Table 1: PrenatalScreen® - List of analysed genes and examined genetic diseases

	DISEASE	PhenoMIM	GENE
1	17-alpha-hydroxylase/17,20-lyase deficiency	202110	CYP17A1
2	17-beta-hydroxysteroid dehydrogenase X deficiency	300438	HSD17B10
3	3-beta-hydroxysteroid dehydrogenase, type II, deficiency	201810	HSD3B2
4	3-hydroxy-3-methylglutaric aciduria	246450	HMGCL
5	3-methylglutaconic aciduria type 1	250950	AUH
6	3-methylglutaconic aciduria type 3	258501	OPA3
7	46XY sex reversal 3	612965	NR5A1
8	4-hydroxybutyric aciduria	271980	ALDH5A1
9	Aarskog-Scott syndrome	305400	FGD1
10	ABCD syndrome	600501	EDNRB
11	Acampomelic campomelic dysplasia	114290	SOX9
12	Achalasia-addisonianism-alacrimia syndrome	231550	AAAS
13	Achondrogenesis type 1B	600972	SLC26A2
14	Achondrogenesis, type IA	200600	TRIP11
15	Achondrogenesis, type II or hypochondrogenesis	200610	COL2A1
16	Achondroplasia	100800	FGFR3
17	Acromicric dysplasia	102370	FBN1
18	Acyl-CoA dehydrogenase 9 deficiency	611126	ACAD9
19	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	202010	CYP11B1
20	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	613743	CYP11A1
21	Adrenocortical insufficiency	612965	NR5A1
22	Adrenoleukodystrophy	300100	ABCD1
23	Adult neuronal ceroid lipofuscinosis	256730	PPT1
24	Adult neuronal ceroid lipofuscinosis 10	610127	CTSD
25	Adult neuronal ceroid lipofuscinosis 4A	204300	CLN6
26	Aicardi-Goutières syndrome	225750	TREX1
27	Aicardi-Goutieres syndrome 2	610181	RNASEH2B
28	Aicardi-Goutieres syndrome 3	610329	RNASEH2C
29	Aicardi-Goutieres syndrome 4	610333	RNASEH2A
30	Aicardi-Goutieres syndrome 5	612952	SAMHD1
31	Aicardi-Goutieres syndrome 6	615010	ADAR
32	Aldosteronism, glucocorticoid-remediable	103900	CYP11B1
33	Allan-Herndon-Dudley syndrome	300523	SLC16A2
34	Alpers syndrome	203700	POLG
35	Alpha-methylacyl-Coa Racemase deficiency	614307	AMACR
36	Alpha-thalassemia	604131	HBA1- HBA2

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37	Alpha-thalassemia myelodysplasia syndrome, somatic	300448	ATRX
38	Alpha-thalassemia/mental retardation syndrome	301040	ATRX
39	Alport syndrome X-Linked	301050	COL4A5
40	Alport syndrome autosomal recessive	203780	COL4A3 - COL4A4
41	Alström syndrome	203800	ALMS1
42	Amish infantile epilepsy syndrome	609056	ST3GAL5
43	Amyotrophic lateral sclerosis 11	612577	FIG4
44	Amyotrophic lateral sclerosis 2, juvenile	205100	ALS2
45	Anauxetic dysplasia	607095	RMRP
46	Andersen syndrome	170390	KCNJ2
47	Angelman syndrome	105830	UBE3A
48	Antenatal Bartter syndrome	241200	KCNJ1
49	Antenatal Bartter syndrome type 1	601678	SLC12A1
50	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	201750	POR
51	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis	207410	FGFR2
52	Apert syndrome	101200	FGFR2
53	Aplasia/hypoplasia of limbs and pelvis	276820	WNT7A
54	Aplastic anemia	609135	NBN
55	Apparent mineralocorticoid excess	218030	HSD11B2
56	Argininosuccinic aciduria	207900	ASL
57	Aromatic L-amino acid decarboxylase deficiency	608643	DDC
58	Arrhythmogenic right ventricular dysplasia 2	600996	RYR2
59	Arthrogryposis - renal dysfunction - cholestasis	208085	VPS33B
60	Arthrogryposis multiplex congenita, distal, type 1	108120	TPM2
61	Arthrogryposis multiplex congenita, distal, type 2B	601680	TNNI2
62	Arthrogryposis, distal, type 2B	601680	TPM2
63	Arthrogryposis, renal dysfunction, and cholestasis 2	613404	VIPAR
64	Asperger syndrome susceptibility, X-linked 1	300494	NLGN3
65	Ataxia - oculomotor apraxia type 1	208920	APTX
66	Ataxia with vitamin E deficiency	277460	TTPA
67	Ataxia-telangiectasia	208900	ATM
68	Atelosteogenesis type II	256050	SLC26A2
69	Atrial fibrillation, familial, 16	613120	SCN3B
70	Atrial fibrillation, familial, 17	611819	SCN4B
71	Atrial fibrillation, familial, 3	607554	KCNQ1
72	Atrial fibrillation, familial, 4	611493	KCNE2
73	Atrial fibrillation, familial, 9	613980	KCNJ2
74	Autism, susceptibility to, X-linked 5	300847	RPL10

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75	Autoimmune lymphoproliferative syndrome type IV	614470	NRAS
76	Autoimmune lymphoproliferative syndrome, type IA	601859	FAS
77	Autoimmune lymphoproliferative syndrome, type IB	601859	FASLG
78	Autoimmune lymphoproliferative syndrome, type II	603909	CASP10
79	Autoimmune polyendocrinopathy syndrome , type I	240300	AIRE
80	Autosomal dominant Charcot-Marie-Tooth disease type 2K	607831	GDAP1
81	Autosomal recessive ataxia due to ubiquinone deficiency	612016	ADCK3
82	Autosomal recessive Charcot-Marie-Tooth disease with hoarseness	607706	GDAP1
83	Autosomal recessive distal spinal muscular atrophy type 4	611067	PLEKHG5
84	Autosomal recessive dopa-responsive dystonia	605407	TH
85	Autosomal recessive hypophosphatemic rickets 1	241520	DMP1
86	Autosomal recessive hypophosphatemic rickets 2	613312	ENPP1
87	Autosomal recessive intermediate Charcot-Marie-Tooth disease type A	608340	GDAP1
88	Autosomal recessive limb-girdle muscular dystrophy type 2I	607155	FKRP
89	Autosomal recessive limb-girdle muscular dystrophy type C	609308 - 613157 - 613158	POMT1 - POMGNT1 - POMT2
90	Autosomal recessive malignant osteopetrosis 1	259700	TCIRG1
91	Autosomal recessive malignant osteopetrosis 4	611490	CLCN7
92	Autosomal recessive nonsyndromic sensorineural deafness type DFNB12	601386	CDH23
93	Autosomal recessive nonsyndromic sensorineural deafness type DFNB18	602092	USH1C
94	Autosomal recessive nonsyndromic sensorineural deafness type DFNB1A	220290	GJB2
95	Autosomal recessive nonsyndromic sensorineural deafness type DFNB2	600060	MYO7A
96	Autosomal recessive polycystic kidney disease	263200	PKHD1
97	Autosomal recessive progressive external ophthalmoplegia	258450	POLG
98	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	270550	SACS
99	Autosomal recessive spondylocostal dysostosis 1	277300	DLL3
100	Avascular necrosis of the femoral head	608805	COL2A1
101	Bannayan-Riley-Ruvalcaba syndrome	153480	PTEN
102	Bardet-Biedl syndrome 11	615988	TRIM32
103	Basal cell carcinoma, somatic	605462	PTCH1
104	Basal cell nevus syndrome	109400	PTCH1
105	Beare-Stevenson cutis gyrata syndrome	123790	FGFR2
106	Becker muscular dystrophy	300376	DMD
107	Beckwith-Wiedemann syndrome	130650	CDKN1C - H19 - NSD1
108	Bent bone dysplasia syndrome	614592	FGFR2
109	Beta-thalassemia	613985	HBB
110	Bethlem myopathy	158810	COL6A1 - COL6A2 - COL6A3

