747	IGF1
585	Guttmacher syndrome 176305	HOXA13
586	Gyrate atrophy of choroid and retina with or without ornithinemia 258870	OAT
587	HARP syndrome 607236	PANK2
588	HDL deficiency type 2 604091	ABCA1
588	{HDL response to hormone replacement, augmented}	ESR1
589	HELIX syndrome 617671	CLDN10
590	HELLP syndrome maternal of pregnancy 609016	HADHA
591	HMG-CoA lyase deficiency 246450	HMGCL
591	HMG-CoA synthase-2 deficiency 605911	HMGCS2
592	HPRT-related gout 300323	HPRT1
593	HSD10 mitochondrial disease 300438	HSD17B10
594	Hailey-Hailey disease 169600	ATP2C1
595	Haim-Munk syndrome 245010	CTSC
596	Hajdu-Cheney syndrome 102500	NOTCH2
597	Hamamy syndrome 611174	IRX5
598	Hand-foot-uterus syndrome 140000	HOXA13
599	Coproporphyria 121300	CPOX
599	Harderoporphyria 121300	CPOX
600	Harel-Yoon syndrome 617183	ATAD3A
601	Hartnup disorder 234500	SLC6A19
602	Hartsfield syndrome 615465	FGFR1
603	Hawkinsinuria 140350	HPD
604	Hay-Wells syndrome 106260	TP63
605	Heart and brain malformation syndrome 616920	C19orf61
606	Heart block nonprogressive 113900	SCN5A

606	Heart block progressive type IA 113900	SCN5A
607	Heart-hand syndrome Slovenian type 610140	LMNA
608	Heimler syndrome 1 234580	PEX1
608	Heimler syndrome 2 616617	PEX6
609	Heinz body anemia 140700	HBA2
609	Heinz body anemias alpha- 140700	HBA1
609	Heinz body anemias beta- 140700	HBB
610	Helsmoortel-van der Aa syndrome 615873	ADNP
611	Hemangioma capillary infantile somatic 602089	FLT4
611	Hemangioma capillary infantile somatic 602089	KDR
611	{Hemangioma capillary infantile susceptibility to} 602089	KDR
612	Hematuria benign familial 141200	COL4A3
613	Heme oxygenase-1 deficiency 614034	HMOX1
614	Hemochromatosis 235200	HFE
614	{HFE hemochromatosis modifier of} 235200	BMP2
614	Hemochromatosis type 2A 602390	HJV
614	Hemochromatosis type 2B 613313	HAMP
614	Hemochromatosis type 3 604250	TFR2
614	Hemochromatosis type 4 606069	SLC40A1
615	Hemoglobin H disease nondeletional 613978	HBA1
615	Hemoglobin H disease nondeletional 613978	HBA2
616	Hemolytic anemia CD59-mediated with or without immune-mediated polyneuropathy 612300	CD59
616	Hemolytic anemia due to G6PD deficiency 300908	G6PD
616	Hemolytic anemia due to adenylate kinase deficiency 612631	AK1
616	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency 230450	GCLC
616	Hemolytic anemia due to glutathione synthetase deficiency 231900	GSS
616	Hemolytic anemia due to hexokinase deficiency 235700	HK1
616	Hemolytic anemia due to triosephosphate isomerase deficiency 615512	TPI1
616	Hemolytic anemia nonspherocytic due to glucose phosphate isomerase deficiency 613470	GPI
617	Hemophagocytic lymphohistiocytosis familial 2 603553	PRF1
617	Hemophagocytic lymphohistiocytosis familial 3 608898	UNC13D
617	Hemophagocytic lymphohistiocytosis familial 4 603552	STX11
617	Hemophagocytic lymphohistiocytosis familial 5 613101	STXBP2
618	Hemophilia A 306700	F8
618	Hemophilia B 306900	F9
619	Hemorrhagic destruction of the brain subependymal calcification and cataracts 613730	JAM3
620	Hennekam lymphangiectasia-lymphedema syndrome 1 235510	CCBE1
620	Hennekam lymphangiectasia-lymphedema syndrome 2 616006	FAT4
621	Hepatic lipase deficiency 614025	LIPC

622	Hepatic venoocclusive disease with immunodeficiency 235550	SP110
623	Hepatic adenoma somatic 142330	HNF1A
623	Hepatoblastoma somatic 114550	APC
623	Hepatocellular cancer somatic 114550	PDGFRL
623	Hepatocellular carcinoma 114550	TP53
623	Hepatocellular carcinoma childhood type somatic 114550	MET
623	Hepatocellular carcinoma somatic 114550	AXIN1
623	Hepatocellular carcinoma somatic 114550	CASP8
623	Hepatocellular carcinoma somatic 114550	CTNNB1
623	Hepatocellular carcinoma somatic 114550	IGF2R
623	Hepatocellular carcinoma somatic 114550	PIK3CA
624	Hereditary motor and sensory neuropathy Okinawa type 604484	TFG
624	Hereditary motor and sensory neuropathy VIA 601152	MFN2
624	Hereditary motor and sensory neuropathy type IIc 606071	TRPV4
625	Hermansky-Pudlak syndrome 1 203300	HPS1
625	Hermansky-Pudlak syndrome 2 608233	AP3B1
625	Hermansky-Pudlak syndrome 3 614072	HPS3
625	Hermansky-Pudlak syndrome 4 614073	HPS4
625	Hermansky-Pudlak syndrome 5 614074	HPS5
625	Hermansky-Pudlak syndrome 6 614075	HPS6
625	Hermansky-Pudlak syndrome 7 614076	DTNBP1
625	Hermansky-Pudlak syndrome 8 614077	BLOC1S3
626	Heterotaxy visceral 1 X-linked 306955	ZIC3
626	Heterotaxy visceral 2 autosomal 605376	CFC1
626	Heterotaxy visceral 4 autosomal 613751	ACVR2B
626	Heterotaxy visceral 5 270100	NODAL
626	Heterotaxy visceral 6 autosomal recessive 614779	CFAP53
626	Heterotaxy visceral 7 autosomal 616749	MMP21
626	Heterotaxy visceral 8 autosomal 617205	PKD1L1
627	Heterotopia periventricular 300049	FLNA
628	Histiocytoma angiomatoid fibrous somatic 612160	CREB1
629	Histiocytosis-lymphadenopathy plus syndrome 602782	SLC39A5
629	Histiocytosis-lymphadenopathy plus syndrome 602782	SLC29A3
630	Holocarboxylase synthetase deficiency 253270	HLCS
631	Holoprosencephaly 11 614226	CDON
631	Holoprosencephaly 2 157170	SIX3
631	Holoprosencephaly 3 142945	SHH
631	Holoprosencephaly 4 142946	TGIF1
631	Holoprosencephaly 5 609637	ZIC2
631	Holoprosencephaly 7 610828	PTCH1
631	Holoprosencephaly 9 610829	GLI2
632	Holt-Oram syndrome 142900	TBX5
633	Homocystinuria B6-responsive and nonresponsive types 236200	CBS
633	Thrombosis hyperhomocysteinemic 236200	CBS
633	Homocystinuria cblD type variant 1 277410	MMADHC

633	Homocystinuria due to MTHFR deficiency 236250	MTHFR
633	Homocystinuria-megaloblastic anemia cbl E type 236270	MTRR
633	Homocystinuria-megaloblastic anemia cblG complementation type 250940	MTR
634	Huntington disease 143100	HTT
634	Huntington disease-like 1 603218	PRNP
635	Hutchinson-Gilford progeria 176670	LMNA
636	Hyaline fibromatosis syndrome 228600	ANTXR2
637	Hydatidiform mole recurrent 1 231090	NLRP7
637	Hydatidiform mole recurrent 2 614293	KHDC3L
638	Hydrocephalus due to aqueductal stenosis 307000	L1CAM
638	Hydrocephalus nonsyndromic autosomal recessive 2 615219	MPDZ
638	Hydrocephalus nonsyndromic autosomal recessive 236600	CCDC88C
638	Hydrocephalus with Hirschsprung disease 307000	L1CAM
638	Hydrocephalus with congenital idiopathic intestinal pseudoobstruction 307000	L1CAM
639	Hydrolethalus syndrome 236680	HYLS1
640	Hyper-IgD syndrome 260920	MVK
641	Hyper-IgE recurrent infection syndrome 147060	STAT3
641	Hyper-IgE recurrent infection syndrome autosomal recessive 243700	DOCK8
642	Hyperaldosteronism familial type III 613677	KCNJ5
642	Hyperaldosteronism familial type IV 617027	CACNA1H
643	Hyperalphalipoproteinemia 143470	CETP
644	Hyperammonemia due to carbonic anhydrase VA deficiency 615751	CA5A
645	Hyperbilirubinemia Rotor type digenic 237450	SLCO1B1
645	Hyperbilirubinemia Rotor type digenic 237450	SLCO1B3
645	Hyperbilirubinemia familial transient neonatal 237900	UGT1A1
646	Hyperbiliverdinemia 614156	BLVRA
647	Hypercalcemia infantile 1 143880	CYP24A1
647	Hypercalcemia infantile 2 616963	SLC34A1
648	Hypercarotenemia and vitamin A deficiency autosomal dominant 115300	BCMO1
649	Hyperchlorhidrosis isolated 143860	CA12
650	Hypercholanemia familial 607748	BAAT
650	Hypercholanemia familial 607748	EPHX1
650	Hypercholanemia familial 607748	TJP2
651	Hypercholesterolemia due to ligand-defective apo B 144010	APOB
651	Hypercholesterolemia familial 143890	LDLR
651	Hypercholesterolemia familial 3 603776	PCSK9
651	{Low density lipoprotein cholesterol level QTL 1} 603776	PCSK9
651	Hypercholesterolemia familial autosomal recessive 603813	LDLRAP1
651	{Hypercholesterolemia familial due to LDLR defect modifier of} 143890	EPHX2

651	{Hypercholesterolemia familial modifier of} 143890	APOA2
651	{Hypercholesterolemia familial modifier of} 143890	GHR
651	{Hypercholesterolemia susceptibility to} 143890	PPP1R17
651	{Hypercholesterolemia susceptibility to} 143890	ITIH4
652	Hyperchylomicronemia late-onset 144650	APOA5
653	Hyperekplexia 2 autosomal recessive 614619	GLRB
653	Hyperekplexia 3 614618	SLC6A5
653	Hyperekplexia hereditary 1 autosomal dominant or recessive 149400	GLRA1
654	Hypereosinophilic syndrome idiopathic resistant to imatinib 607685	PDGFRA
655	Hyperferritinemia-cataract syndrome 600886	FTL
656	Hyperglycinemia lactic acidosis and seizures 614462	LIAS
656	Hyperglycinuria 138500	SLC36A2
656	Hyperglycinuria 138500	SLC6A19
656	Hyperglycinuria 138500	SLC6A20
657	Hyperinsulinemic hypoglycemia familial 1 256450	ABCC8
657	Hyperinsulinemic hypoglycemia familial 2 601820	KCNJ11
657	Hyperinsulinemic hypoglycemia familial 3 602485	GCK
657	Hyperinsulinemic hypoglycemia familial 4 609975	HADH
657	Hyperinsulinemic hypoglycemia familial 5 609968	INSR
657	Hyperinsulinemic hypoglycemia familial 7 610021	SLC16A1
658	Hyperinsulinism-hyperammonemia syndrome 606762	GLUD1
659	Hyperkalemic periodic paralysis type 2 170500	SCN4A
659	Hypokalemic periodic paralysis type 1 170400	CACNA1S
659	Hypokalemic periodic paralysis type 2 613345	SCN4A
660	Hyperlipoproteinemia type 1D 615947	GPIHBP1
660	Hyperlipoproteinemia type III 617347	APOE
660	Hyperlipoproteinemia type Ib 207750	APOC2
661	Hyperlysinemia 238700	AASS
662	Hypermanganesemia with dystonia 1 613280	SLC30A10
662	Hypermanganesemia with dystonia 2 617013	SLC39A14
663	Hypermethioninemia due to adenosine kinase deficiency 614300	ADK
663	Hypermethioninemia persistent autosomal dominant due to methionine adenosyltransferase I or III deficiency 250850	MAT1A
663	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase 613752	AHCY
663	Methionine adenosyltransferase deficiency autosomal recessive 250850	MAT1A
664	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome 238970	SLC25A15
665	Hyperoxaluria primary type 1 259900	AGXT
665	Hyperoxaluria primary type II 260000	GRHPR
665	Hyperoxaluria primary type III 613616	HOGA1
666	Hyperparathyroidism 4 617343	GCM2

666	Hyperparathyroidism familial primary 145000	CDC73
666	Hyperparathyroidism neonatal 239200	CASR
666	Hyperparathyroidism-jaw tumor syndrome 145001	CDC73
667	Hyperphenylalaninemia BH4-deficient A 261640	PTS
667	Hyperphenylalaninemia BH4-deficient B 233910	GCH1
667	Hyperphenylalaninemia BH4-deficient C 261630	QDPR
667	Hyperphenylalaninemia BH4-deficient D 264070	PCBD1
667	Hyperphenylalaninemia mild non-BH4-deficient 617384	DNAJC12
668	Hyperphosphatasia with mental retardation syndrome 1 239300	PIGV
668	Hyperphosphatasia with mental retardation syndrome 2 614749	PIGO
668	Hyperphosphatasia with mental retardation syndrome 3 614207	PGAP2
668	Hyperphosphatasia with mental retardation syndrome 4 615716	PGAP3
668	Hyperphosphatasia with mental retardation syndrome 6 616809	PIGY
669	Hyperpigmentation with or without hypopigmentation 145250	KITLG
670	Hyperproinsulinemia 616214	INS
671	Hyperprolinemia type I 239500	PRODH
671	Hyperprolinemia type II 239510	ALDH4A1
672	Hypertension and brachydactyly syndrome 112410	PDE3A
672	Hypertension early-onset autosomal dominant with exacerbation in pregnancy 605115	NR3C2
672	Hypertension essential 145500	PTGIS
672	{Hypertension essential salt-sensitive} 145500	ADD1
672	{Hypertension essential susceptibility to} 145500	AGT
672	{Hypertension essential susceptibility to} 145500	ECE1
672	{Hypertension essential susceptibility to} 145500	GNB3
672	{Hypertension essential} 145500	AGTR1
672	{Hypertension salt-sensitive essential susceptibility to} 145500	CYP3A5
672	{Hypertension susceptibility to} 145500	NOS3
672	{Hypertension diastolic resistance to} 608622	KCNMB1
672	{Hypertension insulin resistance-related susceptibility to} 125853	RETN
672	{Hypertension pregnancy-induced} 189800	NOS3
673	Hyperthyroidism familial gestational 603373	TSHR
673	Hyperthyroidism nonautoimmune 609152	TSHR
674	Hypertrichotic osteochondrodysplasia 239850	ABCC9
675	Hypertriglyceridemia transient infantile 614480	GPD1
675	{Hypertriglyceridemia susceptibility to} 145750	APOA5
675	{Hypertriglyceridemia susceptibility to} 145750	LIPI
676	Cranioosteoarthropathy 259100	HPGD
676	Hypertrophic osteoarthropathy primary autosomal recessive 1 259100	HPGD
676	Hypertrophic osteoarthropathy primary autosomal recessive 2 614441	SLCO2A1
677	Hyperuricemia pulmonary hypertension renal failure and alkalosis 613845	SARS2
678	Hyperuricemic nephropathy familial juvenile 1 162000	UMOD

678	Hyperuricemic nephropathy familial juvenile 2 613092	REN
678	Hyperuricemic nephropathy familial juvenile 4 617056	SEC61A1
679	Hypoaldosteronism congenital due to CMO I deficiency 203400	CYP11B2
679	Hypoaldosteronism congenital due to CMO II deficiency 610600	CYP11B2
680	Hypoalphalipoproteinemia 604091	APOA1
680	Hypoalphalipoproteinemia 604091	ABCA1
681	Hypobetalipoproteinemia 615558	APOB
681	Hypobetalipoproteinemia familial 2 605019	ANGPTL3
682	Hypocalcemia autosomal dominant 2 615361	GNA11
682	Hypocalcemia autosomal dominant 601198	CASR
682	Hypocalcemia autosomal dominant with Bartter syndrome 601198	CASR
683	Hypocalciuric hypercalcemia type I 145980	CASR
683	Hypocalciuric hypercalcemia type II 145981	GNA11
683	Hypocalciuric hypercalcemia type III 600740	AP2S1
684	Hypochondroplasia 146000	FGFR3
685	Hypoglycemia of infancy leucine-sensitive 240800	ABCC8
686	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1) 308700	ANOS1
686	Hypogonadotropic hypogonadism 10 with or without anosmia 614839	TAC3
686	Hypogonadotropic hypogonadism 11 with or without anosmia 614840	TACR3
686	Hypogonadotropic hypogonadism 14 with or without anosmia 614858	WDR11
686	Hypogonadotropic hypogonadism 17 with or without anosmia 615266	SPRY4
686	Hypogonadotropic hypogonadism 18 with or without anosmia 615267	IL17RD
686	Hypogonadotropic hypogonadism 19 with or without anosmia 615269	DUSP6
686	Hypogonadotropic hypogonadism 2 with or without anosmia 147950	FGFR1
686	Hypogonadotropic hypogonadism 20 with or without anosmia 615270	FGF17
686	Hypogonadotropic hypogonadism 21 with anosmia 615271	FLRT3
686	Hypogonadotropic hypogonadism 22 with or without anosmia 616030	FEZF1
686	Hypogonadotropic hypogonadism 23 with or without anosmia 228300	LHB
686	Hypogonadotropic hypogonadism 24 without anosmia 229070	FSHB
686	Hypogonadotropic hypogonadism 3 with or without anosmia 244200	PROKR2
686	Hypogonadotropic hypogonadism 4 with or without anosmia 610628	PROK2

686	Hypogonadotropic hypogonadism 5 with or without anosmia 612370	CHD7
686	Hypogonadotropic hypogonadism 6 with or without anosmia 612702	FGF8
686	Hypogonadotropic hypogonadism 7 without anosmia 146110	GNRHR
686	Hypogonadotropic hypogonadism 8 with or without anosmia 614837	KISS1R
686	Hypogonadotropic hypogonadism 9 with or without anosmia 614838	NSMF
686	{Hypogonadotropic hypogonadism 15 with or without anosmia} 614880	HS6ST1
686	{Hypogonadotropic hypogonadism 16 with or without anosmia} 614897	SEMA3A
687	Hypoinsulinemic hypoglycemia with hemihypertrophy 240900	AKT2
688	Hypomagnesemia 1 intestinal 602014	TRPM6
688	Hypomagnesemia 2 renal 154020	FXYD2
688	Hypomagnesemia 3 renal 248250	CLDN16
688	Hypomagnesemia 4 renal 611718	EGF
688	Hypomagnesemia 5 renal with ocular involvement 248190	CLDN19
688	Hypomagnesemia 6 renal 613882	CNNM2
688	Hypomagnesemia seizures and mental retardation 616418	CNNM2
689	Hypomyelination with brainstem and spinal cord involvement and leg spasticity 615281	DARS
690	Hypoparathyroidism autosomal dominant 146200	PTH
690	Hypoparathyroidism autosomal recessive 146200	PTH
690	Hypoparathyroidism familial isolated 146200	GCM2
690	Hypoparathyroidism sensorineural deafness and renal dysplasia 146255	GATA3
691	Hypoparathyroidism-retardation-dysmorphism syndrome 241410	TBCE
692	Hypophosphatasia adult 146300	ALPL
692	Odontohypophosphatasia 146300	ALPL
692	Hypophosphatasia childhood 241510	ALPL
692	Hypophosphatasia infantile 241500	ALPL
693	Hypophosphatemic rickets 300554	CLCN5
693	Hypophosphatemic rickets AR 241520	DMP1
693	Hypophosphatemic rickets X-linked dominant 307800	PHEX
693	Hypophosphatemic rickets autosomal dominant 193100	FGF23
693	Hypophosphatemic rickets autosomal recessive 2 613312	ENPP1
693	Hypophosphatemic rickets with hypercalciuria 241530	SLC34A3
694	Hypoplastic left heart syndrome 1 241550	GJA1
694	Hypoplastic left heart syndrome 2 614435	NKX2-5
695	Hypoplastic or aplastic tibia with polydactyly 188740	LMBR1
696	Hypoprothrombinemia 613679	F2
696	Dysprothrombinemia 613679	F2
697	Hypospadias 1 X-linked 300633	AR

697	Hypospadias 2 X-linked 300758	MAMLD1
698	Hypothyroidism central and testicular enlargement 300888	IGSF1
698	Hypothyroidism congenital due to thyroid dysgenesis or hypoplasia 218700	PAX8
698	Hypothyroidism congenital nongoitrous 1 275200	TSHR
698	Hypothyroidism congenital nongoitrous 4 275100	TSHB
698	Hypothyroidism congenital nongoitrous 5 225250	NKX2-5
698	Hypothyroidism congenital nongoitrous 6 614450	THRA
699	Hypotonia ataxia and delayed development syndrome 617330	EBF3
700	Hypotonia infantile with psychomotor retardation 616816	CCDC174
700	Hypotonia infantile with psychomotor retardation and characteristic facies 1 615419	NALCN
700	Hypotonia infantile with psychomotor retardation and characteristic facies 2 616801	UNC80
700	Hypotonia infantile with psychomotor retardation and characteristic facies 3 616900	TBCK
701	Hypotrichosis 1 605389	APCDD1
701	Hypotrichosis 11 615059	SNRPE
701	Hypotrichosis 12 615885	RPL21
701	Hypotrichosis 2 146520	CDSN
701	Hypotrichosis 4 146550	HR
701	Hypotrichosis 6 607903	DSG4
701	Hypotrichosis 7 604379	LIPH
701	Hypotrichosis 8 278150	LPAR6
701	Hypotrichosis congenital with juvenile macular dystrophy 601553	CDH3
701	Hypotrichosis-lymphedema-telangiectasia syndrome 607823	SOX18
701	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome 137940	SOX18
702	Hypouricemia renal 2 612076	SLC2A9
702	{Uric acid concentration serum QTL 2} 612076	SLC2A9
702	Hypouricemia renal 220150	SLC22A12
703	Hystrix-like ichthyosis with deafness 602540	GJB2
704	IFAP syndrome with or without BRESHECK syndrome 308205	MBTPS2
705	IMAGE syndrome 614732	CDKN1C
706	IRAK4 deficiency 607676	IRAK4
707	IVIC syndrome 147750	SALL4
708	Ichthyosis X-linked 308100	STS
708	Ichthyosis bullosa of Siemens 146800	KRT2
708	Ichthyosis congenital autosomal recessive 1 242300	TGM1
708	Ichthyosis congenital autosomal recessive 10 615024	PNPLA1
708	Ichthyosis congenital autosomal recessive 11 602400	ST14
708	Ichthyosis congenital autosomal recessive 12 617320	CASP14
708	Ichthyosis congenital autosomal recessive 13 617574	SDR9C7
708	Ichthyosis congenital autosomal recessive 14 617571	SULT2B1
708	Ichthyosis congenital autosomal recessive 2 242100	ALOX12B

708	Ichthyosis congenital autosomal recessive 3 606545	ALOXE3
708	Ichthyosis congenital autosomal recessive 4A 601277	ABCA12
708	Ichthyosis congenital autosomal recessive 4B (harlequin) 242500	ABCA12
708	Ichthyosis congenital autosomal recessive 5 604777	CYP4F22
708	Ichthyosis congenital autosomal recessive 6 612281	NIPAL4
708	Ichthyosis congenital autosomal recessive 8 613943	LIPN
708	Ichthyosis congenital autosomal recessive 9 615023	CERS3
708	Ichthyosis cyclic with epidermolytic hyperkeratosis 607602	KRT1
708	Ichthyosis cyclic with epidermolytic hyperkeratosis 607602	KRT10
708	Ichthyosis histrix Curth-Macklin type 146590	KRT1
708	Ichthyosis leukocyte vacuoles alopecia and sclerosing cholangitis 607626	CLDN1
708	Ichthyosis prematurity syndrome 608649	SLC27A4
708	Ichthyosis spastic quadriplegia and mental retardation 614457	ELOVL4
708	Ichthyosis vulgaris 146700	FLG
708	Ichthyosis with confetti 609165	KRT10
709	Iminoglycinuria digenic 242600	SLC36A2
709	Iminoglycinuria digenic 242600	SLC6A19
709	Iminoglycinuria digenic 242600	SLC6A20
710	Immunodeficiency 10 612783	STIM1
710	Immunodeficiency 11A 615206	CARD11
710	Immunodeficiency 11B with atopic dermatitis 617638	CARD11
710	Immunodeficiency 12 615468	MALT1
710	Immunodeficiency 14 615513	PIK3CD
710	Immunodeficiency 15 615592	IKBKB
710	Immunodeficiency 17 CD3 gamma deficient 615607	CD3G
710	Immunodeficiency 18 615615	CD3E
710	Immunodeficiency 18 SCID variant 615615	CD3E
710	Immunodeficiency 19 615617	CD3D
710	Immunodeficiency 20 615707	FCGR3A
710	Immunodeficiency 21 614172	GATA2
710	Immunodeficiency 23 615816	PGM3
710	Immunodeficiency 24 615897	CTPS1
710	Immunodeficiency 26 with or without neurologic abnormalities 615966	PRKDC
710	Immunodeficiency 27A mycobacteriosis AR 209950	IFNGR1
710	Immunodeficiency 27B mycobacteriosis AD 615978	IFNGR1
710	Immunodeficiency 28 mycobacteriosis 614889	IFNGR2
710	Immunodeficiency 29 mycobacteriosis 614890	IL12B
710	Immunodeficiency 30 614891	IL12RB1
710	Immunodeficiency 31A mycobacteriosis autosomal dominant 614892	STAT1
710	Immunodeficiency 31B mycobacterial and viral infections autosomal recessive 613796	STAT1
710	Immunodeficiency 31C autosomal dominant 614162	STAT1

710	Immunodeficiency 32A mycobacteriosis autosomal dominant 614893	IRF8
710	Immunodeficiency 32B monocyte and dendritic cell deficiency autosomal recessive 614894	IRF8
710	Immunodeficiency 33 300636	IKBKG
710	Immunodeficiency 34 mycobacteriosis X-linked 300645	CYBB
710	Immunodeficiency 35 611521	TYK2
710	Immunodeficiency 36 616005	PIK3R1
710	Immunodeficiency 38 616126	ISG15
710	Immunodeficiency 40 616433	DOCK2
710	Immunodeficiency 41 with lymphoproliferation and autoimmunity 606367	IL2RA
710	Immunodeficiency 42 616622	RORC
710	Immunodeficiency 43 241600	B2M
710	Immunodeficiency 44 616636	STAT2
710	Immunodeficiency 46 616740	TFRC
710	Immunodeficiency 47 300972	ATP6AP1
710	Immunodeficiency 48 269840	ZAP70
710	Immunodeficiency 50 300988	MSN
710	Immunodeficiency 51 613953	IL17RA
710	Immunodeficiency 52 617514	LAT
710	Immunodeficiency 7 TCR-alpha or beta deficient 615387	TRAC
710	Immunodeficiency 8 615401	CORO1A
710	Immunodeficiency 9 612782	ORAI1
710	Immunodeficiency X-linked with hyper-IgM 308230	CD40LG
710	Immunodeficiency X-linked with magnesium defect Epstein-Barr virus infection and neoplasia 300853	MAGT1
710	Immunodeficiency common variable 1 607594	ICOS
710	Immunodeficiency common variable 10 615577	NFKB2
710	Immunodeficiency common variable 12 616576	NFKB1
710	Immunodeficiency common variable 13 616873	IKZF1
710	Immunodeficiency common variable 2 240500	TNFRSF13B
710	Immunodeficiency common variable 3 613493	CD19
710	Immunodeficiency common variable 4 613494	TNFRSF13C
710	Immunodeficiency common variable 5 613495	MS4A1
710	Immunodeficiency common variable 6 613496	CD81
710	Immunodeficiency common variable 7 614699	CR2
710	Immunodeficiency common variable 8 with autoimmunity 614700	LRBA
710	Immunodeficiency developmental delay and hypohomocysteinemia 617744	NFE2L2
710	Immunodeficiency due to defect in MAPBP-interacting protein 610798	LAMTOR2
710	Immunodeficiency due to ficolin 3 deficiency 613860	FCN3

