

## GeneScreen®: Elenco dei geni analizzati e malattie genetiche investigate

	DISEASE NAME	PhenoMIM	GENE
1	17-alpha-hydroxylase/17,20-lyase deficiency	202110	<i>CYP17A1</i>
2	17-beta-hydroxysteroid dehydrogenase X deficiency	300438	<i>HSD17B10</i>
3	3-beta-hydroxysteroid dehydrogenase, type II, deficiency	201810	<i>HSD3B2</i>
4	3-hydroxy-3-methylglutaric aciduria	246450	<i>HMGCL</i>
5	3-methylglutaconic aciduria type 1	250950	<i>AUH</i>
6	3-methylglutaconic aciduria type 3	258501	<i>OPA3</i>
7	46XY sex reversal 3	612965	<i>NR5A1</i>
8	4-hydroxybutyric aciduria	271980	<i>ALDH5A1</i>
9	Aarskog-Scott syndrome	305400	<i>FGD1</i>
10	ABCD syndrome	600501	<i>EDNRB</i>
11	Achalasia-addisonianism-alacrimia syndrome	231550	<i>AAAS</i>
12	Achondrogenesis type 1B	600972	<i>SLC26A2</i>
13	Acyl-CoA dehydrogenase 9 deficiency	611126	<i>ACAD9</i>
14	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	202010	<i>CYP11B1</i>
15	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	613743	<i>CYP11A1</i>
16	Adrenocortical insufficiency	612965	<i>NR5A1</i>
17	Adrenoleukodystrophy	300100	<i>ABCD1</i>
18	Adult neuronal ceroid lipofuscinosis	256730	<i>PPT1</i>
19	Adult neuronal ceroid lipofuscinosis 10	610127	<i>CTSD</i>
20	Adult neuronal ceroid lipofuscinosis 4A	204300	<i>CLN6</i>
21	Aicardi-Goutières syndrome	225750	<i>TREX1</i>
22	Aicardi-Goutieres syndrome 2	610181	<i>RNASEH2B</i>
23	Aicardi-Goutieres syndrome 3	610329	<i>RNASEH2C</i>
24	Aicardi-Goutieres syndrome 4	610333	<i>RNASEH2A</i>
25	Aicardi-Goutieres syndrome 5	612952	<i>SAMHD1</i>
26	Aldosteronism, glucocorticoid-remediable	103900	<i>CYP11B1</i>
27	Allan-Herndon-Dudley syndrome	300523	<i>SLC16A2</i>
28	Alpers syndrome	203700	<i>POLG</i>
29	Alpha-methylacyl-Coa Racemase deficiency	614307	<i>AMACR</i>
30	Alpha-thalassemia	604131	<i>HBA1</i>
31	Alpha-thalassemia myelodysplasia syndrome, somatic	300448	<i>ATRX</i>
32	Alpha-thalassemia/mental retardation syndrome	301040	<i>ATRX</i>
33	Alport syndrome	301050	<i>COL4A5</i>
34	Alport syndrome autosomal recessive (gene COL4A3)	203780	<i>COL4A3</i>
35	Alport syndrome autosomal recessive (gene COL4A4)	203780	<i>COL4A4</i>
36	Alström syndrome	203800	<i>ALMS1</i>

37	Amish infantile epilepsy syndrome	609056	<i>ST3GAL5</i>
38	Amyotrophic lateral sclerosis 2, juvenile	205100	<i>ALS2</i>
39	Anauxetic dysplasia	607095	<i>RMRP</i>
40	Angelman syndrome	105830	<i>UBE3A</i>
41	Antenatal Bartter syndrome	241200	<i>KCNJ1</i>
42	Antenatal Bartter syndrome type 1	601678	<i>SLC12A1</i>
43	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	201750	<i>POR</i>
44	Aplasia/hypoplasia of limbs and pelvis	276820	<i>WNT7A</i>
45	Aplastic anemia	609135	<i>NBN</i>
46	Apparent mineralocorticoid excess	218030	<i>HSD11B2</i>
47	Argininosuccinic aciduria	207900	<i>ASL</i>
48	Aromatic L-amino acid decarboxylase deficiency	608643	<i>DDC</i>
49	Arthrogyrosis - renal dysfunction - cholestasis	208085	<i>VPS33B</i>
50	Arthrogyrosis, renal dysfunction, and cholestasis 2	613404	<i>VIPAR</i>
51	Ataxia - oculomotor apraxia type 1	208920	<i>APTX</i>
52	Ataxia with vitamin E deficiency	277460	<i>TTPA</i>
53	Ataxia-telangiectasia	208900	<i>ATM</i>
54	Atelosteogenesis type II	256050	<i>SLC26A2</i>
55	Autism, susceptibility to, X-linked 5	300847	<i>RPL10</i>
56	Autoimmune lymphoproliferative syndrome, type IA	601859	<i>FAS</i>
57	Autoimmune lymphoproliferative syndrome, type IB	601859	<i>FASLG</i>
58	Autoimmune lymphoproliferative syndrome, type II	603909	<i>CASPI0</i>
59	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia	240300	<i>AIRE</i>
60	Autosomal dominant Charcot-Marie-Tooth disease type 2K	607831	<i>GDAPI</i>
61	Autosomal recessive ataxia due to ubiquinone deficiency	612016	<i>ADCK3</i>
62	Autosomal recessive Charcot-Marie-Tooth disease with hoarseness	607706	<i>GDAPI</i>
63	Autosomal recessive distal spinal muscular atrophy type 4	611067	<i>PLEKHG5</i>
64	Autosomal recessive dopa-responsive dystonia	605407	<i>TH</i>
65	Autosomal recessive hypophosphatemic rickets 1	241520	<i>DMP1</i>
66	Autosomal recessive hypophosphatemic rickets 2	613312	<i>ENPP1</i>
67	Autosomal recessive intermediate Charcot-Marie-Tooth disease type A	608340	<i>GDAPI</i>
68	Autosomal recessive limb-girdle muscular dystrophy type 2I	607155	<i>FKRP</i>
69	Autosomal recessive limb-girdle muscular dystrophy type 2M	611588	<i>FKTN</i>
70	Autosomal recessive limb-girdle muscular dystrophy type C	613157	<i>POMGNT1</i>
71	Autosomal recessive limb-girdle muscular dystrophy type C	609308	<i>POMT1</i>
72	Autosomal recessive limb-girdle muscular dystrophy type C	613158	<i>POMT2</i>
73	Autosomal recessive malignant osteopetrosis 1	259700	<i>TCIRG1</i>
74	Autosomal recessive malignant osteopetrosis 4	611490	<i>CLCN7</i>