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111	Bifunctional enzyme deficiency	261515	HSD17B4
112	Biotinidase deficiency	253260	BTD
113	Björnstad syndrome	262000	BCS1L
114	Bloom syndrome	210900	BLM
115	Borjeson-Forssman-Lehmann syndrome	301900	PHF6
116	Brachyolmia type 3	113500	TRPV4
117	Brachytelephalangic chondrodysplasia punctata	302950	ARSE
118	Brittle cornea syndrome	229200	ZNF469
119	Brugada syndrome 2	611777	GPD1L
120	Brugada syndrome 7	613120	SCN3B
121	Brunner syndrome	300615	MAOA
122	Buschke-Ollendorff syndrome	166700	LEMD3
123	Caffey disease	114000	COL1A1
124	Campomelic dysplasia with autosomal sex reversal	114290	SOX9
125	Canavan disease	271900	ASPA
126	CAP myopathy 1	609284	TPM3
127	CAP myopathy 2	609285	TPM2
128	Carbamoylphosphate synthetase deficiency	237300	CPS1
129	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	604377	SCO2
130	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2	615119	COX15
131	Cardiofaciocutaneous syndrome	115150	BRAF
132	Cardiofaciocutaneous syndrome 2	615278	KRAS
133	Cardiofaciocutaneous syndrome 3	615279	MAP2K1
134	Cardiofaciocutaneous syndrome 4	615280	MAP2K2
135	Cardiomyopathy, familial hypertrophic	192600	CAV3
136	Carnitine deficiency, systemic primary	212140	SLC22A5
137	Carnitine palmitoyl transferase 1A deficiency	255120	CPT1A
138	Carnitine palmitoyl transferase II deficiency, infantile form	600649	CPT2
139	Carnitine palmitoyl transferase II deficiency, neonatal form	608836	CPT2
140	Carnitine-acylcarnitine translocase deficiency	212138	SLC25A20
141	Carpenter syndrome	201000	RAB23
142	Cartilage-hair hypoplasia	250250	RMRP
143	Cataract - intellectual deficit - hypogonadism	212720	RAB3GAP2
144	Cataract 40, X-linked	302200	NHS
145	CATSHL syndrome	610474	FGFR3
146	Central core disease	117000	RYR1
147	Central hypoventilation syndrome, congenital	209880	RET
148	Cerebellar ataxia - intellectual deficit - dysequilibrium syndrome	224050	VLDLR

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149	Cerebral dysgenesis-neuropathy-ichthyosis-palmoplantar keratoderma syndrome	609528	SNAP29
150	Cerebrotendinous xanthomatosis	213700	CYP27A1
151	Charcot-Marie-Tooth disease axonal type 2B1	605588	LMNA
152	Charcot-Marie-Tooth disease type 4A	214400	GDAP1
153	Charcot-Marie-Tooth disease type 4E	605253	EGR2
154	Charcot-Marie-Tooth disease type 4F	614895	PRX
155	Charcot-Marie-Tooth disease type 4H	609311	FGD4
156	Charcot-Marie-Tooth disease, axonal, type 2M	606482	DNM2
157	Charcot-Marie-Tooth disease, dominant intermediate B	606482	DNM2
158	Charcot-Marie-Tooth disease, type 1A	118220	PMP22
159	Charcot-Marie-Tooth disease, type 1B	118200	MPZ
160	Charcot-Marie-Tooth disease, type 1C	601098	LITAF
161	Charcot-Marie-Tooth disease, type 1E	118300	PMP22
162	Charcot-Marie-Tooth disease, type 1F	607734	NEFL
163	Charcot-Marie-Tooth disease, type 2A1	118210	KIF1B
164	Charcot-Marie-Tooth disease, type 2A2	609260	MFN2
165	Charcot-Marie-Tooth disease, type 2B	600882	RAB7A
166	Charcot-Marie-Tooth disease, type 2D	601472	GARS
167	Charcot-Marie-Tooth disease, type 2E	607684	NEFL
168	Charcot-Marie-Tooth disease, type 2I	607677	MPZ
169	Charcot-Marie-Tooth disease, type 2J	607736	MPZ
170	Charcot-Marie-Tooth disease, type 4B1	601382	MTMR2
171	Charcot-Marie-Tooth disease, type 4B2	604563	SBF2
172	Charcot-Marie-Tooth disease, type 4C	601596	SH3TC2
173	Charcot-Marie-Tooth disease, type 4D	601455	NDRG1
174	Charcot-Marie-Tooth disease, type 4J	611228	FIG4
175	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1	302800	GJB1
176	CHARGE syndrome	214800	CHD7
177	Chediak-Higashi syndrome	214500	LYST
178	Chilblain lupus 2	614415	SAMHD1
179	CHILD syndrome	308050	NSDHL
180	Childhood-onset hypophosphatasia	241510	ALPL
181	Cholestasis, benign recurrent intrahepatic	243300	ATP8B1
182	Cholestasis, benign recurrent intrahepatic, 2	605479	ABCB11
183	Cholestasis, intrahepatic, of pregnancy, 1	147480	ATP8B1
184	Cholestasis, intrahepatic, of pregnancy, 3	614972	ABCB4
185	Cholestasis, progressive familial intrahepatic 1	211600	ATP8B1
186	Cholestasis, progressive familial intrahepatic 2	601847	ABCB11
187	Cholestasis, progressive familial intrahepatic 3	602347	ABCB4

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188	Chondrodysplasia, Blomstrand type	215045	PTH1R
189	Ciliary dyskinesia, primary, 1	244400	DNAI1
190	Ciliary dyskinesia, primary, 3	608644	DNAH5
191	Citrullinemia type I	215700	ASS1
192	CK syndrome	300831	NSDHL
193	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	201910	CYP21A2
194	Classic galactosemia	230400	GALT
195	Classic maple syrup urine disease	248600	DBT
196	Classical homocystinuria	236200	CBS
197	COACH syndrome	216360	TMEM67
198	Cockayne syndrome type A	216400	ERCC8
199	Cockayne syndrome type B	133540	ERCC6
200	Coenzyme Q10 deficiency, primary, 5	614654	COQ9
201	Coffin-Lowry syndrome	303600	RPS6KA3
202	COFS syndrome 1	214150	ERCC6
203	Cohen Syndrome type 1	216550	VPS13B
204	Combined immunodeficiency with skin granulomas	233650	RAG1 - RAG2
205	Combined oxidative phosphorylation defect type 2	610498	MRPS16
206	Combined oxidative phosphorylation defect type 5	611719	MRPS22
207	Combined oxidative phosphorylation deficiency 4	610678	TUFM
208	Combined pituitary hormone deficiencies, genetic forms	182230 - 262600 - 613038	HESX1 - PROP1 - POU1F1
209	Combined pituitary hormone deficiency with spine abnormalities	221750	LHX3
210	Complete androgen insensitivity syndrome	300068	AR
211	Complex I, mitochondrial respiratory chain, deficiency of	252010	NDUFS6
212	Congenital bile acid synthesis defect type 4	214950	AMACR
213	Congenital disorder of glycosylation type 1a	212065	PMM2
214	Congenital disorder of glycosylation type 1b	602579	MPI
215	Congenital disorder of glycosylation type 1e	608799	DPM1
216	Congenital disorder of glycosylation type 1j	608093	DPAGT1
217	Congenital disorder of glycosylation type 2a	212066	MGAT2
218	Congenital disorder of glycosylation type 2c	266265	SLC35C1
219	Congenital disorder of glycosylation type 2d	607091	B4GALT1
220	Congenital disorder of glycosylation type 2f	603585	SLC35A1
221	Congenital disorder of glycosylation type Ic	603147	ALG6
222	Congenital disorder of glycosylation type Ik	608540	ALG1
223	Congenital disorder of glycosylation, type Id	601110	ALG3
224	Congenital disorder of glycosylation, type If	609180	MPDU1
225	Congenital disorder of glycosylation, type Ig	607143	ALG12

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226	Congenital disorder of glycosylation, type Ih	608104	ALG8
227	Congenital disorder of glycosylation, type Ii	607906	ALG2
228	Congenital disorder of glycosylation, type IIb	606056	MOGS
229	Congenital disorder of glycosylation, type IIe	608779	COG7
230	Congenital disorder of glycosylation, type IIg	611209	COG1
231	Congenital disorder of glycosylation, type IIh	611182	COG8
232	Congenital disorder of glycosylation, type Il	608776	ALG9
233	Congenital disorder of glycosylation, type Im	610768	DOLK
234	Congenital disorder of glycosylation, type In	612015	RFT1
235	Congenital disorder of glycosylation, type Iq	612379	SRD5A3
236	Congenital fibrinogen deficiency	202400	FGA
237	Congenital heart defects, nonsyndromic, 1, X-linked	306955	ZIC3
238	Congenital hereditary endothelial dystrophy type II	217700	SLC4A11
239	Congenital lipoid adrenal hyperplasia	201710	STAR
240	Congenital malabsorptive diarrhea due to paucity of enteroendocrine cells	610370	NEUROG3
241	Congenital muscular dystrophy type 1A	607855	LAMA2
242	Congenital muscular dystrophy type 1D	608840	LARGE
243	Congenital muscular dystrophy type 5B	606612	FKRP
244	Congenital muscular dystrophy with cerebellar involvement	613151 - 613155 - 613156	POMGNT1 - POMT1 - POMT2
245	Congenital myopathy with excess of muscle spindles	218040	HRAS
246	Corneal dystrophy - perceptive deafness	217400	SLC4A11
247	Cornelia de Lange syndrome 1	122470	NIPBL
248	Cornelia de Lange syndrome 2	300590	SMC1A
249	Cornelia de Lange syndrome 3	610759	SMC3
250	Corpus callosum agenesis - neuronopathy	218000	SLC12A6
251	Corpus callosum hypoplasia-retardation-adducted thumbs-spasticity-hydrocephalus syndrome	307000	L1CAM
252	Corpus callosum, agenesis of, with mental retardation	300472	IGBP1
253	Costello syndrome	218040	HRAS
254	Cowden syndrome 1	158350	PTEN
255	Craniofacial-deafness-hand syndrome	122880	PAX3
256	Craniofacial-skeletal-dermatologic dysplasia	101600	FGFR2
257	Craniofrontonasal dysplasia	304110	EFNB1
258	Craniosynostosis, type 1	123100	TWIST1
259	Creatine phosphokinase, elevated serum	123320	CAV3
260	Crouzon syndrome	123500	FGFR2
261	Crouzon syndrome with acanthosis nigricans	612247	FGFR3
262	Culler-Jones syndrome	615849	GLI2