710	Immunodeficiency due to purine nucleoside phosphorylase deficiency 613179	PNP
710	Immunodeficiency isolated 300584	IKBKG
710	Immunodeficiency primary autosomal recessive IL21R-related 615207	IL21R
710	Immunodeficiency with hyper IgM type 5 608106	UNG
710	Immunodeficiency with hyper-IgM type 2 605258	AICDA
710	Immunodeficiency with hyper-IgM type 3 606843	CD40
711	Immunodeficiency-centromeric instability-facial anomalies syndrome 1 242860	DNMT3B
711	Immunodeficiency-centromeric instability-facial anomalies syndrome 3 616910	CDCA7
711	Immunodeficiency-centromeric instability-facial anomalies syndrome 4 616911	HELLS
711	Immunodeficiency-centromeric instability-facial anomalies syndrome-2 614069	ZBTB24
712	Immunodysregulation polyendocrinopathy and enteropathy X-linked 304790	FOXP3
713	Immunoglobulin A deficiency 2 609529	TNFRSF13B
714	Immunoskeletal dysplasia with neurodevelopmental abnormalities 617425	EXTL3
715	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1 167320	VCP
716	Incontinentia pigmenti 308300	IKBKG
717	Infantile cerebellar-retinal degeneration 614559	ACO2
718	Infantile liver failure syndrome 2 616483	NBAS
719	Infantile neuroaxonal dystrophy 1 256600	PLA2G6
720	Infantile-onset multisystem neurologic endocrine and pancreatic disease 616263	PTRH2
721	Infections recurrent with encephalopathy hepatic dysfunction and cardiovascular malformations 613759	FADD
722	Inflammatory bowel disease 25 early onset autosomal recessive 612567	IL10RB
722	Inflammatory bowel disease 28 early onset autosomal recessive 613148	IL10RA
722	{Inflammatory bowel disease (Crohn disease) 10} 611081	ATG16L1
722	{Inflammatory bowel disease (Crohn disease) 19} 612278	IRGM
722	{Crohn disease-associated growth failure} 266600	IL6
722	{Inflammatory bowel disease 1 Crohn disease} 266600	NOD2
722	{Inflammatory bowel disease 13} 612244	ABCB1
722	{Inflammatory bowel disease 14} 612245	IRF5
722	{Inflammatory bowel disease 17 protection against} 612261	IL23R
723	Insensitivity to pain congenital 243000	SCN9A
723	Insensitivity to pain congenital with anhidrosis 256800	NTRK1
723	HSAN2D autosomal recessive 243000	SCN9A

724	Insomnia fatal familial 600072	PRNP
725	Insulin-like growth factor I resistance to 270450	IGF1R
726	Intellectual developmental disorder with cardiac arrhythmia 617173	GNB5
726	Intellectual developmental disorder with dysmorphic facies and ptosis 617333	BRPF1
726	Intellectual developmental disorder with dysmorphic facies seizures and distal limb anomalies 617452	OTUD6B
726	Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold 617450	PPM1D
726	Intellectual developmental disorder with neuropsychiatric features 617532	SLC45A1
727	Interleukin 1 receptor antagonist deficiency 612852	IL1RN
728	Interstitial lung and liver disease 615486	MARS
728	Interstitial lung disease nephrotic syndrome and epidermolysis bullosa congenital 614748	ITGA3
729	Interstitial nephritis karyomegalic 614817	FAN1
730	Intrinsic factor deficiency 261000	GIF
731	Invasive pneumococcal disease recurrent isolated 1 610799	IRAK4
731	{Pneumococcal disease invasive protection against} 610799	TIRAP
731	Invasive pneumococcal disease recurrent isolated 2 300640	IKBKG
732	Iron-refractory iron deficiency anemia 206200	TMPRSS6
733	Ischiocoxopodopatellar syndrome 147891	TBX4
734	Isobutyryl-CoA dehydrogenase deficiency 611283	ACAD8
735	Isovaleric acidemia 243500	IVD
736	Jackson-Weiss syndrome 123150	FGFR1
736	Jackson-Weiss syndrome 123150	FGFR2
737	Jalili syndrome 217080	CNNM4
738	Jawad syndrome 251255	RBBP8
739	Jervell and Lange-Nielsen syndrome 2 612347	KCNE1
739	Jervell and Lange-Nielsen syndrome 220400	KCNQ1
740	Johanson-Blizzard syndrome 243800	UBR1
741	Joint laxity short stature and myopia 617662	GZF1
742	Joubert syndrome 1 213300	INPP5E
742	Joubert syndrome 10 300804	OFD1
742	Joubert syndrome 12 200990	KIF7
742	Acrocallosal syndrome 200990	KIF7
742	Joubert syndrome 13 614173	TCTN1
742	Joubert syndrome 14 614424	TMEM237
742	Joubert syndrome 15 614464	CEP41
742	Joubert syndrome 16 614465	TMEM138
742	Joubert syndrome 17 614615	C5orf42
742	Joubert syndrome 18 614815	TCTN3
742	Joubert syndrome 19 614844	ZNF423
742	Joubert syndrome 2 608091	TMEM216

742	Joubert syndrome 20 614970	TMEM231
742	Joubert syndrome 21 615636	CSPP1
742	Joubert syndrome 23 616490	KIAA0586
742	Joubert syndrome 24 616654	TCTN2
742	Joubert syndrome 25 616781	CEP104
742	Joubert syndrome 27 617120	B9D1
742	Joubert syndrome 28 617121	MKS1
742	Joubert syndrome 3 608629	AHI1
742	Joubert syndrome 30 617622	ARMC9
742	Joubert syndrome 31 617761	CEP120
742	Joubert syndrome 32 617757	SUFU
742	Joubert syndrome 33 617767	PIBF1
742	Joubert syndrome 34 614175	B9D2
742	Joubert syndrome 4 609583	NPHP1
742	Joubert syndrome 5 610188	CEP290
742	Joubert syndrome 6 610688	TMEM67
742	Joubert syndrome 7 611560	RPGRIP1L
742	Joubert syndrome 8 612291	ARL13B
742	Joubert syndrome 9 612285	CC2D2A
743	Juvenile polyposis syndrome infantile form 174900	BMPR1A
743	Juvenile polyposis or hereditary hemorrhagic telangiectasia syndrome 175050	SMAD4
743	Polyposis juvenile intestinal 174900	BMPR1A
743	Polyposis juvenile intestinal 174900	SMAD4
743	Polyposis syndrome hereditary mixed 2 610069	BMPR1A
744	KBG syndrome 148050	ANKRD11
745	Kabuki syndrome 1 147920	KMT2D
745	Kabuki syndrome 2 300867	KDM6A
746	Kahrizi syndrome 612713	SRD5A3
747	Kanzaki disease 609242	NAGA
748	Kappa light chain deficiency 614102	IGKC
749	Kaufman oculocerebrofacial syndrome 244450	UBE3B
750	Kenny-Caffey syndrome type 1 244460	TBCE
750	Kenny-Caffey syndrome type 2 127000	FAM111A
751	Keppen-Lubinsky syndrome 614098	KCNJ6
752	Keratitis 148190	PAX6
753	Keratitis-ichthyosis-deafness syndrome 148210	GJB2
754	Keratoconus 1 148300	VSX1
755	Keratoderma palmoplantar punctate type IA 148600	AAGAB
755	Keratoderma palmoplantar with deafness 148350	GJB2
756	Keratosis follicularis spinulosa decalvans X-linked 308800	MBTPS2
756	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma 601952	POMP
756	Keratosis palmoplantaris striata I AD 148700	DSG1
756	Keratosis palmoplantaris striata II 612908	DSP

756	Keratosis palmoplantaris striata III 607654	KRT1
756	Keratosis seborrheic somatic 182000	PIK3CA
757	Keutel syndrome 245150	MGP
758	Kindler syndrome 173650	KIND1
759	King-Denborough syndrome 145600	RYR1
759	{Malignant hyperthermia susceptibility 1} 145600	RYR1
759	{Malignant hyperthermia susceptibility 5} 601887	CACNA1S
760	Kleefstra syndrome 1 610253	EHMT1
760	Kleefstra syndrome 2 617768	KMT2C
761	Klippel-Feil syndrome 1 autosomal dominant 118100	GDF6
761	Klippel-Feil syndrome 2 214300	MEOX1
761	Klippel-Feil syndrome 3 autosomal dominant 613702	GDF3
761	Klippel-Feil syndrome 4 autosomal recessive with myopathy and facial dysmorphism 616549	MYO18B
762	Kniest dysplasia 156550	COL2A1
763	Knobloch syndrome type 1 267750	COL18A1
764	Kohlschutter-Tonz syndrome 226750	ROGDI
765	Koolen-De Vries syndrome 610443	KANSL1
766	Kosaki overgrowth syndrome 616592	PDGFRB
767	Kowarski syndrome 262650	GH1
768	Krabbe disease 245200	GALC
768	Krabbe disease atypical 611722	PSAP
769	Kufor-Rakeb syndrome 606693	ATP13A2
770	L-2-hydroxyglutaric aciduria 236792	L2HGDH
771	L-ferritin deficiency dominant and recessive 615604	FTL
772	LADD syndrome 149730	FGF10
772	LADD syndrome 149730	FGFR2
772	LADD syndrome 149730	FGFR3
773	LDL cholesterol level QTL2 143890	LDLR
774	LEOPARD syndrome 1 151100	PTPN11
774	LEOPARD syndrome 2 611554	RAF1
774	LEOPARD syndrome 3 613707	BRAF
775	LIG4 syndrome 606593	LIG4
776	Lactase deficiency congenital 223000	LCT
776	Lactase persistence or nonpersistence 223100	MCM6
777	Lacticacidemia due to PDX1 deficiency 245349	PDX1
778	Laing distal myopathy 160500	MYH7
779	Lamb-Shaffer syndrome 616803	SOX5
780	Langer mesomelic dysplasia 249700	SHOX
780	Langer mesomelic dysplasia 249700	SHOX
781	Language delay and ADHD or cognitive impairment with or without cardiac arrhythmia 617182	GNB5
782	Laron dwarfism 262500	GHR
783	Larsen syndrome 150250	FLNB
784	Laryngoonychocutaneous syndrome 245660	LAMA3

785	Lateral meningocele syndrome 130720	NOTCH3
786	Lathosterolosis 607330	SC5DL
787	Laurin-Sandrow syndrome 135750	LMBR1
788	Leber congenital amaurosis 1 204000	GUCY2D
788	Leber congenital amaurosis 10 611755	CEP290
788	Leber congenital amaurosis 11 613837	IMPDH1
788	Leber congenital amaurosis 12 610612	RD3
788	Leber congenital amaurosis 13 612712	RDH12
788	Leber congenital amaurosis 14 613341	LRAT
788	Leber congenital amaurosis 15 613843	TULP1
788	Leber congenital amaurosis 16 614186	KCNJ13
788	Leber congenital amaurosis 17 615360	GDF6
788	Leber congenital amaurosis 18 608133	PRPH2
788	Leber congenital amaurosis 2 204100	RPE65
788	Leber congenital amaurosis 3 604232	SPATA7
788	Leber congenital amaurosis 5 604537	LCA5
788	Leber congenital amaurosis 6 613826	RPGRIP1
788	Leber congenital amaurosis 7 613829	CRX
788	Leber congenital amaurosis 8 613835	CRB1
788	Leber congenital amaurosis 9 608553	NMNAT1
789	Left ventricular noncompaction 1 with or without congenital heart defects 604169	DTNA
789	Left ventricular noncompaction 10 615396	MYBPC3
789	Left ventricular noncompaction 3 601493	LDB3
789	Left ventricular noncompaction 4 613424	ACTC1
789	Left ventricular noncompaction 5 613426	MYH7
789	Left ventricular noncompaction 6 601494	TNNT2
789	Left ventricular noncompaction 7 615092	MIB1
789	Left ventricular noncompaction 8 615373	PRDM16
789	Left ventricular noncompaction 9 611878	TPM1
790	Legg-Calve-Perthes disease 150600	COL2A1
791	Legius syndrome 611431	SPRED1
792	Leigh syndrome 256000	BCS1L
792	Leigh syndrome 256000	NDUFA10
792	Leigh syndrome 256000	NDUFAF2
792	Leigh syndrome 256000	NDUFS4
792	Leigh syndrome 256000	NDUFS7
792	Leigh syndrome 256000	SDHA
792	Leigh syndrome French-Canadian type 220111	LRPPRC
792	Leigh syndrome due to COX IV deficiency 256000	SURF1
792	Leigh syndrome due to cytochrome c oxidase deficiency 256000	COX15
792	Leigh syndrome due to mitochondrial COX4 deficiency 256000	COX10
792	Leigh syndrome due to mitochondrial complex 1 deficiency 256000	NDUFA12

792	Leigh syndrome due to mitochondrial complex I deficiency 256000	FOXRED1
792	Leigh syndrome due to mitochondrial complex I deficiency 256000	NDUFA2
792	Leigh syndrome due to mitochondrial complex I deficiency 256000	NDUFA9
792	Leigh syndrome due to mitochondrial complex I deficiency 256000	NDUFAF6
792	Leigh syndrome due to mitochondrial complex I deficiency 256000	NDUFS3
792	Leigh syndrome due to mitochondrial complex I deficiency 256000	NDUFS8
793	Leiomyomatosis and renal cell cancer 150800	FH
794	Lenz-Majewski hyperostotic dwarfism 151050	PTDSS1
795	Leprechaunism 246200	INSR
796	Leri-Weill dyschondrosteosis 127300	SHOX
796	Leri-Weill dyschondrosteosis 127300	SHOX
797	Lesch-Nyhan syndrome 300322	HPRT1
798	Lethal congenital contractural syndrome 2 607598	ERBB3
798	Lethal congenital contractural syndrome 3 611369	PIP5K1C
798	Lethal congenital contracture syndrome 1 253310	GLE1
798	Lethal congenital contracture syndrome 10 617022	NEK9
798	Lethal congenital contracture syndrome 11 617194	GLDN
798	Lethal congenital contracture syndrome 4 614915	MYBPC1
798	Lethal congenital contracture syndrome 5 615368	DNM2
798	Lethal congenital contracture syndrome 7 616286	CNTNAP1
798	Lethal congenital contracture syndrome 9 616503	ADGRG6
799	T-cell acute lymphoblastic leukemia somatic 613065	BAX
799	Leukemia T-cell acute lymphoblastic somatic 613065	NUP214
799	Leukemia T-cell acute lymphocytic somatic 613065	TAL1
799	Leukemia T-cell acute lymphocytic somatic 613065	TAL2
799	Leukemia acute lymphoblastic 613065	NBN
799	Leukemia acute lymphoblastic somatic 613065	FLT3
799	Leukemia acute lymphoblastic somatic 613065	GNB1
799	{Leukemia acute lymphoblastic susceptibility to 3} 615545	PAX5
799	Leukemia acute lymphocytic somatic 613065	BCR
799	Leukemia acute myeloid 601626	MLLT10
799	Leukemia acute myeloid 601626	KIT
799	Leukemia acute myeloid 601626	KRAS
799	Leukemia acute myeloid 601626	LPP
799	Leukemia acute myeloid 601626	NSD3
799	Leukemia acute myeloid 601626	RUNX1
799	Leukemia acute myeloid reduced survival in somatic 601626	FLT3
799	Leukemia acute myeloid somatic 601626	CEBPA
799	Leukemia acute myeloid somatic 601626	ETV6
799	Leukemia acute myeloid somatic 601626	FLT3

799	Leukemia acute myeloid somatic 601626	JAK2
799	Leukemia acute myeloid somatic 601626	NPM1
799	Leukemia acute myeloid somatic 601626	NUP214
799	Leukemia acute myeloid somatic 601626	PICALM
799	{Leukemia acute myeloid susceptibility to} 601626	GATA2
799	{Leukemia acute myeloid} 601626	CHIC2
799	{Leukemia acute myeloid} 601626	TERT
799	Leukemia acute promyelocytic somatic 102578	STAT5B
799	Leukemia acute promyelocytic somatic 612376	NUMA1
799	Leukemia chronic myeloid somatic 608232	BCR
799	Leukemia juvenile myelomonocytic 607785	NF1
799	Leukemia juvenile myelomonocytic somatic 607785	ARHGAP26
799	Leukemia juvenile myelomonocytic somatic 607785	PTPN11
799	Leukemia megakaryoblastic with or without Down syndrome somatic 190685	GATA1
799	Leukemia or lymphoma B-cell 2	BCL2
799	Leukemia Philadelphia chromosome-positive resistant to imatinib	ABL1
799	Leukemia acute promyelocytic PL2F/RARA type	ZBTB16
799	Leukemia acute promyelocytic PML/RARA type	PML
799	Lymphocytic leukemia, acute T-cell	RAP1GDS1
799	Megakaryoblastic leukemia, acute	MKL1
799	Myelogenous leukemia, acute	ACSL6
799	Myelogenous leukemia, acute	IRF1
799	T-cell prolymphocytic leukemia, somatic	ATM
799	{Leukemia, post-chemotherapy, susceptibility to}	NQO1
799	{T-cell acute lymphoblastic leukemia}	MYB
800	Leukocyte adhesion deficiency 116920	ITGB2
800	Leukocyte adhesion deficiency type III 612840	FERMT3
801	Leukodystrophy adult-onset autosomal dominant 169500	LMNB1
801	Leukodystrophy and acquired microcephaly with or without dystonia 616763	PLEKHG2
801	Leukodystrophy hypomyelinating 10 616420	PYCR2
801	Leukodystrophy hypomyelinating 11 616494	POLR1C
801	Leukodystrophy hypomyelinating 12 616683	VPS11
801	Leukodystrophy hypomyelinating 13 616881	HIKESHI
801	Leukodystrophy hypomyelinating 2 608804	GJC2
801	Leukodystrophy hypomyelinating 3 260600	AIMP1
801	Leukodystrophy hypomyelinating 4 612233	HSPD1
801	Leukodystrophy hypomyelinating 5 610532	FAM126A
801	Leukodystrophy hypomyelinating 6 612438	TUBB4A
801	Leukodystrophy hypomyelinating 7 with or without oligodontia and or hypogonadotropic hypogonadism 607694	POLR3A
801	Leukodystrophy hypomyelinating 8 with or without oligodontia and or hypogonadotropic hypogonadism 614381	POLR3B

801	Leukodystrophy hypomyelinating 9 616140	RARS
802	Leukoencephalopathy brain calcifications and cysts 614561	SNORD118
802	Leukoencephalopathy cystic without megalencephaly 612951	RNASET2
802	Leukoencephalopathy diffuse hereditary with spheroids 221820	CSF1R
802	Leukoencephalopathy progressive with ovarian failure 615889	AARS2
802	Leukoencephalopathy with ataxia 615651	CLCN2
802	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation 611105	DARS2
802	Leukoencephalopathy with vanishing white matter 603896	EIF2B1
802	Leukoencephalopathy with vanishing white matter 603896	EIF2B2
802	Leukoencephalopathy with vanishing white matter 603896	EIF2B3
802	Leukoencephalopathy with vanishing white matter 603896	EIF2B4
802	Leukoencephalopathy with vanishing white matter 603896	EIF2B5
803	Leydig cell hypoplasia with hypergonadotropic hypogonadism 238320	LHCGR
803	Leydig cell hypoplasia with pseudohermaphroditism 238320	LHCGR
803	Luteinizing hormone resistance female 238320	LHCGR
804	Li-Fraumeni syndrome 151623	TP53
804	Li-Fraumeni syndrome 609265	CHEK2
805	Liddle syndrome 177200	SCNN1B
805	Liddle syndrome 177200	SCNN1G
806	Limb-mammary syndrome 603543	TP63
807	Linear skin defects with multiple congenital anomalies 1 309801	HCCS
807	Linear skin defects with multiple congenital anomalies 2 300887	COX7B
807	Linear skin defects with multiple congenital anomalies 3 300952	NDUFB11
808	Lipase deficiency combined 246650	LMF1
809	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency 255100	FLAD1
810	Lipodystrophy congenital generalized type 1 608594	AGPAT2
810	Lipodystrophy congenital generalized type 2 269700	BSCL2
810	Lipodystrophy congenital generalized type 4 613327	CAVIN1
810	Lipodystrophy familial partial type 2 151660	LMNA
810	Lipodystrophy familial partial type 3 604367	PPARG
810	Lipodystrophy familial partial type 4 613877	PLIN1
810	Lipodystrophy familial partial type 6 615980	LIPE
810	{Lipodystrophy partial acquired susceptibility to} 608709	LMNB2
811	Insulin resistance severe digenic 125853	PPP1R3A
811	{Insulin resistance susceptibility to} 125853	PTPN1
811	Insulin resistance severe digenic 604367	PPARG
812	Lipoid adrenal hyperplasia 201710	STAR
813	Lipoprotein glomerulopathy 611771	APOE
814	Lipoprotein lipase deficiency 238600	LPL
815	Lipoyltransferase 1 deficiency 616299	LIPT1
816	Lissencephaly 1 607432	PAFAH1B1
816	Subcortical laminar heterotopia 607432	PAFAH1B1

816	Lissencephaly 2 (Norman-Roberts type) 257320	RELN
816	Lissencephaly 3 611603	TUBA1A
816	Lissencephaly 4 (with microcephaly) 614019	NDE1
816	Lissencephaly 5 615191	LAMB1
816	Lissencephaly 6 with microcephaly 616212	KATNB1
816	Lissencephaly 8 617255	TMTC3
816	Lissencephaly X-linked 2 300215	ARX
816	Hydranencephaly with abnormal genitalia 300215	ARX
816	Lissencephaly X-linked 300067	DCX
816	Subcortical laminal heterotopia X-linked 300067	DCX
817	Liver failure transient infantile 613070	TRMU
818	Loeys-Dietz syndrome 1 609192	TGFBR1
818	Loeys-Dietz syndrome 2 610168	TGFBR2
818	Loeys-Dietz syndrome 3 613795	SMAD3
818	Loeys-Dietz syndrome 4 614816	TGFB2
818	Loeys-Dietz syndrome 5 615582	TGFB3
819	Long QT syndrome 1 192500	KCNQ1
819	{Long QT syndrome 1 acquired susceptibility to} 192500	KCNQ1
819	Long QT syndrome 12 612955	SNTA1
819	Long QT syndrome 13 613485	KCNJ5
819	Long QT syndrome 14 616247	CALM1
819	Long QT syndrome 15 616249	CALM2
819	Long QT syndrome 2 613688	KCNH2
819	{Long QT syndrome 2 acquired susceptibility to} 613688	KCNH2
819	{Long QT syndrome acquired reduced susceptibility to} 613688	ALG10
819	Long QT syndrome 4 600919	ANK2
819	Long QT syndrome 5 613695	KCNE1
819	Long QT syndrome 6 613693	KCNE2
819	Long QT syndrome 9 611818	CAV3
819	Long QT syndrome-10 611819	SCN4B
819	Long QT syndrome-3 603830	SCN5A
819	Cardiac arrhythmia ankyrin-B-related 600919	ANK2
820	Lopes-Maciel-Rodan syndrome 617435	HTT
821	Lowe syndrome 309000	OCRL
822	Lujan-Fryns syndrome 309520	MED12
823	Lung cancer 211980	PPP2R1B
823	Lung cancer somatic 211980	KRAS
823	Lung cancer somatic 211980	MAP3K8
823	Lung cancer somatic 211980	SLC22A1
823	{Lung cancer protection against} 211980	CASP8
823	{Lung cancer resistance to} 211980	CYP2A6
823	{Lung cancer susceptibility to} 211980	ERCC6
823	{Lung cancer susceptibility to} 211980	FASLG
823	Adenocarcinoma of lung response to tyrosine kinase inhibitor in 211980	EGFR

823	Adenocarcinoma of lung somatic 211980	BRAF
823	Adenocarcinoma of lung somatic 211980	ERBB2
823	Adenocarcinoma of lung somatic 211980	PRKN
823	Nonsmall cell lung cancer response to tyrosine kinase inhibitor in 211980	EGFR
823	Nonsmall cell lung cancer somatic 211980	IRF1
823	Nonsmall cell lung cancer somatic 211980	PIK3CA
823	{Nonsmall cell lung cancer susceptibility to} 211980	EGFR
823	Small cell cancer of the lung somatic 182280	RB1
823	Nonsmall cell lung cancer, somatic	BRAF
823	{Lung cancer, protection against, in smokers}	MPO
824	Lung disease immunodeficiency and chromosome breakage syndrome 617241	NSMCE3
825	Luscan-Lumish syndrome 616831	SETD2
826	Lymphangioleiomyomatosis 606690	TSC1
826	Lymphangioleiomyomatosis somatic 606690	TSC2
827	Lymphedema hereditary IA 153100	FLT4
827	Lymphedema hereditary IC 613480	GJC2
827	Lymphedema hereditary ID 615907	VEGFC
827	Lymphedema hereditary III 616843	PIEZO1
827	Lymphedema-distichiasis syndrome 153400	FOXC2
827	Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus 153400	FOXC2
828	Lymphoma MALT somatic 137245	BCL10
828	Lymphoma non-Hodgkin 605027	PRF1
828	Lymphoma non-Hodgkin somatic 605027	CASP10
828	Lymphoma non-Hodgkin somatic 605027	RAD54B
828	Lymphoma non-Hodgkin somatic 605027	RAD54L
828	{Lymphoma follicular somatic} 605027	BCL10
828	Lymphoma, B-cell non-Hodgkin, somatic	ATM
828	Lymphoma, mantle cell, somatic	ATM
828	Lymphoma, somatic	MAD1L1
828	{Hodgkin lymphoma susceptibility to} 236000	KLHDC8B
828	B-cell non-Hodgkin lymphoma high-grade	BCL7A
828	Burkitt lymphoma 113970	MYC
829	Lymphoproliferative syndrome 1 613011	ITK
829	Lymphoproliferative syndrome 2 615122	CD27
829	Lymphoproliferative syndrome X-linked 1 308240	SH2D1A
829	Lymphoproliferative syndrome X-linked 2 300635	XIAP
830	Lysinuric protein intolerance 222700	SLC7A7
831	Lysyl hydroxylase 3 deficiency 612394	PLOD3
832	MASA syndrome 303350	L1CAM
832	CRASH syndrome 303350	L1CAM
833	MASP2 deficiency 613791	MASP2
834	MASS syndrome 604308	FBN1

835	MEDNIK syndrome 609313	AP1S1
836	MEND syndrome 300960	EBP
837	MIRAGE syndrome 617053	SAMD9
838	MODY type I 125850	HNF4A
838	MODY type II 125851	GCK
838	MODY type III 600496	HNF1A
838	MODY type IV 606392	PDX1
839	Machado-Joseph disease 109150	ATXN3
840	Macrocephaly alopecia cutis laxa and scoliosis 613075	RIN2
840	Macrocephaly dysmorphic facies and psychomotor retardation 617011	HERC1
840	Macrocephaly macrosomia facial dysmorphism syndrome 614192	RNF135
840	Macrocephaly or autism syndrome 605309	PTEN
840	Macrocephaly or megalencephaly syndrome autosomal recessive 248000	TBC1D7
841	Macrocytic anemia refractory due to 5q deletion somatic 153550	RPS14
842	Macroglobulinemia Waldenstrom somatic 153600	MYD88
843	Macrothrombocytopenia and progressive sensorineural deafness 600208	MYH9
843	Macrothrombocytopenia autosomal dominant TUBB1-related 613112	TUBB1
844	Macular degeneration X-linked atrophic 300834	RPGR
844	Macular degeneration age-related 3 608895	FBLN5
844	Macular degeneration early-onset 616118	FBN2
844	Macular degeneration juvenile 248200	CNGB3
844	{Macular degeneration age-related 11} 611953	CST3
844	{Macular degeneration age-related 12} 613784	CX3CR1
844	{Macular degeneration age-related 13 susceptibility to} 615439	CFI
844	{Macular degeneration age-related 14 reduced risk of} 615489	C2
844	{Macular degeneration age-related 14 reduced risk of} 615489	CFB
844	{Macular degeneration age-related 15 susceptibility to} 615591	C9
844	{Macular degeneration age-related 1} 603075	HMCN1
844	{Macular degeneration age-related 2} 153800	ABCA4
844	{Macular degeneration age-related 4} 610698	CFH
844	{Macular degeneration age-related 7} 610149	HTRA1
844	{Macular degeneration age-related 8} 613778	ARMS2
844	{Macular degeneration age-related 9} 611378	C3
844	{Macular degeneration age-related neovascular type} 610149	HTRA1
844	{Macular degeneration age-related reduced risk of} 603075	CFHR1
844	{Macular degeneration age-related reduced risk of} 603075	CFHR3
844	{Macular degeneration age-related susceptibility to 5} 613761	ERCC6
845	Macular corneal dystrophy 217800	CHST6
845	Macular dystrophy North Carolina type 136550	DHS6S1
845	Macular dystrophy patterned 1 169150	PRPH2
845	Macular dystrophy patterned 2 608970	CTNNA1