75	Autosomal recessive nonsyndromic sensorineural deafness type DFNB12	601386	<i>CDH23</i>
76	Autosomal recessive nonsyndromic sensorineural deafness type DFNB18	602092	<i>USH1C</i>
77	Autosomal recessive nonsyndromic sensorineural deafness type DFNB1A (gene GJB2)	220290	<i>GJB2</i>
78	Autosomal recessive nonsyndromic sensorineural deafness type DFNB2	600060	<i>MYO7A</i>
79	Autosomal recessive polycystic kidney disease	263200	<i>PKHD1</i>
80	Autosomal recessive progressive external ophthalmoplegia	258450	<i>POLG</i>
81	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	270550	<i>SACS</i>
82	Autosomal recessive spondylocostal dysostosis 1	277300	<i>DLL3</i>
83	Bannayan-Riley-Ruvalcaba syndrome	153480	<i>PTEN</i>
84	Barth syndrome	302060	<i>TAZ</i>
85	Becker muscular dystrophy	300376	<i>DMD</i>
86	Beckwith-Wiedemann syndrome	130650	<i>NSD1</i>
87	Beta-thalassemia	613985	<i>HBB</i>
88	Bethlem myopathy	158810	<i>COL6A1</i>
89	Bethlem myopathy	158810	<i>COL6A2</i>
90	Bethlem myopathy	158810	<i>COL6A3</i>
91	Bifunctional enzyme deficiency	261515	<i>HSD17B4</i>
92	Biotinidase deficiency	253260	<i>BTBD</i>
93	Björnstad syndrome	262000	<i>BCS1L</i>
94	Bloom syndrome	210900	<i>BLM</i>
95	Brachytelephalangic chondrodysplasia punctata	302950	<i>ARSE</i>
96	Brittle cornea syndrome	229200	<i>ZNF469</i>
97	Caffey disease	114000	<i>COL1A1</i>
98	Canavan disease	271900	<i>ASPA</i>
99	Carbamoylphosphate synthetase deficiency	237300	<i>CPS1</i>
100	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	604377	<i>SCO2</i>
101	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2	615119	<i>COX15</i>
102	Carnitine deficiency, systemic primary	212140	<i>SLC22A5</i>
103	Carnitine palmitoyl transferase 1A deficiency	255120	<i>CPT1A</i>
104	Carnitine palmitoyl transferase II deficiency, infantile form	600649	<i>CPT2</i>
105	Carnitine palmitoyl transferase II deficiency, neonatal form	608836	<i>CPT2</i>
106	Carnitine-acylcarnitine translocase deficiency	212138	<i>SLC25A20</i>
107	Carpenter syndrome	201000	<i>RAB23</i>
108	Cartilage-hair hypoplasia	250250	<i>RMRP</i>
109	Cataract - intellectual deficit - hypogonadism	212720	<i>RAB3GAP2</i>
110	Cataract 40, X-linked	302200	<i>NHS</i>
111	Cerebellar ataxia - intellectual deficit - dysequilibrium syndrome	224050	<i>VLDLR</i>
112	Cerebral dysgenesis-neuropathy-ichthyosis-palmoplantar keratoderma	609528	<i>SNAP29</i>

	syndrome		
113	Cerebrotendinous xanthomatosis	213700	<i>CYP27A1</i>
114	Charcot-Marie-Tooth disease axonal type 2B1	605588	<i>LMNA</i>
115	Charcot-Marie-Tooth disease type 4A	214400	<i>GDAP1</i>
116	Charcot-Marie-Tooth disease type 4E	605253	<i>EGR2</i>
117	Charcot-Marie-Tooth disease type 4F	614895	<i>PRX</i>
118	Charcot-Marie-Tooth disease type 4H	609311	<i>FGD4</i>
119	Charcot-Marie-Tooth disease, type 1A	118220	<i>PMP22</i>
120	Charcot-Marie-Tooth disease, type 1B	118200	<i>MPZ</i>
121	Charcot-Marie-Tooth disease, type 1E	118300	<i>PMP22</i>
122	Charcot-Marie-Tooth disease, type 2I	607677	<i>MPZ</i>
123	Charcot-Marie-Tooth disease, type 2J	607736	<i>MPZ</i>
124	Chediak-Higashi syndrome	214500	<i>LYST</i>
125	Chilblain lupus 2	614415	<i>SAMHD1</i>
126	Childhood-onset hypophosphatasia	241510	<i>ALPL</i>
127	Cholestasis, benign recurrent intrahepatic	243300	<i>ATP8B1</i>
128	Cholestasis, benign recurrent intrahepatic, 2	605479	<i>ABCB11</i>
129	Cholestasis, intrahepatic, of pregnancy, 1	147480	<i>ATP8B1</i>
130	Cholestasis, intrahepatic, of pregnancy, 3	614972	<i>ABCB4</i>
131	Cholestasis, progressive familial intrahepatic 1	211600	<i>ATP8B1</i>
132	Cholestasis, progressive familial intrahepatic 2	601847	<i>ABCB11</i>
133	Cholestasis, progressive familial intrahepatic 3	602347	<i>ABCB4</i>
134	Chondrodysplasia, Blomstrand type	215045	<i>PTH1R</i>
135	Citrullinemia type I	215700	<i>ASS1</i>
136	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	201910	<i>CYP21A2</i>
137	Classic galactosemia	230400	<i>GALT</i>
138	Classic maple syrup urine disease	248600	<i>DBT</i>
139	Classical homocystinuria	236200	<i>CBS</i>
140	COACH syndrome	216360	<i>TMEM67</i>
141	Cockayne syndrome type A	216400	<i>ERCC8</i>
142	Cockayne syndrome type B	133540	<i>ERCC6</i>
143	Coenzyme Q10 deficiency, primary, 5	614654	<i>COQ9</i>
144	Coffin-Lowry syndrome	303600	<i>RPS6KA3</i>
145	COFS syndrome 1	214150	<i>ERCC6</i>
146	Cohen Syndrome type 1	216550	<i>VPS13B</i>
147	Cold-induced sweating syndrome	272430	<i>CRLF1</i>
148	Combined immunodeficiency with skin granulomas	233650	<i>RAG1</i>
149	Combined immunodeficiency with skin granulomas	233650	<i>RAG2</i>
150	Combined oxidative phosphorylation defect type 2	610498	<i>MRPS16</i>