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263	Cutis laxa, autosomal dominant 2	614434	FBLN5
264	Cutis laxa, autosomal recessive, type IA	219100	FBLN5
265	Cutis laxa, autosomal recessive, type IB	614437	EFEMP2
266	Cutis laxa, autosomal recessive, type IIA	219200	ATP6V0A2
267	Cystic fibrosis; mucoviscidosis	219700	CFTR
268	Cystinosis	219800	CTNS
269	Czech dysplasia	609162	COL2A1
270	Danon disease	300257	LAMP2
271	Deafness - encephaloneuropathy - obesity - valvulopathy	614651	PDSS1
272	Deafness, autosomal dominant 13	601868	COL11A2
273	Deafness, autosomal dominant 3B	612643	GJB6
274	Deafness, autosomal dominant type 1	124900	DIAPH1
275	Deafness, autosomal dominant type 10	601316	EYA4
276	Deafness, autosomal dominant type 12	601543	TECTA
277	Deafness, autosomal dominant type 15	602459	POU4F3
278	Deafness, autosomal dominant type 17	603622	MYH9
279	Deafness, autosomal dominant type 20	604717	ACTG1
280	Deafness, autosomal dominant type 22	606346	MYO6
281	Deafness, autosomal dominant type 23	605192	SIX1
282	Deafness, autosomal dominant type 25	605583	SLC17A8
283	Deafness, autosomal dominant type 28	608641	GRHL2
284	Deafness, autosomal dominant type 2A	600101	KCNQ4
285	Deafness, autosomal dominant type 2B	612644	GJB3
286	Deafness, autosomal dominant type 36	606705	TMC1
287	Deafness, autosomal dominant type 4	600652	MYH14
288	Deafness, autosomal dominant type 40	123740	CRYM
289	Deafness, autosomal dominant type 48	607841	MYO1A
290	Deafness, autosomal dominant type 5	600994	DFNA5
291	Deafness, autosomal dominant type 50	613074	MIR96
292	Deafness, autosomal dominant type 6	600965	WFS1
293	Deafness, autosomal dominant type 64	614152	DIABLO
294	Deafness, autosomal dominant type 9	601369	COCH
295	Deafness, autosomal recessive 1B	612645	GJB6
296	Deafness, autosomal recessive 53	609706	COL11A2
297	Deafness, digenic GJB2/GJB6	220290	GJB6
298	Dejerine-Sottas disease	145900	MPZ - PMP22
299	Dent disease	300009	CLCN5
300	Dent disease 2	300555	OCRL
301	Denys-Drash syndrome	194080	WT1

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CHIAMATA GRATUITA
NUMEROVERDE
800-501651

302	Dermatopathia pigmentosa reticularis	125595	KRT14
303	Desmosterolosis	602398	DHCR24
304	Diabetes mellitus, noninsulin-dependent	125853	ABCC8
305	Diabetes mellitus, permanent neonatal	606176	ABCC8
306	Diabetes mellitus, transient neonatal 2	610374	ABCC8
307	Diastrophic dwarfism	222600	SLC26A2
308	Digital arthropathy-brachydactyly, familial	606835	TRPV4
309	Dihydropyrimidine dehydrogenase deficiency	274270	DPYD
310	Dilated cardiomyopathy with ataxia	610198	DNAJC19
311	Donnai-Barrow syndrome	222448	LRP2
312	Dowling-Degos disease 1	179850	KRT5
313	Duchenne muscular dystrophy	310200	DMD
314	Dyschromatosis symmetrica hereditaria	127400	ADAR
315	Dyskeratosis congenita X-linked	305000	DKC1
316	Dystonia-1, torsion	128100	TOR1A
317	Dystonia-11, myoclonic	159900	SGCE
318	Dystrophic epidermolysis bullosa pruriginosa	604129	COL7A1
319	Early infantile epileptic encephalopathy	308350 - 609304	ARX - SLC25A22
320	Ectodermal dysplasia 1, hypohidrotic, X-linked	305100	EDA
321	Ectodermal dysplasia 2, Clouston type	129500	GJB6
322	Ectodermal dysplasia, hypohidrotic, with immune deficiency	300291	IKBKG
323	Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency	300301	IKBKG
324	Ectopia lentis, familial	129600	FBN1
325	Ehlers-Danlos syndrome type 6	225400	PLOD1
326	Ehlers-Danlos syndrome, cardiac valvular type	225320	COL1A2
327	Ehlers-Danlos syndrome, type I	130000	COL5A1 - COL5A2 - COL1A1
328	Ehlers-Danlos syndrome, type II	130010	COL5A1
329	Ehlers-Danlos syndrome, type III	130020	COL3A1
330	Ehlers-Danlos syndrome, type IV	130050	COL3A1
331	Ehlers-Danlos syndrome, type VIIA	130060	COL1A1
332	Eiken syndrome	600002	PTH1R
333	Elliptocytosis-1	611804	EPB41
334	Ellis-van Creveld syndrome	225500	EVC2
335	Emery-Dreifuss muscular dystrophy 1, X-linked	310300	EMD
336	Encephalopathy due to prosaposin deficiency	611721	PSAP
337	Encephalopathy, progressive, with or without lipodystrophy	615924	BSCL2
338	Epidermolysis bullosa simplex with muscular dystrophy	226670	PLEC

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339	Epidermolysis bullosa simplex with pyloric atresia	612138	PLEC
340	Epidermolysis bullosa simplex, Dowling-Meara type	131760	KRT14 - KRT5
341	Epidermolysis bullosa simplex, Koebner type	131900	KRT14 - KRT5
342	Epidermolysis bullosa simplex, recessive 1	601001	KRT14 - KRT5
343	Epidermolysis bullosa simplex, Weber-Cockayne type	131800	KRT14 - KRT5
344	Epidermolysis bullosa simplex-MP	131960	KRT5
345	Epidermylysis bullosa simplex-MCR	609352	KRT5
346	Epilepsy, familial temporal lobe, 1	600512	LGI1
347	Epilepsy, progressive myoclonic 2A (Lafora)	254780	EPM2A
348	Epilepsy, progressive myoclonic 2B (Lafora)	254780	NHLRC1
349	Epilepsy, pyridoxine-dependent	266100	ALDH7A1
350	Epilepsy, X-linked, with variable learning disabilities and behavior disorders	300491	SYN1
351	Epileptic encephalopathy, early infantile, 15	615006	ST3GAL3
352	Epileptic encephalopathy, early infantile, 2	300672	CDKL5
353	Epileptic encephalopathy, early infantile, 8	300607	ARHGEF9
354	Epileptic encephalopathy, early infantile, 9	300088	PCDH19
355	Epiphyseal dysplasia, multiple 1	132400	COMP
356	Epiphyseal dysplasia, multiple, with myopia and deafness	132450	COL2A1
357	Erythrocytosis, familial, 2	263400	VHL
358	Escobar syndrome	265000	CHRNG
359	Ethylmalonic encephalopathy	602473	ETHE1
360	Exostoses, multiple, type 1	133700	EXT1
361	Exostoses, multiple, type 2	133701	EXT2
362	Exudative vitreoretinopathy 2, X-linked	305390	NDP
363	Fabry disease	301500	GLA
364	Failure of tooth eruption, primary	125350	PTH1R
365	Familial dysautonomia	223900	IKBKAP
366	Familial hypomagnesemia - hypercalciuria - nephrocalcinosis - severe ocular	248190	CLDN19
367	Familial Mediterranean fever	249100	MEFV
368	Fanconi anemia complementation group C	227645	FANCC
369	Fanconi anemia, complementation group A	227650	FANCA
370	Fanconi anemia, complementation group B	300514	FANCB
371	Fanconi anemia, complementation group G	614082	FANCG
372	Fatal infantile lactic acidosis with methylmalonic aciduria	245400	SUCLG1
373	Fatal mitochondrial disease due to combined oxidative phosphorylation deficiency 3	610505	TSFM
374	Favism	134700	G6PD
375	Fertile eunuch syndrome	228300	GNRHR