845	Macular dystrophy retinal 2 608051	PROM1
845	Macular dystrophy vitelliform 2 153700	BEST1
845	Macular dystrophy vitelliform 3 608161	PRPH2
845	Macular dystrophy vitelliform 4 616151	IMPG1
845	Macular dystrophy vitelliform 5 616152	IMPG2
845	Macular dystrophy with central cone involvement 616170	MFSD8
846	Majeed syndrome 609628	LPIN2
847	Malignant melanoma somatic 155600	PTEN
847	{Melanoma cutaneous malignant 2} 155601	CDKN2A
847	{Melanoma cutaneous malignant 3} 609048	CDK4
847	{Melanoma cutaneous malignant 5} 613099	MC1R
847	{Melanoma cutaneous malignant 6} 613972	XRCC3
847	{Melanoma cutaneous malignant 9} 615134	TERT
847	{Melanoma cutaneous malignant susceptibility to 10} 615848	POT1
847	{Melanoma cutaneous malignant susceptibility to 8} 601800	TYR
847	{Melanoma cutaneous malignant susceptibility to 8} 614456	MITF
847	Melanoma and neural system tumor syndrome 155755	CDKN2A
847	Melanoma, malignant, somatic	BRAF
847	Melanoma, malignant, somatic	STK11
848	Malonyl-CoA decarboxylase deficiency 248360	MLYCD
849	Malouf syndrome 212112	LMNA
850	Mandibular hypoplasia deafness progeroid features and lipodystrophy syndrome 615381	POLD1
851	Mandibuloacral dysplasia 248370	LMNA
851	Mandibuloacral dysplasia with type B lipodystrophy 608612	ZMPSTE24
852	Mandibulofacial dysostosis Guion-Almeida type 610536	EFTUD2
852	Mandibulofacial dysostosis with alopecia 616367	EDNRA
853	Manitoba oculotrichoanal syndrome 248450	FREM1
854	Mannosidosis alpha- types I and II 248500	MAN2B1
854	Mannosidosis beta 248510	MANBA
855	Maple syrup urine disease type II 248600	DBT
855	Maple syrup urine disease type Ia 248600	BCKDHA
855	Maple syrup urine disease type Ib 248600	BCKDHB
856	Marfan lipodystrophy syndrome 616914	FBN1
856	Marfan syndrome 154700	FBN1
857	Marinesco-Sjogren syndrome 248800	SIL1
858	Marshall syndrome 154780	COL11A1
859	Marshall-Smith syndrome 602535	NFIX
860	Martsolf syndrome 212720	RAB3GAP2
861	Mast cell disease 154800	KIT
862	Mast syndrome 248900	ACP33
863	May-Hegglin anomaly 155100	MYH9
864	McArdle disease 232600	PYGM
865	McCune-Albright syndrome somatic mosaic 174800	GNAS
866	McKusick-Kaufman syndrome 236700	MKKS

867	McLeod syndrome with or without chronic granulomatous disease 300842	XK
868	Meacham syndrome 608978	WT1
869	Meckel syndrome 1 249000	MKS1
869	Meckel syndrome 11 615397	TMEM231
869	Meckel syndrome 13 617562	TMEM107
869	Meckel syndrome 2 603194	TMEM216
869	Meckel syndrome 3 607361	TMEM67
869	Meckel syndrome 4 611134	CEP290
869	Meckel syndrome 5 611561	RPGRIP1L
869	Meckel syndrome 6 612284	CC2D2A
869	Meckel syndrome 7 267010	NPHP3
870	Meconium ileus 614665	GUCY2C
871	Medullary cystic kidney disease 1 174000	MUC1
871	Medullary cystic kidney disease 2 603860	UMOD
872	Medulloblastoma desmoplastic 155255	SUFU
872	Medulloblastoma somatic 155255	CTNNB1
872	Medulloblastoma somatic 155255	PTCH2
872	{Medulloblastoma} 155255	BRCA2
873	Meesmann corneal dystrophy 122100	KRT12
873	Meesmann corneal dystrophy 122100	KRT3
874	Meester-Loeys syndrome 300989	BGN
875	Megalencephalic leukoencephalopathy with subcortical cysts 2A 613925	HEPACAM
875	Megalencephalic leukoencephalopathy with subcortical cysts 2B remitting with or without mental retardation 613926	HEPACAM
875	Megalencephalic leukoencephalopathy with subcortical cysts 604004	MLC1
876	Megalencephaly-capillary malformation-polymicrogyria syndrome somatic 602501	PIK3CA
877	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1 603387	PIK3R2
877	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2 615937	AKT3
877	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3 615938	CCND2
878	Megaloblastic anemia due to dihydrofolate reductase deficiency 613839	DHFR
878	Megaloblastic anemia-1 Finnish type 261100	CUBN
878	Megaloblastic anemia-1 Norwegian type 261100	AMN
879	Megalocornea 1 X-linked 309300	CHRDL1
880	Meier-Gorlin syndrome 1 224690	ORC1
880	Meier-Gorlin syndrome 2 613800	ORC4
880	Meier-Gorlin syndrome 3 613803	ORC6
880	Meier-Gorlin syndrome 4 613804	CDT1

880	Meier-Gorlin syndrome 6 616835	GMNN
880	Meier-Gorlin syndrome 7 617063	CDC45
881	Melanocytic nevus syndrome congenital somatic 137550	NRAS
881	{Spitz nevus or nevus spilus somatic} 137550	HRAS
882	Meleda disease 248300	SLURP1
883	Melnick-Needles syndrome 309350	FLNA
884	Meningioma 607174	MN1
884	Meningioma NF2-related somatic 607174	NF2
884	Meningioma SIS-related 607174	PDGFB
884	{Meningioma familial susceptibility to} 607174	SMARCE1
884	{Meningioma familial susceptibility to} 607174	SUFU
884	{Meningioma} 607174	PTEN
885	Menkes disease 309400	ATP7A
886	Mental retardation 105 300984	USP27X
886	Mental retardation FRA12A type 136630	DIP2B
886	Mental retardation X-linked 1 or 78 309530	IQSEC2
886	Mental retardation X-linked 102 300958	DDX3X
886	Mental retardation X-linked 103 300982	KLHL15
886	Mental retardation X-linked 104 300983	FRMPD4
886	Mental retardation X-linked 106 300997	OGT
886	Mental retardation X-linked 12 or 35 300957	THOC2
886	Mental retardation X-linked 19 300844	RPS6KA3
886	Mental retardation X-linked 21 or 34 300143	IL1RAPL1
886	Mental retardation X-linked 29 and others 300419	ARX
886	Mental retardation X-linked 3 (methylmalonic acidemia and homocysteinemia cblX type) 309541	HCFC1
886	Mental retardation X-linked 30 or 47 300558	PAK3
886	Mental retardation X-linked 300495	NLGN4X
886	Mental retardation X-linked 41 300849	GDI1
886	Mental retardation X-linked 46 300436	ARHGEF6
886	Mental retardation X-linked 49 or 15 300114	CLCN4
886	Mental retardation X-linked 58 300210	TSPAN7
886	Mental retardation X-linked 61 300978	RLIM
886	Mental retardation X-linked 63 300387	ACSL4
886	Mental retardation X-linked 72 300271	RAB39B
886	Mental retardation X-linked 9 or 44 309549	FTSJ1
886	Mental retardation X-linked 90 300850	DLG3
886	Mental retardation X-linked 93 300659	BRWD3
886	Mental retardation X-linked 94 300699	GRIA3
886	Mental retardation X-linked 96 300802	SYP
886	Mental retardation X-linked 97 300803	ZNF711
886	Mental retardation X-linked 98 300912	NEXMIF
886	Mental retardation X-linked 99 300919	USP9X
886	Mental retardation X-linked 99 syndromic female-restricted 300968	USP9X

886	Mental retardation X-linked FRAXE type 309548	AFF2
886	Mental retardation X-linked Snyder-Robinson type 309583	SMS
886	Mental retardation X-linked syndromic 13 300055	MECP2
886	Mental retardation X-linked syndromic 14 300676	UPF3B
886	Mental retardation X-linked syndromic 15 (Cabezas type) 300354	CUL4B
886	Mental retardation X-linked syndromic 16 305400	FGD1
886	Aarskog-Scott syndrome 305400	FGD1
886	Mental retardation X-linked syndromic 33 300966	TAF1
886	Mental retardation X-linked syndromic 34 300967	NONO
886	Mental retardation X-linked syndromic 35 300998	RPL10
886	Mental retardation X-linked syndromic 5 304340	AP1S2
886	Mental retardation X-linked syndromic Bain type 300986	HNRNPH2
886	Mental retardation X-linked syndromic Borck type 300987	EIF2S3
886	Mental retardation X-linked syndromic Christianson type 300243	SLC9A6
886	Mental retardation X-linked syndromic Claes-Jensen type 300534	KDM5C
886	Mental retardation X-linked syndromic Hedera type 300423	ATP6AP2
886	Mental retardation X-linked syndromic Lubs type 300260	MECP2
886	Mental retardation X-linked syndromic Nascimento-type 300860	UBE2A
886	Mental retardation X-linked syndromic Raymond type 300799	ZDHHC9
886	Mental retardation X-linked syndromic Turner type 300706	HUWE1
886	Mental retardation X-linked with cerebellar hypoplasia and distinctive facial appearance 300486	OPHN1
886	Mental retardation X-linked with isolated growth hormone deficiency 300123	SOX3
886	Mental retardation and distinctive facial features with or without cardiac defects 616789	MED13L
886	Mental retardation and microcephaly with pontine and cerebellar hypoplasia 300749	CASK
886	Mental retardation anterior maxillary protrusion and strabismus 613671	SOBP
886	Mental retardation autosomal dominant 1 156200	MBD5
886	Mental retardation autosomal dominant 13 614563	DYNC1H1
886	Mental retardation autosomal dominant 18 615074	GATAD2B
886	Mental retardation autosomal dominant 19 615075	CTNNB1
886	Mental retardation autosomal dominant 21 615502	CTCF
886	Mental retardation autosomal dominant 22 612337	ZBTB18
886	Mental retardation autosomal dominant 23 615761	SETD5
886	Mental retardation autosomal dominant 24 615828	DEAF1
886	Mental retardation autosomal dominant 26 615834	AUTS2
886	Mental retardation autosomal dominant 27 615866	SOX11
886	Mental retardation autosomal dominant 29 616078	SETBP1
886	Mental retardation autosomal dominant 3 612580	CDH15
886	Mental retardation autosomal dominant 30 616083	ZMYND11
886	Mental retardation autosomal dominant 31 616158	PURA
886	Mental retardation autosomal dominant 32 616268	KAT6A

886	Mental retardation autosomal dominant 33 616311	DPP6
886	Mental retardation autosomal dominant 34 616351	COL4A3BP
886	Mental retardation autosomal dominant 35 616355	PPP2R5D
886	Mental retardation autosomal dominant 36 616362	PPP2R1A
886	Mental retardation autosomal dominant 38 616393	EEF1A2
886	Mental retardation autosomal dominant 39 616521	MYT1L
886	Mental retardation autosomal dominant 4 612581	KIRREL3
886	Mental retardation autosomal dominant 40 616579	CHAMP1
886	Mental retardation autosomal dominant 41 616944	TBL1XR1
886	Mental retardation autosomal dominant 42 616973	GNB1
886	Mental retardation autosomal dominant 43 616977	HIVEP2
886	Mental retardation autosomal dominant 44 617061	TRIO
886	Mental retardation autosomal dominant 45 617600	CIC
886	Mental retardation autosomal dominant 46 617601	KCNQ5
886	Mental retardation autosomal dominant 47 617635	STAG1
886	Mental retardation autosomal dominant 48 617751	RAC1
886	Mental retardation autosomal dominant 49 617752	TRIP12
886	Mental retardation autosomal dominant 5 612621	SYNGAP1
886	Mental retardation autosomal dominant 6 613970	GRIN2B
886	Mental retardation autosomal dominant 7 614104	DYRK1A
886	Mental retardation autosomal dominant 8 614254	GRIN1
886	Mental retardation autosomal dominant 9 614255	KIF1A
886	Mental retardation autosomal recessive 1 249500	PRSS12
886	Mental retardation autosomal recessive 12 611090	ST3GAL3
886	Mental retardation autosomal recessive 13 613192	TRAPPC9
886	Mental retardation autosomal recessive 14 614020	TECR
886	Mental retardation autosomal recessive 15 614202	MAN1B1
886	Mental retardation autosomal recessive 18 614249	MED23
886	Mental retardation autosomal recessive 2 607417	CRBN
886	Mental retardation autosomal recessive 27 614340	LINS1
886	Mental retardation autosomal recessive 3 608443	CC2D1A
886	Mental retardation autosomal recessive 34 with variant lissencephaly 614499	CRADD
886	Mental retardation autosomal recessive 36 615286	ADAT3
886	Mental retardation autosomal recessive 38 615516	HERC2
886	Mental retardation autosomal recessive 39 615541	TTI2
886	Mental retardation autosomal recessive 40 615599	TAF2
886	Mental retardation autosomal recessive 41 615637	KPTN
886	Mental retardation autosomal recessive 42 615802	PGAP1
886	Mental retardation autosomal recessive 44 615942	METTL23
886	Mental retardation autosomal recessive 46 616116	NDST1
886	Mental retardation autosomal recessive 47 616193	FMN2
886	Mental retardation autosomal recessive 48 616269	SLC6A17
886	Mental retardation autosomal recessive 49 616281	GPT2
886	Mental retardation autosomal recessive 5 611091	NSUN2

886	Mental retardation autosomal recessive 51 616739	HNMT
886	Mental retardation autosomal recessive 53 616917	PIGG
886	Mental retardation autosomal recessive 54 617028	TNIK
886	Mental retardation autosomal recessive 56 617125	ZC3H14
886	Mental retardation autosomal recessive 57 617188	MBOAT7
886	Mental retardation autosomal recessive 58 617270	ELP2
886	Mental retardation autosomal recessive 59 617323	IMPA1
886	Mental retardation autosomal recessive 6 611092	GRIK2
886	Mental retardation autosomal recessive 60 617432	TAF13
886	Mental retardation autosomal recessive 61 617773	RUSC2
886	Mental retardation autosomal recessive 7 611093	TUSC3
886	Mental retardation stereotypic movements epilepsy and or cerebral malformations 613443	MEF2C
886	Mental retardation syndrome X-linked Siderius type 300263	PHF8
886	Mental retardation truncal obesity retinal dystrophy and micropenis 610156	INPP5E
886	Mental retardation with language impairment and with or without autistic features 613670	FOXP1
886	Mental retardation with or without nystagmus 300422	CASK
886	Mental retardation-hypotonic facies syndrome X-linked 309580	ATRX
887	Mephenytoin poor metabolizer 609535	CYP2C19
888	Mesothelioma somatic 156240	WT1
888	{Mesothelioma somatic} 156240	BCL10
889	Metabolic encephalomyopathic crises recurrent with rhabdomyolysis cardiac arrhythmias and neurodegeneration 616878	TANGO2
890	Metacarpal 4-5 fusion 309630	FGF16
891	Metachondromatosis 156250	PTPN11
892	Metachromatic leukodystrophy 250100	ARSA
892	Metachromatic leukodystrophy due to SAP-b deficiency 249900	PSAP
893	Metaphyseal anadysplasia 1 602111	MMP13
893	Metaphyseal anadysplasia 2 613073	MMP9
893	Metaphyseal chondrodysplasia Murk Jansen type 156400	PTH1R
893	Metaphyseal chondrodysplasia Schmid type 156500	COL10A1
893	Metaphyseal dysplasia Spahr type 250400	MMP13
893	Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly 156510	RUNX2
893	Metaphyseal dysplasia without hypotrichosis 250460	RMRP
893	Metatropic dysplasia 156530	TRPV4
894	Methemoglobinemia type I 250800	CYB5R3
894	Methemoglobinemia type II 250800	CYB5R3
894	Methemoglobinemias, alpha-	HBA1
894	Methemoglobinemias, beta-	HBB
895	Methylmalonate semialdehyde dehydrogenase deficiency 614105	ALDH6A1
896	Methylmalonic aciduria and homocystinuria cblC type 277400	MMACHC

896	Methylmalonic aciduria and homocystinuria cblD type 277410	MMADHC
896	Methylmalonic aciduria and homocystinuria cblF type 277380	LMBRD1
896	Methylmalonic aciduria and homocystinuria cblJ type 614857	ABCD4
896	Methylmalonic aciduria cblD type variant 2 277410	MMADHC
896	Methylmalonic aciduria mut(0) type 251000	MUT
896	Methylmalonic aciduria transient due to transcobalamin receptor defect 613646	CD320
896	Methylmalonic aciduria vitamin B12-responsive 251100	MMAA
896	Methylmalonic aciduria vitamin B12-responsive due to defect in synthesis of adenosylcobalamin cblB complementation type 251110	MMAB
897	Methylmalonyl-CoA epimerase deficiency 251120	MCEE
898	Mevalonic aciduria 610377	MVK
899	Microcephalic osteodysplastic primordial dwarfism type I 210710	RNU4ATAC
899	Microcephalic osteodysplastic primordial dwarfism type II 210720	PCNT
900	Microcephaly 1 primary autosomal recessive 251200	MCPH1
900	Microcephaly 15 primary autosomal recessive 616486	MFSD2A
900	Microcephaly 17 primary autosomal recessive 617090	CIT
900	Microcephaly 2 primary autosomal recessive with or without cortical malformations 604317	WDR62
900	Microcephaly 3 primary autosomal recessive 604804	CDK5RAP2
900	Microcephaly 4 primary autosomal recessive 604321	KNL1
900	Microcephaly 5 primary autosomal recessive 608716	ASPM
900	Microcephaly 6 primary autosomal recessive 608393	CENPJ
900	Microcephaly 7 primary autosomal recessive 612703	STIL
900	Microcephaly 8 primary autosomal recessive 614673	CEP135
900	Microcephaly 9 primary autosomal recessive 614852	CEP152
900	Microcephaly Amish type 607196	SLC25A19
900	Microcephaly and chorioretinopathy autosomal recessive 1 251270	TUBGCP6
900	Microcephaly and chorioretinopathy autosomal recessive 2 616171	PLK4
900	Microcephaly and chorioretinopathy autosomal recessive 3 616335	TUBGCP4
900	Microcephaly congenital cataract and psoriasiform dermatitis 616834	MSMO1
900	Microcephaly epilepsy and diabetes syndrome 614231	IER3IP1
900	Microcephaly postnatal progressive with seizures and brain atrophy 613668	MED17
900	Microcephaly progressive seizures and cerebral and cerebellar atrophy 615760	QARS
900	Microcephaly seizures and developmental delay 613402	PNKP
900	Microcephaly seizures spasticity and brain calcification 605622	PCDH12
900	Microcephaly short stature and impaired glucose metabolism 1 616033	TRMT10A

900	Microcephaly short stature and impaired glucose metabolism 2 616817	PPP1R15B
900	Microcephaly short stature and limb abnormalities 617604	DONSON
900	Microcephaly short stature and polymicrogyria with seizures 614833	RTTN
900	Microcephaly with or without chorioretinopathy lymphedema or mental retardation 152950	KIF11
900	Microcephaly-capillary malformation syndrome 614261	STAMBP
900	Microcephaly-micromelia syndrome 251230	DONSON
901	Microcornea myopic chorioretinal atrophy and telecanthus 615458	ADAMTS18
901	Microcornea rod-cone dystrophy cataract and posterior staphyloma 193220	BEST1
901	Vitreoretinochoroidopathy 193220	BEST1
902	Microphthalmia isolated 2 610093	CHX10
902	Microphthalmia isolated 3 611038	RAX
902	Microphthalmia isolated 4 613094	GDF6
902	Microphthalmia isolated 5 611040	MFRP
902	Microphthalmia isolated 6 613517	PRSS56
902	Microphthalmia isolated 7 613704	GDF3
902	Microphthalmia isolated 8 615113	ALDH1A3
902	Microphthalmia isolated with coloboma 10 616428	RBP4
902	Microphthalmia isolated with coloboma 7 614497	ABCB6
902	Microphthalmia isolated with coloboma 8 601186	STRA6
902	Microphthalmia isolated with coloboma 9 615145	TENM3
902	Microphthalmia syndromic 12 615524	RARB
902	Microphthalmia syndromic 2 300166	BCOR
902	Microphthalmia syndromic 3 206900	SOX2
902	Microphthalmia syndromic 5 610125	OTX2
902	Microphthalmia syndromic 6 607932	BMP4
902	Microphthalmia syndromic 9 601186	STRA6
902	Microphthalmia with coloboma 3 610092	CHX10
902	Microphthalmia with coloboma 5 611638	SHH
902	Microphthalmia with coloboma 6 613703	GDF3
902	Microphthalmia with coloboma 6 digenic 613703	GDF6
902	Microphthalmia with limb anomalies 206920	SMOC1
902	Microphthalmia or coloboma and skeletal dysplasia syndrome 615877	MAB21L2
902	Optic nerve hypoplasia and abnormalities of the central nervous system 206900	SOX2
903	Microspherophakia and or megalocornea with ectopia lentis and with or without secondary glaucoma 251750	LTBP2
904	Microvillus inclusion disease 251850	MYO5B
905	Midface hypoplasia hearing impairment elliptocytosis and nephrocalcinosis 300990	AMMECR1

906	Migraine familial basilar 602481	ATP1A2
906	Migraine familial hemiplegic 1 141500	CACNA1A
906	Migraine familial hemiplegic 1 with progressive cerebellar ataxia 141500	CACNA1A
906	Migraine familial hemiplegic 2 602481	ATP1A2
906	Migraine familial hemiplegic 3 609634	SCN1A
906	{Migraine resistance to} 157300	EDNRA
906	{Migraine susceptibility to} 157300	ESR1
906	{Migraine with or without aura susceptibility to 13} 613656	KCNK18
906	{Migraine without aura susceptibility to} 157300	TNF
907	Miller syndrome 263750	DHODH
908	Minicore myopathy with external ophthalmoplegia 255320	RYR1
909	Mirror movements 1 and or agenesis of the corpus callosum 157600	DCC
909	Mirror movements 2 614508	RAD51
910	Mismatch repair cancer syndrome 276300	MLH1
910	Mismatch repair cancer syndrome 276300	MSH2
910	Mismatch repair cancer syndrome 276300	MSH6
910	Mismatch repair cancer syndrome 276300	PMS2
911	Mitchell-Riley syndrome 615710	RFX6
912	Mitochondrial DNA depletion syndrome 1 (MNGIE type) 603041	TYMP
912	Mitochondrial DNA depletion syndrome 11 615084	MGME1
912	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD 617184	SLC25A4
912	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR 615418	SLC25A4
912	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type) 615471	FBXL4
912	Mitochondrial DNA depletion syndrome 2 (myopathic type) 609560	TK2
912	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type) 251880	DGUOK
912	Mitochondrial DNA depletion syndrome 4A (Alpers type) 203700	POLG
912	Mitochondrial DNA depletion syndrome 4B (MNGIE type) 613662	POLG
912	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria) 612073	SUCLA2
912	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type) 256810	MPV17
912	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type) 271245	TWNK
912	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy) 612075	RRM2B
912	Mitochondrial DNA depletion syndrome 8B (MNGIE type) 612075	RRM2B
912	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria) 245400	SUCLG1

913	Mitochondrial complex 1 deficiency 252010	NDUFAF5
913	Mitochondrial complex I deficiency 252010	FOXRED1
913	Mitochondrial complex I deficiency 252010	NDUFA1
913	Mitochondrial complex I deficiency 252010	NDUFA11
913	Mitochondrial complex I deficiency 252010	NDUFAF1
913	Mitochondrial complex I deficiency 252010	NDUFAF2
913	Mitochondrial complex I deficiency 252010	NDUFAF3
913	Mitochondrial complex I deficiency 252010	NDUFAF4
913	Mitochondrial complex I deficiency 252010	NDUFB3
913	Mitochondrial complex I deficiency 252010	NDUFS1
913	Mitochondrial complex I deficiency 252010	NDUFS2
913	Mitochondrial complex I deficiency 252010	NDUFS3
913	Mitochondrial complex I deficiency 252010	NDUFS4
913	Mitochondrial complex I deficiency 252010	NDUFS6
913	Mitochondrial complex I deficiency 252010	NDUFV1
913	Mitochondrial complex I deficiency 252010	NDUFV2
913	Mitochondrial complex I deficiency 252010	NUBPL
913	Mitochondrial complex I deficiency 252010	TIMMDC1
913	Mitochondrial complex I deficiency 252010	TMEM126B
913	Mitochondrial complex I deficiency due to ACAD9 deficiency 611126	ACAD9
913	Mitochondrial complex II deficiency 252011	SDHAF1
913	Mitochondrial complex II deficiency 252011	SDHD
913	Mitochondrial respiratory chain complex II deficiency 252011	SDHA
913	Mitochondrial complex III deficiency nuclear type 1 124000	BCS1L
913	Mitochondrial complex III deficiency nuclear type 2 615157	TTC19
913	Mitochondrial complex III deficiency nuclear type 3 615158	UQCRB
913	Mitochondrial complex III deficiency nuclear type 4 615159	UQCRQ
913	Mitochondrial complex III deficiency nuclear type 5 615160	UQCRC2
913	Mitochondrial complex III deficiency nuclear type 6 615453	CYC1
913	Mitochondrial complex III deficiency nuclear type 8 615838	LYRM7
913	Mitochondrial complex IV deficiency 220110	APOPT1
913	Mitochondrial complex IV deficiency 220110	COX10
913	Mitochondrial complex IV deficiency 220110	COX20
913	Mitochondrial complex IV deficiency 220110	COX6B1
913	Mitochondrial complex IV deficiency 220110	PET100
913	Mitochondrial complex IV deficiency 220110	SCO1
913	Mitochondrial complex IV deficiency 220110	TACO1
913	Mitochondrial complex V (ATP synthase) deficiency nuclear type 2 614052	TMEM70
914	Mitochondrial phosphate carrier deficiency 610773	SLC25A3
915	Mitochondrial pyruvate carrier deficiency 614741	BRP44L
916	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE) 607459	POLG

917	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency 616277	ECHS1
918	Mitral valve prolapse 2 607829	DCHS1
919	Miyoshi muscular dystrophy 1 254130	DYSF
919	Miyoshi muscular dystrophy 3 613319	ANO5
920	Mohr-Tranebjaerg syndrome 304700	TIMM8A
921	Molybdenum cofactor deficiency A 252150	MOCS1
921	Molybdenum cofactor deficiency B 252160	MOCS2
921	Molybdenum cofactor deficiency C 615501	GPHN
922	Monilethrix 158000	KRT81
922	Monilethrix 158000	KRT83
922	Monilethrix 158000	KRT86
923	Monocarboxylate transporter 1 deficiency 616095	SLC16A1
924	Mononeuropathy of the median nerve mild 613353	SH3TC2
925	Morbid obesity and spermatogenic failure 615703	CEP19
926	Mosaic variegated aneuploidy syndrome 1 257300	BUB1B
926	Mosaic variegated aneuploidy syndrome 2 614114	CEP57
926	Mosaic variegated aneuploidy syndrome 3 617598	TRIP13
927	Mowat-Wilson syndrome 235730	ZEB2
928	Moyamoya 6 with achalasia 615750	GUCY1A3
928	Moyamoya disease 5 614042	ACTA2
928	{Moyamoya disease 2 susceptibility to} 607151	RNF213
929	Muckle-Wells syndrome 191900	NLRP3
930	Mucolipidosis II alpha or beta 252500	GNPTAB
930	Mucolipidosis III alpha or beta 252600	GNPTAB
930	Mucolipidosis III gamma 252605	GNPTG
930	Mucolipidosis IV 252650	MCOLN1
931	Mucopolysaccharidosis II 309900	IDS
931	Mucopolysaccharidosis IVA 253000	GALNS
931	Mucopolysaccharidosis Ih 607014	IDUA
931	Mucopolysaccharidosis Ih or s 607015	IDUA
931	Mucopolysaccharidosis Is 607016	IDUA
931	Mucopolysaccharidosis VII 253220	GUSB
931	Mucopolysaccharidosis type IIIA (Sanfilippo A) 252900	SGSH
931	Mucopolysaccharidosis type IIIB (Sanfilippo B) 252920	NAGLU
931	Mucopolysaccharidosis type IIIC (Sanfilippo C) 252930	HGSNAT
931	Mucopolysaccharidosis type IIID 252940	GNS
931	Mucopolysaccharidosis type IVB (Morquio) 253010	GLB1
931	Mucopolysaccharidosis type VI (Maroteaux-Lamy) 253200	ARSB
931	Mucopolysaccharidosis-plus syndrome 617303	VPS33A
932	Muenke syndrome 602849	FGFR3
933	Muir-Torre syndrome 158320	MLH1
933	Muir-Torre syndrome 158320	MSH2
934	Mulchandani-Bhoj-Conlin syndrome 617352	MBCS
935	Mulibrey nanism 253250	TRIM37