151	Combined oxidative phosphorylation defect type 5	611719	<i>MRPS22</i>
152	Combined oxidative phosphorylation deficiency 4	610678	<i>TUFM</i>
153	Combined pituitary hormone deficiencies, genetic forms	182230	<i>HESX1</i>
154	Combined pituitary hormone deficiencies, genetic forms	613038	<i>POU1F1</i>
155	Combined pituitary hormone deficiencies, genetic forms	262600	<i>PROPI</i>
156	Combined pituitary hormone deficiency with spine abnormalities	221750	<i>LHX3</i>
157	Complete androgen insensitivity syndrome	300068	<i>AR</i>
158	Complex I, mitochondrial respiratory chain, deficiency of	252010	<i>NDUFS6</i>
159	Congenital bile acid synthesis defect type 4	214950	<i>AMACR</i>
160	Congenital disorder of glycosylation type 1a	212065	<i>PMM2</i>
161	Congenital disorder of glycosylation type 1b	602579	<i>MPI</i>
162	Congenital disorder of glycosylation type 1e	608799	<i>DPMI</i>
163	Congenital disorder of glycosylation type 1j	608093	<i>DPAGT1</i>
164	Congenital disorder of glycosylation type 2a	212066	<i>MGAT2</i>
165	Congenital disorder of glycosylation type 2c	266265	<i>SLC35C1</i>
166	Congenital disorder of glycosylation type 2d	607091	<i>B4GALT1</i>
167	Congenital disorder of glycosylation type 2f	603585	<i>SLC35A1</i>
168	Congenital disorder of glycosylation type 1c	603147	<i>ALG6</i>
169	Congenital disorder of glycosylation type 1k	608540	<i>ALG1</i>
170	Congenital disorder of glycosylation, type 1d	601110	<i>ALG3</i>
171	Congenital disorder of glycosylation, type 1f	609180	<i>MPDU1</i>
172	Congenital disorder of glycosylation, type 1g	607143	<i>ALG12</i>
173	Congenital disorder of glycosylation, type 1h	608104	<i>ALG8</i>
174	Congenital disorder of glycosylation, type 1i	607906	<i>ALG2</i>
175	Congenital disorder of glycosylation, type 1Ib	606056	<i>MOGS</i>
176	Congenital disorder of glycosylation, type 1Ie	608779	<i>COG7</i>
177	Congenital disorder of glycosylation, type 1Ig	611209	<i>COG1</i>
178	Congenital disorder of glycosylation, type 1Ih	611182	<i>COG8</i>
179	Congenital disorder of glycosylation, type 1I	608776	<i>ALG9</i>
180	Congenital disorder of glycosylation, type 1Im	610768	<i>DOLK</i>
181	Congenital disorder of glycosylation, type 1In	612015	<i>RFT1</i>
182	Congenital disorder of glycosylation, type 1Iq	612379	<i>SRD5A3</i>
183	Congenital fibrinogen deficiency (gene FGA)	202400	<i>FGA</i>
184	Congenital heart defects, nonsyndromic, 1, X-linked	306955	<i>ZIC3</i>
185	Congenital hereditary endothelial dystrophy type II	217700	<i>SLC4A11</i>
186	Congenital lipoid adrenal hyperplasia	201710	<i>STAR</i>
187	Congenital malabsorptive diarrhea due to paucity of enteroendocrine cells	610370	<i>NEUROG3</i>
188	Congenital muscular dystrophy type 1A	607855	<i>LAMA2</i>
189	Congenital muscular dystrophy type 1D	608840	<i>LARGE</i>

190	Congenital muscular dystrophy type 4B	613152	<i>FKTN</i>
191	Congenital muscular dystrophy type 5B	606612	<i>FKRP</i>
192	Congenital muscular dystrophy with cerebellar involvement	613151	<i>POMGNT1</i>
193	Congenital muscular dystrophy with cerebellar involvement	613155	<i>POMT1</i>
194	Congenital muscular dystrophy with cerebellar involvement	613156	<i>POMT2</i>
195	Corneal dystrophy - perceptive deafness	217400	<i>SLC4A11</i>
196	Corpus callosum agenesis - neuronopathy	218000	<i>SLC12A6</i>
197	Corpus callosum hypoplasia-retardation-adducted thumbs-spasticity-hydrocephalus syndrome	307000	<i>LICAM</i>
198	Cowden syndrome 1	158350	<i>PTEN</i>
199	Craniofrontonasal dysplasia	304110	<i>EFNB1</i>
200	Cutis laxa, autosomal dominant 2	614434	<i>FBLN5</i>
201	Cutis laxa, autosomal recessive, type IA	219100	<i>FBLN5</i>
202	Cutis laxa, autosomal recessive, type IB	614437	<i>EFEMP2</i>
203	Cutis laxa, autosomal recessive, type IIA	219200	<i>ATP6V0A2</i>
204	Cystic fibrosis; mucoviscidosis	219700	<i>CFTR</i>
205	Cystinosis	219800	<i>CTNS</i>
206	Deafness - encephaloneuropathy - obesity - valvulopathy	614651	<i>PDSS1</i>
207	Dejerine-Sottas disease	145900	<i>MPZ</i>
208	Dejerine-Sottas disease	145900	<i>PMP22</i>
209	Dent disease	300009	<i>CLCN5</i>
210	Dent disease 2	300555	<i>OCRL</i>
211	Desmosterolosis	602398	<i>DHCR24</i>
212	Diabetes mellitus, noninsulin-dependent	125853	<i>ABCC8</i>
213	Diabetes mellitus, permanent neonatal	606176	<i>ABCC8</i>
214	Diabetes mellitus, transient neonatal 2	610374	<i>ABCC8</i>
215	Diastrophic dwarfism	222600	<i>SLC26A2</i>
216	Dihydropyrimidine dehydrogenase deficiency	274270	<i>DPYD</i>
217	Dilated cardiomyopathy with ataxia	610198	<i>DNAJC19</i>
218	Donnai-Barrow syndrome	222448	<i>LRP2</i>
219	Duchenne muscular dystrophy	310200	<i>DMD</i>
220	Dyskeratosis congenita X-linked	305000	<i>DKC1</i>
221	Dystrophic epidermolysis bullosa pruriginosa	604129	<i>COL7A1</i>
222	Early infantile epileptic encephalopathy	308350	<i>ARX</i>
223	Early infantile epileptic encephalopathy	609304	<i>SLC25A22</i>
224	Ectodermal dysplasia 1, hypohidrotic, X-linked	305100	<i>EDA</i>
225	Ectodermal dysplasia, hypohidrotic, with immune deficiency	300291	<i>IKBK</i>
226	Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency	300301	<i>IKBK</i>
227	Ehlers-Danlos syndrome type 6	225400	<i>PLOD1</i>
228	Ehlers-Danlos syndrome, cardiac valvular type	225320	<i>COL1A2</i>