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376	Foetal akinesia deformation sequence	208150	RAPSN DOK7
377	Foetal Gaucher disease	608013	GBA
378	FG syndrome 4	300422	CASK
379	Fibrochondrogenesis	228520	COL11A1
380	Fibrochondrogenesis 2	614524	COL11A2
381	Fibromatosis, gingival	135300	SOS1
382	Fibular hypoplasia or aplasia - femoral bowing - oligodactyly	228930	WNT7A
383	Focal cortical dysplasia, Taylor balloon cell type	607341	TSC1
384	Focal dermal hypoplasia	305600	PORCN
385	Fraser syndrome (gene FRAS1)	219000	FRAS1 - FREM2
386	Frasier syndrome	136680	WT1
387	Free sialic acid storage disease, infantile form	269920	SLC17A5
388	French-Canadian type Leigh syndrome	220111	LRPPRC
389	Fucosidosis	230000	FUCA1
390	Fumaric aciduria	606812	FH
391	Galactokinase deficiency with cataracts	230200	GALK1
392	Gallbladder disease 1	600803	ABCB4
393	Gaucher disease type 2	230900	GBA
394	Gaucher disease type 3	231000	GBA
395	Gaucher disease type 3C	231005	GBA
396	Geleophysic dysplasia 1	231050	ADAMTSL2
397	Geleophysic dysplasia 2	614185	FBN1
398	Generalized junctional epidermolysis bullosa, non-Herlitz type	226650	COL17A1
399	Glutaric acidemia type 2	231680	ETFA - ETFB - ETFDH
400	Glutaryl-CoA dehydrogenase deficiency	231670	GCDH
401	Glutathione synthetase deficiency with 5-oxoprolinuria	266130	GSS
402	Glycerol kinase deficiency	307030	GK
403	Glycine encephalopathy	605899	AMT - GCSH - GLDC
404	Glycogen storage disease due to acid maltase deficiency	232300	GAA
405	Glycogen storage disease due to glucose-6-phosphatase deficiency type 1a	232200	G6PC
406	Glycogen storage disease due to glucose-6-phosphatase deficiency type b	232220	SLC37A4
407	Glycogen storage disease due to glucose-6-phosphatase deficiency type c	232240	SLC37A4
408	Glycogen storage disease due to glycogen branching enzyme deficiency, childhood combined hepatic and myopathic form	232500	GBE1
409	Glycogen storage disease due to glycogen debranching enzyme deficiency	232400	AGL
410	Glycogen storage disease due to muscle glycogen phosphorylase deficiency	232600	PYGM
411	GM1 gangliosidosis type 1	230500	GLB1
412	GM1 gangliosidosis type 2	230600	GLB1

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413	GM1 gangliosidosis type 3	230650	GLB1
414	Gnathodiaphyseal dysplasia	166260	ANO5
415	GRACILE syndrome	603358	BCS1L
416	Greenberg dysplasia	215140	LBR
417	Griscelli disease type 1	214450	MYO5A
418	Griscelli disease type 2	607624	RAB27A
419	Guanidinoacetate methyltransferase deficiency	612736	GAMT
420	Heinz body anemia	140700	HBA2
421	Hemochromatosis	235200	HFE
422	Hemochromatosis, type 2A	602390	HFE2
423	Hemoglobin H disease, nondeletional	613978	HBA2
424	Hemolytic anemia due to G6PD deficiency	300908	G6PD
425	Hemolytic anemia due to red cell pyruvate kinase deficiency	266200	PKLR
426	Hemophagocytic lymphohistiocytosis, familial, 2	603553	PRF1
427	Hemophagocytic lymphohistiocytosis, familial, 3	608898	UNC13D
428	Hemophagocytic lymphohistiocytosis, familial, 4	603552	STX11
429	Hemophagocytic lymphohistiocytosis, familial, 5	613101	STXBP2
430	Hemophilia A	306700	F8
431	Hemophilia B	306900	F9
432	Hepatic venoocclusive disease with immunodeficiency	235550	SP110
433	Hepatoencephalopathy due to combined oxidative phosphorylation deficiency type 1	609060	GFM1
434	Hereditary fructose intolerance	229600	ALDOB
435	Hereditary motor and sensory neuropathy VI	601152	MFN2
436	Hereditary motor and sensory neuropathy, type IIc	606071	TRPV4
437	Hereditary sensory and autonomic neuropathy type 4	256800	NTRK1
438	Hermansky-Pudlak syndrome 2	608233	AP3B1
439	Hermansky-pudlak syndrome 9	614171	PLDN
440	Heterotaxy, visceral, 1, X-linked	306955	ZIC3
441	Heterotopia, periventricular	300049	FLNA
442	Histidinemia	235800	HAMP
443	Holocarboxylase synthetase deficiency	253270	HLCS
444	Holoprosencephaly	607502	DISP1
445	Holoprosencephaly-2	157170	SIX3
446	Holoprosencephaly-3	142945	SHH
447	Holoprosencephaly-5	609637	ZIC2
448	Holoprosencephaly-7	610828	PTCH1
449	Holoprosencephaly-9	610829	GLI2
450	Homocystinuria due to MTHFR deficienc	236250	MTHFR
451	Hoyeraal-Hreidarsson syndrome	300240	DKC1

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452	Hyaline fibromatosis syndrome	228600	ANTXR2
453	Hyperammonemia due to N-acetylglutamate synthetase deficiency	237310	NAGS
454	Hyper-IgE recurrent infection syndrome, autosomal recessive	243700	DOCK8
455	Hyperinsulinemic hypoglycemia, familial, 1	256450	ABCC8
456	Hyperornithinemia-hyperammonemia-homocitrullinuria	238970	SLC25A15
457	Hyperthyroidism, nonautoimmune	609152	TSHR
458	Hypochondroplasia	146000	FGFR3
459	Hypoglycemia of infancy, leucine-sensitive	240800	ABCC8
460	Hypogonadotropic hypogonadism 5	612370	CHD7
461	Hypogonadotropic hypogonadism 7 without anosmia	146110	GNRHR
462	Hypomyelination - congenital cataract	610532	FAM126A
463	Hypoparathyroidism - intellectual deficit - dysmorphism syndrome	241410	TBCE
464	Hypophosphatemic rickets	300554	CLCN5
465	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia	218700	PAX8
466	Hypothyroidism, congenital, nongoitrous, 1	275200	TSHR
467	Ichthyosis follicularis - alopecia - photophobia	308205	MBTPS2
468	Ichthyosis, autosomal recessive 4B (harlequin)	242500	ABCA12
469	Ichthyosis, congenital, autosomal recessive 1	242300	TGM1
470	Ichthyosis, congenital, autosomal recessive 4A	601277	ABCA12
471	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	607626	CLDN1
472	IMAGE syndrome	614732	CDKN1C
473	Immunodeficiency 10	612783	STIM1
474	Immunodeficiency 17, CD3 gamma deficient	615607	CD3G
475	Immunodeficiency 18, SCID variant	615615	CD3E
476	Immunodeficiency 19	615617	CD3D
477	Immunodeficiency 27A, mycobacteriosis, AR	209950	IFNGR1
478	Immunodeficiency 28, mycobacteriosis	614889	IFNGR2
479	Immunodeficiency 29, mycobacteriosis	614890	IL12B
480	Immunodeficiency 30	614891	IL12RB1
481	Immunodeficiency 31A, mycobacteriosis, autosomal dominant	614892	STAT1
482	Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive	613796	STAT1
483	Immunodeficiency 31C, autosomal dominant	614162	STAT1
484	Immunodeficiency 33	300636	IKBKG
485	Immunodeficiency 35	611521	TYK2
486	Immunodeficiency 9	612782	ORAI1
487	Immunodeficiency, common variable, 1	607594	ICOS
488	Immunodeficiency, common variable, 3	613493	CD19
489	Immunodeficiency, X-linked	300853	MAGT1
490	Immunodeficiency-centromeric instability-facial anomalies syndrome 1	242860	DNMT3B

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491	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	304790	FOXP3
492	Incontinentia pigmenti, type II	308300	IKBKG
493	Infantile bilateral striatal necrosis	271930	NUP62
494	Infantile hypophosphatasia	241500	ALPL
495	Infantile neuroaxonal dystrophy 2A	256600	PLA2G6
496	Infantile neuroaxonal dystrophy 2B	610217	PLA2G6
497	Infantile onset spinocerebellar ataxia	271245	C10orf2
498	Interleukin 1 receptor antagonist deficiency	612852	IL1RN
499	Isolated CoQ-cytochrome C reductase deficiency	124000	BCS1L
500	Isolated growth hormone deficiency type III	307200	BTK
501	Isolated thyroid-stimulating hormone deficiency	275100	TSHB
502	Isovaleric acidemia	243500	IVD
503	Jackson-Weiss syndrome	123150	FGFR2
504	Jervell and Lange-Nielsen syndrome	220400	KCNQ1
505	Jervell and Lange-Nielsen syndrome 2	612347	KCNE1
506	Jeune syndrome	611263	IFT80
507	Johanson-Blizzard syndrome	243800	UBR1
508	Joubert syndrome 4	609583	NPHP1
509	Joubert syndrome 6	610688	TMEM67
510	Joubert syndrome with hepatic defect	216360	RPGRIP1L
511	Joubert syndrome with ocular defect	608629	AHI1
512	Joubert syndrome with oculorenal defect 5	610188	CEP290
513	Junctional epidermolysis bullosa - pyloric atresia	226730	ITGA6 - ITGB4
514	Junctional epidermolysis bullosa, Herlitz type	226700	LAMA3- LAMB3 - LAMC2
515	Junctional epidermolysis bullosa, non-Herlitz type	226650	ITGB4 - LAMA3- LAMB3- LAMC2
516	Juvenile neuronal ceroid lipofuscinosis 3	204200	CLN3
517	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome	175050	SMAD4
518	Kabuki syndrome 1	147920	KMT2D (MLL2)
519	Kabuki syndrome 2	300867	KDM6A
520	Kahrizi syndrome	612713	SRD5A3
521	Kelley-Seegmiller syndrome	300323	HPRT1
522	Ketoacidosis due to beta-ketothiolase deficiency	203750	ACAT1
523	King-Denborough syndrome	145600	RYR1
524	Kniest dysplasia	156550	COL2A1
525	Krabbe disease	245200 -	GALC - PSAP