936	Mullerian aplasia and hyperandrogenism 158330	WNT4
937	Multicentric carpotarsal osteolysis syndrome 166300	MAFB
938	Multicentric osteolysis nodulosis and arthropathy 259600	MMP2
939	Multinucleated neurons anhydramnios renal dysplasia cerebellar hypoplasia and hydranencephaly 236500	CEP55
940	Multiple congenital anomalies-hypotonia-seizures syndrome 1 614080	PIGN
940	Multiple congenital anomalies-hypotonia-seizures syndrome 2 300868	PIGA
940	Multiple congenital anomalies-hypotonia-seizures syndrome 3 615398	PIGT
941	Multiple endocrine neoplasia 1 131100	MEN1
941	Multiple endocrine neoplasia IIA 171400	RET
941	Multiple endocrine neoplasia IIB 162300	RET
941	Multiple endocrine neoplasia type IV 610755	CDKN1B
942	Multiple fibroadenomas of the breast 615554	PRLR
943	Multiple joint dislocations short stature craniofacial dysmorphism with or without congenital heart defects 245600	B3GAT3
944	Multiple mitochondrial dysfunctions syndrome 1 605711	NFU1
944	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia 614299	BOLA3
944	Multiple mitochondrial dysfunctions syndrome 4 616370	ISCA2
944	Multiple mitochondrial dysfunctions syndrome 5 617613	ISCA1
945	Multiple pterygium syndrome lethal type 253290	CHRNA1
945	Multiple pterygium syndrome lethal type 253290	CHRND
945	Multiple pterygium syndrome lethal type 253290	CHRNG
946	Multiple sulfatase deficiency 272200	SUMF1
947	Multiple synostoses syndrome 1 186500	NOG
947	Multiple synostoses syndrome 2 610017	GDF5
947	Multiple synostoses syndrome 3 612961	FGF9
948	Multisystemic smooth muscle dysfunction syndrome 613834	ACTA2
949	Muscle glycogenosis 300559	PHKA1
949	Muscle hypertrophy 614160	MSTN
950	Muscular dystrophy congenital 613205	LMNA
950	Muscular dystrophy congenital due to ITGA7 deficiency 613204	ITGA7
950	Muscular dystrophy congenital due to partial LAMA2 deficiency 607855	LAMA2
950	Muscular dystrophy congenital megaconial type 602541	CHKB
950	Muscular dystrophy congenital merosin-deficient 607855	LAMA2
950	Muscular dystrophy congenital with cataracts and intellectual disability 617404	INPP5K
950	Muscular dystrophy limb-girdle type 1A 159000	MYOT
950	Muscular dystrophy limb-girdle type 1B 159001	LMNA
950	Muscular dystrophy limb-girdle type 1E 603511	DNAJB6
950	Muscular dystrophy limb-girdle type 1F 608423	TNPO3

950	Muscular dystrophy limb-girdle type 1G 609115	HNRNPDL
950	Muscular dystrophy limb-girdle type 2A 253600	CAPN3
950	Muscular dystrophy limb-girdle type 2B 253601	DYSF
950	Muscular dystrophy limb-girdle type 2C 253700	SGCG
950	Muscular dystrophy limb-girdle type 2D 608099	SGCA
950	Muscular dystrophy limb-girdle type 2E 604286	SGCB
950	Muscular dystrophy limb-girdle type 2F 601287	SGCD
950	Muscular dystrophy limb-girdle type 2G 601954	TCAP
950	Muscular dystrophy limb-girdle type 2H 254110	TRIM32
950	Muscular dystrophy limb-girdle type 2J 608807	TTN
950	Muscular dystrophy limb-girdle type 2L 611307	ANO5
950	Muscular dystrophy limb-girdle type 2Q 613723	PLEC
950	Muscular dystrophy limb-girdle type 2S 615356	TRAPPC11
950	Muscular dystrophy limb-girdle type 2W 616827	LIMS2
950	Muscular dystrophy limb-girdle type IC 607801	CAV3
950	Muscular dystrophy rigid spine 1 602771	SELENON
950	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies type A 11 615181	B3GALNT2
950	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies type A 8 614830	POMGNT2
950	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 1 236670	POMT1
950	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 10 615041	TMEM5
950	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 12 615249	POMK
950	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 13 615287	B3GNT2
950	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 14 615350	GMPPB
950	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 2 613150	POMT2
950	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 3 253280	POMGNT1
950	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 4 253800	FKTN
950	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 5 613153	FKRP
950	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 6 613154	LARGE1
950	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 7 614643	ISPD
950	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 9 616538	DAG1

950	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation) type B 1 613155	POMT1
950	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation) type B 14 615351	GMPPB
950	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation) type B 2 613156	POMT2
950	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation) type B 3 613151	POMGNT1
950	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation) type B 6 608840	LARGE1
950	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation) type B 5 606612	FKRP
950	Muscular dystrophy-dystroglycanopathy (congenital without mental retardation) type B 4 613152	FKTN
950	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 1 609308	POMT1
950	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 14 615352	GMPPB
950	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 2 613158	POMT2
950	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 3 613157	POMGNT1
950	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 4 611588	FKTN
950	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 5 607155	FKRP
950	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 7 616052	ISPD
950	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 9 613818	DAG1
951	Myasthenia congenital 12 with tubular aggregates 610542	GFPT1
952	Myasthenic syndrome congenital 10 254300	DOK7
952	Myasthenic syndrome congenital 11 associated with acetylcholine receptor deficiency 616326	RAPSN
952	Myasthenic syndrome congenital 13 with tubular aggregates 614750	DPAGT1
952	Myasthenic syndrome congenital 14 with tubular aggregates 616228	ALG2
952	Myasthenic syndrome congenital 16 614198	SCN4A
952	Myasthenic syndrome congenital 19 616720	COL13A1
952	Myasthenic syndrome congenital 1A slow-channel 601462	CHRNA1
952	Myasthenic syndrome congenital 1B fast-channel 608930	CHRNA1
952	Myasthenic syndrome congenital 20 presynaptic 617143	SLC5A7
952	Myasthenic syndrome congenital 21 presynaptic 617239	SLC18A3
952	Myasthenic syndrome congenital 2A slow-channel 616313	CHRNB1

952	Myasthenic syndrome congenital 3B fast-channel 616322	CHRND
952	Myasthenic syndrome congenital 4A slow-channel 605809	CHRNE
952	Myasthenic syndrome congenital 4B fast-channel 616324	CHRNE
952	Myasthenic syndrome congenital 4C associated with acetylcholine receptor deficiency 608931	CHRNE
952	Myasthenic syndrome congenital 5 603034	COLQ
952	Myasthenic syndrome congenital 6 presynaptic 254210	CHAT
952	Myasthenic syndrome congenital 7 presynaptic 616040	SYT2
952	Myasthenic syndrome congenital 8 with pre- and postsynaptic defects 615120	AGRN
952	Myasthenic syndrome congenital 9 associated with acetylcholine receptor deficiency 616325	MUSK
953	Myelodysplastic syndrome somatic 614286	ASXL1
953	Myelodysplastic syndrome somatic 614286	SF3B1
953	Myelodysplastic syndrome somatic 614286	TET2
953	{Myelodysplastic syndrome susceptibility to} 614286	GATA2
954	Myelofibrosis somatic 254450	CALR
954	Myelofibrosis somatic 254450	JAK2
954	Myelofibrosis somatic 254450	SH2B3
954	Myelofibrosis with myeloid metaplasia somatic 254450	MPL
955	Myeloperoxidase deficiency 254600	MPO
956	Myhre syndrome 139210	SMAD4
957	Myoclonic epilepsy infantile familial 605021	TBC1D24
957	Myoclonic-atonic epilepsy 616421	SLC6A1
957	{Myoclonic epilepsy juvenile susceptibility to 1} 254770	EFHC1
958	Myoclonus familial cortical 614937	NOL3
958	Myoclonus intractable neonatal 617235	KIF5A
959	Myofibromatosis infantile 1 228550	PDGFRB
960	Myoglobinuria acute recurrent autosomal recessive 268200	LPIN1
961	Myopathy X-linked with excessive autophagy 310440	VMA21
961	Myopathy areflexia respiratory distress and dysphagia early-onset 614399	MEGF10
961	Myopathy areflexia respiratory distress and dysphagia early-onset mild variant 614399	MEGF10
961	Myopathy congenital with fiber-type disproportion 1 255310	ACTA1
961	Myopathy congenital with fiber-type disproportion 255310	SELENON
961	Myopathy congenital with fiber-type disproportion 255310	TPM3
961	Myopathy distal 4 614065	FLNC
961	Myopathy distal 5 617030	ADSL
961	Myopathy distal Tateyama type 614321	CAV3
961	Myopathy distal with anterior tibial onset 606768	DYSF
961	Myopathy distal with rimmed vacuoles 617158	SQSTM1
961	Myopathy due to myoadenylate deaminase deficiency 615511	AMPD1
961	Myopathy lactic acidosis and sideroblastic anemia 1 600462	PUS1
961	Myopathy lactic acidosis and sideroblastic anemia 2 613561	YARS2

961	Myopathy mitochondrial and ataxia 617675	MSTO1
961	Myopathy mitochondrial progressive with congenital cataract hearing loss and developmental delay 613076	GFER
961	Myopathy myofibrillar 1 601419	DES
961	Myopathy myofibrillar 2 608810	CRYAB
961	Myopathy myofibrillar 3 609200	MYOT
961	Myopathy myofibrillar 4 609452	LDB3
961	Myopathy myofibrillar 5 609524	FLNC
961	Myopathy myofibrillar 6 612954	BAG3
961	Myopathy myofibrillar 7 617114	KY
961	Myopathy myofibrillar 8 617258	PYROXD1
961	Myopathy myofibrillar fatal infantile hypertonic alpha-B crystallin-related 613869	CRYAB
961	Myopathy myosin storage autosomal dominant 608358	MYH7
961	Myopathy myosin storage autosomal recessive 255160	MYH7
961	Myopathy proximal with early respiratory muscle involvement 603689	TTN
961	Myopathy spheroid body 182920	MYOT
961	Myopathy tubular aggregate 1 160565	STIM1
961	Myopathy tubular aggregate 2 615883	ORAI1
961	Myopathy vacuolar with CASQ1 aggregates 616231	CASQ1
961	Myopathy with extrapyramidal signs 615673	MICU1
961	Myopathy with lactic acidosis hereditary 255125	ISCU
962	Nemaline myopathy 1 autosomal dominant or recessive 609284	TPM3
962	Nemaline myopathy 10 616165	LMOD3
962	Nemaline myopathy 11 autosomal recessive 617336	MYPN
962	Nemaline myopathy 2 autosomal recessive 256030	NEB
962	Nemaline myopathy 3 autosomal dominant or recessive 161800	ACTA1
962	Nemaline myopathy 4 autosomal dominant 609285	TPM2
962	Nemaline myopathy 5 Amish type 605355	TNNT1
962	Nemaline myopathy 6 autosomal dominant 609273	KBTBD13
962	Nemaline myopathy 7 autosomal recessive 610687	CFL2
962	Nemaline myopathy 8 autosomal recessive 615348	KLHL40
962	Nemaline myopathy 9 615731	KLHL41
962	Myopathy actin congenital with cores 161800	ACTA1
962	Myopathy actin congenital with excess of thin myofilaments 161800	ACTA1
963	Myopia 21 autosomal dominant 614167	ZNF644
963	Myopia 22 autosomal dominant 615420	PRIMPOL
963	Myopia 23 autosomal recessive 615431	LRPAP1
963	Myopia 24 autosomal dominant 615946	SLC39A5
963	Myopia 25 autosomal dominant 617238	P4HA2
963	Myopia 6 608908	SCO2
963	Myopia high with cataract and vitreoretinal degeneration 614292	P3H2
964	Myotonia congenita atypical acetazolamide-responsive 608390	SCN4A

964	Myotonia congenita dominant 160800	CLCN1
964	Myotonia congenita recessive 255700	CLCN1
964	Myotonic dystrophy 1 160900	DMPK
964	Myotonic dystrophy 2 602668	CNBP
964	Myotonia levior, recessive	CLCN1
965	Myotubular myopathy X-linked 310400	MTM1
966	Myxoma intracardiac 255960	PRKAR1A
967	N-acetylglutamate synthase deficiency 237310	NAGS
968	NOR polyagglutination syndrome 111400	A4GALT
969	Naegeli-Franceschetti-Jadassohn syndrome 161000	KRT14
970	Nail disorder nonsyndromic congenital 10 (claw-shaped nails) 614157	FZD6
970	Nail disorder nonsyndromic congenital 3 (leukonychia) 151600	PLCD1
971	Nail-patella syndrome 161200	LMX1B
972	Nance-Horan syndrome 302350	NHS
973	Nanophthalmos 2 609549	MFRP
973	Nanophthalmos 4 615972	TMEM98
974	Nasopharyngeal carcinoma 607107	TP53
974	{Nasopharyngeal carcinoma susceptibility to 3} 617075	MST1R
975	Nasu-Hakola disease 221770	TREM2
975	Nasu-Hakola disease 221770	TYROBP
976	Native American myopathy 255995	STAC3
977	Natural killer cell and glucocorticoid deficiency with DNA repair defect 609981	MCM4
978	Naxos disease 601214	JUP
979	CAP myopathy 1 609284	TPM3
979	CAP myopathy 2 609285	TPM2
980	Nephrogenic syndrome of inappropriate antidiuresis 300539	AVPR2
981	Nephrolithiasis type I 310468	CLCN5
981	Nephrolithiasis or osteoporosis hypophosphatemic 1 612286	SLC34A1
981	Nephrolithiasis or osteoporosis hypophosphatemic 2 612287	SLC9A3R1
981	{Nephrolithiasis uric acid susceptibility to} 605990	ZNF365
982	Nephronophthisis 1 juvenile 256100	NPHP1
982	Nephronophthisis 11 613550	TMEM67
982	Nephronophthisis 12 613820	TTC21B
982	Nephronophthisis 13 614377	WDR19
982	Nephronophthisis 14 614844	ZNF423
982	Nephronophthisis 15 614845	CEP164
982	Nephronophthisis 16 615382	ANKS6
982	Nephronophthisis 18 615862	CEP83
982	Nephronophthisis 19 616217	DCDC2
982	Nephronophthisis 2 infantile 602088	INVS
982	Nephronophthisis 20 617271	MAPKBP1
982	Nephronophthisis 3 604387	NPHP3
982	Nephronophthisis 4 606966	NPHP4

982	Nephronophthisis 7 611498	GLIS2
982	Nephronophthisis-like nephropathy 1 613159	XPNPEP3
982	Nephropathy due to CFHR5 deficiency 614809	CFHR5
982	Nephropathy with pretibial epidermolysis bullosa and deafness 609057	CD151
982	Nephrotic syndrome 14 617575	SGPL1
982	Nephrotic syndrome 15 617609	MAGI2
982	Nephrotic syndrome 16 617783	KANK2
982	Nephrotic syndrome type 1 256300	NPHS1
982	Nephrotic syndrome type 10 615861	EMP2
982	Nephrotic syndrome type 11 616730	NUP107
982	Nephrotic syndrome type 12 616892	NUP93
982	Nephrotic syndrome type 2 600995	NPHS2
982	Nephrotic syndrome type 3 610725	PLCE1
982	Nephrotic syndrome type 4 256370	WT1
982	Nephrotic syndrome type 5 with or without ocular abnormalities 614199	LAMB2
982	Nephrotic syndrome type 6 614196	PTPRO
982	Nephrotic syndrome type 7 615008	DGKE
982	Nephrotic syndrome type 8 615244	ARHGDIA
982	Nephrotic syndrome type 9 615573	COQ8B
983	{Hemolytic uremic syndrome atypical susceptibility to 1} 235400	CFH
983	{Hemolytic uremic syndrome atypical susceptibility to 2} 612922	MCP
983	{Hemolytic uremic syndrome atypical susceptibility to 3} 612923	CFI
983	{Hemolytic uremic syndrome atypical susceptibility to 4} 612924	CFB
983	{Hemolytic uremic syndrome atypical susceptibility to 5} 612925	C3
983	{Hemolytic uremic syndrome atypical susceptibility to 6} 612926	THBD
983	{Hemolytic uremic syndrome atypical susceptibility to} 235400	CFHR1
983	{Hemolytic uremic syndrome atypical susceptibility to} 235400	CFHR3
983	{Hemolytic uremic syndrome atypical susceptibility to 7} 615008	DGKE
984	Nestor-Guillermo progeria syndrome 614008	BANF1
985	Netherton syndrome 256500	SPINK5
986	Neu-Laxova syndrome 1 256520	PHGDH
986	Neu-Laxova syndrome 2 616038	PSAT1
987	Neural tube defects 182940	FUZ
987	Neural tube defects 182940	VANGL2
987	{Neural tube defects susceptibility to} 182940	T
987	{Neural tube defects susceptibility to} 182940	VANGL1
987	{Spina bifida susceptibility to} 182940	CCL2
987	{Neural tube defects folate-sensitive susceptibility to} 601634	MTHFD1
987	{Neural tube defects folate-sensitive susceptibility to} 601634	MTR
987	{Neural tube defects folate-sensitive susceptibility to} 601634	MTRR
987	{Neural tube defects susceptibility to} 601634	MTHFR
988	Neuroblastoma 256700	NME1
988	{Neuroblastoma susceptibility to 1} 256700	KIF1B

988	Neuroblastoma with Hirschsprung disease 613013	PHOX2B
988	{Neuroblastoma susceptibility to 2} 613013	PHOX2B
988	{Neuroblastoma susceptibility to 3} 613014	ALK
989	Neurocutaneous melanosis somatic 249400	NRAS
990	Neurodegeneration childhood-onset with brain atrophy 617672	UBTF
990	Neurodegeneration due to cerebral folate transport deficiency 613068	FOLR1
990	Neurodegeneration with ataxia dystonia and gaze palsy childhood-onset 617145	SQSTM1
990	Neurodegeneration with brain iron accumulation 1 234200	PANK2
990	Neurodegeneration with brain iron accumulation 2B 610217	PLA2G6
990	Neurodegeneration with brain iron accumulation 3 606159	FTL
990	Neurodegeneration with brain iron accumulation 4 614298	C19orf12
990	Neurodegeneration with brain iron accumulation 5 300894	WDR45
990	Neurodegeneration with brain iron accumulation 6 615643	COASY
991	Neurodevelopmental disorder mitochondrial with abnormal movements and lactic acidosis with or without seizures 617710	WARS2
991	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies 617755	BPTF
991	Neurodevelopmental disorder with epilepsy cataracts feeding difficulties and delayed brain myelination 617393	NACC1
991	Neurodevelopmental disorder with hypotonia seizures and absent language 617268	HECW2
991	Neurodevelopmental disorder with involuntary movements 617493	GNAO1
991	Neurodevelopmental disorder with microcephaly hypotonia and variable brain anomalies 617481	PRUNE1
991	Neurodevelopmental disorder with or without anomalies of the brain eye or heart 616975	RERE
991	Neurodevelopmental disorder with progressive microcephaly spasticity and brain anomalies 617527	PLAA
992	Neurofibromatosis familial spinal 162210	NF1
992	Neurofibromatosis type 1 162200	NF1
992	Neurofibromatosis type 2 101000	NF2
992	Neurofibromatosis-Noonan syndrome 601321	NF1
993	Neuromyotonia and axonal neuropathy autosomal recessive 137200	HINT1
994	Neuronopathy distal hereditary motor type IID 615575	FBXO38
994	Neuronopathy distal hereditary motor type IX 617721	WARS
994	Neuronopathy distal hereditary motor type VI 604320	IGHMBP2
994	Neuronopathy distal hereditary motor type VIIA 158580	SLC5A7
995	Neuropathy congenital hypomyelinating 1 605253	EGR2
995	Neuropathy congenital hypomyelinating 605253	MPZ
995	Neuropathy distal hereditary motor type IIA 158590	HSPB8
995	Neuropathy distal hereditary motor type IIB 608634	HSPB1

995	Neuropathy distal hereditary motor type VA 600794	BSCL2
995	Neuropathy distal hereditary motor type VA 600794	GARS
995	Neuropathy distal hereditary motor type VIIB 607641	DCTN1
995	Neuropathy hereditary motor and sensory Russe type 605285	HK1
995	Neuropathy hereditary motor and sensory type VIB 616505	SLC25A46
995	Neuropathy hereditary sensory and autonomic type IA 162400	SPTLC1
995	Neuropathy hereditary sensory and autonomic type IC 613640	SPTLC2
995	Neuropathy hereditary sensory and autonomic type II 201300	WNK1
995	Neuropathy hereditary sensory and autonomic type IIB 613115	FAM134B
995	Neuropathy hereditary sensory and autonomic type V 608654	NGF
995	Neuropathy hereditary sensory and autonomic type VII 615548	SCN11A
995	Neuropathy hereditary sensory and autonomic type VIII 616488	PRDM12
995	Neuropathy hereditary sensory type ID 613708	ATL1
995	Neuropathy hereditary sensory type IE 614116	DNMT1
995	Neuropathy hereditary sensory type IF 615632	ATL3
995	Neuropathy hereditary sensory type IIC 614213	KIF1A
995	Neuropathy hereditary sensory with spastic paraplegia 256840	CCT5
995	Neuropathy hereditary with or without age-related macular degeneration 608895	FBLN5
995	Neuropathy recurrent with pressure palsies 162500	PMP22
996	Neutral lipid storage disease with myopathy 610717	PNPLA2
997	Neutropenia cyclic 162800	ELANE
997	Neutropenia severe congenital 1 autosomal dominant 202700	ELANE
997	Neutropenia severe congenital 3 autosomal recessive 610738	HAX1
997	Neutropenia severe congenital 4 autosomal recessive 612541	G6PC3
997	Neutropenia severe congenital 5 autosomal recessive 615285	VPS45
997	Neutropenia severe congenital 6 autosomal recessive 616022	JAGN1
997	Neutropenia severe congenital 7 autosomal recessive 617014	CSF3R
997	Neutropenia severe congenital X-linked 300299	WAS
997	Neutropenia, alloimmune neonatal	FCGR3B
997	Dursun syndrome 612541	G6PC3
998	Neutrophil immunodeficiency syndrome 608203	RAC2
999	Nevus comedonicus somatic 617025	NEK9
1000	Nevus epidermal somatic 162900	FGFR3
1000	Nevus epidermal somatic 162900	PIK3CA
1000	{Nevus sebaceous or woolly hair nevus somatic} 162900	HRAS
1000	Epidermal nevus somatic 162900	NRAS
1001	Newfoundland rod-cone dystrophy 607476	RLBP1
1002	Nicolaides-Baraitser syndrome 601358	SMARCA2
1003	Niemann-Pick disease type A 257200	SMPD1
1003	Niemann-Pick disease type B 607616	SMPD1
1003	Niemann-Pick disease type C1 257220	NPC1
1003	Niemann-Pick disease type D 257220	NPC1
1003	Niemann-pick disease type C2 607625	NPC2

1004	Night blindness congenital stationary (complete) 1A X-linked 310500	NYX
1004	Night blindness congenital stationary (complete) 1B autosomal recessive 257270	GRM6
1004	Night blindness congenital stationary (complete) 1C autosomal recessive 613216	TRPM1
1004	Night blindness congenital stationary (complete) 1D autosomal recessive 613830	SLC24A1
1004	Night blindness congenital stationary (complete) 1E autosomal recessive 614565	GPR179
1004	Night blindness congenital stationary (complete) 1F autosomal recessive 615058	LRIT3
1004	Night blindness congenital stationary (incomplete) 2A X-linked 300071	CACNA1F
1004	Night blindness congenital stationary autosomal dominant 1 610445	RHO
1004	Night blindness congenital stationary autosomal dominant 2 163500	PDE6B
1004	Night blindness congenital stationary autosomal dominant 3 610444	GNAT1
1004	Night blindness congenital stationary type 1H 617024	GNB3
1005	Nijmegen breakage syndrome 251260	NBN
1005	Nijmegen breakage syndrome-like disorder 613078	RAD50
1006	Nonaka myopathy 605820	GNE
1007	Noonan syndrome 1 163950	PTPN11
1007	Noonan syndrome 10 616564	LZTR1
1007	Noonan syndrome 3 609942	KRAS
1007	Noonan syndrome 4 610733	SOS1
1007	Noonan syndrome 5 611553	RAF1
1007	Noonan syndrome 6 613224	NRAS
1007	Noonan syndrome 7 613706	BRAF
1007	Noonan syndrome 8 615355	RIT1
1007	Noonan syndrome 9 616559	SOS2
1007	Noonan syndrome-like disorder with loose anagen hair 2 617506	PPP1CB
1007	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia 613563	CBL
1007	Noonan-like syndrome with loose anagen hair 607721	SHOC2
1008	Norrie disease 310600	NDP
1009	Norum disease 245900	LCAT
1010	Nystagmus 1 congenital X-linked 310700	FRMD7
1010	Nystagmus 6 congenital X-linked 300814	GPR143
1010	Nystagmus infantile periodic alternating X-linked 310700	FRMD7
1011	OKT4 epitope deficiency 613949	CD4
1012	Obesity adrenal insufficiency and red hair due to POMC deficiency 609734	POMC

1012	Obesity autosomal dominant 601665	MC4R
1012	Obesity hyperphagia and developmental delay 613886	NTRK2
1012	Obesity mild early-onset 601665	NR0B2
1012	Obesity morbid due to leptin deficiency 614962	LEP
1012	Obesity morbid due to leptin receptor deficiency 614963	LEPR
1012	Obesity severe 601665	PPARG
1012	Obesity severe 601665	SIM1
1012	Obesity with impaired prohormone processing 600955	PCSK1
1012	{Obesity association with} 601665	SDC3
1012	{Obesity early-onset susceptibility to} 601665	POMC
1012	{Obesity late-onset} 601665	AGRP
1012	{Obesity severe and type II diabetes} 601665	UCP3
1012	{Obesity susceptibility to} 601665	ADRB2
1012	{Obesity susceptibility to} 601665	ADRB3
1012	{Obesity susceptibility to} 601665	ENPP1
1012	{Obesity susceptibility to} 601665	GHRL
1012	{Obesity susceptibility to} 601665	UCP1
1012	{Obesity variation in} 601665	PPARGC1B
1012	{Obesity severe susceptibility to BMIQ9} 602025	MC3R
1012	{Obesity susceptibility to BMIQ11} 300306	SLC6A14
1012	{Obesity susceptibility to BMIQ12} 612362	PCSK1
1012	{Obesity susceptibility to BMIQ14} 612460	FTO
1012	{Obesity susceptibility to BMIQ4} 607447	UCP2
1012	{Obesity susceptibility to} 607514	FFAR4
1012	Obesity, hyperphagia, and developmental delay	AKR1C2
1013	Occipital horn syndrome 304150	ATP7A
1014	Occult macular dystrophy 613587	RP1L1
1015	Ocular albinism type I Nettleship-Falls type 300500	GPR143
1016	Oculoauricular syndrome 612109	HMX1
1017	Oculodentodigital dysplasia 164200	GJA1
1017	Oculodentodigital dysplasia autosomal recessive 257850	GJA1
1018	Oculopharyngeal muscular dystrophy 164300	PABPN1
1019	Odontoonychodermal dysplasia 257980	WNT10A
1020	Ogden syndrome 300855	NAA10
1021	Oguchi disease-1 258100	SAG
1021	Oguchi disease-2 613411	GRK1
1022	Ohdo syndrome X-linked 300895	MED12
1023	Okur-Chung neurodevelopmental syndrome 617062	CSNK2A1
1024	Oligodontia-colorectal cancer syndrome 608615	AXIN2
1025	Oliver-McFarlane syndrome 275400	PNPLA6
1026	Olmsted syndrome 614594	TRPV3
1027	Omenn syndrome 603554	DCLRE1C
1027	Omenn syndrome 603554	RAG1
1027	Omenn syndrome 603554	RAG2
1028	Omeprazole poor metabolizer 609535	CYP2C19