229	Ehlers-Danlos syndrome, type I	130000	<i>COL1A1</i>
230	Ehlers-Danlos syndrome, type VIIA	130060	<i>COL1A1</i>
231	Eiken syndrome	600002	<i>PTH1R</i>
232	Ellis-van Creveld syndrome	225500	<i>EVC2</i>
233	Ellis-van Creveld syndrome	225500	<i>EVC</i>
234	Encephalopathy due to prosaposin deficiency	611721	<i>PSAP</i>
235	Epidermolysis bullosa simplex with muscular dystrophy	226670	<i>PLEC</i>
236	Epidermolysis bullosa simplex with pyloric atresia	612138	<i>PLEC</i>
237	Epilepsy, progressive myoclonic 2A (Lafora)	254780	<i>EPM2A</i>
238	Epilepsy, progressive myoclonic 2B (Lafora)	254780	<i>NHLRC1</i>
239	Epilepsy, pyridoxine-dependent	266100	<i>ALDH7A1</i>
240	Epileptic encephalopathy, early infantile, 15	615006	<i>ST3GAL3</i>
241	Epileptic encephalopathy, early infantile, 2	300672	<i>CDKL5</i>
242	Epileptic encephalopathy, early infantile, 8	300607	<i>ARHGEF9</i>
243	Epileptic encephalopathy, early infantile, 9	300088	<i>PCDH19</i>
244	Escobar syndrome	265000	<i>CHRNA3</i>
245	Ethylmalonic encephalopathy	602473	<i>ETHE1</i>
246	Exudative vitreoretinopathy 2, X-linked	305390	<i>NDP</i>
247	Fabry disease	301500	<i>GLA</i>
248	Failure of tooth eruption, primary	125350	<i>PTH1R</i>
249	Familial dysautonomia	223900	<i>IKBKAP</i>
250	Familial hypomagnesemia - hypercalciuria - nephrocalcinosis - severe ocular involvement	248190	<i>CLDN19</i>
251	Familial Mediterranean fever	249100	<i>MEFV</i>
252	Fanconi anemia complementation group C	227645	<i>FANCC</i>
253	Fatal infantile lactic acidosis with methylmalonic aciduria	245400	<i>SUCLG1</i>
254	Fatal mitochondrial disease due to combined oxidative phosphorylation deficiency 3	610505	<i>TSMF</i>
255	Favism	134700	<i>G6PD</i>
256	Fertile eunuch syndrome	228300	<i>GNRHR</i>
257	Fetal akinesia deformation sequence	208150	<i>RAPSN</i>
258	Fetal akinesia deformation sequence	208150	<i>DOK7</i>
259	Fetal Gaucher disease	608013	<i>GBA</i>
260	FG syndrome 4	300422	<i>CASK</i>
261	Fibular hypoplasia or aplasia - femoral bowing - oligodactyly	228930	<i>WNT7A</i>
262	Fraser syndrome (gene FRAS1)	219000	<i>FRAS1</i>
263	Fraser syndrome (gene FRAS2)	219000	<i>FREM2</i>
264	Free sialic acid storage disease, infantile form	269920	<i>SLC17A5</i>
265	French-Canadian type Leigh syndrome	220111	<i>LRPPRC</i>
266	Fucosidosis	230000	<i>FUCA1</i>

267	Fukuyama congenital muscular dystrophy	253800	<i>FKTN</i>
268	Fumaric aciduria	606812	<i>FH</i>
269	Galactokinase deficiency with cataracts	230200	<i>GALK1</i>
270	Gallbladder disease 1	600803	<i>ABCB4</i>
271	Gaucher disease type 2	230900	<i>GBA</i>
272	Gaucher disease type 3	231000	<i>GBA</i>
273	Gaucher disease type 3C	231005	<i>GBA</i>
274	Geleophysic dysplasia 1	231050	<i>ADAMTSL2</i>
275	Generalized junctional epidermolysis bullosa, non-Herlitz type	226650	<i>COL17A1</i>
276	Glutaric acidemia type 2 (gene ETFA)	231680	<i>ETF A</i>
277	Glutaric acidemia type 2 (gene ETFB)	231680	<i>ETFB</i>
278	Glutaric acidemia type 2 (gene ETFDH)	231680	<i>ETFDH</i>
279	Glutaryl-CoA dehydrogenase deficiency	231670	<i>GCDH</i>
280	Glutathione synthetase deficiency with 5-oxoprolinuria	266130	<i>GSS</i>
281	Glycine encephalopathy	605899	<i>AMT</i>
282	Glycine encephalopathy	605899	<i>GCSH</i>
283	Glycine encephalopathy	605899	<i>GLDC</i>
284	Glycogen storage disease due to acid maltase deficiency	232300	<i>GAA</i>
285	Glycogen storage disease due to glucose-6-phosphatase deficiency type 1a	232200	<i>G6PC</i>
286	Glycogen storage disease due to glucose-6-phosphatase deficiency type b	232220	<i>SLC37A4</i>
287	Glycogen storage disease due to glucose-6-phosphatase deficiency type c	232240	<i>SLC37A4</i>
288	Glycogen storage disease due to glycogen branching enzyme deficiency, childhood combined hepatic and myopathic form	232500	<i>GBE1</i>
289	Glycogen storage disease due to glycogen debranching enzyme deficiency	232400	<i>AGL</i>
290	Glycogen storage disease due to muscle glycogen phosphorylase deficiency	232600	<i>PYGM</i>
291	GM1 gangliosidosis type 1	230500	<i>GLB1</i>
292	GM1 gangliosidosis type 2	230600	<i>GLB1</i>
293	GM1 gangliosidosis type 3	230650	<i>GLB1</i>
294	GRACILE syndrome	603358	<i>BCS1L</i>
295	Greenberg dysplasia	215140	<i>LBR</i>
296	GrisCELLI disease type 1	214450	<i>MYO5A</i>
297	GrisCELLI disease type 2	607624	<i>RAB27A</i>
298	Guanidinoacetate methyltransferase deficiency	612736	<i>GAMT</i>
299	Hemochromatosis, type 2A	602390	<i>HFE2</i>
300	Hemolytic anemia due to G6PD deficiency	300908	<i>G6PD</i>
301	Hemolytic anemia due to red cell pyruvate kinase deficiency	266200	<i>PKLR</i>
302	Hemophagocytic lymphohistiocytosis, familial, 2	603553	<i>PRF1</i>
303	Hemophagocytic lymphohistiocytosis, familial, 3	608898	<i>UNC13D</i>
304	Hemophagocytic lymphohistiocytosis, familial, 4	603552	<i>STX11</i>
305	Hemophagocytic lymphohistiocytosis, familial, 5	613101	<i>STXBP2</i>