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		611722	
526	Lacticacidemia due to PDX1 deficiency	245349	PDHX
527	LADD syndrome	149730	FGFR2 - FGFR3
528	Late infantile neuronal ceroid lipofuscinosis	610951	MFSD8
529	Late infantile neuronal ceroid lipofuscinosis 5	256731	CLN5
530	Late infantile neuronal ceroid lipofuscinosis 6	601780	CLN6
531	Late infantile neuronal ceroid lipofuscinosis 8	600143	CLN8
532	Lathosterolosis	607330	SC5DL
533	Legg-Calve-Perthes disease	150600	COL2A1
534	Leigh syndrome	256000	BCS1L - DLD - NDUFAF2 - NDUFS4 - NDUFS7 - NDUFS3 - NDUFS8
535	Leigh syndrome due to cytochrome c oxidase deficiency	256000	COX15
536	Leigh syndrome due to mitochondrial COX4 deficiency	256000	COX10
537	Leigh syndrome with nephrotic syndrome	607426 - 614652	COQ2 - PDSS2
538	Leigh syndrome, due to COX deficiency	256000	SURF1
539	Leigh syndrome, X-linked	308930	PDHA1
540	LEOPARD syndrome 1	151100	PTPN11
541	LEOPARD syndrome 2	611554	RAF1
542	LEOPARD syndrome 3	613707	BRAF
543	Leprechaunism	246200	INSR
544	Lesch-Nyhan syndrome	300322	HPRT1
545	Lethal acantholytic epidermolysis bullosa	609638	DSP
546	Lethal ataxia with deafness and optic atrophy	301835	PRPS1
547	Lethal congenital contractural syndrome 2	607598	ERBB3
548	Lethal congenital contracture syndrome 5	615368	DNM2
549	Lethal congenital contracture syndrome type 1	253310	GLE1
550	Lethal osteosclerotic bone dysplasia	259775	FAM20C
551	Lethal restrictive dermopathy	275210	LMNA - ZMPSTE24
552	Leukemia, juvenile myelomonocytic	607785	PTPN11
553	Leukocyte adhesion deficiency, type III	612840	FERMT3
554	Leydig cell adenoma, somatic, with precocious puberty	176410	LHCGR
555	Leydig cell hypoplasia with hypergonadotropic hypogonadism	238320	LHCGR
556	Leydig cell hypoplasia with pseudohermaphroditism	238320	LHCGR
557	Lhermitte-Duclos syndrome	158350	PTEN
558	Limb girdle dystrophy with epidermolysis bullosa simplex	613723	PLEC

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559	Lipodystrophy, congenital generalized, type 2	269700	BSCL2
560	Lissencephaly 3	611603	TUBA1A
561	Lissencephaly syndrome, Norman-Roberts type	257320	RELN
562	Lissencephaly, X-linked	300067	DCX
563	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	609016	HADH
564	Long QT syndrome 1	192500	KCNQ1
565	Long QT syndrome 12	612955	SNTA1
566	Long QT syndrome 2	613688	KCNH2
567	Long QT syndrome 4	600919	ANK2
568	Long QT syndrome 5	613695	KCNE1
569	Long QT syndrome 6	613693	KCNE2
570	Long QT syndrome 9	611818	CAV3
571	Long QT syndrome-10	611819	SCN4B
572	Long QT syndrome-11	611820	AKAP9
573	Luteinizing hormone resistance, female	238320	LHCGR
574	Lymphangioleiomyomatosis	606690	TSC1
575	Lymphoproliferative syndrome, X-linked, 2	300635	XIAP
576	Macrocephaly/autism syndrome	605309	PTEN
577	Macroglobulinemia, Waldenstrom	153600	MYD88
578	Macular degeneration, age-related, 3	608895	FBLN5
579	Mandibuloacral dysplasia with type A lipodystrophy	248370	LMNA
580	Mandibuloacral dysplasia with type B lipodystrophy	608612	ZMPSTE24
581	Mannosidosis, alpha-, types I and II	248500	MAN2B1
582	Maple syrup urine disease	248600	DLD - BCKDHA - BCKDHB
583	Marfan syndrome	154700	FBN1
584	Marinesco-Sjögren syndrome	248800	SIL1
585	Marshall syndrome	154780	COL11A1
586	Masa syndrome	303350	L1CAM
587	MASS syndrome	604308	FBN1
588	Meacham syndrome	608978	WT1
589	Meckel syndrome type 1	249000	MKS1
590	Meckel syndrome, type 5	611561	RPGRIP1L
591	Medium chain acyl-CoA dehydrogenase deficiency	201450	ACADM
592	Medullary thyroid carcinoma	155240	RET
593	Megalencephalic leukoencephalopathy with subcortical cysts	604004	MLC1
594	Melorheostosis with osteopoikilosis	155950	LEMD3
595	Menkes disease	309400	ATP7A
596	Mental retardation and microcephaly and cerebellar hypoplasia	300749	CASK

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597	Mental retardation syndrome, X-linked, Siderius type	300263	PHF8
598	Mental retardation, autosomal dominant type 1	156200	MBD5
599	Mental retardation, autosomal dominant type 12	614562	ARID1B
600	Mental retardation, autosomal dominant type 14	614607	ARID1A
601	Mental retardation, autosomal dominant type 15	614608	SMARCB1
602	Mental retardation, autosomal dominant type 16	614609	SMARCA4
603	Mental retardation, autosomal dominant type 20	613443	MEF2C
604	Mental retardation, autosomal dominant type 5	612621	SYNGAP1
605	Mental retardation, autosomal dominant type 6	613970	GRIN2B
606	Mental retardation, autosomal dominant type 9	614255	KIF1A
607	Mental retardation, autosomal recessive 1	249500	PRSS12
608	Mental retardation, autosomal recessive 12	611090	ST3GAL3
609	Mental retardation, autosomal recessive 13	613192	TRAPPC9
610	Mental retardation, autosomal recessive 5	611091	NSUN2
611	Mental retardation, autosomal recessive 7	611093	TUSC3
612	Mental retardation, autosomal recessive, 6	611092	GRIK2
613	Mental retardation, with or without nystagmus	300422	CASK
614	Mental retardation, X-linked	300034	AGTR2
615	Mental retardation, X-linked	311040	ELK1
616	Mental retardation, X-linked	300495	NLGN4X
617	Mental retardation, X-linked 19	300844	RPS6KA3
618	Mental retardation, X-linked 21/34	300143	IL1RAPL1
619	Mental retardation, X-linked 30/47	300558	PAK3
620	Mental retardation, X-linked 41	300849	GDI1
621	Mental retardation, X-linked 45	300498	ZNF81
622	Mental retardation, X-linked 46	300436	ARHGEF6
623	Mental retardation, X-linked 58	300210	TSPAN7
624	Mental retardation, X-linked 63	300387	ACSL4
625	Mental retardation, X-linked 72	300271	RAB39B
626	Mental retardation, X-linked 9	309549	FTSJ1
627	Mental retardation, X-linked 91	300577	ZDHHC15
628	Mental retardation, X-linked 93	300659	BRWD3
629	Mental retardation, X-linked 94	300699	GRIA3
630	Mental retardation, X-linked 96	300802	SYP
631	Mental retardation, X-linked 97	300803	ZNF711
632	Mental retardation, X-linked 98	300912	KIAA2022
633	Mental retardation, X-linked syndromic 16	305400	FGD1
634	Mental retardation, X-linked syndromic 5	304340	AP1S2
635	Mental retardation, X-linked syndromic, Christianson type	300243	SLC9A6