1029	Omodysplasia 1 258315	GPC6
1030	Oocyte maturation defect 1 615774	ZP1
1030	Oocyte maturation defect 2 616780	TUBB8
1030	Oocyte maturation defect 3 617712	ZP3
1030	Oocyte maturation defect 4 617743	PATL2
1031	Opitz GBBB syndrome type I 300000	MID1
1031	Opitz GBBB syndrome type II 145410	SPECC1L
1032	Opitz-Kaveggia syndrome 305450	MED12
1033	Opsismodysplasia 258480	INPPL1
1034	Optic atrophy 1 165500	OPA1
1034	Optic atrophy 10 with or without ataxia mental retardation and seizures 616732	RTN4IP1
1034	Optic atrophy 3 with cataract 165300	OPA3
1034	Optic atrophy 5 610708	DNM1L
1034	Optic atrophy 7 612989	TMEM126A
1034	Optic atrophy plus syndrome 125250	OPA1
1035	Optic disc anomalies with retinal and or macular dystrophy 212550	SIX6
1036	Optic nerve hypoplasia 165550	PAX6
1037	Ornithine transcarbamylase deficiency 311250	OTC
1038	Orofacial cleft 10 613705	SUMO1
1038	Orofacial cleft 11 600625	BMP4
1038	Orofacial cleft 5 608874	MSX1
1038	Orofacial cleft 7 225060	NECTIN1
1038	Orofacial cleft 8 129400	TP63
1038	{Orofacial cleft 6} 608864	IRF6
1038	Rapp-Hodgkin syndrome 129400	TP63
1039	Orofaciodigital syndrome I 311200	OFD1
1039	Orofaciodigital syndrome IV 258860	TCTN3
1039	Orofaciodigital syndrome V 174300	DDX59
1039	Orofaciodigital syndrome VI 277170	C5orf42
1039	Orofaciodigital syndrome XVI 617563	TMEM107
1040	Orotic aciduria 258900	UMPS
1041	Orthostatic intolerance 604715	SLC6A2
1042	Osseous heteroplasia progressive 166350	GNAS
1043	Osteoarthritis with mild chondrodysplasia 604864	COL2A1
1043	{Osteoarthritis susceptibility 1} 165720	FRZB
1043	{Osteoarthritis susceptibility 2} 140600	MATN3
1043	{Osteoarthritis susceptibility 3} 607850	ASPN
1043	{Osteoarthritis-5} 612400	GDF5
1044	Osteochondrodysplasia complex lethal Symoens-Barnes-Gistelinck type 616897	TAPT1
1045	Osteogenesis imperfecta type I 166200	COL1A1
1045	Osteogenesis imperfecta type II 166210	COL1A1
1045	Osteogenesis imperfecta type II 166210	COL1A2

1045	Osteogenesis imperfecta type III 259420	COL1A1
1045	Osteogenesis imperfecta type III 259420	COL1A2
1045	Osteogenesis imperfecta type IV 166220	COL1A1
1045	Osteogenesis imperfecta type IV 166220	COL1A2
1045	Osteogenesis imperfecta type IX 259440	PPIB
1045	Osteogenesis imperfecta type V 610967	IFITM5
1045	Osteogenesis imperfecta type VI 613982	SERPINF1
1045	Osteogenesis imperfecta type VII 610682	CRTAP
1045	Osteogenesis imperfecta type VIII 610915	P3H1
1045	Osteogenesis imperfecta type X 613848	SERPINH1
1045	Osteogenesis imperfecta type XI 610968	FKBP10
1045	Osteogenesis imperfecta type XIII 614856	BMP1
1045	Osteogenesis imperfecta type XIV 615066	TMEM38B
1045	Osteogenesis imperfecta type XV 615220	WNT1
1045	Osteogenesis imperfecta type XVII 616507	SPARC
1046	Osteoglophonic dysplasia 166250	FGFR1
1047	Osteolysis familial expansile 174810	TNFRSF11A
1048	Osteopathia striata with cranial sclerosis 300373	AMER1
1049	Osteopetrosis autosomal dominant 1 607634	LRP5
1049	Osteopetrosis autosomal dominant 2 166600	CLCN7
1049	Osteopetrosis autosomal recessive 1 259700	TCIRG1
1049	Osteopetrosis autosomal recessive 2 259710	TNFSF11
1049	Osteopetrosis autosomal recessive 3 with renal tubular acidosis 259730	CA2
1049	Osteopetrosis autosomal recessive 4 611490	CLCN7
1049	Osteopetrosis autosomal recessive 5 259720	OSTM1
1049	Osteopetrosis autosomal recessive 6 611497	PLEKHM1
1049	Osteopetrosis autosomal recessive 7 612301	TNFRSF11A
1049	Osteopetrosis autosomal recessive 8 615085	SNX10
1050	Osteopoikilosis with or without melorheostosis 166700	LEMD3
1050	Buschke-Ollendorff syndrome 166700	LEMD3
1051	Osteosclerosis 144750	LRP5
1051	Hyperostosis endosteal 144750	LRP5
1052	Osteoporosis-pseudoglioma syndrome 259770	LRP5
1053	Osteosarcoma 259500	TP53
1053	Osteosarcoma somatic 259500	CHEK2
1053	Osteosarcoma somatic 259500	RB1
1054	Otopalatodigital syndrome type I 311300	FLNA
1054	Otopalatodigital syndrome type II 304120	FLNA
1055	Otospondylomegaepiphyseal dysplasia autosomal dominant 184840	COL11A2
1055	Otospondylomegaepiphyseal dysplasia autosomal recessive 215150	COL11A2
1056	Ovalocytosis SA type 166900	SLC4A1
1057	Ovarian cancer somatic 167000	AKT1

1057	Ovarian cancer somatic 167000	CTNNB1
1057	Ovarian cancer somatic 167000	PIK3CA
1057	Ovarian carcinoma somatic 167000	CDH1
1057	Adenocarcinoma ovarian somatic 167000	PRKN
1057	{Ovarian cancer somatic} 167000	OPCML
1057	{Breast-ovarian cancer familial 1} 604370	BRCA1
1057	{Breast-ovarian cancer familial 2} 612555	BRCA2
1057	{Breast-ovarian cancer familial susceptibility to 3} 613399	RAD51C
1057	{Breast-ovarian cancer familial susceptibility to 4} 614291	RAD51D
1057	Ovarian cancer, somatic	ERBB2
1057	Ovarian carcinoma	RRAS2
1058	Ovarian dysgenesis 1 233300	FSHR
1058	Ovarian dysgenesis 2 300510	BMP15
1058	Ovarian dysgenesis 3 614324	PSMC3IP
1058	Ovarian dysgenesis 4 616185	MCM9
1058	Ovarian dysgenesis 5 617690	SOHLH1
1059	Ovarian hyperstimulation syndrome 608115	FSHR
1060	Ovarian response to FSH stimulation 276400	FSHR
1061	Ovarioleukodystrophy 603896	EIF2B2
1061	Ovarioleukodystrophy 603896	EIF2B4
1061	Ovarioleukodystrophy 603896	EIF2B5
1062	Overhydrated hereditary stomatocytosis 185000	RHAG
1063	PCWH syndrome 609136	SOX10
1064	PEHO syndrome 260565	ZNHIT3
1065	Pachyonychia congenita 1 167200	KRT16
1065	Pachyonychia congenita 2 167210	KRT17
1065	Pachyonychia congenita 3 615726	KRT6A
1065	Pachyonychia congenita 4 615728	KRT6B
1066	Paget disease of bone 3 167250	SQSTM1
1066	Paget disease of bone 5 juvenile-onset 239000	TNFRSF11B
1066	Paget disease of bone 6 616833	ZNF687
1067	Pallister-Hall syndrome 146510	GLI3
1068	Palmoplantar carcinoma multiple self-healing 615255	NLRP1
1069	Palmoplantar hyperkeratosis and true hermaphroditism 610644	RSPO1
1069	Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal 610644	RSPO1
1070	Palmoplantar keratoderma Bothnian type 600231	AQP5
1070	Palmoplantar keratoderma Nagashima type 615598	SERPINB7
1070	Palmoplantar keratoderma and woolly hair 616099	KANK2
1070	Palmoplantar keratoderma epidermolytic 144200	KRT1
1070	Palmoplantar keratoderma epidermolytic 144200	KRT9
1070	Palmoplantar keratoderma nonepidermolytic 600962	KRT1
1070	Palmoplantar keratoderma nonepidermolytic focal 613000	KRT16
1070	Palmoplantar keratoderma nonepidermolytic focal or diffuse 615735	KRT6C

1070	Palmoplantar keratoderma with congenital alopecia 104100	GJA1
1071	Pancreatic agenesis 1 260370	PDX1
1071	Pancreatic agenesis 2 615935	PTF1A
1071	Pancreatic agenesis and congenital heart defects 600001	GATA6
1071	Pancreatic and cerebellar agenesis 609069	PTF1A
1072	Pancreatic cancer 260350	STK11
1072	Pancreatic cancer 260350	TP53
1072	Pancreatic cancer somatic 260350	SMAD4
1072	Pancreatic cancer or melanoma syndrome 606719	CDKN2A
1072	Pancreatic carcinoma somatic 260350	KRAS
1072	{Pancreatic cancer 2} 613347	BRCA2
1072	{Pancreatic cancer susceptibility to 1} 606856	PALLD
1072	{Pancreatic cancer susceptibility to 3} 613348	PALB2
1072	{Pancreatic cancer susceptibility to 4} 614320	BRCA1
1072	Pancreatic cancer, somatic	ACVR1B
1072	Pancreatic carcinoma, somatic	RBBP8
1073	Pancreatitis hereditary 167800	PRSS1
1073	Pancreatitis hereditary 167800	SPINK1
1073	{Pancreatitis chronic protection against} 167800	PRSS2
1073	{Pancreatitis chronic susceptibility to} 167800	CTRC
1073	{Pancreatitis idiopathic} 167800	CFTR
1074	Panhypopituitarism X-linked 312000	SOX3
1075	Papillon-Lefevre syndrome 245000	CTSC
1076	Papillorenal syndrome 120330	PAX2
1077	Paraganglioma and gastric stromal sarcoma 606864	SDHB
1077	Paraganglioma and gastric stromal sarcoma 606864	SDHC
1077	Paraganglioma and gastric stromal sarcoma 606864	SDHD
1077	Paragangliomas 1 with or without deafness 168000	SDHD
1077	Paragangliomas 2 601650	SDHAF2
1077	Paragangliomas 3 605373	SDHC
1077	Paragangliomas 4 115310	SDHB
1077	Paragangliomas 5 614165	SDHA
1078	Paramyotonia congenita 168300	SCN4A
1079	Parastremmatic dwarfism 168400	TRPV4
1080	Parietal foramina 1 168500	MSX2
1080	Parietal foramina 2 609597	ALX4
1080	Parietal foramina with cleidocranial dysplasia 168550	MSX2
1081	Parkes Weber syndrome 608355	RASA1
1082	Parkinson disease 1 168601	SNCA
1082	Parkinson disease 14 autosomal recessive 612953	PLA2G6
1082	Parkinson disease 15 autosomal recessive 260300	FBXO7
1082	Parkinson disease 19a juvenile-onset 615528	DNAJC6
1082	Parkinson disease 19b early-onset 615528	DNAJC6
1082	Parkinson disease 20 early-onset 615530	SYNJ1
1082	Parkinson disease 22 autosomal dominant 616710	CHCHD2

1082	Parkinson disease 23 autosomal recessive early onset 616840	VPS13C
1082	Parkinson disease 4 605543	SNCA
1082	Parkinson disease 6 early onset 605909	PINK1
1082	Parkinson disease 7 autosomal recessive early-onset 606324	PARK7
1082	Parkinson disease juvenile type 2 600116	PRKN
1082	Parkinsonism-dystonia infantile 613135	SLC6A3
1082	{Parkinson disease 11} 607688	GIGYF2
1082	{Parkinson disease 13} 610297	HTRA2
1082	{Parkinson disease 17} 614203	VPS35
1082	{Parkinson disease 18} 614251	EIF4G1
1082	{Parkinson disease 8} 607060	LRRK2
1082	{Parkinson disease age of onset modifier} 168600	GLUD2
1082	{Parkinson disease late-onset susceptibility to} 168600	ATXN2
1082	{Parkinson disease late-onset susceptibility to} 168600	GBA
1082	{Parkinson disease susceptibility to} 168600	ADH1C
1082	{Parkinson disease susceptibility to} 168600	MAPT
1082	{Parkinson disease susceptibility to} 168600	TBP
1083	Paroxysmal nocturnal hemoglobinuria somatic 300818	PIGA
1084	Paroxysmal nonkinesigenic dyskinesia 1 118800	MR1
1084	Paroxysmal nonkinesigenic dyskinesia 3 with or without generalized epilepsy 609446	KCNMA1
1085	Partington syndrome 309510	ARX
1086	Patent ductus arteriosus 2 617035	TFAP2B
1086	Patent ductus arteriosus 3 617039	PRDM6
1087	Peeling skin syndrome 1 270300	CDSN
1087	Peeling skin syndrome 2 609796	TGM5
1087	Peeling skin syndrome 4 607936	CSTA
1087	Peeling skin syndrome 5 617115	SERPINB8
1087	Peeling skin with leukonychia acral punctate keratoses cheilitis and knuckle pads 616295	CAST
1088	Pelger-Huet anomaly 169400	LBR
1089	Pelizaeus-Merzbacher disease 312080	PLP1
1090	Pendred syndrome 274600	SLC26A4
1091	Periodic fever familial 142680	TNFRSF1A
1091	Periodic fever menstrual cycle dependent 614674	HTR1A
1092	Periodontitis 1 juvenile 170650	CTSC
1093	Periampullary Adenoma somatic	APC
1094	Periventricular heterotopia with microcephaly 608097	ARFGEF2
1095	Periventricular nodular heterotopia 7 617201	NEDD4L
1096	Perlman syndrome 267000	DIS3L2
1097	Peroxisomal acyl-CoA oxidase deficiency 264470	ACOX1
1098	Peroxisomal fatty acyl-CoA reductase 1 disorder 616154	FAR1
1099	Peroxisome biogenesis disorder 10A (Zellweger) 614882	PEX3
1099	Peroxisome biogenesis disorder 11A (Zellweger) 614883	PEX13
1099	Peroxisome biogenesis disorder 11B 614885	PEX13

1099	Peroxisome biogenesis disorder 12A (Zellweger) 614886	PEX19
1099	Peroxisome biogenesis disorder 13A (Zellweger) 614887	PEX14
1099	Peroxisome biogenesis disorder 1A (Zellweger) 214100	PEX1
1099	Peroxisome biogenesis disorder 1B (NALD or IRD) 601539	PEX1
1099	Peroxisome biogenesis disorder 2A (Zellweger) 214110	PEX5
1099	Peroxisome biogenesis disorder 2B 202370	PEX5
1099	Peroxisome biogenesis disorder 3A (Zellweger) 614859	PEX12
1099	Peroxisome biogenesis disorder 3B 266510	PEX12
1099	Peroxisome biogenesis disorder 4A (Zellweger) 614862	PEX6
1099	Peroxisome biogenesis disorder 4B 614863	PEX6
1099	Peroxisome biogenesis disorder 5A (Zellweger) 614866	PEX2
1099	Peroxisome biogenesis disorder 5B 614867	PEX2
1099	Peroxisome biogenesis disorder 6A (Zellweger) 614870	PEX10
1099	Peroxisome biogenesis disorder 6B 614871	PEX10
1099	Peroxisome biogenesis disorder 7A (Zellweger) 614872	PEX26
1099	Peroxisome biogenesis disorder 7B 614873	PEX26
1099	Peroxisome biogenesis disorder 8A (Zellweger) 614876	PEX16
1099	Peroxisome biogenesis disorder 8B 614877	PEX16
1099	Peroxisome biogenesis disorder 9B 614879	PEX7
1100	Perrault syndrome 1 233400	HSD17B4
1100	Perrault syndrome 3 614129	CLPP
1100	Perrault syndrome 4 615300	LARS2
1100	Perrault syndrome 5 616138	TWNK
1100	Perrault syndrome 6 617565	ERAL1
1101	Perry syndrome 168605	DCTN1
1102	Persistent Mullerian duct syndrome type I 261550	AMH
1102	Persistent Mullerian duct syndrome type II 261550	AMHR2
1103	Persistent hyperplastic primary vitreous autosomal recessive 221900	ATOH7
1104	Peters-plus syndrome 261540	B3GLCT
1105	Peutz-Jeghers syndrome 175200	STK11
1106	Pfeiffer syndrome 101600	FGFR1
1106	Pfeiffer syndrome 101600	FGFR2
1106	Craniofacial-skeletal-dermatologic dysplasia 101600	FGFR2
1107	Phelan-McDermid syndrome 606232	SHANK3
1108	Phenylketonuria 261600	PAH
1109	Pheochromocytoma 171300	KIF1B
1109	Pheochromocytoma 171300	RET
1109	Pheochromocytoma 171300	SDHB
1109	Pheochromocytoma 171300	SDHD
1109	Pheochromocytoma 171300	VHL
1109	{Pheochromocytoma modifier of} 171300	GDNF
1109	{Pheochromocytoma susceptibility to} 171300	MAX
1109	{Pheochromocytoma susceptibility to} 171300	TMEM127
1110	Phosphoglycerate dehydrogenase deficiency 601815	PHGDH

1110	Phosphoglycerate kinase 1 deficiency 300653	PGK1
1111	Phospholipid phosphatase 6 611666	PLPP6
1112	Phosphoribosylpyrophosphate synthetase superactivity 300661	PRPS1
1112	Gout PRPS-related 300661	PRPS1
1113	Phosphorylase kinase deficiency of liver and muscle autosomal recessive 261750	PHKB
1114	Phosphoserine phosphatase deficiency 614023	PSPH
1115	Pick disease 172700	MAPT
1115	Pick disease 172700	PSEN1
1116	Piebaldism 172800	KIT
1116	Piebaldism 172800	SNAI2
1117	Pierpont syndrome 602342	TBL1XR1
1118	Pierson syndrome 609049	LAMB2
1119	Pigmentary disorder reticulate with systemic manifestations X-linked 301220	POLA1
1120	Pigmented nodular adrenocortical disease primary 1 610489	PRKAR1A
1120	Pigmented nodular adrenocortical disease primary 2 610475	PDE11A
1120	Pigmented nodular adrenocortical disease primary 3 614190	PDE8B
1121	Pigmented paravenous chorioretinal atrophy 172870	CRB1
1122	Pilarowski-Bjornsson syndrome 617682	CHD1
1123	Pitt-Hopkins like syndrome 1 610042	CNTNAP2
1123	Pitt-Hopkins syndrome 610954	TCF4
1123	Pitt-Hopkins-like syndrome 2 614325	NRXN1
1124	Pituitary adenoma 1 multiple types 102200	AIP
1124	Pituitary adenoma 2 GH-secreting 300943	GPR101
1124	Pituitary adenoma 3 multiple types somatic 617686	GNAS
1124	Pituitary adenoma 4 ACTH-secreting somatic 219090	USP8
1124	Pituitary adenoma predisposition 102000	AIP
1124	{Pituitary adenoma 5 multiple types} 617540	CDH23
1124	Pituitary ACTH-secreting adenoma	GNAI2
1124	Pituitary tumor, invasive	PRKCA
1125	Pituitary hormone deficiency combined 1 613038	POU1F1
1125	Pituitary hormone deficiency combined 2 262600	PROP1
1125	Pituitary hormone deficiency combined 3 221750	LHX3
1125	Pituitary hormone deficiency combined 4 262700	LHX4
1125	Pituitary hormone deficiency combined 5 182230	HESX1
1125	Pituitary hormone deficiency combined 6 613986	OTX2
1126	Pityriasis rubra pilaris 173200	CARD14
1127	Plasma triglyceride level QTL low 615881	ANGPTL4
1128	Plasminogen activator inhibitor-1 deficiency 613329	SERPINE1
1129	Plasminogen deficiency type I 217090	PLG
1129	Dysplasminogenemia 217090	PLG
1130	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease 617718	ARPC1B

1130	Platelet disorder familial with associated myeloid malignancy 601399	RUNX1
1130	Platelet glycoprotein IV deficiency 608404	CD36
1130	Platelet-activating factor acetylhydrolase deficiency 614278	PLA2G7
1131	Platyspondylic skeletal dysplasia Torrance type 151210	COL2A1
1132	Pleuropulmonary blastoma 601200	DICER1
1133	Pneumothorax primary spontaneous 173600	FLCN
1134	Poikiloderma hereditary fibrosing with tendon contractures myopathy and pulmonary fibrosis 615704	FAM111B
1134	Poikiloderma with neutropenia 604173	USB1
1135	Polyarteritis nodosa childhood-onset 615688	CECR1
1136	Polycystic kidney disease 1 173900	PKD1
1136	Polycystic kidney disease 2 613095	PKD2
1136	Polycystic kidney disease 3 600666	GANAB
1136	Polycystic kidney disease 4 with or without hepatic disease 263200	PKHD1
1136	Polycystic kidney disease 5 617610	DZIP1L
1137	Polycystic liver disease 1 174050	PRKCSH
1137	Polycystic liver disease 2 617004	SEC63
1138	Polycythemia vera somatic 263300	JAK2
1139	Polydactyly postaxial types A1 and B 174200	GLI3
1139	Polydactyly preaxial type II 174500	LMBR1
1139	Polydactyly preaxial type IV 174700	GLI3
1140	Polyglucosan body disease adult form 263570	GBE1
1140	Polyglucosan body myopathy 1 with or without immunodeficiency 615895	RBCK1
1140	Polyglucosan body myopathy 2 616199	GYG1
1141	Polyhydramnios megalencephaly and symptomatic epilepsy 611087	STRADA
1142	Polymicrogyria bilateral frontoparietal 606854	ADGRG1
1142	Polymicrogyria bilateral perisylvian 615752	ADGRG1
1142	Polymicrogyria perisylvian with cerebellar hypoplasia and arthrogryposis 616531	PI4KA
1143	Polyneuropathy hearing loss ataxia retinitis pigmentosa and cataract 612674	ABHD12
1144	Pontocerebellar hypoplasia type 10 615803	CLP1
1144	Pontocerebellar hypoplasia type 11 617695	TBC1D23
1144	Pontocerebellar hypoplasia type 1A 607596	VRK1
1144	Pontocerebellar hypoplasia type 1B 614678	EXOSC3
1144	Pontocerebellar hypoplasia type 1C 616081	EXOSC8
1144	Pontocerebellar hypoplasia type 2A 277470	TSEN54
1144	Pontocerebellar hypoplasia type 2B 612389	TSEN2
1144	Pontocerebellar hypoplasia type 2D 613811	SEPSECS
1144	Pontocerebellar hypoplasia type 2E 615851	VPS53
1144	Pontocerebellar hypoplasia type 2F 617026	TSEN15

1144	Pontocerebellar hypoplasia type 4 225753	TSEN54
1144	Pontocerebellar hypoplasia type 6 611523	RARS2
1144	Pontocerebellar hypoplasia type 7 614969	TOE1
1144	Pontocerebellar hypoplasia type 8 614961	CHMP1A
1144	Pontocerebellar hypoplasia type 9 615809	AMPD2
1145	Popliteal pterygium syndrome 1 119500	IRF6
1145	Popliteal pterygium syndrome Bartsocas-Papas type 263650	RIPK4
1146	Porencephaly 1 175780	COL4A1
1146	Porencephaly 2 614483	COL4A2
1147	Poretti-Boltshauser syndrome 615960	LAMA1
1148	Porokeratosis 1 multiple types 175800	PMVK
1148	Porokeratosis 3 multiple types 175900	MVK
1148	Porokeratosis 7 multiple types 614714	MVD
1148	Porokeratosis 8 disseminated superficial actinic type 616063	SLC17A9
1148	Porokeratosis 9 multiple types 616631	FDPS
1149	Porphyria acute hepatic 612740	ALAD
1149	Porphyria acute intermittent 176000	HMBS
1149	Porphyria acute intermittent nonerythroid variant 176000	HMBS
1149	Porphyria congenital erythropoietic 263700	UROS
1149	Porphyria cutanea tarda 176100	UROD
1149	Porphyria hepatoerythropoietic 176100	UROD
1149	Porphyria variegata 176200	PPOX
1149	{Porphyria cutanea tarda susceptibility to} 176100	HFE
1149	{Porphyria variegata susceptibility to} 176200	HFE
1149	{Lead poisoning susceptibility to} 612740	ALAD
1150	Portal hypertension noncirrhotic 617068	DGUOK
1151	Prader-Willi syndrome 176270	NDN
1151	Prader-Willi syndrome 176270	SNRPN
1152	Precocious puberty central 2 615346	MKRN3
1152	Precocious puberty male 176410	LHCGR
1152	Leydig cell adenoma somatic with precocious puberty 176410	LHCGR
1153	Preeclampsia or eclampsia 4 609404	STOX1
1153	Preeclampsia or eclampsia 5 614595	CORIN
1153	{Preeclampsia, susceptibility to}	AGT
1154	Pregnancy loss recurrent 4 270960	SYCP3
1154	{Pregnancy loss recurrent susceptibility to 1} 614389	F5
1154	{Pregnancy loss recurrent susceptibility to 2} 614390	F2
1154	{Pregnancy loss recurrent susceptibility to 3} 614391	ANXA5
1155	Preimplantation embryonic lethality 2 617234	PADI6
1155	Preimplantation embryonic lethality 616814	TLE6
1156	Premature aging syndrome Penttinen type 601812	PDGFRB
1157	Premature ovarian failure 1 311360	FMR1
1157	Premature ovarian failure 11 616946	ERCC6
1157	Premature ovarian failure 3 608996	FOXL2
1157	Premature ovarian failure 4 300510	BMP15

1157	Premature ovarian failure 5 611548	NOBOX
1157	Premature ovarian failure 6 612310	FIGLA
1157	Premature ovarian failure 7 612964	NR5A1
1157	Premature ovarian failure 8 615723	STAG3
1157	Premature ovarian failure 9 615724	HFM1
1157	Adrenocortical insufficiency 612964	NR5A1
1158	Primary aldosteronism seizures and neurologic abnormalities 615474	CACNA1D
1159	Primary lateral sclerosis juvenile 606353	ALS2
1160	Primrose syndrome 259050	ZBTB20
1161	Prion disease with protracted course 606688	PRNP
1162	Progressive external ophthalmoplegia autosomal dominant 1 157640	POLG
1162	Progressive external ophthalmoplegia autosomal recessive 1 258450	POLG
1162	Progressive external ophthalmoplegia with mitochondrial DNA deletions autosomal dominant 2 609283	SLC25A4
1162	Progressive external ophthalmoplegia with mitochondrial DNA deletions autosomal dominant 3 609286	TWNK
1162	Progressive external ophthalmoplegia with mitochondrial DNA deletions autosomal dominant 4 610131	POLG2
1162	Progressive external ophthalmoplegia with mitochondrial DNA deletions autosomal dominant 5 613077	RRM2B
1162	Progressive external ophthalmoplegia with mitochondrial DNA deletions autosomal dominant 6 615156	DNA2
1162	Progressive external ophthalmoplegia with mitochondrial DNA deletions autosomal recessive 2 616479	RNASEH1
1162	Progressive external ophthalmoplegia with mitochondrial DNA deletions autosomal recessive 4 617070	DGUOK
1163	Progressive familial heart block type IB 604559	TRPM4
1164	Prolidase deficiency 170100	PEPD
1165	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome 225790	FLVCR2
1166	Properdin deficiency X-linked 312060	PFC
1167	Propionicacidemia 606054	PCCA
1167	Propionicacidemia 606054	PCCB
1168	Prostate cancer 1 601518	RNASEL
1168	Prostate cancer hereditary 176807	MSR1
1168	Prostate cancer somatic 176807	KLF6
1168	Prostate cancer somatic 176807	MAD1L1
1168	{Prostate cancer familial susceptibility to} 176807	CHEK2
1168	{Prostate cancer somatic} 176807	PTEN
1168	{Prostate cancer susceptibility to somatic} 176807	ZFHX3
1168	{Prostate cancer susceptibility to} 176807	AR
1168	{Prostate cancer susceptibility to} 176807	CDH1