306	Hemophilia A	306700	<i>F8</i>
307	Hemophilia B	306900	<i>F9</i>
308	Hepatic venoocclusive disease with immunodeficiency	235550	<i>SP110</i>
309	Hepatoencephalopathy due to combined oxidative phosphorylation deficiency type 1	609060	<i>GFMI</i>
310	Hereditary fructose intolerance	229600	<i>ALDOB</i>
311	Hereditary sensory and autonomic neuropathy type 4	256800	<i>NTRK1</i>
312	Hermansky-Pudlak syndrome 2	608233	<i>AP3BI</i>
313	Hermansky-pudlak syndrome 9	614171	<i>PLDN</i>
314	Heterotaxy, visceral, 1, X-linked	306955	<i>ZIC3</i>
315	Histidinemia	235800	<i>HAMP</i>
316	Holocarboxylase synthetase deficiency	253270	<i>HLCS</i>
317	Hoyeraal-Hreidarsson syndrome	300240	<i>DKC1</i>
318	Hyaline fibromatosis syndrome	228600	<i>ANTXR2</i>
319	Hyperammonemia due to N-acetylglutamate synthetase deficiency	237310	<i>NAGS</i>
320	Hyper-IgE recurrent infection syndrome, autosomal recessive	243700	<i>DOCK8</i>
321	Hyperinsulinemic hypoglycemia, familial, 1	256450	<i>ABCC8</i>
322	Hyperornithinemia-hyperammonemia-homocitrullinuria	238970	<i>SLC25A15</i>
323	Hypoglycemia of infancy, leucine-sensitive	240800	<i>ABCC8</i>
324	Hypogonadotropic hypogonadism 7 without anosmia	146110	<i>GNRHR</i>
325	Hypomyelination - congenital cataract	610532	<i>FAM126A</i>
326	Hypoparathyroidism - intellectual deficit - dysmorphism syndrome	241410	<i>TBCE</i>
327	Hypophosphatemic rickets	300554	<i>CLCN5</i>
328	Ichthyosis follicularis - alopecia - photophobia	308205	<i>MBTPS2</i>
329	Ichthyosis, autosomal recessive 4B (harlequin)	242500	<i>ABCA12</i>
330	Ichthyosis, congenital, autosomal recessive 1	242300	<i>TGMI</i>
331	Ichthyosis, congenital, autosomal recessive 4A	601277	<i>ABCA12</i>
332	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis 607626		<i>CLDN1</i>
333	Immunodeficiency 10	612783	<i>STIM1</i>
334	Immunodeficiency 17, CD3 gamma deficient	615607	<i>CD3G</i>
335	Immunodeficiency 18, SCID variant	615615	<i>CD3E</i>
336	Immunodeficiency 19	615617	<i>CD3D</i>
337	Immunodeficiency 27A, mycobacteriosis, AR	209950	<i>IFNGR1</i>
338	Immunodeficiency 28, mycobacteriosis	614889	<i>IFNGR2</i>
339	Immunodeficiency 29, mycobacteriosis	614890	<i>IL12B</i>
340	Immunodeficiency 30	614891	<i>IL12RB1</i>
341	Immunodeficiency 31A, mycobacteriosis, autosomal dominant	614892	<i>STAT1</i>
342	Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive	613796	<i>STAT1</i>
343	Immunodeficiency 31C, autosomal dominant	614162	<i>STAT1</i>

344	Immunodeficiency 33	300636	<i><b>IKBK</b></i>
345	Immunodeficiency 35	611521	<i><b>TYK2</b></i>
346	Immunodeficiency 9	612782	<i><b>ORAI1</b></i>
347	Immunodeficiency, common variable, 1	607594	<i><b>ICOS</b></i>
348	Immunodeficiency, common variable, 3	613493	<i><b>CD19</b></i>
349	Immunodeficiency-centromeric instability-facial anomalies syndrome 1	242860	<i><b>DNMT3B</b></i>
350	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	304790	<i><b>FOXP3</b></i>
351	Incontinentia pigmenti, type II	308300	<i><b>IKBK</b></i>
352	Infantile bilateral striatal necrosis	271930	<i><b>NUP62</b></i>
353	Infantile hypophosphatasia	241500	<i><b>ALPL</b></i>
354	Infantile neuroaxonal dystrophy 2A	256600	<i><b>PLA2G6</b></i>
355	Infantile neuroaxonal dystrophy 2B	610217	<i><b>PLA2G6</b></i>
356	Infantile onset spinocerebellar ataxia	271245	<i><b>C10orf2</b></i>
357	Interleukin 1 receptor antagonist deficiency	612852	<i><b>IL1RN</b></i>
358	Isolated CoQ-cytochrome C reductase deficiency	124000	<i><b>BCS1L</b></i>
359	Isolated growth hormone deficiency type III	307200	<i><b>BTK</b></i>
360	Isolated thyroid-stimulating hormone deficiency	275100	<i><b>TSHB</b></i>
361	Isovaleric acidemia	243500	<i><b>IVD</b></i>
362	Jeune syndrome	611263	<i><b>IFT80</b></i>
363	Johanson-Blizzard syndrome	243800	<i><b>UBR1</b></i>
364	Joubert syndrome 4	609583	<i><b>NPHP1</b></i>
365	Joubert syndrome 6	610688	<i><b>TMEM67</b></i>
366	Joubert syndrome with hepatic defect	216360	<i><b>RPGRIP1L</b></i>
367	Joubert syndrome with ocular defect	608629	<i><b>AH11</b></i>
368	Joubert syndrome with oculorenal defect 5	610188	<i><b>CEP290</b></i>
369	Junctional epidermolysis bullosa - pyloric atresia	226730	<i><b>ITGA6</b></i>
370	Junctional epidermolysis bullosa with piloric atresia	226730	<i><b>ITGB4</b></i>
371	Junctional epidermolysis bullosa, Herlitz type (gene LAMA3)	226700	<i><b>LAMA3</b></i>
372	Junctional epidermolysis bullosa, Herlitz type (gene LAMB3)	226700	<i><b>LAMA3</b></i>
373	Junctional epidermolysis bullosa, Herlitz type (gene LAMC2)	226700	<i><b>LAMC2</b></i>
374	Junctional epidermolysis bullosa, non-Herlitz type	226650	<i><b>ITGB4</b></i>
375	Junctional epidermolysis bullosa, non-Herlitz type (gene LAMA3)	226650	<i><b>LAMA3</b></i>
376	Junctional epidermolysis bullosa, non-Herlitz type (gene LAMB3)	226650	<i><b>LAMB3</b></i>
377	Junctional epidermolysis bullosa, non-Herlitz type (gene LAMC2)	226650	<i><b>LAMC2</b></i>
378	Juvenile neuronal ceroid lipofuscinosis 3	204200	<i><b>CLN3</b></i>
379	Kahrizi syndrome	612713	<i><b>SRD5A3</b></i>
380	Kelley-Seegmiller syndrome	300323	<i><b>HPRT1</b></i>
381	Kennedy disease	313200	<i><b>AR</b></i>
382	Ketoacidosis due to beta-ketothiolase deficiency	203750	<i><b>ACAT1</b></i>