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636	Mental retardation, X-linked syndromic, Nascimento-type	300860	UBE2A
637	Mental retardation, X-linked syndromic, Raymond type	300799	ZDHHC9
638	Mental retardation, X-linked syndromic, Turner type	300706	HUWE1
639	Mental retardation, X-linked, Snyder-Robinson type	309583	SMS
640	Mental retardation, X-linked, syndromic 14	300676	UPF3B
641	Mental retardation, X-linked, syndromic 15 (Cabezas type)	300354	CUL4B
642	Mental retardation, X-linked, syndromic, Claes-Jensen type	300534	KDM5C
643	Mental retardation, X-linked, syndromic, Hedera type	300423	ATP6AP2
644	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	300486	OPHN1
645	Mental retardation, X-linked, with isolated growth hormone deficiency	300123	SOX3
646	Mental retardation-hypotonic facies syndrome, X-linked	309580	ATRX
647	Mesothelioma, somatic	156240	WT1
648	Metachondromatosis	156250	PTPN11
649	Metachromatic leukodystrophy	249900 - 250100	PSAP - ARSA
650	Metaphyseal chondrodysplasia, Murk Jansen type	156400	PTH1R
651	Metaphyseal dysplasia without hypotrichosis	250460	RMRP
652	Metatropic dysplasia	156530	TRPV4
653	Methylmalonic acidemia with homocystinuria, type cblC	277400	MMACHC
654	Methylmalonic acidemia with homocystinuria, type cblD	277410	MMACHC
655	Mevalonic aciduria	610377	MVK
656	Micro syndrome	600118	RAB3GAP1
657	Microcephaly 5, primary, autosomal recessive	608716	ASPM
658	Microphthalmia with coloboma 5	611638	SHH
659	Microphthalmia, syndromic 2	300166	BCOR
660	Microphthalmia, syndromic 7	309801	HCCS
661	Minicore myopathy with external ophthalmoplegia	255320	RYR1
662	Mitochondrial complex I deficiency	252010	NDUFA1 - NDUFAF2 - NDUFAF4 - NDUFS3 - NDUFS4 - NDUFV1
663	Mitochondrial complex IV deficiency	220110 - 603644	COX10 - COX6B1 - FASTKD2 - SCO1
664	Mitochondrial DNA depletion syndrome 1 (MNGIE type)	603041	TYMP
665	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	612073	SUCLA2
666	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)	612075	RRM2B
667	Mitochondrial DNA depletion syndrome 8B (MNGIE type)	612075	RRM2B

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668	Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency 3	251880	DGUOK
669	Mitochondrial DNA depletion syndrome, myopathic form	609560	TK2
670	Mitochondrial neurogastrointestinal encephalomyopathy	613662	POLG
671	Mitochondrial respiratory chain complex III deficiency	124000	UQCRB - UQCRQ
672	Mitochondrial trifunctional protein deficiency	609015	HADHA - HADHB
673	Miyoshi muscular dystrophy 1	254130	DYSF
674	Miyoshi muscular dystrophy 3	613319	ANO5
675	Mohr-Tranebjaerg syndrome	304700	TIMM8A
676	Mononeuropathy of the median nerve, mild	613353	SH3TC2
677	Mowat-Wilson syndrome	235730	ZEB2
678	Mucolipidosis type 2	252500	GNPTAB
679	Mucolipidosis type 3	252600	GNPTAB
680	Mucolipidosis type 4	252650	MCOLN1
681	Mucopolysaccharidosis Ih	607014	IDUA
682	Mucopolysaccharidosis Ih/s	607015	IDUA
683	Mucopolysaccharidosis Is	607016	IDUA
684	Mucopolysaccharidosis type 2	309900	IDS
685	Mucopolysaccharidosis type 3A (Sanfilippo syndrome type A)	252900	SGSH
686	Mucopolysaccharidosis type 4B	253010	GLB1
687	Mucopolysaccharidosis type 6	253200	ARSB
688	Mucopolysaccharidosis type 7	253220	GUSB
689	Mucopolysaccharidosis type IIIB (Sanfilippo B)	252920	NAGLU
690	Muenke syndrome	602849	FGFR3
691	MULIBREY nanism	253250	TRIM37
692	Mullerian aplasia and hyperandrogenism	158330	WNT4
693	Multiple endocrine neoplasia 1	131100	MEN1
694	Multiple endocrine neoplasia IIA	171400	RET
695	Multiple endocrine neoplasia IIB	162300	RET
696	Multiple endocrine neoplasia, type IV	610755	CDKN1B
697	Multiple epiphyseal dysplasia type 4	226900	SLC26A2
698	Multiple pterygium syndrome, lethal type	253290	CHRNA1 - CHRND - CHRNG
699	Muscle-eye-brain disease	613153 - 613154	FKRP - LARGE
700	Muscular dystrophy, limb-girdle, type 1A	159000	MYOT
701	Muscular dystrophy, limb-girdle, type 1E	603511	DNAJB6
702	Muscular dystrophy, limb-girdle, type 2A	253600	CAPN3
703	Muscular dystrophy, limb-girdle, type 2B	253601	DYSF

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704	Muscular dystrophy, limb-girdle, type 2C	253700	SGCG
705	Muscular dystrophy, limb-girdle, type 2D	608099	SGCA
706	Muscular dystrophy, limb-girdle, type 2E	604286	SGCB
707	Muscular dystrophy, limb-girdle, type 2H	254110	TRIM32
708	Muscular dystrophy, limb-girdle, type 2L	611307	ANO5
709	Muscular dystrophy, limb-girdle, type IC	607801	CAV3
710	Myasthenia gravis, neonatal transient	100730	CHRNG
711	Myasthenia, limb-girdle, familial	254300	DOK7
712	Myasthenic syndrome, fast-channel congenital	608930	CHRNA1 - CHRND
713	Myasthenic syndrome, slow-channel congenital	601462	CHRNA1 - CHRNB1- CHRND
714	Myhre syndrome	139210	SMAD4
715	Myopathy due to myoadenylate deaminase deficiency	615511	AMPD1
716	Myopathy, actin, congenital	161800	ACTA1
717	Myopathy, centronuclear	160150	DNM2
718	Myopathy, congenital, with fiber-type disproportion	255310	TPM3
719	Myopathy, congenital, with fiber-type disproportion 1	255310	ACTA1
720	Myopathy, distal, Tateyama type	614321	CAV3
721	Myopathy, distal, with anterior tibial onset	606768	DYSF
722	Myopathy, myofibrillar, 3	609200	MYOT
723	Myopathy, spheroid body	182920	MYOT
724	Myopathy, tubular aggregate, 1	160565	STIM1
725	Myopathy, tubular aggregate, 2	615883	ORAI1
726	Naegeli-Franceschetti-Jadassohn syndrome	161000	KRT14
727	Nail-patella syndrome	161200	LMX1B
728	Nance-Horan syndrome	302350	NHS
729	Navajo neurohepatopathy	256810	MPV17
730	Nemaline myopathy 1, autosomal dominant or recessive	609284	TPM3
731	Nemaline myopathy 2	256030	NEB
732	Nemaline myopathy 3, autosomal dominant or recessive	161800	ACTA1
733	Nemaline myopathy 4, autosomal dominant	609285	TPM2
734	Nemaline myopathy 5, Amish type	605355	TNNT1
735	Neonatal adrenoleukodystrophy (gene PEX12)	266510	PEX12
736	Neonatal adrenoleukodystrophy (gene PEX26)	614873	PEX26
737	Neonatal adrenoleukodystrophy (gene PEX5)	202370	PEX5
738	Nephrolithiasis, type I	310468	CLCN5
739	Nephronophthisis 2, infantile	602088	INVS
740	Nephrotic syndrome, tupe 3	610725	PLCE1

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741	Nephrotic syndrome, type 1	256300	NPHS1
742	Nephrotic syndrome, type 2	600995	NPHS2
743	Nephrotic syndrome, type 4	256370	WT1
744	Nephrotic syndrome, type 5, with or without ocular abnormalities	614199	LAMB2
745	Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency	250620	HIBCH
746	Neurodegeneration due to cerebral folate transport deficiency	613068	FOLR1
747	Neurofibromatosis, type 1	162200	NF1
748	Neurofibromatosis, type 2	101000	NF2
749	Neuromuscular disease, congenital, with uniform type 1 fiber	117000	RYR1
750	Neuronal ceroid lipofuscinosis 2	204500	TPP1
751	Neuropathy, congenital hypomyelinating	605253	MPZ
752	Neuropathy, distal hereditary motor, type VA	600794	BSCL2 - GARS
753	Neutropenia, severe congenital 3, autosomal recessive	610738	HAX1
754	Niemann-Pick disease type A	257200	SMPD1
755	Niemann-Pick disease type B	607616	SMPD1
756	Niemann-Pick disease type C1	257220	NPC1
757	Niemann-Pick disease type C2	607625	NPC2
758	Night blindness, autosomal dominant type 2	163500	PDE6B
759	Nijmegen breakage syndrome	251260	NBN
760	Noonan syndrome 1	163950	PTPN11
761	Noonan syndrome 3	609942	KRAS
762	Noonan syndrome 4	610733	SOS1
763	Noonan syndrome 5	611553	RAF1
764	Noonan syndrome 6	613224	NRAS
765	Noonan syndrome 7	613706	BRAF
766	Noonan syndrome-like disorder	613563	CBL
767	Noonan-like syndrome with loose anagen hair	607721	SHOC2
768	Norrie disease	310600	NDP
769	Occipital horn syndrome	304150	ATP7A
770	Oculocerebrorenal syndrome	309000	OCRL
771	Omenn syndrome	603554	DCLRE1C
772	Omenn syndrome (gene RAG1)	603554	RAG1 - RAG2
773	Opitz GBBB syndrome, type I	300000	MID1
774	Ornithine transcarbamylase deficiency	311250	OTC
775	Osteoarthritis with mild chondrodysplasia	604864	COL2A1
776	Osteochondritis dissecans	165800	ACAN
777	Osteogenesis imperfecta type 8	610915	LEPRE1
778	Osteogenesis imperfecta type VII	610682	CRTAP
779	Osteogenesis imperfecta, type I	166200	COL1A1