1168	{Prostate cancer susceptibility to} 176807	MXI1
1168	{Prostate cancer} 176807	BRCA2
1168	{Prostate cancer hereditary 12} 611868	EHBP1
1168	{Prostate cancer hereditary 13} 611928	MSMB
1168	{Prostate cancer hereditary 2 susceptibility to} 614731	ELAC2
1168	{Prostate cancer or brain cancer susceptibility somatic} 603688	EPHB2
1169	Proteinuria low molecular weight with hypercalciuric nephrocalcinosis 308990	CLCN5
1170	Proteus syndrome somatic 176920	AKT1
1171	Protoporphyria erythropoietic X-linked 300752	ALAS2
1171	Protoporphyria erythropoietic autosomal recessive 177000	FECH
1172	Proud syndrome 300004	ARX
1173	Proximal myopathy and ophthalmoplegia 605637	MYH2
1174	Pseudo-TORCH syndrome 1 251290	OCLN
1174	Pseudo-TORCH syndrome 2 617397	USP18
1175	Pseudoachondroplasia 177170	COMP
1176	Pseudohermaphroditism male with gynecomastia 264300	HSD17B3
1177	Pseudohyperkalemia familial 2 due to red cell leak 609153	ABCB6
1178	Pseudohypoaldosteronism type I 264350	SCNN1A
1178	Pseudohypoaldosteronism type I 264350	SCNN1B
1178	Pseudohypoaldosteronism type I 264350	SCNN1G
1178	Pseudohypoaldosteronism type I autosomal dominant 177735	NR3C2
1178	Pseudohypoaldosteronism type IIB 614491	WNK4
1178	Pseudohypoaldosteronism type IIC 614492	WNK1
1178	Pseudohypoaldosteronism type IID 614495	KLHL3
1178	Pseudohypoaldosteronism type IIE 614496	CUL3
1179	Pseudohypoparathyroidism Ia 103580	GNAS
1179	Pseudohypoparathyroidism Ib 603233	GNAS
1179	Pseudohypoparathyroidism Ic 612462	GNAS
1179	Pseudohypoparathyroidism type IB 603233	GNAS-AS1
1179	Pseudohypoparathyroidism type IB 603233	STX16
1180	Pseudopseudohypoparathyroidism 612463	GNAS
1181	Pseudovaginal perineoscrotal hypospadias 264600	SRD5A2
1182	Pseudoxanthoma elasticum 264800	ABCC6
1182	Pseudoxanthoma elasticum forme fruste 177850	ABCC6
1182	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency 610842	GGCX
1182	{Pseudoxanthoma elasticum modifier of severity of} 264800	XYLT1
1182	{Pseudoxanthoma elasticum modifier of severity of} 264800	XYLT2
1183	Psoriasis 14 pustular 614204	IL36RN
1183	Psoriasis 2 602723	CARD14
1183	{Psoriasis 15 pustular susceptibility to} 616106	AP1S3
1183	{Psoriasis protection against} 605606	IL23R
1183	{Psoriasis susceptibility 13} 614070	TRAF3IP2
1183	{Psoriasis susceptibility 1} 177900	HLA-C

1184	Psychomotor retardation epilepsy and craniofacial dysmorphism 614501	SNIP1
1185	Pulmonary alveolar microlithiasis 265100	SLC34A2
1186	Pulmonary fibrosis and or bone marrow failure telomere-related 3 616373	RTEL1
1186	Pulmonary fibrosis and or bone marrow failure telomere-related 4 616371	PARN
1186	{Pulmonary fibrosis and or bone marrow failure telomere-related 1} 614742	TERT
1186	Pulmonary fibrosis idiopathic 178500	SFTPA2
1186	{Pulmonary fibrosis idiopathic susceptibility to} 178500	MUC5B
1186	{Pulmonary fibrosis idiopathic susceptibility to} 178500	SFTPA1
1186	{Pulmonary fibrosis idiopathic susceptibility to} 614743	TERC
1187	Aplastic anemia 609135	NBN
1187	Aplastic anemia 609135	PRF1
1187	{Aplastic anemia susceptibility to} 609135	SBDS
1187	{Aplastic anemia} 609135	IFNG
1187	{Aplastic anemia} 614743	TERC
1188	Pulmonary hypertension familial primary 1 with or without HHT 178600	BMPR2
1188	Pulmonary hypertension primary 2 615342	SMAD9
1188	Pulmonary hypertension primary 3 615343	CAV1
1188	Pulmonary hypertension primary 4 615344	KCNK3
1188	Pulmonary hypertension primary fenfluramine or dexfenfluramine-associated 178600	BMPR2
1188	{Pulmonary hypertension neonatal susceptibility to} 615371	CPS1
1189	Pulmonary venoocclusive disease 1 265450	BMPR2
1189	Pulmonary venoocclusive disease 2 234810	EIF2AK4
1190	Pycnodysostosis 265800	CTSK
1191	Pyle disease 265900	SFRP4
1192	Pyogenic bacterial infections recurrent due to MYD88 deficiency 612260	MYD88
1193	Pyogenic sterile arthritis pyoderma gangrenosum and acne 604416	PSTPIP1
1194	Pyridoxamine 5'-phosphate oxidase deficiency 610090	PNPO
1195	Pyropoikilocytosis 266140	SPTA1
1196	Pyruvate carboxylase deficiency 266150	PC
1196	Pyruvate dehydrogenase E1-alpha deficiency 312170	PDHA1
1196	Pyruvate dehydrogenase E1-beta deficiency 614111	PDHB
1196	Pyruvate dehydrogenase E2 deficiency 245348	DLAT
1196	Pyruvate dehydrogenase phosphatase deficiency 608782	PDP1
1196	Pyruvate kinase deficiency 266200	PKLR
1197	Quebec platelet disorder 601709	PLAU
1198	Question mark ears isolated 612798	EDN1
1199	RAPADILINO syndrome 266280	RECQL4

1200	RAS-associated autoimmune leukoproliferative disorder 614470	KRAS
1201	RIDDLE syndrome 611943	RNF168
1202	Rabson-Mendenhall syndrome 262190	INSR
1203	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1 605432	HOXA11
1203	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2 616738	MECOM
1204	Rahman syndrome 617537	HIST1H1E
1205	Raine syndrome 259775	FAM20C
1206	Recombination rate QTL 1 612042	RNF212
1207	Reducing body myopathy X-linked 1a severe infantile or early childhood onset 300717	FHL1
1207	Reducing body myopathy X-linked 1b with late childhood or adult onset 300718	FHL1
1208	Refsum disease 266500	PHYH
1209	Renal carcinoma chromophobe somatic 144700	FLCN
1209	Renal cell carcinoma 144700	HNF1A
1209	Renal cell carcinoma 144700	RNF139
1209	Renal cell carcinoma clear cell somatic 144700	OGG1
1209	Renal cell carcinoma papillary 1 300854	TFE3
1209	Renal cell carcinoma papillary 1 familial and somatic 605074	MET
1209	Renal cell carcinoma papillary 605074	PRCC
1209	Renal cell carcinoma somatic 144700	VHL
1209	{Renal cell carcinoma} 144700	HNF1B
1210	Renal cysts and diabetes syndrome 137920	HNF1B
1211	Renal glucosuria 233100	SLC5A2
1212	Renal hypodysplasia or aplasia 1 191830	ITGA8
1212	Renal-hepatic-pancreatic dysplasia 1 208540	NPHP3
1212	{Renal dysplasia cystic susceptibility to} 601331	BICC1
1213	Renal tubular acidosis distal AD 179800	SLC4A1
1213	Renal tubular acidosis distal AR 611590	SLC4A1
1213	Renal tubular acidosis distal autosomal recessive 602722	ATP6V0A4
1213	Renal tubular acidosis proximal with ocular abnormalities 604278	SLC4A4
1213	Renal tubular acidosis with deafness 267300	ATP6V1B1
1214	Renal tubular dysgenesis 267430	ACE
1214	Renal tubular dysgenesis 267430	AGT
1214	Renal tubular dysgenesis 267430	AGTR1
1214	Renal tubular dysgenesis 267430	REN
1215	Renpenning syndrome 309500	PQBP1
1216	Restrictive dermopathy lethal 275210	LMNA
1216	Restrictive dermopathy lethal 275210	ZMPSTE24
1217	Reticular dysgenesis 267500	AK2
1218	Reticulate acropigmentation of Kitamura 615537	ADAM10
1219	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis 614224	IGFBP7

1220	Retinal degeneration, autosomal recessive, clumped pigment type	NRL
1220	Retinal degeneration late-onset autosomal dominant 605670	C1QTNF5
1220	Retinal cone dystrophy 3 610024	PDE6H
1220	Retinal cone dystrophy 3B 610356	KCNV2
1220	Retinal cone dystrophy 4 610478	CACNA2D4
1220	Retinal dystrophy early-onset severe 248200	ABCA4
1220	Retinal dystrophy early-onset severe 613341	LRAT
1220	Retinal dystrophy early-onset with or without pituitary dysfunction 610125	OTX2
1220	Retinal dystrophy iris coloboma and comedogenic acne syndrome 615147	RBP4
1220	Retinal dystrophy with macular staphyloma 617547	C21orf2
1220	Retinal dystrophy with or without extraocular anomalies 617175	RCBTB1
1221	Retinitis pigmentosa 1 180100	RP1
1221	Retinitis pigmentosa 10 180105	IMPDH1
1221	Retinitis pigmentosa 11 600138	PRPF31
1221	Retinitis pigmentosa 13 600059	PRPF8
1221	Retinitis pigmentosa 14 600132	TULP1
1221	Retinitis pigmentosa 17 600852	CA4
1221	Retinitis pigmentosa 18 601414	PRPF3
1221	Retinitis pigmentosa 19 601718	ABCA4
1221	Retinitis pigmentosa 2 312600	RP2
1221	Retinitis pigmentosa 20 613794	RPE65
1221	Retinitis pigmentosa 25 602772	EYS
1221	Retinitis pigmentosa 26 608380	CERKL
1221	Retinitis pigmentosa 27 613750	NRL
1221	Retinitis pigmentosa 28 606068	FAM161A
1221	Retinitis pigmentosa 3 300029	RPGR
1221	Retinitis pigmentosa 30 607921	FSCN2
1221	Retinitis pigmentosa 31 609923	TOPORS
1221	Retinitis pigmentosa 33 610359	SNRNP200
1221	Retinitis pigmentosa 35 610282	SEMA4A
1221	Retinitis pigmentosa 36 610599	PRCD
1221	Retinitis pigmentosa 37 611131	NR2E3
1221	Retinitis pigmentosa 38 613862	MERTK
1221	Retinitis pigmentosa 39 613809	USH2A
1221	Retinitis pigmentosa 4 autosomal dominant or recessive 613731	RHO
1221	Retinitis pigmentosa 41 612095	PROM1
1221	Retinitis pigmentosa 42 612943	KLHL7
1221	Retinitis pigmentosa 43 613810	PDE6A
1221	Retinitis pigmentosa 44 613769	RGR
1221	Retinitis pigmentosa 45 613767	CNGB1
1221	Retinitis pigmentosa 46 612572	IDH3B
1221	Retinitis pigmentosa 47 613758	SAG

1221	Retinitis pigmentosa 48 613827	GUCA1B
1221	Retinitis pigmentosa 49 613756	CNGA1
1221	Retinitis pigmentosa 54 613428	C2orf71
1221	Retinitis pigmentosa 56 613581	IMPG2
1221	Retinitis pigmentosa 57 613582	PDE6G
1221	Retinitis pigmentosa 59 613861	DHDDS
1221	Retinitis pigmentosa 60 613983	PRPF6
1221	Retinitis pigmentosa 61 614180	CLRN1
1221	Retinitis pigmentosa 62 614181	MAK
1221	Retinitis pigmentosa 64 614500	C8orf37
1221	Retinitis pigmentosa 65 613660	CDHR1
1221	Retinitis pigmentosa 68 615725	SLC7A14
1221	Retinitis pigmentosa 69 615780	KIZ
1221	Retinitis pigmentosa 7 and digenic 608133	PRPH2
1221	Retinitis pigmentosa 7 digenic 608133	ROM1
1221	Retinitis pigmentosa 70 615922	PRPF4
1221	Retinitis pigmentosa 71 616394	IFT172
1221	Retinitis pigmentosa 72 616469	ZNF408
1221	Retinitis pigmentosa 73 616544	HGSNAT
1221	Retinitis pigmentosa 74 616562	BBS2
1221	Retinitis pigmentosa 75 617023	AGBL5
1221	Retinitis pigmentosa 76 617123	POMGNT1
1221	Retinitis pigmentosa 77 617304	REEP6
1221	Retinitis pigmentosa 78 617433	ARHGEF18
1221	Retinitis pigmentosa 79 617460	HK1
1221	Retinitis pigmentosa 80 617781	IFT140
1221	Retinitis pigmentosa X-linked and sinorespiratory infections with or without deafness 300455	RPGR
1221	Retinitis pigmentosa and erythrocytic microcytosis 616959	TRNT1
1221	Retinitis pigmentosa concentric 613194	BEST1
1221	Retinitis pigmentosa juvenile 613341	LRAT
1221	Retinitis pigmentosa juvenile autosomal recessive 604232	SPATA7
1221	Retinitis pigmentosa with or without situs inversus 615434	ARL2BP
1221	Retinitis pigmentosa with or without skeletal anomalies 250410	CWC27
1221	Retinitis pigmentosa-12 autosomal recessive 600105	CRB1
1221	Retinitis pigmentosa-40 613801	PDE6B
1221	Retinitis pigmentosa-50 613194	BEST1
1221	Retinitis punctata albescens 136880	PRPH2
1221	Retinitis punctata albescens 136880	RHO
1221	Retinitis punctata albescens 136880	RLBP1
1222	Retinoblastoma 180200	RB1
1222	Retinoblastoma trilateral 180200	RB1
1223	Retinoschisis 312700	RS1
1224	Rett syndrome 312750	MECP2
1224	Rett syndrome atypical 312750	MECP2

1224	Rett syndrome congenital variant 613454	FOXG1
1224	Rett syndrome preserved speech variant 312750	MECP2
1225	Revesz syndrome 268130	TINF2
1226	Rhabdoid tumors somatic 609322	SMARCB1
1226	{Rhabdoid predisposition syndrome 1} 609322	SMARCB1
1226	{Rhabdoid tumor predisposition syndrome 2} 613325	SMARCA4
1227	Rhabdomyosarcoma 2 alveolar 268220	PAX3
1227	Rhabdomyosarcoma 2 alveolar 268220	PAX7
1227	Rhabdomyosarcoma alveolar 268220	FOXO1
1227	Rhabdomyosarcoma embryonal 2 180295	DICER1
1227	Rhabdomyosarcoma somatic 268210	SLC22A1
1228	Rhizomelic chondrodysplasia punctata type 1 215100	PEX7
1228	Rhizomelic chondrodysplasia punctata type 2 222765	GNPAT
1228	Rhizomelic chondrodysplasia punctata type 3 600121	AGPS
1228	Rhizomelic chondrodysplasia punctata type 5 616716	PEX5
1229	Riboflavin deficiency 615026	SLC52A1
1230	Rickets due to defect in vitamin D 25-hydroxylation 600081	CYP2R1
1230	Rickets vitamin D-resistant type IIA 277440	VDR
1231	Right atrial isomerism 208530	GDF1
1232	Rigidity and multifocal seizure syndrome lethal neonatal 614498	BRAT1
1233	Ring dermoid of cornea 180550	PITX2
1234	Rippling muscle disease 606072	CAV3
1235	Ritscher-Schinzel syndrome 1 220210	WSHC5
1235	Ritscher-Schinzel syndrome 2 300963	CCDC22
1236	Roberts syndrome 268300	ESCO2
1237	Robin sequence with cleft mandible and limb anomalies 268305	DDX48
1238	Robinow syndrome autosomal dominant 1 180700	WNT5A
1238	Robinow syndrome autosomal dominant 2 616331	DVL1
1238	Robinow syndrome autosomal dominant 3 616894	DVL3
1238	Robinow syndrome autosomal recessive 268310	ROR2
1239	Robinow-Sorauf syndrome 180750	TWIST1
1240	Roifman syndrome 616651	RNU4ATAC
1241	Rothmund-Thomson syndrome 268400	RECQL4
1242	Roussy-Levy syndrome 180800	MPZ
1242	Roussy-Levy syndrome 180800	PMP22
1243	Rubinstein-Taybi syndrome 1 180849	CREBBP
1243	Rubinstein-Taybi syndrome 2 613684	EP300
1244	Ruijs-Aalfs syndrome 616200	SPRTN
1245	SADDAN 616482	FGFR3
1246	SBBYSS syndrome 603736	KAT6B
1247	SC phocomelia syndrome 269000	ESCO2
1248	SCID autosomal recessive T-negative or B-positive type 600802	JAK3
1249	SED Maroteaux type 184095	TRPV4
1249	SED congenita 183900	COL2A1
1250	SESAME syndrome 612780	KCNJ10

1251	SHORT syndrome 269880	PIK3R1
1252	SMED Strudwick type 184250	COL2A1
1253	STAR syndrome 300707	FAM58A
1254	STING-associated vasculopathy infantile-onset 615934	TMEM173
1255	Sacral agenesis with vertebral anomalies 615709	T
1256	Saethre-Chotzen syndrome 101400	FGFR2
1256	Saethre-Chotzen syndrome with or without eyelid anomalies 101400	TWIST1
1257	Salih myopathy 611705	TTN
1258	Salla disease 604369	SLC17A5
1259	Salt and pepper developmental regression syndrome 609056	SIAT9
1260	Sandhoff disease infantile juvenile and adult forms 268800	HEXB
1261	Scalp-ear-nipple syndrome 181270	KCTD1
1262	Scaphocephaly maxillary retrusion and mental retardation 609579	FGFR2
1263	Scapuloperoneal myopathy X-linked dominant 300695	FHL1
1263	Scapuloperoneal spinal muscular atrophy 181405	TRPV4
1263	Scapuloperoneal syndrome myopathic type 181430	MYH7
1263	Scapuloperoneal syndrome neurogenic Kaeser type 181400	DES
1264	Schaaf-Yang syndrome 615547	MAGEL2
1265	Schimke immunoosseous dysplasia 242900	SMARCAL1
1266	Schimmelpenning-Feuerstein-Mims syndrome somatic mosaic 163200	HRAS
1266	Schimmelpenning-Feuerstein-Mims syndrome somatic mosaic 163200	KRAS
1266	Schimmelpenning-Feuerstein-Mims syndrome somatic mosaic 163200	NRAS
1267	Schindler disease type I 609241	NAGA
1267	Schindler disease type III 609241	NAGA
1268	Schinzel-Giedion midface retraction syndrome 269150	SETBP1
1269	Schizencephaly 269160	COL4A1
1269	Schizencephaly 269160	EMX2
1269	Schizencephaly 269160	SHH
1269	Schizencephaly 269160	SIX3
1270	Schneckenbecken dysplasia 269250	SLC35D1
1271	Schopf-Schulz-Passarge syndrome 224750	WNT10A
1272	Schuurs-Hoeijmakers syndrome 615009	PACS1
1273	Schwannomatosis 162091	NF2
1273	{Schwannomatosis-1 susceptibility to} 162091	SMARCB1
1273	{Schwannomatosis-2 susceptibility to} 615670	LZTR1
1274	Schwartz-Jampel syndrome type 1 255800	HSPG2
1275	Sclerosing cholangitis neonatal 617394	DCDC2
1276	Sclerosteosis 1 269500	SOST
1276	Sclerosteosis 2 614305	LRP4
1277	Scott syndrome 262890	ANO6

1278	Sea-blue histiocyte disease 269600	APOE
1279	Sebastian syndrome 605249	MYH9
1280	Seborrhea-like dermatitis with psoriasiform elements 610227	ZNF750
1281	Seckel syndrome 1 210600	ATR
1281	Seckel syndrome 10 617253	NSMCE2
1281	Seckel syndrome 2 606744	RBBP8
1281	Seckel syndrome 5 613823	CEP152
1281	Seckel syndrome 9 616777	TRAIP
1282	Segawa syndrome recessive 605407	TH
1283	Seizures benign familial infantile 2 605751	PRRT2
1283	Seizures benign familial infantile 3 607745	SCN2A
1283	Seizures benign familial infantile 5 617080	SCN8A
1283	Seizures benign neonatal 1 121200	KCNQ2
1283	Myokymia 121200	KCNQ2
1283	Seizures benign neonatal type 2 121201	KCNQ3
1283	Seizures cortical blindness microcephaly syndrome 616632	DIAPH1
1284	Sengers syndrome 212350	AGK
1285	Senior-Loken syndrome 4 606996	NPHP4
1285	Senior-Loken syndrome 5 609254	IQCB1
1285	Senior-Loken syndrome 6 610189	CEP290
1285	Senior-Loken syndrome 7 613615	SDCCAG8
1285	Senior-Loken syndrome 8 616307	WDR19
1285	Senior-Loken syndrome 9 616629	TRAF3IP1
1285	Senior-Loken syndrome-1 266900	NPHP1
1286	Sessile serrated polyposis cancer syndrome 617108	RNF43
1287	Severe combined immunodeficiency Athabascan type 602450	DCLRE1C
1287	Severe combined immunodeficiency B cell-negative 601457	RAG1
1287	Severe combined immunodeficiency B cell-negative 601457	RAG2
1287	Severe combined immunodeficiency T cell-negative B-cell or natural killer-cell positive 608971	PTPRC
1287	Severe combined immunodeficiency T-cell negative B-cell or natural killer cell-positive type 608971	IL7R
1287	Severe combined immunodeficiency X-linked 300400	IL2RG
1287	Severe combined immunodeficiency with microcephaly growth retardation and sensitivity to ionizing radiation 611291	NHEJ1
1288	Shaheen syndrome 615328	COG6
1289	Shashi-Pena syndrome 617190	ASXL2
1290	Short QT syndrome 1 609620	KCNH2
1290	Short QT syndrome 2 609621	KCNQ1
1290	Short QT syndrome 3 609622	KCNJ2
1291	Short stature and advanced bone age with or without early-onset osteoarthritis and or osteochondritis dissecans 165800	ACAN
1291	Short stature auditory canal atresia mandibular hypoplasia skeletal abnormalities 602471	GSC

1291	Short stature brachydactyly intellectual developmental disability and seizures 617157	PRMT7
1291	Short stature developmental delay and congenital heart defects 617044	TKT
1291	Short stature hearing loss retinitis pigmentosa and distinctive facies 617763	EXOSC2
1291	Short stature idiopathic familial 300582	SHOX
1291	Short stature idiopathic familial 300582	SHOX
1291	Short stature microcephaly and endocrine dysfunction 616541	XRCC4
1291	Short stature onychodysplasia facial dysmorphism and hypotrichosis 614813	POC1A
1291	Short stature optic nerve atrophy and Pelger-Huet anomaly 614800	NBAS
1291	Short stature rhizomelic with microcephaly micrognathia and developmental delay 617164	ARCN1
1291	Short stature with microcephaly and distinctive facies 615789	CRIPT
1291	Short stature with nonspecific skeletal abnormalities 616255	NPR2
1292	Short-rib thoracic dysplasia 10 with or without polydactyly 615630	IFT172
1292	Short-rib thoracic dysplasia 11 with or without polydactyly 615633	WDR34
1292	Short-rib thoracic dysplasia 13 with or without polydactyly 616300	CEP120
1292	Short-rib thoracic dysplasia 14 with polydactyly 616546	KIAA0586
1292	Short-rib thoracic dysplasia 15 with polydactyly 617088	DYNC2LI1
1292	Short-rib thoracic dysplasia 16 with or without polydactyly 617102	IFT52
1292	Short-rib thoracic dysplasia 17 with or without polydactyly 617405	TCTEX1D2
1292	Short-rib thoracic dysplasia 2 with or without polydactyly 611263	IFT80
1292	Short-rib thoracic dysplasia 3 with or without polydactyly 613091	DYNC2H1
1292	Short-rib thoracic dysplasia 4 with or without polydactyly 613819	TTC21B
1292	Short-rib thoracic dysplasia 6 with or without polydactyly 263520	NEK1
1292	Short-rib thoracic dysplasia 7 with or without polydactyly 614091	WDR35
1292	Short-rib thoracic dysplasia 8 with or without polydactyly 615503	WDR60
1292	Short-rib thoracic dysplasia 9 with or without polydactyly 266920	IFT140
1293	Shprintzen-Goldberg syndrome 182212	SKI
1294	Shwachman-Diamond syndrome 260400	SBDS
1295	Sialic acid storage disorder infantile 269920	SLC17A5
1295	Sialidosis type I 256550	NEU1
1295	Sialidosis type II 256550	NEU1
1296	Sialuria 269921	GNE
1297	Sick sinus syndrome 1 608567	SCN5A
1297	Sick sinus syndrome 2 163800	HCN4
1297	{Sick sinus syndrome 3} 614090	MYH6
1298	Sickle cell anemia 603903	HBB

1299	Sideroblastic anemia with B-cell immunodeficiency periodic fevers and developmental delay 616084	TRNT1
1300	Sifrim-Hitz-Weiss syndrome 617159	CHD4
1301	Silver spastic paraplegia syndrome 270685	BSCL2
1302	Silver-Russell syndrome 180860	H19
1303	Simpson-Golabi-Behmel syndrome type 1 312870	GPC3
1303	Simpson-Golabi-Behmel syndrome type 2 300209	OFD1
1304	Single median maxillary central incisor 147250	SHH
1305	Singleton-Merten syndrome 1 182250	IFIH1
1305	Singleton-Merten syndrome 2 616298	DDX58
1306	Sinoatrial node dysfunction and deafness 614896	CACNA1D
1307	Sitosterolemia 210250	ABCG5
1307	Sitosterolemia 210250	ABCG8
1308	Sjogren-Larsson syndrome 270200	ALDH3A2
1309	Skeletal defects genital hypoplasia and mental retardation 612447	ZBTB16
1310	Skin fragility-woolly hair syndrome 607655	DSP
1311	Skraban-Deardorff syndrome 617616	WDR26
1312	Small fiber neuropathy 133020	SCN9A
1312	Erythermalgia primary 133020	SCN9A
1313	Smith-Kingsmore syndrome 616638	MTOR
1314	Smith-Lemli-Opitz syndrome 270400	DHCR7
1315	Smith-Magenis syndrome 182290	RAI1
1316	Smith-McCort dysplasia 2 615222	RAB33B
1316	Smith-McCort dysplasia 607326	DYM
1317	Snowflake vitreoretinal degeneration 193230	KCNJ13
1318	Sorsby fundus dystrophy 136900	TIMP3
1319	Sotos syndrome 1 117550	NSD1
1319	Sotos syndrome 2 614753	NFIX
1320	Spastic ataxia 1 autosomal dominant 108600	VAMP1
1320	Spastic ataxia 2 autosomal recessive 611302	KIF1C
1320	Spastic ataxia 3 autosomal recessive 611390	MARS2
1320	Spastic ataxia 5 autosomal recessive 614487	AFG3L2
1320	Spastic ataxia 8 autosomal recessive with hypomyelinating leukodystrophy 617560	NKX6-2
1320	Spastic ataxia Charlevoix-Saguenay type 270550	SACS
1320	Spastic paralysis infantile onset ascending 607225	ALS2
1320	Spastic paraplegia 10 autosomal dominant 604187	KIF5A
1320	Spastic paraplegia 11 autosomal recessive 604360	SPG11
1320	Spastic paraplegia 12 autosomal dominant 604805	RTN2
1320	Spastic paraplegia 13 autosomal dominant 605280	HSPD1
1320	Spastic paraplegia 15 autosomal recessive 270700	ZFYVE26
1320	Spastic paraplegia 18 autosomal recessive 611225	ERLIN2
1320	Spastic paraplegia 2 X-linked 312920	PLP1
1320	Spastic paraplegia 23 270750	DSTYK

1320	Spastic paraplegia 26 autosomal recessive 609195	B4GALNT1
1320	Spastic paraplegia 28 autosomal recessive 609340	DDHD1
1320	Spastic paraplegia 30 autosomal recessive 610357	KIF1A
1320	Spastic paraplegia 31 autosomal dominant 610250	REEP1
1320	Spastic paraplegia 33 autosomal dominant 610244	ZFYVE27
1320	Spastic paraplegia 35 autosomal recessive 612319	FA2H
1320	Spastic paraplegia 39 autosomal recessive 612020	PNPLA6
1320	Spastic paraplegia 3A autosomal dominant 182600	ATL1
1320	Spastic paraplegia 4 autosomal dominant 182601	SPAST
1320	Spastic paraplegia 42 autosomal dominant 612539	SLC33A1
1320	Spastic paraplegia 44 autosomal recessive 613206	GJC2
1320	Spastic paraplegia 45 autosomal recessive 613162	NT5C2
1320	Spastic paraplegia 46 autosomal recessive 614409	GBA2
1320	Spastic paraplegia 47 autosomal recessive 614066	AP4B1
1320	Spastic paraplegia 48 autosomal recessive 613647	AP5Z1
1320	Spastic paraplegia 49 autosomal recessive 615031	TECPR2
1320	Spastic paraplegia 50 autosomal recessive 612936	AP4M1
1320	Spastic paraplegia 51 autosomal recessive 613744	AP4E1
1320	Spastic paraplegia 52 autosomal recessive 614067	AP4S1
1320	Spastic paraplegia 53 autosomal recessive 614898	VPS37A
1320	Spastic paraplegia 54 autosomal recessive 615033	DDHD2
1320	Spastic paraplegia 55 autosomal recessive 615035	C12orf65
1320	Spastic paraplegia 56 autosomal recessive 615030	CYP2U1
1320	Spastic paraplegia 5A autosomal recessive 270800	CYP7B1
1320	Spastic paraplegia 6 autosomal dominant 600363	NIPA1
1320	Spastic paraplegia 62 615681	ERLIN1
1320	Spastic paraplegia 64 autosomal recessive 615683	ENTPD1
1320	Spastic paraplegia 7 autosomal recessive 607259	PGLYRP1
1320	Spastic paraplegia 75 autosomal recessive 616680	MAG
1320	Spastic paraplegia 76 autosomal recessive 616907	CAPN1
1320	Spastic paraplegia 78 autosomal recessive 617225	ATP13A2
1320	Spastic paraplegia 79 autosomal recessive 615491	UCHL1
1320	Spastic paraplegia 8 autosomal dominant 603563	WSHC5
1320	Spastic paraplegia 9A autosomal dominant 601162	ALDH18A1
1320	Spastic paraplegia 9B autosomal recessive 616586	ALDH18A1
1320	Spastic paraplegia and psychomotor retardation with or without seizures 616756	HACE1
1320	Spastic paraplegia intellectual disability nystagmus and obesity 617296	KIDINS220
1320	Spastic paraplegia optic atrophy and neuropathy 609541	KLC2
1320	Spastic tetraplegia thin corpus callosum and progressive microcephaly 616657	SLC1A4
1321	Spasticity childhood-onset with hyperglycinemia 616859	GLRX5
1322	Specific granule deficiency 2 617475	SMARCD2
1322	Specific granule deficiency 245480	CEBPE