383	Krabbe disease	245200	<i>GALC</i>
384	Krabbe disease	611722	<i>PSAP</i>
385	Lacticacidemia due to PDX1 deficiency	245349	<i>PDHX</i>
386	Late infantile neuronal ceroid lipofuscinosis	610951	<i>MFSD8</i>
387	Late infantile neuronal ceroid lipofuscinosis 5	256731	<i>CLN5</i>
388	Late infantile neuronal ceroid lipofuscinosis 6	601780	<i>CLN6</i>
389	Late infantile neuronal ceroid lipofuscinosis 8	600143	<i>CLN8</i>
390	Lathosterolosis	607330	<i>SC5DL</i>
391	Leigh syndrome	256000	<i>BCS1L</i>
392	Leigh syndrome	256000	<i>DLD</i>
393	Leigh syndrome	256000	<i>NDUFAF2</i>
394	Leigh syndrome	256000	<i>NDUFS4</i>
395	Leigh syndrome	256000	<i>NDUFS7</i>
396	Leigh syndrome due to cytochrome c oxidase deficiency	256000	<i>COX15</i>
397	Leigh syndrome due to mitochondrial complex I deficiency	256000	<i>NDUFS3</i>
398	Leigh syndrome due to mitochondrial complex I deficiency	256000	<i>NDUFS8</i>
399	Leigh syndrome due to mitochondrial COX4 deficiency	256000	<i>COX10</i>
400	Leigh syndrome with nephrotic syndrome	607426	<i>COQ2</i>
401	Leigh syndrome with nephrotic syndrome	614652	<i>PDSS2</i>
402	Leigh syndrome, due to COX deficiency	256000	<i>SURF1</i>
403	Leigh syndrome, X-linked	308930	<i>PDHA1</i>
404	Leprechaunism	246200	<i>INSR</i>
405	Lesch-Nyhan syndrome	300322	<i>HPRT1</i>
406	Lethal acantholytic epidermolysis bullosa	609638	<i>DSP</i>
407	Lethal ataxia with deafness and optic atrophy	301835	<i>PRPS1</i>
408	Lethal congenital contractural syndrome 2	607598	<i>ERBB3</i>
409	Lethal congenital contracture syndrome type 1	253310	<i>GLE1</i>
410	Lethal osteosclerotic bone dysplasia	259775	<i>FAM20C</i>
411	Lethal restrictive dermopathy	275210	<i>LMNA</i>
412	Lethal restrictive dermopathy	275210	<i>ZMPSTE24</i>
413	Leukocyte adhesion deficiency, type III	612840	<i>FERMT3</i>
414	Leydig cell adenoma, somatic, with precocious puberty	176410	<i>LHCGR</i>
415	Leydig cell hypoplasia with hypergonadotropic hypogonadism	238320	<i>LHCGR</i>
416	Leydig cell hypoplasia with pseudohermaphroditism	238320	<i>LHCGR</i>
417	Lhermitte-Duclos syndrome	158350	<i>PTEN</i>
418	Limb girdle dystrophy with epidermolysis bullosa simplex	613723	<i>PLEC</i>
419	Lissencephaly 3	611603	<i>TUBA1A</i>
420	Lissencephaly syndrome, Norman-Roberts type	257320	<i>RELN</i>
421	Lissencephaly, X-linked	300067	<i>DCX</i>

422	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	609016	<i>HADH</i>
423	Luteinizing hormone resistance, female	238320	<i>LHCGR</i>
424	Lymphoproliferative syndrome, X-linked, 2	300635	<i>XIAP</i>
425	Macrocephaly/autism syndrome	605309	<i>PTEN</i>
426	Macroglobulinemia, Waldenstrom	153600	<i>MYD88</i>
427	Macular degeneration, age-related, 3	608895	<i>FBLN5</i>
428	Mandibuloacral dysplasia with type A lipodystrophy	248370	<i>LMNA</i>
429	Mandibuloacral dysplasia with type B lipodystrophy	608612	<i>ZMPSTE24</i>
430	Mannosidosis, alpha-, types I and II	248500	<i>MAN2B1</i>
431	Maple syrup urine disease	248600	<i>DLD</i>
432	Maple syrup urine disease (gene BCKDHA)	248600	<i>BCKDHA</i>
433	Maple syrup urine disease (gene BCKDHB)	248600	<i>BCKDHB</i>
434	Marinesco-Sjögren syndrome	248800	<i>SIL1</i>
435	Masa syndrome	303350	<i>LICAM</i>
436	Meckel syndrome type 1	249000	<i>MKS1</i>
437	Meckel syndrome, type 5	611561	<i>RPGRIPL</i>
438	Medium chain acyl-CoA dehydrogenase deficiency	201450	<i>ACADM</i>
439	Megalencephalic leukoencephalopathy with subcortical cysts	604004	<i>MLC1</i>
440	Menkes disease	309400	<i>ATP7A</i>
441	Mental retardation and microcephaly with pontine and cerebellar hypoplasia	300749	<i>CASK</i>
442	Mental retardation, autosomal recessive 1	249500	<i>PRSS12</i>
443	Mental retardation, autosomal recessive 12	611090	<i>ST3GAL3</i>
444	Mental retardation, autosomal recessive 13	613192	<i>TRAPPC9</i>
445	Mental retardation, autosomal recessive 5	611091	<i>NSUN2</i>
446	Mental retardation, autosomal recessive, 6	611092	<i>GRIK2</i>
447	Mental retardation, with or without nystagmus	300422	<i>CASK</i>
448	Mental retardation, X-linked	300495	<i>NLGN4X</i>
449	Mental retardation, X-linked 19	300844	<i>RPS6KA3</i>
450	Mental retardation, X-linked 21/34	300143	<i>ILIRAPL1</i>
451	Mental retardation, X-linked 30/47	300558	<i>PAK3</i>
452	Mental retardation, X-linked 41	300849	<i>GDI1</i>
453	Mental retardation, X-linked 46	300436	<i>ARHGEF6</i>
454	Mental retardation, X-linked 63	300387	<i>ACSL4</i>
455	Mental retardation, X-linked 72	300271	<i>RAB39B</i>
456	Mental retardation, X-linked 9	309549	<i>FTSJ1</i>
457	Mental retardation, X-linked 90	300850	<i>DLG3</i>
458	Mental retardation, X-linked 93	300659	<i>BRWD3</i>
459	Mental retardation, X-linked 96	300802	<i>SYP</i>
460	Mental retardation, X-linked 97	300803	<i>ZNF711</i>