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780	Osteogenesis imperfecta, type II	166210	COL1A1
781	Osteogenesis imperfecta, type III	259420	COL1A1
782	Osteogenesis imperfecta, type IV	166220	COL1A1
783	Osteopetrosis with renal tubular acidosis	259730	CA2
784	Osteopetrosis, autosomal recessive 5	259720	OSTM1
785	Osteopoikilosis	166700	LEMD3
786	Otospondylomegaepiphyseal dysplasia	215150	COL11A2 - COL2A1
787	Paget disease, juvenile	239000	TNFRSF11B
788	Pancreatic cancer, somatic	260350	SMAD4
789	Panhypopituitarism, X-linked	312000	SOX3
790	Pantothenate kinase-associated neurodegeneration	234200	PANK2
791	Parastremmatic dwarfism	168400	TRPV4
792	Partial androgen insensitivity syndrome	312300	AR
793	PCWH syndrome	609136	SOX10
794	Pelizaeus-Merzbacher-like due to GJC2 mutation	608804	GJC2
795	Peroxisomal acyl-CoA oxidase deficiency	264470	ACOX1
796	Peroxisome biogenesis disorder 11A (Zellweger)	614883	PEX13
797	Peroxisome biogenesis disorder 11B	614885	PEX13
798	Peroxisome biogenesis disorder 13A (Zellweger)	614887	PEX14
799	Peroxisome biogenesis disorder 4A (Zellweger)	614862	PEX6
800	Peroxisome biogenesis disorder 4B	614863	PEX6
801	Peroxisome biogenesis disorder 5A (Zellweger)	614866	PEX2
802	Peroxisome biogenesis disorder 5B	614867	PEX2
803	Peroxisome biogenesis disorder 6A (Zellweger)	614870	PEX10
804	Peroxisome biogenesis disorder 6B	614871	PEX10
805	Perrault syndrome	233400	HSD17B4
806	Phenylketonuria	261600	PAH
807	Pheochromocytoma	171300	KIF1B - RET -VHL
808	Phosphoglycerate kinase 1 deficiency	300653	PGK1
809	Pierson syndrome	609049	LAMB2
810	Pitt-Hopkins syndrome	610954	TCF4
811	Plasminogen deficiency type 1	217090	PLG
812	Platyspondylic skeletal dysplasia, Torrance type	151210	COL2A1
813	Polycystic kidney disease 2	613095	PKD2
814	Polyposis, juvenile intestinal	174900	SMAD4
815	Porphyria, congenital erythropoietic	263700	UROS
816	Precocious puberty, male	176410	LHCGR
817	Primary lateral sclerosis, juvenile	606353	ALS2

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818	Progressive epilepsy - intellectual deficit, Finnish type	610003	CLN8
819	Properdin deficiency, X-linked	312060	CFP
820	Propionic acidemia (gene PCCA)	606054	PCCA - PCCB
821	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis	308990	CLCN5
822	Proximal spinal muscular atrophy type 1	253300	SMN1
823	Proximal spinal muscular atrophy type 2	253550	SMN1
824	Proximal spinal muscular atrophy type 3	253400	SMN1
825	Proximal spinal muscular atrophy type 4	271150	SMN1
826	Pseudoachondroplasia	177170	COMP
827	Pseudohermaphroditism, male, with gynecomastia	264300	HSD17B3
828	Pseudohypoaldosteronism type 1, autosomal recessive	264350	SCNN1A - SCNN1B - SCNN1G
829	Pseudovaginal perineoscrotal hypospadias	264600	SRD5A2
830	Pycnodysostosis	265800	CTSK
831	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency	612260	MYD88
832	Pyridoxal phosphate-responsive seizures	610090	PNPO
833	Pyruvate carboxylase deficiency	266150	PC
834	Pyruvate dehydrogenase phosphatase deficiency	608782	PDP1
835	Renal-hepatic-pancreatic dysplasia	208540	NPHP3
836	Renpenning syndrome	309500	PQBP1
837	Retinitis pigmentosa type 1,autosomal dominant	180100	RP1
838	Retinitis pigmentosa type 10, autosomal dominant	180105	IMPDH1
839	Retinitis pigmentosa type 11, autosomal dominant	600138	PRPF31
840	Retinitis pigmentosa type 13, autosomal dominant	600059	PRPF8
841	Retinitis pigmentosa type 17, autosomal dominant	600852	CA4
842	Retinitis pigmentosa type 18, autosomal dominant	601414	PRPF3
843	Retinitis pigmentosa type 19, autosomal dominant	601718	ABCA4
844	Retinitis pigmentosa type 27, autosomal dominant	613750	NRL
845	Retinitis pigmentosa type 30, autosomal dominant	607921	FSCN2
846	Retinitis pigmentosa type 31, autosomal dominant	609923	TOPORS
847	Retinitis pigmentosa type 33, autosomal dominant	610359	SNRNP200
848	Retinitis pigmentosa type 35, autosomal dominant	610282	SEMA4A
849	Retinitis pigmentosa type 4, autosomal dominant	613731	RHO
850	Retinitis pigmentosa type 42, autosomal dominant	612943	KLHL7
851	Retinitis pigmentosa type 48, autosomal dominant	613827	GUCA1B
852	Retinitis pigmentosa type 50, autosomal dominant	613194	BEST1
853	Retinitis pigmentosa type 7, autosomal dominant	608133	PRPH2
854	Retinitis pigmentosa type 9, autosomal dominant	180104	RP9
855	Rett syndrome, congenital variant	613454	FOXG1

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856	Rhabdomyosarcoma 2, alveolar	268220	PAX3
857	Rhizomelic chondrodysplasia punctata type 1	215100	PEX7
858	Rhizomelic chondrodysplasia punctata type 3	600121	AGPS
859	Rippling muscle disease	606072	CAV3
860	Roberts syndrome	269000	ESCO2
861	Robinow-Sorauf syndrome	180750	TWIST1
862	Rolandic epilepsy, mental retardation, and speech dyspraxia	300643	SRPX2
863	Roussy-Levy syndrome	180800	MPZ - PMP22
864	Saethre-Chotzen syndrome	101400	FGFR2 - TWIST1
865	Sandhoff disease	268800	HEXB
866	Sanfilippo syndrome type C	252930	HGSNAT
867	Scaphocephaly, maxillary retrusion, and mental retardation	609579	FGFR2
868	Scapuloperoneal spinal muscular atrophy	181405	TRPV4
869	Schizencephaly	269160	SHH - SIX3
870	Schneckenbecken dysplasia	269250	SLC35D1
871	Schwannomatosis	162091	NF2
872	Schwartz-Jampel syndrome	255800	HSPG2
873	Seckel syndrome	210600	ATR
874	SED congenita	183900	COL2A1
875	SED, Maroteaux type	184095	TRPV4
876	Senior-Loken syndrome	606996	NPHP4
877	Senior-Loken syndrome	610189	CEP290
878	Senior-Loken syndrome 1	266900	NPHP1
879	Senior-Loken syndrome 5	609254	IQCB1
880	Sensory ataxic neuropathy - dysarthria - ophthalmoparesis	607459	POLG
881	SERKAL syndrome	611812	WNT4
882	Severe combined immunodeficiency due to complete RAG1/2 deficiency	601457	RAG1 - RAG2
883	Severe combined immunodeficiency due to DCLRE1C deficiency	602450	DCLRE1C
884	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	611291	NHEJ1
885	Severe combined immunodeficiency with sensitivity to ionizing radiation	602450	LIG4
886	Severe combined immunodeficiency. deaminase deficiency	102700	ADA
887	Severe generalized recessive dystrophic epidermolysis bullosa	226600	COL7A1
888	Severe neonatal-onset encephalopathy with microcephaly	300673	MECP2
889	Severe T-cell immunodeficiency - congenital alopecia - nail dystrophy	601705	FOXN1
890	Short QT syndrome 1	609620	KCNH2
891	Short QT syndrome 2	609621	KCNQ1
892	Short QT syndrome 3	609622	KCNJ2
893	Short-rib thoracic dysplasia 3 with or without polydactyly	613091	DYNC2H1