1323	Speech-language disorder-1 602081	FOXP2
1324	Spermatocytic seminoma somatic 273300	FGFR3
1324	Germ cell tumors somatic 273300	KIT
1324	Testicular tumor somatic 273300	STK11
1324	{Male germ cell tumor, somatic} 273300	BCL10
1325	Spermatogenic failure 10 614822	12-Sep
1325	Spermatogenic failure 11 615081	KLHL10
1325	Spermatogenic failure 12 615413	NANOS1
1325	Spermatogenic failure 16 617187	SUN5
1325	Spermatogenic failure 18 617576	DNAH1
1325	Spermatogenic failure 19 617592	CFAP43
1325	Spermatogenic failure 3 606766	SLC26A8
1325	Spermatogenic failure 4 270960	SYCP3
1325	Spermatogenic failure 5 243060	AURKC
1325	Spermatogenic failure 7 612997	CATSPER1
1325	Spermatogenic failure 8 613957	NR5A1
1325	Spermatogenic failure 9 613958	DPY19L2
1325	Spermatogenic failure X-linked 2 309120	TEX11
1325	Spermatogenic failure Y-linked 2 415000	USP9Y
1326	Spherocytosis type 1 182900	ANK1
1326	Spherocytosis type 2 616649	SPTB
1326	Spherocytosis type 3 270970	SPTA1
1326	Spherocytosis type 4 612653	SLC4A1
1326	Spherocytosis type 5 612690	EPB42
1327	Spinal and bulbar muscular atrophy of Kennedy 313200	AR
1327	Spinal muscular atrophy Jokela type 615048	CHCHD10
1327	Spinal muscular atrophy X-linked 2 infantile 301830	UBA1
1327	Spinal muscular atrophy distal X-linked 3 300489	ATP7A
1327	Spinal muscular atrophy distal autosomal recessive 4 611067	PLEKHG5
1327	Spinal muscular atrophy distal autosomal recessive 5 614881	DNAJB2
1327	Spinal muscular atrophy distal congenital nonprogressive 600175	TRPV4
1327	Spinal muscular atrophy late-onset Finkel type 182980	VAPB
1327	Spinal muscular atrophy lower extremity-predominant 1 AD 158600	DYNC1H1
1327	Spinal muscular atrophy lower extremity-predominant 2 AD 615290	BICD2
1327	Spinal muscular atrophy with congenital bone fractures 1 616866	TRIP4
1327	Spinal muscular atrophy with progressive myoclonic epilepsy 159950	ASAH1
1327	Spinal muscular atrophy-1 253300	SMN1
1327	Spinal muscular atrophy-2 253550	SMN1
1327	Spinal muscular atrophy-3 253400	SMN1
1327	{Spinal muscular atrophy type III modifier of} 253400	SMN2
1327	Spinal muscular atrophy-4 271150	SMN1
1328	Spinocerebellar ataxia 1 164400	ATXN1

1328	Spinocerebellar ataxia 10 603516	ATXN10
1328	Spinocerebellar ataxia 11 604432	TTBK2
1328	Spinocerebellar ataxia 12 604326	PPP2R2B
1328	Spinocerebellar ataxia 13 605259	KCNC3
1328	Spinocerebellar ataxia 14 605361	PRKCG
1328	Spinocerebellar ataxia 15 606658	ITPR1
1328	Spinocerebellar ataxia 17 607136	TBP
1328	Spinocerebellar ataxia 19 607346	KCND3
1328	Spinocerebellar ataxia 2 183090	ATXN2
1328	Spinocerebellar ataxia 21 607454	TMEM240
1328	Spinocerebellar ataxia 23 610245	PDYN
1328	Spinocerebellar ataxia 27 609307	FGF14
1328	Spinocerebellar ataxia 28 610246	AFG3L2
1328	Spinocerebellar ataxia 29 congenital nonprogressive 117360	ITPR1
1328	Spinocerebellar ataxia 31 117210	BEAN1
1328	Spinocerebellar ataxia 34 133190	ELOVL4
1328	Spinocerebellar ataxia 35 613908	TGM6
1328	Spinocerebellar ataxia 36 614153	NOP56
1328	Spinocerebellar ataxia 37 615945	DAB1
1328	Spinocerebellar ataxia 38 615957	ELOVL5
1328	Spinocerebellar ataxia 42 616795	CACNA1G
1328	Spinocerebellar ataxia 44 617691	GRM1
1328	Spinocerebellar ataxia 45 617769	FAT2
1328	Spinocerebellar ataxia 5 600224	SPTBN2
1328	Spinocerebellar ataxia 6 183086	CACNA1A
1328	Spinocerebellar ataxia 7 164500	ATXN7
1328	Spinocerebellar ataxia 8 608768	ATXN8
1328	Spinocerebellar ataxia 8 608768	ATXN8OS
1328	Spinocerebellar ataxia autosomal recessive 1 606002	SETX
1328	Spinocerebellar ataxia autosomal recessive 10 613728	ANO10
1328	Spinocerebellar ataxia autosomal recessive 11 614229	SYT14
1328	Spinocerebellar ataxia autosomal recessive 12 614322	WWOX
1328	Spinocerebellar ataxia autosomal recessive 13 614831	GRM1
1328	Spinocerebellar ataxia autosomal recessive 14 615386	SPTBN2
1328	Spinocerebellar ataxia autosomal recessive 16 615768	STUB1
1328	Spinocerebellar ataxia autosomal recessive 17 616127	CWF19L1
1328	Spinocerebellar ataxia autosomal recessive 18 616204	GRID2
1328	Spinocerebellar ataxia autosomal recessive 2 213200	PMPCA
1328	Spinocerebellar ataxia autosomal recessive 20 616354	SNX14
1328	Spinocerebellar ataxia autosomal recessive 21 616719	SCYL1
1328	Spinocerebellar ataxia autosomal recessive 23 616949	TDP2
1328	Spinocerebellar ataxia autosomal recessive 7 609270	TPP1
1328	Spinocerebellar ataxia autosomal recessive 8 610743	SYNE1
1328	Spinocerebellar ataxia autosomal recessive with axonal neuropathy 607250	TDP1

1329	Split-foot malformation with mesoaxial polydactyly 616890	ZAK
1329	Split-hand or foot malformation 4 605289	TP63
1329	Split-hand or foot malformation 6 225300	WNT10B
1330	Spondylo-megaepiphyseal-metaphyseal dysplasia 613330	NKX3-2
1331	Spondylocarpotarsal synostosis syndrome 272460	FLNB
1332	Spondylocheirodysplasia Ehlers-Danlos syndrome-like 612350	SLC39A13
1333	Spondylocostal dysostosis 1 autosomal recessive 277300	DLL3
1333	Spondylocostal dysostosis 2 autosomal recessive 608681	MESP2
1333	Spondylocostal dysostosis 4 autosomal recessive 613686	HES7
1333	Spondylocostal dysostosis 5 122600	TBX6
1334	Spondyloenchondrodysplasia with immune dysregulation 607944	ACP5
1335	Spondyloepimetaphyseal dysplasia 608728	MATN3
1335	Spondyloepimetaphyseal dysplasia Camera-Genevieve type 610442	NANS
1335	Spondyloepimetaphyseal dysplasia Faden-Alkuraya type 616723	RSPRY1
1335	Spondyloepimetaphyseal dysplasia Missouri type 602111	MMP13
1335	Spondyloepimetaphyseal dysplasia X-linked 300106	BGN
1335	Spondyloepimetaphyseal dysplasia with joint laxity type 1 with or without fractures 271640	B3GALT6
1335	Spondyloepimetaphyseal dysplasia with joint laxity type 2 603546	KIF22
1335	Spondyloepiphyseal dysplasia Stanescu type 616583	COL2A1
1335	Spondyloepiphyseal dysplasia tarda 313400	TRAPPC2
1335	Spondyloepiphyseal dysplasia tarda with progressive arthropathy 208230	WISP3
1335	Spondyloepiphyseal dysplasia with congenital joint dislocations 143095	CHST3
1335	Arthropathy progressive pseudorheumatoid of childhood 208230	WISP3
1336	Spondylometaepiphyseal dysplasia short limb-hand type 271665	DDR2
1337	Spondylometaphyseal dysplasia Kozlowski type 184252	TRPV4
1337	Spondylometaphyseal dysplasia Megarbane-Dagher-Melike type 613320	PAM16
1337	Spondylometaphyseal dysplasia Sedaghatian type 250220	GPX4
1337	Spondylometaphyseal dysplasia axial 602271	C21orf2
1337	Spondylometaphyseal dysplasia with cone-rod dystrophy 608940	PCYT1A
1338	Spondyloocular syndrome 605822	XYLT2
1339	Spondyloperipheral dysplasia 271700	COL2A1
1340	Squamous cell carcinoma head and neck 275355	TNFRSF10B
1340	Squamous cell carcinoma head and neck somatic 275355	ING1
1340	Squamous cell carcinoma head and neck somatic 275355	PTEN
1341	Squamous cell carcinoma, burn scar-related, somatic	FAS
1342	Stankiewicz-Isidor syndrome 617516	PSMD12
1343	Stapes ankylosis with broad thumb and toes 184460	NOG
1344	Stargardt disease 1 248200	ABCA4
1344	Stargardt disease 3 600110	ELOVL4
1344	Stargardt disease 4 603786	PROM1

1345	Steatocystoma multiplex 184500	KRT17
1346	Stickler syndrome type I nonsyndromic ocular 609508	COL2A1
1346	Stickler syndrome type I 108300	COL2A1
1346	Stickler syndrome type II 604841	COL11A1
1346	Stickler syndrome type IV 614134	COL9A1
1347	Stiff skin syndrome 184900	FBN1
1348	Stocco dos Santos X-linked mental retardation syndrome 300434	SHROOM4
1349	Stomatin-deficient cryohydrocytosis with neurologic defects 608885	SLC2A1
1350	Stormorken syndrome 185070	STIM1
1351	Striatal degeneration autosomal dominant 609161	PDE8B
1351	Striatal degeneration autosomal dominant 616922	PDE10A
1352	Striatonigral degeneration childhood-onset 617054	VAC14
1352	Striatonigral degeneration infantile 271930	NUP62
1353	Stromme syndrome 243605	CENPF
1354	Structural heart defects and renal anomalies syndrome 617478	TMEM260
1355	Sturge-Weber syndrome somatic mosaic 185300	GNAQ
1356	Stuttering familial persistent 1 184450	AP4E1
1357	Stuve-Wiedemann syndrome or Schwartz-Jampel type 2 syndrome 601559	LIFR
1358	Succinic semialdehyde dehydrogenase deficiency 271980	ALDH5A1
1359	Succinyl CoA:3-oxoacid CoA transferase deficiency 245050	OXCT1
1360	Sucrase-isomaltase deficiency congenital 222900	SI
1361	Sudden cardiac failure infantile 617222	PPA2
1362	Sudden infant death with dysgenesis of the testes syndrome 608800	TSPYL1
1362	{Sudden infant death syndrome susceptibility to} 272120	SCN5A
1363	Sulfite oxidase deficiency 272300	SUOX
1364	Supranuclear palsy progressive 601104	MAPT
1364	Supranuclear palsy progressive atypical 260540	MAPT
1365	Supravalvar aortic stenosis 185500	ELN
1366	Surfactant metabolism dysfunction pulmonary 1 265120	SFTPB
1366	Surfactant metabolism dysfunction pulmonary 2 610913	SFTPC
1366	Surfactant metabolism dysfunction pulmonary 3 610921	ABCA3
1366	Surfactant metabolism dysfunction pulmonary 4 300770	CSF2RA
1366	Surfactant metabolism dysfunction pulmonary 5 614370	CSF2RB
1367	Sveinsson chorioretinal atrophy 108985	TEAD1
1368	Sweeney-Cox syndrome 617746	TWIST1
1369	Symmetric circumferential skin creases congenital 1 156610	TUBB
1369	Symmetric circumferential skin creases congenital 2 616734	MAPRE2
1370	Symphalangism proximal 1A 185800	NOG
1370	Symphalangism proximal 1B 615298	GDF5
1371	Syndactyly mesoaxial synostotic with phalangeal reduction 609432	BHLHA9
1371	Syndactyly type III 186100	GJA1

1371	Syndactyly type IV 186200	LMBR1
1371	Syndactyly type V 186300	HOXD13
1372	Synpolydactyly 1 186000	HOXD13
1373	Systemic lupus erythematosus 16 614420	DNASE1L3
1373	{Systemic lupus erythematosus resistance to} 601744	TLR5
1373	{Systemic lupus erythematosus susceptibility to 10} 612251	IRF5
1373	{Systemic lupus erythematosus susceptibility to 11} 612253	STAT4
1373	{Systemic lupus erythematosus susceptibility to 1} 601744	TLR5
1373	{Systemic lupus erythematosus susceptibility to 2} 605218	PDCD1
1373	{Systemic lupus erythematosus susceptibility to 9} 610927	CR2
1373	{Systemic lupus erythematosus susceptibility to} 152700	PTPN22
1373	{Systemic lupus erythematosus susceptibility to} 152700	CTLA4
1373	{Systemic lupus erythematosus susceptibility to} 152700	DNASE1
1373	{Systemic lupus erythematosus susceptibility to} 152700	FCGR2B
1373	{Systemic lupus erythematosus susceptibility to} 152700	TREX1
1373	{Lupus nephritis susceptibility to} 152700	FCGR2A
1373	{Systemic lupus erythematous association with susceptibility to 6} 609939	ITGAM
1374	T-cell immunodeficiency congenital alopecia and nail dystrophy 601705	FOXN1
1374	T-cell immunodeficiency recurrent infections autoimmunity and cardiac malformations 614868	STK4
1375	TARP syndrome 311900	RBM10
1376	Takenouchi-Kosaki syndrome 616737	CDC42
1377	Tangier disease 205400	ABCA1
1378	Tarsal-carpal coalition syndrome 186570	NOG
1379	Tatton-Brown-Rahman syndrome 615879	DNMT3A
1380	Telangiectasia hereditary hemorrhagic type 1 187300	ENG
1380	Telangiectasia hereditary hemorrhagic type 2 600376	ACVRL1
1380	Telangiectasia hereditary hemorrhagic type 5 615506	GDF2
1381	Temple-Baraitser syndrome 611816	KCNH1
1382	Temtamy preaxial brachydactyly syndrome 605282	CHSY1
1383	Temtamy syndrome 218340	C12orf57
1384	Tenorio syndrome 616260	RNF125
1385	Terminal osseous dysplasia 300244	FLNA
1386	Tetralogy of Fallot 187500	GATA4
1386	Tetralogy of Fallot 187500	GATA6
1386	Tetralogy of Fallot 187500	GDF1
1386	Tetralogy of Fallot 187500	JAG1
1386	Tetralogy of Fallot 187500	NKX2-5
1386	Tetralogy of Fallot 187500	TBX1
1386	Tetralogy of Fallot 187500	ZFPM2
1387	Thalassemia Hispanic gamma-delta-beta 613985	HBB-LCR
1387	Thalassemia alpha- 604131	HBA2
1387	Thalassemia-beta dominant inclusion-body 603902	HBB

1387	Thalassemias alpha- 604131	HBA1
1387	Thalassemias beta- 613985	HBB
1387	Thalassemia due to Hb Lepore	HBD
1387	Thalassemia, delta-	HBD
1387	Delta-beta thalassemia 141749	HBB
1388	Thanatophoric dysplasia type I 187600	FGFR3
1388	Thanatophoric dysplasia type II 187601	FGFR3
1389	Thauvin-Robinet-Faivre syndrome 617107	FIBP
1390	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2) 607483	SLC19A3
1390	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type) 613710	SLC25A19
1390	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type) 614458	TPK1
1390	Thiamine-responsive megaloblastic anemia syndrome 249270	SLC19A2
1391	Thrombocythemia 1 187950	THPO
1391	Thrombocythemia 2 601977	MPL
1391	Thrombocythemia 3 614521	JAK2
1391	Thrombocythemia somatic 187950	CALR
1391	Thrombocythemia somatic 187950	SH2B3
1392	Thrombocytopenia 2 188000	ANKRD26
1392	Thrombocytopenia 3 273900	FYB
1392	Thrombocytopenia 4 612004	CYCS
1392	Thrombocytopenia 5 616216	ETV6
1392	Thrombocytopenia X-linked 313900	WAS
1392	Thrombocytopenia X-linked intermittent 313900	WAS
1392	Thrombocytopenia X-linked with or without dyserythropoietic anemia 300367	GATA1
1392	Thrombocytopenia congenital amegakaryocytic 604498	MPL
1392	Thrombocytopenia with beta-thalassemia X-linked 314050	GATA1
1392	Thrombocytopenia-absent radius syndrome 274000	RBM8A
1392	Thrombocytopenia, neonatal alloimmune	ITGB3
1392	Thrombocytopenia, neonatal alloimmune, BAK antigen related	ITGA2B
1393	{Deep venous thrombosis protection against} 300807	F9
1393	Thrombophilia X-linked due to factor IX defect 300807	F9
1393	Thrombophilia due to HRG deficiency 613116	HRG
1393	Thrombophilia due to activated protein C resistance 188055	F5
1393	{Thrombophilia susceptibility to due to factor V Leiden} 188055	F5
1393	Thrombophilia due to antithrombin III deficiency 613118	SERPINC1
1393	Thrombophilia due to heparin cofactor II deficiency 612356	SERPIND1
1393	Thrombophilia due to protein C deficiency autosomal dominant 176860	PROC
1393	Thrombophilia due to protein C deficiency autosomal recessive 612304	PROC

1393	Thrombophilia due to protein S deficiency autosomal dominant 612336	PROS1
1393	Thrombophilia due to protein S deficiency autosomal recessive 614514	PROS1
1393	Thrombophilia due to thrombin defect 188050	F2
1393	{Venous thromboembolism susceptibility to} 188050	HABP2
1393	{Venous thrombosis protection against} 188050	F13A1
1393	{Thromboembolism susceptibility to} 188050	MTHFR
1393	Thrombophilia due to thrombomodulin defect 614486	THBD
1394	Thrombotic thrombocytopenic purpura familial 274150	ADAMTS13
1394	Purpura, posttransfusion	ITGB3
1395	Thyroid carcinoma follicular 188470	MINPP1
1395	Thyroid carcinoma follicular somatic 188470	NRAS
1395	{Thyroid cancer nonmedullary 2} 188470	SRGAP1
1395	{Thyroid carcinoma follicular somatic} 188470	HRAS
1395	{Thyroid cancer nonmedullary 1} 188550	NKX2-1
1395	{Thyroid cancer nonmedullary 4} 616534	FOXE1
1395	{Thyroid carcinoma Hurthle cell} 607464	NDUFA13
1395	Medullary thyroid carcinoma 155240	RET
1395	Medullary thyroid carcinoma familial 155240	NTRK1
1395	Parathyroid adenoma with cystic changes 145001	CDC73
1395	Parathyroid carcinoma 608266	CDC73
1395	Parathyroid adenoma, somatic	MEN1
1395	Thyroid adenoma, hyperfunctioning, somatic	TSHR
1395	Thyroid carcinoma with thyrotoxicosis	TSHR
1396	Thyroid dyshormonogenesis 1 274400	SLC5A5
1396	Thyroid dyshormonogenesis 2A 274500	TPO
1396	Thyroid dyshormonogenesis 3 274700	TG
1396	Thyroid dyshormonogenesis 4 274800	IYD
1396	Thyroid dyshormonogenesis 5 274900	DUOXA2
1396	Thyroid dyshormonogenesis 6 607200	DUOX2
1397	Thyroid hormone metabolism abnormal 609698	SECISBP2
1397	Thyroid hormone resistance 188570	THRB
1397	Thyroid hormone resistance autosomal recessive 274300	THRB
1397	Thyroid hormone resistance selective pituitary 145650	THRB
1398	Tibial muscular dystrophy tardive 600334	TTN
1399	Tietz albinism-deafness syndrome 103500	MITF
1400	Timothy syndrome 601005	CACNA1C
1401	Tn polyagglutination syndrome somatic 300622	C1GALT1C1
1402	Toenail dystrophy isolated 607523	COL7A1
1403	Tooth agenesis selective 1 with or without orofacial cleft 106600	MSX1
1403	Tooth agenesis selective 3 604625	PAX9
1403	Tooth agenesis selective 4 150400	WNT10A
1403	Tooth agenesis selective 7 616724	LRP6
1403	Tooth agenesis selective 8 617073	WNT10B

1403	Tooth agenesis selective 9 617275	GREM2
1403	Tooth agenesis selective X-linked 1 313500	EDA
1404	Tourette syndrome 137580	SLITRK1
1404	{Gilles de la Tourette syndrome susceptibility to} 137580	HDC
1405	Townes-Brocks branchiootorenal-like syndrome 107480	SALL1
1405	Townes-Brocks syndrome 1 107480	SALL1
1406	Traboulsi syndrome 601552	ASPH
1407	Transaldolase deficiency 606003	TALDO1
1408	Transcobalamin II deficiency 275350	TCN2
1409	Transient bullous of the newborn 131705	COL7A1
1410	Transposition of great arteries dextro-looped 3 613854	GDF1
1410	Transposition of the great arteries dextro-looped 1 608808	MED13L
1411	Treacher Collins syndrome 1 154500	TCOF1
1411	Treacher Collins syndrome 2 613717	POLR1D
1411	Treacher Collins syndrome 3 248390	POLR1C
1412	Trichodontoosseous syndrome 190320	DLX3
1413	Trichoepithelioma multiple familial 1 601606	CYLD
1414	Trichohepatoenteric syndrome 1 222470	TTC37
1414	Trichohepatoenteric syndrome 2 614602	SKIV2L
1415	Trichomegaly 190330	FGF5
1416	Trichorhinophalangeal syndrome type I 190350	TRPS1
1416	Trichorhinophalangeal syndrome type III 190351	TRPS1
1417	Trichothiodystrophy 1 photosensitive 601675	ERCC2
1417	Trichothiodystrophy 2 photosensitive 616390	ERCC3
1417	Trichothiodystrophy 3 photosensitive 616395	GTF2H5
1417	Trichothiodystrophy 4 nonphotosensitive 234050	MPLKIP
1417	Trichothiodystrophy 6 nonphotosensitive 616943	GTF2E2
1418	Trifunctional protein deficiency 609015	HADHA
1418	Trifunctional protein deficiency 609015	HADHB
1419	Trigonocephaly 1 190440	FGFR1
1419	Trigonocephaly 2 614485	FREM1
1420	Trimethylaminuria 602079	FMO3
1421	Triphalangeal thumb type I 174500	LMBR1
1421	Triphalangeal thumb-polysyndactyly syndrome 174500	LMBR1
1422	Trismus-pseudocamptodactyly syndrome 158300	MYH8
1423	Tropical calcific pancreatitis 608189	SPINK1
1423	{Fibrocalculous pancreatic diabetes susceptibility to} 608189	SPINK1
1424	Troyer syndrome 275900	SPG20
1425	Tuberous sclerosis-1 191100	TSC1
1425	Tuberous sclerosis-2 613254	TSC2
1425	{TSC2 angiomyolipomas renal modifier of} 613254	IFNG
1426	Tumor predisposition syndrome 614327	BAP1
1427	Tumoral calcinosis familial normophosphatemic 610455	SAMD9
1427	Tumoral calcinosis hyperphosphatemic 211900	KL
1427	Tumoral calcinosis hyperphosphatemic familial 211900	FGF23

1427	Tumoral calcinosis hyperphosphatemic familial 211900	GALNT3
1428	Tylosis with esophageal cancer 148500	RHBDF2
1429	Tyrosinemia type I 276700	FAH
1429	Tyrosinemia type II 276600	TAT
1429	Tyrosinemia type III 276710	HPD
1430	UV-sensitive syndrome 1 600630	ERCC6
1430	UV-sensitive syndrome 2 614621	ERCC8
1430	UV-sensitive syndrome 3 614640	UVSSA
1431	Ullrich congenital muscular dystrophy 1 254090	COL6A1
1431	Ullrich congenital muscular dystrophy 1 254090	COL6A2
1431	Ullrich congenital muscular dystrophy 1 254090	COL6A3
1432	Ulna and fibula absence of with severe limb deficiency 276820	WNT7A
1433	Ulnar-mammary syndrome 181450	TBX3
1434	Uncombable hair syndrome 191480	PADI3
1435	Urbach-Wiethe disease 247100	ECM1
1436	Urofacial syndrome 1 236730	HPSE2
1436	Urofacial syndrome 2 615112	LRIG2
1437	Usher syndrome type 1B 276900	MYO7A
1437	Usher syndrome type 1C 276904	USH1C
1437	Usher syndrome type 1D 601067	CDH23
1437	Usher syndrome type 1D or F digenic 601067	CDH23
1437	Usher syndrome type 1D or F digenic 601067	PCDH15
1437	Usher syndrome type 1F 602083	PCDH15
1437	Usher syndrome type 1G 606943	USH1G
1437	Usher syndrome type 2A 276901	USH2A
1437	{Retinal disease in Usher syndrome type IIA modifier of} 276901	PDZD7
1437	Usher syndrome type 2C 605472	ADGRV1
1437	Usher syndrome type 2C GPR98 or PDZD7 digenic 605472	ADGRV1
1437	Usher syndrome type 2D 611383	WHRN
1437	Usher syndrome type 3A 276902	CLRN1
1437	Usher syndrome type 3B 614504	HARS
1437	Usher syndrome type IIC GPR98 or PDZD7 digenic 605472	PDZD7
1437	Usher syndrome type IJ 614869	CIB2
1438	VACTERL association X-linked 314390	ZIC3
1439	VATER association with macrocephaly and ventriculomegaly 276950	PTEN
1440	VLCAD deficiency 201475	ACADVL
1441	Van Buchem disease 239100	SOST
1442	Van Maldergem syndrome 1 601390	DCHS1
1442	Van Maldergem syndrome 2 615546	FAT4
1443	Van den Ende-Gupta syndrome 600920	SCARF2
1444	Van der Woude syndrome 2 606713	GRHL3
1445	Vas deferens congenital bilateral aplasia of X-linked 300985	ADGRG2
1446	Vascular malformation primary intraosseous 606893	ELMO2
1447	Vasculopathy retinal with cerebral leukodystrophy 192315	TREX1