461	Mental retardation, X-linked syndromic 16	305400	<i>FGD1</i>
462	Mental retardation, X-linked syndromic 5	304340	<i>APIS2</i>
463	Mental retardation, X-linked syndromic, Christianson type	300243	<i>SLC9A6</i>
464	Mental retardation, X-linked syndromic, Nascimento-type	300860	<i>UBE2A</i>
465	Mental retardation, X-linked syndromic, Raymond type	300799	<i>ZDHHC9</i>
466	Mental retardation, X-linked syndromic, Turner type	300706	<i>HUWE1</i>
467	Mental retardation, X-linked, FRAXE type	309548	<i>AFF2</i>
468	Mental retardation, X-linked, Snyder-Robinson type	309583	<i>SMS</i>
469	Mental retardation, X-linked, syndromic 14	300676	<i>UPF3B</i>
470	Mental retardation, X-linked, syndromic 15 (Cabezas type)	300354	<i>CULAB</i>
471	Mental retardation, X-linked, syndromic, Claes-Jensen type	300534	<i>KDM5C</i>
472	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	300486	<i>OPHN1</i>
473	Mental retardation, X-linked, with isolated growth hormone deficiency	300123	<i>SOX3</i>
474	Mental retardation-hypotonic facies syndrome, X-linked	309580	<i>ATRX</i>
475	Metachromatic leukodystrophy	250100	<i>ARSA</i>
476	Metachromatic leukodystrophy	249900	<i>PSAP</i>
477	Metaphyseal chondrodysplasia, Murk Jansen type	156400	<i>PTHIR</i>
478	Metaphyseal dysplasia without hypotrichosis	250460	<i>RMRP</i>
479	Methylmalonic acidemia with homocystinuria, type cblC	277400	<i>MMACHC</i>
480	Methylmalonic acidemia with homocystinuria, type cblD	277410	<i>MMACHC</i>
481	Mevalonic aciduria	610377	<i>MVK</i>
482	Micro syndrome	600118	<i>RAB3GAP1</i>
483	Microphthalmia, syndromic 2	300166	<i>BCOR</i>
484	Mitochondrial complex I deficiency	252010	<i>NDUFA1</i>
485	Mitochondrial complex I deficiency	252010	<i>NDUFAF2</i>
486	Mitochondrial complex I deficiency	252010	<i>NDUFAF4</i>
487	Mitochondrial complex I deficiency	252010	<i>NDUFS3</i>
488	Mitochondrial complex I deficiency	252010	<i>NDUFS4</i>
489	Mitochondrial complex I deficiency	252010	<i>NDUFV1</i>
490	Mitochondrial complex IV deficiency	220110	<i>COX10</i>
491	Mitochondrial complex IV deficiency	220110	<i>COX6B1</i>
492	Mitochondrial complex IV deficiency	220110	<i>FASTKD2</i>
493	Mitochondrial complex IV deficiency		<i>SCO1</i>
494	Mitochondrial DNA depletion syndrome 1 (MNGIE type)	603041	<i>TYMP</i>
495	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	612073	<i>SUCLA2</i>
496	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)	612075	<i>RRM2B</i>
497	Mitochondrial DNA depletion syndrome 8B (MNGIE type)	612075	<i>RRM2B</i>
498	Mitochondrial DNA depletion syndrome, hepatocerebral form due to	251880	<i>DGUOK</i>

	<b>DGUOK deficiency 3</b>		
499	Mitochondrial DNA depletion syndrome, myopathic form	609560	<i>TK2</i>
500	Mitochondrial neurogastrointestinal encephalomyopathy	613662	<i>POLG</i>
501	Mitochondrial respiratory chain complex III deficiency	124000	<i>UQCRB</i>
502	Mitochondrial respiratory chain complex III deficiency	124000	<i>UQCRQ</i>
503	Mitochondrial trifunctional protein deficiency	609015	<i>HADHA</i>
504	Mitochondrial trifunctional protein deficiency	609015	<i>HADHB</i>
505	Mohr-Tranebjaerg syndrome	304700	<i>TIMM8A</i>
506	Mowat-Wilson syndrome	235730	<i>ZEB2</i>
507	Mucopolipidosis type 2	252500	<i>GNPTAB</i>
508	Mucopolipidosis type 3	252600	<i>GNPTAB</i>
509	Mucopolipidosis type 4	252650	<i>MCOLN1</i>
510	Mucopolysaccharidosis Ih	607014	<i>IDUA</i>
511	Mucopolysaccharidosis Ih/s	607015	<i>IDUA</i>
512	Mucopolysaccharidosis Is	607016	<i>IDUA</i>
513	Mucopolysaccharidosis type 2	309900	<i>IDS</i>
514	Mucopolysaccharidosis type 3A (Sanfilippo syndrome type A)	252900	<i>SGSH</i>
515	Mucopolysaccharidosis type 4B	253010	<i>GLBI</i>
516	Mucopolysaccharidosis type 6	253200	<i>ARSB</i>
517	Mucopolysaccharidosis type 7	253220	<i>GUSB</i>
518	Mucopolysaccharidosis type IIIB (Sanfilippo B)	252920	<i>NAGLU</i>
519	MULIBREY nanism	253250	<i>TRIM37</i>
520	Multiple epiphyseal dysplasia type 4	226900	<i>SLC26A2</i>
521	Multiple pterygium syndrome, lethal type	253290	<i>CHRNA1</i>
522	Multiple pterygium syndrome, lethal type	253290	<i>CHRND</i>
523	Multiple pterygium syndrome, lethal type	253290	<i>CHRNA1</i>
524	Muscle-eye-brain disease	613153	<i>FKRP</i>
525	Muscle-eye-brain disease	613154	<i>LARGE</i>
526	Myasthenia gravis, neonatal transient	100730	<i>CHRNA1</i>
527	Myasthenia, limb-girdle, familial	254300	<i>DOK7</i>
528	Myasthenic syndrome, fast-channel congenital	608930	<i>CHRNA1</i>
529	Myasthenic syndrome, fast-channel congenital	608930	<i>CHRND</i>
530	Myasthenic syndrome, slow-channel congenital	601462	<i>CHRNA1</i>
531	Myasthenic syndrome, slow-channel congenital	601462	<i>CHRND</i>
532	Myopathy, tubular aggregate, 1	160565	<i>STIM1</i>
533	Myopathy, tubular aggregate, 2	615883	<i>ORAI1</i>
534	Nance-Horan syndrome	302350	<i>NHS</i>
535	Navajo neurohepatopathy	256810	<i>MPV17</i>
536	Nemaline myopathy 2	256030	<i>NEB</i>