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894	Shwachman-Diamond syndrome	260400	SBDS
895	Sialidosis, type I	256550	NEU1
896	Sialidosis, type II	256550	NEU1
897	Sickle cell anemia	603903	HBB
898	Silver spastic paraplegia syndrome	270685	BSCL2
899	Silver-Russell syndrome	180860	H19
900	Simpson-Golabi-Behmel syndrome type 2	300209	OFD1
901	Simpson-Golabi-Behmel syndrome, type 1	312870	GPC3
902	Síndrome de Dursun	612541	G6PC3
903	Single median maxillary central incisor	147250	SHH
904	Sjogren-Larsson syndrome	270200	ALDH3A2
905	Slowed nerve conduction velocity, AD	608236	ARHGEF10
906	SMED Strudwick type	184250	COL2A1
907	Smith-Lemli-Opitz syndrome	270400	DHCR7
908	Sotos syndrome 1	117550	NSD1
909	Spastic paralysis, infantile onset ascending	607225	ALS2
910	Spastic paraplegia type 2, X-linked	312920	PLP1
911	Spherocytosis, type 1	182900	ANK1
912	Spinal muscular atrophy with respiratory distress	604320	IGHMBP2
913	Spinal muscular atrophy, distal, congenital nonprogressive	600175	TRPV4
914	Spondyloepimetaphyseal dysplasia	612813	ACAN
915	Spondyloepiphyseal dysplasia	608361	ACAN
916	Spondylometaphyseal dysplasia, Kozlowski type	184252	TRPV4
917	Spondyloperipheral dysplasia	271700	COL2A1
918	Stickler sydrome, type I, nonsyndromic ocular	609508	COL2A1
919	Stickler syndrome, type I	108300	COL2A1
920	Stickler syndrome, type II	604841	COL11A1
921	Stickler syndrome, type III	184840	COL11A2
922	Stickler syndrome, type IV	614134	COL9A1
923	Stiff skin syndrome	184900	FBN1
924	Stocco dos Santos X-linked mental retardation syndrome	300434	SHROOM4
925	Stormorken syndrome	185070	STIM1
926	Stüve-Wiedemann syndrome	601559	LIFR
927	Subcortical laminal heteropia, X-linked	300067	DCX
928	Succinyl CoA:3-oxoacid CoA transferase deficiency	245050	OXCT1
929	Sudden infant death with dysgenesis of the testes syndrome	608800	TSPYL1
930	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A	252150	MOCS1 - MOCS2
931	Sulfocysteinuria	272300	SUOX
932	Surfactant metabolism dysfunction, pulmonary, 1	265120	SFTPB

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933	Surfactant metabolism dysfunction, pulmonary, 2	610913	SFTPC
934	Surfactant metabolism dysfunction, pulmonary, 3	610921	ABCA3
935	Syndromic microphthalmia type 9	601186	STRA6
936	Tay-Sachs disease	272800	HEXA
937	T-B+ severe combined immunodeficiency due to gamma chain deficiency	300400	IL2RG
938	T-B+ severe combined immunodeficiency due to JAK3 deficiency	600802	JAK3
939	T-B+ severe combined immunodeficiency, X-linked	312863	IL2RG
940	Telangiectasia, hereditary hemorrhagic, type 1	187300	ENG
941	Telangiectasia, hereditary hemorrhagic, type 2	600376	ACVRL1
942	Tetra-amelia, autosomal recessive	273395	WNT3
943	Thanatophoric dysplasia, type I	187600	FGFR3
944	Thanatophoric dysplasia, type II	187601	FGFR3
945	Thrombocythemia 2	601977	MPL
946	Thrombocytopenia, congenital amegakaryocytic	604498	MPL
947	Thrombocytopenia-absent radius syndrome	274000	RBM8A
948	Thrombotic thrombocytopenic purpura, familial	274150	ADAMTS13
949	Thryoid dyshormonogenesis 6	607200	DUOX2
950	Thyroid dyshormonogenesis 1	274400	SLC5A5
951	Thyroid dyshormonogenesis 2A	274500	TPO
952	Thyroid dyshormonogenesis 3	274700	TG
953	Thyroxine-binding globulin deficiency	314200	SERPINA7
954	Tietz albinism-deafness syndrome	103500	MITF
955	Tooth agenesis, selective, X-linked 1	313500	EDA
956	Treacher Collins syndrome 1	154500	TCOF1
957	Treacher Collins syndrome 3	248390	POLR1C
958	Trichothiodystrophy, complementation group A	601675	GTF2H5
959	Tuberous sclerosis-1	191100	TSC1
960	Tuberous sclerosis-2	613254	TSC2
961	Tyrosinemia type 1	276700	FAH
962	Tyrosinemia type 2	276600	TAT
963	Tyrosinemia type 3	276710	HPD
964	Ullrich congenital muscular dystrophy	254090	COL6A1 - COL6A2 - COL6A3
965	Unverricht-Lundborg disease	254800	CSTB
966	Usher syndrome type 1	276900	MYO7A
967	Usher syndrome type 1C	276904	USH1C
968	Usher syndrome type 1G	606943	USH1G
969	Usher syndrome type 2A	276901	USH2A
970	Usher syndrome type 2C	605472	GPR98

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971	Usher syndrome type 3A	276902	CLRN1
972	Very long chain acyl-CoA dehydrogenase deficiency	201475	ACADVL
973	Vitamin B12-responsive methylmalonic acidemia type cblA	251100	MMAA
974	Vitamin B12-responsive methylmalonic acidemia type cblB	251110	MMAB
975	Vitamin B12-unresponsive methylmalonic acidemia type mut-	251000	MUT
976	Vitamin D-dependent rickets type 2A	277440	VDR
977	Vitamin D-dependent rickets, type I	264700	CYP27B1
978	von Hippel-Lindau syndrome	193300	VHL
979	von Willebrand disease, type 1	193400	VWF
980	von Willebrand disease, types 2A, 2B, 2M, and 2N	613554	VWF
981	von Willibrand disease, type 3	277480	VWF
982	Waardenburg syndrome, type 1	193500	PAX3
983	Waardenburg syndrome, type 2A	193510	MITF
984	Waardenburg syndrome, type 2E, with or without neurologic involvement	611584	SOX10
985	Waardenburg syndrome, type 3	148820	PAX3
986	Waardenburg syndrome, type 4C	613266	SOX10
987	Waardenburg syndrome/ocular albinism, digenic	103470	MITF
988	Waardenburg-Shah syndrome 4A	277580	EDNRB
989	Waardenburg-Shah syndrome 4B	613265	EDN3
990	Walker-Warburg syndrome (gene POMGNT1)	253280	POMGNT1
991	Walker-Warburg syndrome (gene POMT1)	236670	POMT1
992	Walker-Warburg syndrome (gene POMT2)	613150	POMT2
993	Weill-Marchesani syndrome 2, dominant	608328	FBN1
994	Weissenbacher-Zweymuller syndrome	277610	COL11A2
995	Wilms tumor 2	194071	H19
996	Wilms tumor, type 1	194070	WT1
997	Wilson disease	277900	ATP7B
998	Wiskott-Aldrich syndrome	301000	WAS
999	Wolcott-Rallison syndrome	226980	EIF2AK3
1000	Wrinkly skin syndrome	278250	ATP6V0A2
1001	Xeroderma pigmentosum complementation group A	278700	XPA
1002	Xeroderma pigmentosum complementation group E	278740	DDB2
1003	Xeroderma pigmentosum, group C	278720	XPC
1004	Xeroderma pigmentosum/Cockayne syndrome complex complementation group B	610651	ERCC3
1005	Xeroderma pigmentosum/Cockayne syndrome complex complementation group D	278730	ERCC2
1006	Xeroderma pigmentosum/Cockayne syndrome complex complementation group F	278760	ERCC4
1007	Xeroderma pigmentosum/Cockayne syndrome complex complementation group G	278780	ERCC5

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1008	**X-linked agammaglobulinemia**	**300755**	**BTK**
1009	**X-linked centronuclear myopathy**	**310400**	**MTM1**
1010	**X-linked Charcot-Marie-Tooth disease type 5**	**311070**	**PRPS1**
1011	**X-linked creatine transporter deficiency**	**300352**	**SLC6A8**
1012	**X-linked distal spinal muscular atrophy**	**300489**	**ATP7A**
1013	**X-linked hyper-IgM syndrome**	**308230**	**CD40LG**
1014	**X-linked intellectual deficit with marfanoid habitus**	**309520**	**MED12**
1015	**X-linked lymphoproliferative disease**	**308240**	**SH2D1A**
1016	**X-linked severe congenital neutropenia**	**300299**	**WNT10A**
1017	**X-linked spinal muscular atrophy type 2**	**301830**	**UBA1**
1018	**Yunis-Varon syndrome**	**216340**	**FIG4**
1019	**Zellweger syndrome 1A**	**214100**	**PEX1**
1020	**Zellweger syndrome 7A**	**614872**	**PEX26**

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