1448	Velocardiofacial syndrome 192430	TBX1
1449	Venous malformations multiple cutaneous and mucosal 600195	TEK
1450	Ventricular fibrillation familial 1 603829	SCN5A
1450	{Ventricular fibrillation paroxysmal familial 2} 612956	DPP6
1451	Ventricular septal defect 1 614429	GATA4
1451	Ventricular septal defect 2 614431	CITED2
1451	Ventricular septal defect 3 614432	NKX2-5
1452	Ventricular tachycardia catecholaminergic polymorphic 1 604772	RYR2
1452	Ventricular tachycardia catecholaminergic polymorphic 2 611938	CASQ2
1452	Ventricular tachycardia catecholaminergic polymorphic 3 614021	TECRL
1452	Ventricular tachycardia catecholaminergic polymorphic 4 614916	CALM1
1452	Ventricular tachycardia catecholaminergic polymorphic 5 with or without muscle weakness 615441	TRDN
1452	Ventricular tachycardia idiopathic 192605	GNAI2
1453	Ventriculomegaly with cystic kidney disease 219730	CRB2
1454	Verheij syndrome 615583	PUF60
1455	Vertebral cardiac renal and limb defects syndrome 1 617660	HAAO
1455	Vertebral cardiac renal and limb defects syndrome 2 617661	KYNU
1456	Vesicoureteral reflux 2 610878	ROBO2
1456	Vesicoureteral reflux 3 613674	SOX17
1456	Vesicoureteral reflux 8 615963	TNXB
1457	Vibratory urticaria 125630	ADGRE2
1458	Vici syndrome 242840	EPG5
1459	Visceral myopathy 155310	ACTG2
1460	Vitamin D-dependent rickets type I 264700	CYP27B1
1461	Vitamin K-dependent clotting factors combined deficiency of 1 277450	GGCX
1461	Vitamin K-dependent clotting factors combined deficiency of 2 607473	VKORC1
1462	Vohwinkel syndrome 124500	GJB2
1462	Vohwinkel syndrome with ichthyosis 604117	LOR
1463	WHIM syndrome 193670	CXCR4
1464	Waardenburg syndrome type 1 193500	PAX3
1464	Waardenburg syndrome type 2A 193510	MITF
1464	Waardenburg syndrome type 2D 608890	SNAI2
1464	Waardenburg syndrome type 2E with or without neurologic involvement 611584	SOX10
1464	Waardenburg syndrome type 3 148820	PAX3
1464	Waardenburg syndrome type 4A 277580	EDNRB
1464	Waardenburg syndrome type 4B 613265	EDN3
1464	Waardenburg syndrome type 4C 613266	SOX10
1464	Waardenburg syndrome or albinism digenic 103470	TYR
1464	Waardenburg syndrome or ocular albinism digenic 103470	MITF
1465	Wagner syndrome 1 143200	VCAN
1466	Warburg micro syndrome 1 600118	RAB3GAP1

1466	Warburg micro syndrome 2 614225	RAB3GAP2
1466	Warburg micro syndrome 3 614222	RAB18
1466	Warburg micro syndrome 4 615663	TBC1D20
1467	Warfarin resistance 122700	VKORC1
1467	Warfarin sensitivity 122700	CYP2C9
1467	{Warfarin sensitivity} 122700	F9
1467	Coumarin resistance 122700	CYP2A6
1468	Warsaw breakage syndrome 613398	DDX11
1469	Watson syndrome 193520	NF1
1470	Weaver syndrome 277590	EZH2
1471	Weill-Marchesani syndrome 1 recessive 277600	ADAMTS10
1471	Weill-Marchesani syndrome 2 dominant 608328	FBN1
1471	Weill-Marchesani-like syndrome 613195	ADAMTS17
1472	Welander distal myopathy 604454	TIA1
1473	Werner syndrome 277700	RECQL2
1474	Weyers acrofacial dysostosis 193530	EVC2
1475	White sponge nevus 1 193900	KRT4
1475	White sponge nevus 2 615785	KRT13
1476	White-Sutton syndrome 616364	POGZ
1477	Wieacker-Wolff syndrome 314580	ZC4H2
1478	Wiedemann-Steiner syndrome 605130	KMT2A
1479	Wilms tumor 194070	BRCA2
1479	Wilms tumor 2 194071	H19
1479	Wilms tumor somatic 194070	GPC3
1479	Wilms tumor type 1 194070	WT1
1479	{Wilms tumor 6 susceptibility to} 616806	REST
1479	{Wilms tumor susceptibility-5} 601583	POU6F2
1480	Wilson disease 277900	ATP7B
1481	Wilson-Turner syndrome 309585	LAS1L
1482	Wiskott-Aldrich syndrome 301000	WAS
1483	Witteveen-Kolk syndrome 613406	SIN3A
1484	Wolcott-Rallison syndrome 226980	EIF2AK3
1485	Wolff-Parkinson-White syndrome 194200	PRKAG2
1486	Wolfram syndrome 1 222300	WFS1
1486	Wolfram syndrome 2 604928	CISD2
1486	Wolfram-like syndrome autosomal dominant 614296	WFS1
1487	Wolman disease 278000	LIPA
1487	Cholesteryl ester storage disease 278000	LIPA
1488	Woodhouse-Sakati syndrome 241080	DCAF17
1489	Woolly hair autosomal dominant 194300	KRT74
1489	Woolly hair autosomal recessive 1 with or without hypotrichosis 278150	LPAR6
1489	Woolly hair autosomal recessive 2 with or without hypotrichosis 604379	LIPH
1489	Woolly hair autosomal recessive 3 616760	KRT25

1490	Wrinkly skin syndrome 278250	ATP6V0A2
1491	X-inactivation familial skewed 300087	XIC
1492	Xanthinuria type I 278300	XDH
1492	Xanthinuria type II 603592	MOCOS
1493	Xeroderma pigmentosum group A 278700	XPA
1493	Xeroderma pigmentosum group B 610651	ERCC3
1493	Xeroderma pigmentosum group C 278720	XPC
1493	Xeroderma pigmentosum group D 278730	ERCC2
1493	Xeroderma pigmentosum group E DDB-negative subtype 278740	DDB2
1493	Xeroderma pigmentosum group F 278760	ERCC4
1493	Xeroderma pigmentosum group G 278780	ERCC5
1493	Xeroderma pigmentosum group G or Cockayne syndrome 278780	ERCC5
1493	Xeroderma pigmentosum type F or Cockayne syndrome 278760	ERCC4
1493	Xeroderma pigmentosum variant type 278750	POLH
1494	Xia-Gibbs syndrome 615829	AHDC1
1495	You-Hoover-Fong syndrome 616954	TELO2
1496	Yunis-Varon syndrome 216340	FIG4
1497	ZTTK syndrome 617140	SON
1498	Zimmermann-Laband syndrome 1 135500	KCNH1
1498	Zimmermann-Laband syndrome 2 616455	ATP6V1B2
1499	Zinc deficiency transient neonatal 608118	SLC30A2
1500	van Buchem disease type 2 607636	LRP5
1501	van der Woude syndrome 119300	IRF6
1502	von Hippel-Lindau syndrome 193300	VHL
1502	{von Hippel-Lindau syndrome modifier of} 193300	CCND1
1503	von Willebrand disease platelet-type 177820	GP1BA
1503	von Willebrand disease type 1 193400	VWF
1503	von Willebrand disease types 2A 2B 2M and 2N 613554	VWF
1503	von Willibrand disease type 3 277480	VWF
1504	{AIDS delayed or rapid progression to} 609423	KIR3DL1
1504	{AIDS rapid progression to} 609423	IFNG
1504	{AIDS resistance to} 609423	CXCL12
1504	{AIDS slow progression to} 609423	CXCR1
1504	{AIDS slow progression to} 609423	IL4R
1504	{HIV type 1 susceptibility to} 609423	CD209
1504	{HIV-1 resistance to} 609423	CCL2
1504	{HIV-1 susceptibility to} 609423	IL10
1504	{HIV-1 viremia susceptibility to} 609423	HLA-C
1504	{HIV or AIDS susceptibility to} 609423	CCL3L1
1504	{HIV1 infection resistance to} 609423	TLR3
1504	{HIV1 resistance to} 609423	CCL11
1504	{Rapid progression to AIDS from HIV1 infection} 609423	CX3CR1
1504	{HIV infection, susceptibility/resistance to}	CCR2
1504	{HIV infection, susceptibility/resistance to}	CCR5
1504	{HIV-1 disease, delayed progression of}	CCL5

1504	{HIV-1 disease, rapid progression of}	CCL5
1505	{Accelerated tumor formation susceptibility to} 614401	MDM2
1506	{Alcohol dependence protection against} 103780	ADH1B
1506	{Alcohol dependence protection against} 103780	ADH1C
1506	{Alcohol dependence susceptibility to} 103780	GABRA2
1506	{Alcohol dependence susceptibility to} 103780	HTR2A
1506	{Alcohol dependence susceptibility to} 103780	TAS2R16
1507	{Allergic rhinitis susceptibility to} 607154	IL13
1508	{Thiopurines poor metabolism of 1} 610460	TPMT
1508	{Thiopurines poor metabolism of 2} 616903	NUDT15
1509	{Anxiety-related personality traits} 607834	SLC6A4
1510	{Anorexia nervosa susceptibility to} 606788	HTR2A
1510	{Anorexia nervosa susceptibility to} 610269	BDNF
1511	{Asperger syndrome susceptibility X-linked 1} 300494	NLGN3
1511	{Asperger syndrome susceptibility X-linked 2} 300497	NLGN4X
1512	{Aspergillosis susceptibility to} 614079	CLEC7A
1513	{Atopy susceptibility to} 147050	IL4R
1513	{Atopy susceptibility to} 147050	MS4A2
1513	{Atopy susceptibility to} 147050	PLA2G7
1514	{Attention deficit-hyperactivity disorder susceptibility to 7} 613003	TPH2
1514	{Attention deficit-hyperactivity disorder susceptibility to} 143465	DRD5
1514	{Attention deficit-hyperactivity disorder} 143465	DRD4
1515	{Autism susceptibility 15} 612100	CNTNAP2
1515	{Autism susceptibility 17} 613436	SHANK2
1515	{Autism susceptibility X-linked 1} 300425	NLGN3
1515	{Autism susceptibility X-linked 2} 300495	NLGN4X
1515	{Autism susceptibility X-linked 3} 300496	MECP2
1515	{Autism susceptibility to 18} 615032	CHD8
1515	{Autism susceptibility to 19} 615091	EIF4E
1515	{Autism susceptibility to X-linked 4} 300830	PTCHD1
1515	{Autism susceptibility to X-linked 5} 300847	RPL10
1515	{Autism susceptibility to X-linked 6} 300872	TMLHE
1516	{Bacteremia protection against} 614382	TIRAP
1516	{Bacteremia susceptibility to} 614383	CISH
1517	{Blepharospasm primary benign} 606798	DRD5
1518	{Budd-Chiari syndrome somatic} 600880	JAK2
1518	{Budd-Chiari syndrome} 600880	F5
1519	{Bulimia nervosa age of onset of weight loss in} 607499	BDNF
1520	{Buruli ulcer susceptibility to} 610446	SLC11A1
1521	{Celiac disease susceptibility to 3} 609755	CTLA4
1521	{Celiac disease susceptibility to 4} 609753	MYO9B
1521	{Celiac disease susceptibility to} 212750	HLA-DQA1
1521	{Celiac disease susceptibility to} 212750	HLA-DQB1
1522	{Chronic infections due to MBL deficiency} 614372	MBL2

1523	{Codeine sensitivity} 608902	CYP2D6
1523	{Debrisoquine sensitivity} 608902	CYP2D6
1524	{Colchicine resistance} 120080	ABCB1
1525	{Coronary artery disease autosomal dominant 1} 608320	MEF2A
1525	{Coronary artery disease autosomal dominant 2} 610947	LRP6
1525	{Coronary artery disease in familial hypercholesterolemia protection against} 143890	ABCA1
1525	{Coronary artery disease resistance to} 607339	CX3CR1
1525	{Coronary artery disease severe susceptibility to} 617347	APOE
1525	{Coronary heart disease susceptibility to 5} 608901	KALRN
1525	{Coronary heart disease susceptibility to 6} 614466	MMP3
1525	{Coronary heart disease susceptibility to 7} 610938	CD36
1525	{Coronary artery disease, modifier of}	CCL2
1525	{Coronary artery disease, susceptibility to}	IRS1
1525	{Coronary artery disease, susceptibility to}	KL
1525	{Coronary artery disease, susceptibility to}	PON1
1525	{Coronary artery disease, susceptibility to}	PON2
1525	{Coronary artery spasm 1, susceptibility to}	NOS3
1525	{Coronary artery spasm 2, susceptibility to	PON1
1526	{Dengue fever protection against} 614371	CD209
1527	{Dermatitis atopic susceptibility to 2} 605803	FLG
1528	{Drug addiction susceptibility to} 606581	FAAH
1529	{Dyslexia susceptibility to 1} 127700	DNAAF4
1530	{Encephalopathy acute infection-induced 3 susceptibility to} 608033	RANBP2
1530	{Encephalopathy acute infection-induced 4 susceptibility to} 614212	CPT2
1531	{Glaucoma normal tension susceptibility to} 606657	OPA1
1531	{Glaucoma normal tension susceptibility to} 606657	OPTN
1532	{Glucocorticoid therapy response to} 614400	GLCCI1
1533	{Exfoliation syndrome susceptibility to} 177650	LOXL1
1534	{Gout susceptibility 4} 612671	SLC17A3
1535	{Graft-versus-host disease protection against} 614395	IL10
1536	{H. pylori infection susceptibility to} 600263	IFNGR1
1537	{Hashimoto thyroiditis} 140300	CTLA4
1538	{Hemorrhage intracerebral susceptibility to} 614519	COL4A1
1538	{Hemorrhage intracerebral susceptibility to} 614519	COL4A2
1538	{Stroke hemorrhagic} 614519	ACE
1539	{Hepatitis B virus infection susceptibility to} 610424	IFNGR1
1539	{Hepatitis B virus susceptibility to} 610424	IL10RB
1539	{Hepatitis B virus susceptibility to} 610424	IFNAR2
1539	{Hepatitis C virus infection response to therapy of} 609532	IFNL3
1539	{Hepatitis C virus resistance to} 609532	CCR5
1539	{Hepatitis C virus response to therapy of} 609532	IFNG
1539	{Hepatitis C virus susceptibility to} 609532	PTPRC

1540	{Herpes simplex encephalitic susceptibility to 6} 614850	TICAM1
1540	{Herpes simplex encephalitis susceptibility to 1} 610551	UNC93B1
1540	{Herpes simplex encephalitis susceptibility to 2} 613002	TLR3
1541	{Hirschsprung disease susceptibility to 1} 142623	RET
1541	{Hirschsprung disease susceptibility to 2} 600155	EDNRB
1541	{Hirschsprung disease susceptibility to 3} 613711	GDNF
1541	{Hirschsprung disease susceptibility to 4} 613712	EDN3
1542	{Hydrops fetalis nonimmune and or atrial septal defect} 617300	EPHB4
1543	{Hypercalciuria absorptive susceptibility to} 143870	ADCY10
1544	{Hypersensitivity syndrome carbamazepine-induced susceptibility to} 608579	HLA-A
1544	{Abacavir hypersensitivity, susceptibility to}	HLA-B
1544	{Stevens-Johnson syndrome susceptibility to} 608579	HLA-B
1544	{Toxic epidermal necrolysis susceptibility to} 608579	HLA-B
1545	{Hypothalamic hamartomas somatic} 241800	GLI3
1546	{Influenza severe susceptibility to} 614680	IFITM3
1547	{Intracranial hemorrhage in brain cerebrovascular malformations susceptibility to} 108010	IL6
1548	{Kaposi sarcoma susceptibility to} 148000	IL6
1549	{Kawasaki disease susceptibility to} 611775	ITPKC
1550	{Kuru susceptibility to} 245300	PRNP
1551	{Legionnaire disease susceptibility to} 608556	TLR5
1552	{Leprosy protection against} 613223	TLR1
1552	{Leprosy susceptibility to 4} 610988	LTA
1552	{Leprosy susceptibility to 5} 613223	TLR1
1552	{Leprosy susceptibility to} 246300	TLR2
1552	{Leprosy susceptibility to} 607572	PRKN
1553	{Lumbar disc degeneration} 603932	ASPN
1553	{Lumbar disc disease susceptibility to} 603932	CILP
1553	{Lumbar disc herniation susceptibility to} 603932	COL11A1
1553	{Lumbar disc herniation susceptibility to} 603932	THBS2
1553	{Intervertebral disc disease susceptibility to} 603932	COL9A3
1554	{Major affective disorder-7 susceptibility to} 612371	XBP1
1554	{Seasonal affective disorder susceptibility to} 608516	HTR2A
1554	{Major depressive disorder and accelerated response to antidepressant drug treatment} 608516	FKBP5
1554	{Major depressive disorder response to citalopram therapy in} 608516	HTR2A
1554	{Unipolar depression susceptibility to} 608516	TPH2
1555	{Malaria cerebral reduced risk of} 611162	CD36
1555	{Malaria cerebral susceptibility to} 611162	CD36
1555	{Malaria cerebral susceptibility to} 611162	ICAM1
1555	{Malaria cerebral susceptibility to} 611162	TNF
1555	{Malaria mild susceptibility to} 609148	NCR3
1555	{Malaria protection against} 611162	TIRAP

1555	{Malaria resistance to} 611162	FCGR2B
1555	{Malaria resistance to} 611162	GYPA
1555	{Malaria resistance to} 611162	GYPB
1555	{Malaria resistance to} 611162	GYPC
1555	{Malaria resistance to} 611162	HBB
1555	{Malaria resistance to} 611162	NOS2
1555	{Malaria severe resistance to} 611162	CR1
1555	{Malaria severe susceptibility to} 611162	FCGR2A
1555	{Malaria susceptibility to} 611162	CISH
1555	{Malaria vivax protection against} 611162	ACKR1
1555	{Resistance to malaria due to G6PD deficiency} 611162	G6PD
1556	{Melioidosis susceptibility to} 615557	TLR5
1557	{Microvascular complications of diabetes 1} 603933	VEGFA
1557	{Microvascular complications of diabetes 2} 612623	EPO
1557	{Microvascular complications of diabetes 3} 612624	ACE
1557	{Microvascular complications of diabetes 4} 612628	IL1RN
1557	{Microvascular complications of diabetes 5} 612633	PON1
1557	{Microvascular complications of diabetes 6} 612634	SOD2
1557	{Microvascular complications of diabetes 7} 612635	HFE
1558	{Multiple myeloma resistance to} 254500	LIG4
1558	{Multiple myeloma susceptibility to} 254500	CCND1
1559	{Multiple sclerosis disease progression modifier of} 126200	PDCD1
1559	{Multiple sclerosis susceptibility to 1} 126200	HLA-DQB1
1559	{Multiple sclerosis susceptibility to 1} 126200	HLA-DRB1
1559	{Multiple sclerosis susceptibility to 5} 614810	TNFRSF1A
1560	{Multiple self-healing squamous epithelioma susceptibility to} 132800	TGFBR1
1561	{Multiple system atrophy susceptibility to} 146500	COQ2
1562	{Mycobacterium tuberculosis protection against} 607948	IRGM
1562	{Mycobacterium tuberculosis protection against} 607948	MC3R
1562	{Mycobacterium tuberculosis susceptibility to infection by} 607948	SLC11A1
1562	{Mycobacterium tuberculosis susceptibility to} 607948	CCL2
1562	{Mycobacterium tuberculosis susceptibility to} 607948	CD209
1562	{Mycobacterium tuberculosis susceptibility to} 607948	SP110
1562	{Mycobacterium tuberculosis susceptibility to} 607948	TLR2
1562	{Tuberculosis infection protection against} 607948	IFNGR1
1562	{Tuberculosis protection against} 607948	IFNG
1562	{Tuberculosis protection against} 607948	TIRAP
1562	{Tuberculosis susceptibility to} 607948	CISH
1562	{Tuberculosis susceptibility to} 607948	IFNGR1
1563	{Myeloproliferative or lymphoproliferative neoplasms familial (multiple types) susceptibility to} 616871	DDX41
1564	{Myocardial infarction decreased susceptibility to} 608446	F7
1564	{Myocardial infarction protection against} 608446	F13A1

1564	{Myocardial infarction susceptibility to} 608446	ESR1
1564	{Myocardial infarction susceptibility to} 608446	GCLC
1564	{Myocardial infarction susceptibility to} 608446	GCLM
1564	{Myocardial infarction susceptibility to} 608446	ITGB3
1564	{Myocardial infarction susceptibility to} 608446	LGALS2
1564	{Myocardial infarction susceptibility to} 608446	LRP8
1564	{Myocardial infarction susceptibility to} 608446	LTA
1564	{Myocardial infarction susceptibility to} 608446	MIAT
1564	{Myocardial infarction susceptibility to} 608446	OLR1
1564	{Myocardial infarction susceptibility to} 608446	PSMA6
1564	{Myocardial infarction susceptibility to} 608446	TNFSF4
1564	{Myocardial infarction, susceptibility to}	ACE
1565	{Nicotine addiction protection from} 188890	CYP2A6
1565	{Nicotine addiction susceptibility to} 188890	CHRNA4
1565	{Nicotine dependence protection against} 188890	GABBR2
1565	{Nicotine dependence protection against} 188890	SLC6A3
1565	{Nicotine dependence susceptibility to} 188890	GABBR2
1565	{Nicotine dependence susceptibility to} 612052	CHRNA5
1565	{Lung cancer susceptibility 2} 612052	CHRNA3
1565	{Lung cancer susceptibility 2} 612052	CHRNA5
1566	{Nonarteritic anterior ischemic optic neuropathy susceptibility to} 258660	GP1BA
1567	{Obsessive-compulsive disorder protection against} 164230	BDNF
1567	{Obsessive-compulsive disorder susceptibility to} 164230	HTR2A
1567	{Obsessive-compulsive disorder} 164230	SLC6A4
1568	{Osteofibrous dysplasia susceptibility to} 607278	MET
1569	{Paget disease of bone 2 early-onset} 602080	TNFRSF11A
1570	{Panic disorder susceptibility to} 167870	COMT
1571	{Preterm premature rupture of the membranes susceptibility to} 610504	SERPINH1
1572	{Pseudofolliculitis barbae susceptibility to} 612318	KRT75
1573	{Psoriatic arthritis susceptibility to} 607507	LTA
1574	{Rheumatoid arthritis progression of} 180300	IL10
1574	{Rheumatoid arthritis susceptibility to} 180300	CD244
1574	{Rheumatoid arthritis susceptibility to} 180300	CIITA
1574	{Rheumatoid arthritis susceptibility to} 180300	NFKBIL1
1574	{Rheumatoid arthritis susceptibility to} 180300	PADI4
1574	{Rheumatoid arthritis susceptibility to} 180300	PTPN22
1574	{Rheumatoid arthritis susceptibility to} 180300	SLC22A4
1574	{Rheumatoid arthritis systemic juvenile susceptibility to} 604302	MIF
1574	{Rheumatoid arthritis systemic juvenile} 604302	IL6
1575	{Sarcoidosis susceptibility to 1} 181000	HLA-DRB1
1575	{Sarcoidosis susceptibility to 2} 612387	BTNL2
1576	{Schizophrenia 15} 613950	SHANK3
1576	{Schizophrenia 19 susceptibility to} 617629	RBM12

1576	{Schizophrenia 9 susceptibility to} 604906	DISC1
1576	{Schizophrenia susceptibility to 17} 614332	NRXN1
1576	{Schizophrenia susceptibility to 4} 600850	PRODH
1576	{Schizophrenia susceptibility to} 181500	CHI3L1
1576	{Schizophrenia susceptibility to} 181500	COMT
1576	{Schizophrenia susceptibility to} 181500	DRD3
1576	{Schizophrenia susceptibility to} 181500	HTR2A
1576	{Schizophrenia susceptibility to} 181500	MTHFR
1576	{Schizophrenia susceptibility to} 181500	RTN4R
1576	{Schizophrenia susceptibility to} 181500	SYN2
1577	{Specific language impairment 5} 615432	TM4SF20
1578	{Spondyloarthropathy susceptibility to 1} 106300	HLA-B
1579	{Thyrotoxic periodic paralysis susceptibility to 1} 188580	CACNA1S
1579	{Thyrotoxic periodic paralysis susceptibility to 2} 613239	KCNJ18
1580	{UV-induced skin damage} 266300	MC1R
1581	{Vitamin B12 plasma level QTL1} 612542	FUT2
1582	{Vitiligo-associated multiple autoimmune disease susceptibility 1} 606579	NLRP1
1583	{West nile virus susceptibility to} 610379	CCR5
1584	{Yao syndrome} 617321	NOD2
1585	Adrenal adenoma somatic	MEN1
1586	Aldosterone to renin ratio raised	CYP11B2
1587	Alpha-1-antichymotrypsin deficiency	SERPINA3
1588	Angiofibroma somatic	MEN1
1589	Apnea postanesthetic	BCHE
1590	Autonomic nervous system dysfunction	DRD4
1591	Beta-2-adrenoreceptor agonist reduced response to	ADRB2
1592	Carboxylesterase 1 deficiency	CES1
1593	Carcinoid tumor of lung	MEN1
1594	Cerebrovascular disease occlusive	SERPINA3
1595	Cirrhosis due to liver phosphorylase kinase deficiency	PHKG2
1596	DNA ligase I deficiency	LIG1
1597	DNA topoisomerase I camptothecin-resistant	TOP1
1597	DNA topoisomerase II resistance to inhibition of by amsacrine	TOP2A
1598	Dopamine receptor D2, reduced brain density of	ANKK1
1599	Erythremias alpha	HBA1
1599	Erythremias beta	HBB
1600	Forebrain defects	TDGF1
1601	Hemangioblastoma cerebellar somatic	VHL
1602	Hematuria, familial benign	COL4A4
1603	Homocysteine total plasma elevated	CTH
1604	Hypercalciuric hypercalcemia	CASR
1605	Hypochromic microcytic anemia	HBA2
1606	IgG2 deficiency, selective	IGHG2
1607	Left-right axis malformations	EBAF

1608	Merkel cell carcinoma, somatic	SDHD
1609	Mucoepidermoid salivary gland carcinoma	CRTC1
1609	Mucoepidermoid salivary gland carcinoma	MAML2
1610	Myelodysplasia syndrome-1	MECOM
1610	Myelodysplastic syndrome	ACSL6
1610	Myelodysplastic syndrome, preleukemic	IRF1
1611	Myelokathexis, isolated	CXCR4
1612	Neurofibrosarcoma	MXI1
1613	Orolaryngeal cancer, multiple	CDKN2A
1614	PTEN hamartoma tumor syndrome	PTEN
1615	Paroxysmal extreme pain disorder 167400	SCN9A
1616	Phospholipase A2, group IV A, deficiency of	PLA2G4A
1617	Rh-mod syndrome	RHAG
1618	Rh-null disease, amorph type	RHCE
1619	Rhabdomyolysis, cerivastatin-induced	CYP2C8
1620	Scaphocephaly and Axenfeld-Rieger anomaly	FGFR2
1621	Scurvy	GULOP
1622	Sebaceous tumors, somatic	LEF1
1623	{Congestive heart failure and beta-blocker response, modifier of}	ADRA2C
1623	{Congestive heart failure and beta-blocker response, modifier of}	ADRB1
1624	{Cancer progression/metastasis}	FGFR4
1625	{Atherosclerosis, susceptibility to}	ALOX5
1625	{Atherosclerosis, susceptibility to}	ESR1
1626	Somatostatin analog, resistance to	SSTR5
1627	Sweat chloride elevation without CF	CFTR
1628	Thyrotropin-releasing hormone resistance, generalized	TRHR
1629	{Drug-induced liver injury due to flucloxacillin}	HLA-B
1630	{High density lipoprotein cholesterol level QTL 7}	EDN1
1631	Wegener-like granulomatosis	TAP2
1632	Tolbutamide poor metabolizer	CYP2C9
1633	{Aerodigestive tract cancer, squamous cell, alcohol-related, protection against}	ADH1B
1634	{Benzene toxicity, susceptibility to}	NQO1
1635	{Beryllium disease, chronic, susceptibility to}	HLA-DPB1
1636	{SARS, progression of}	ACE
1637	{Norwalk virus infection, resistance to}	FUT2
1638	{Calcium, serum level of}	CASR
1639	{Hyperapobetalipoproteinemia, susceptibility to}	PPARA
1640	{Hypertrypsinemia, neonatal}	CFTR
1641	{Leanness, inherited}	AGRP
1642	{Low renin hypertension, susceptibility to}	CYP11B2
1643	{Memory impairment, susceptibility to}	BDNF
1644	{Organophosphate poisoning, sensitivity to}	PON1
1645	{Placental abruption}	NOS3
1646	{Sepsis, susceptibility to}	CASP12

1646	{Septic shock, susceptibility to}	TNF
1647	{Sezary syndrome, somatic}	BCL10
1648	{Spermatogenic failure, susceptibility to}	DAZL
1649	{Sublingual nitroglycerin, susceptibility to poor response to}	ALDH2
1650	{Synovitis, chronic, susceptibility to}	HLA-B
1651	{Transcription of plasminogen activator inhibitor, modulator of}	SERPINE1
1652	{Vascular disease, susceptibility to}	MTHFR
1653	{Venoocclusive disease after bone marrow transplantation}	CPS1
1654	{Viral infection, susceptibility to}	OAS1