537	Neonatal adrenoleukodystrophy (gene PEX12)	266510	<i>PEX12</i>
538	Neonatal adrenoleukodystrophy (gene PEX26)	614873	<i>PEX26</i>
539	Neonatal adrenoleukodystrophy (gene PEX5)	202370	<i>PEX5</i>
540	Nephrolithiasis, type I	310468	<i>CLCN5</i>
541	Nephronophthisis 2, infantile	602088	<i>INVS</i>
542	Nephrotic syndrome, tupe 3	610725	<i>PLCE1</i>
543	Nephrotic syndrome, type 1	256300	<i>NPHS1</i>
544	Nephrotic syndrome, type 2	600995	<i>NPHS2</i>
545	Nephrotic syndrome, type 5, with or without ocular abnormalities	614199	<i>LAMB2</i>
546	Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency	250620	<i>HIBCH</i>
547	Neurodegeneration due to cerebral folate transport deficiency	613068	<i>FOLR1</i>
548	Neuronal ceroid lipofuscinosis 2	204500	<i>TPPI1</i>
549	Neuropathy, congenital hypomyelinating	605253	<i>MPZ</i>
550	Neutropenia, severe congenital 3, autosomal recessive	610738	<i>HAX1</i>
551	Niemann-Pick disease type A	257200	<i>SMPD1</i>
552	Niemann-Pick disease type B	607616	<i>SMPD1</i>
553	Niemann-Pick disease type C1	257220	<i>NPC1</i>
554	Niemann-Pick disease type C2	607625	<i>NPC2</i>
555	Nijmegen breakage syndrome	251260	<i>NBN</i>
556	Norrie disease	310600	<i>NDP</i>
557	ntal retardation, autosomal recessive 7	611093	<i>TUSC3</i>
558	Occipital horn syndrome	304150	<i>ATP7A</i>
559	Oculocerebrorenal syndrome	309000	<i>OCRL</i>
560	Omenn syndrome	603554	<i>DCLRE1C</i>
561	Omenn syndrome (gene RAG1)	603554	<i>RAG1</i>
562	Omenn syndrome (gene RAG2)	603554	<i>RAG2</i>
563	Opitz GBBB syndrome, type I	300000	<i>MIDI</i>
564	Ornithine transcarbamilase deficiency	311250	<i>OTC</i>
565	Osteogenesis imperfecta type 8	610915	<i>LEPRE1</i>
566	Osteogenesis imperfecta type VII	610682	<i>CRTAP</i>
567	Osteogenesis imperfecta, type I	166200	<i>COL1A1</i>
568	Osteogenesis imperfecta, type II	166210	<i>COL1A1</i>
569	Osteogenesis imperfecta, type III	259420	<i>COL1A1</i>
570	Osteogenesis imperfecta, type IV	166220	<i>COL1A1</i>
571	Osteopetrosis with renal tubular acidosis	259730	<i>CA2</i>
572	Osteopetrosis, autosomal recessive 5	259720	<i>OSTM1</i>
573	Paget disease, juvenile	239000	<i>TNFRSF11B</i>
574	Panhypopituitarism, X-linked	312000	<i>SOX3</i>
575	Pantothenate kinase-associated neurodegeneration	234200	<i>PANK2</i>

576	Partial androgen insensitivity syndrome	312300	<i>AR</i>
577	Pelizaeus-Merzbacher-like due to GJC2 mutation	608804	<i>GJC2</i>
578	Peroxisomal acyl-CoA oxidase deficiency	264470	<i>ACOX1</i>
579	Peroxisome biogenesis disorder 11A (Zellweger)	614883	<i>PEX13</i>
580	Peroxisome biogenesis disorder 11B	614885	<i>PEX13</i>
581	Peroxisome biogenesis disorder 6A (Zellweger)	614870	<i>PEX10</i>
582	Peroxisome biogenesis disorder 6B	614871	<i>PEX10</i>
583	Perrault syndrome	233400	<i>HSD17B4</i>
584	Phenylketonuria	261600	<i>PAH</i>
585	Pierson syndrome	609049	<i>LAMB2</i>
586	Pitt-Hopkins syndrome	610954	<i>TCF4</i>
587	Plasminogen deficiency type 1	217090	<i>PLG</i>
588	Pontocerebellar hypoplasia type 2A	277470	<i>TSEN54</i>
589	Pontocerebellar hypoplasia type 4	225753	<i>TSEN54</i>
590	Porphyria, congenital erythropoietic	263700	<i>UROS</i>
591	Precocious puberty, male	176410	<i>LHCGR</i>
592	Primary lateral sclerosis, juvenile	606353	<i>ALS2</i>
593	Progressive epilepsy - intellectual deficit, Finnish type	610003	<i>CLN8</i>
594	Properdin deficiency, X-linked	312060	<i>CFP</i>
595	Propionic acidemia (gene PCCA)	606054	<i>PCCA</i>
596	Propionic acidemia (gene PCCB)	606054	<i>PCCB</i>
597	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis 308990		<i>CLCN5</i>
598	Proximal spinal muscular atrophy type 1	253300	<i>SMN1</i>
599	Proximal spinal muscular atrophy type 2	253550	<i>SMN1</i>
600	Proximal spinal muscular atrophy type 3	253400	<i>SMN1</i>
601	Proximal spinal muscular atrophy type 4	271150	<i>SMN1</i>
602	Pseudohermaphroditism, male, with gynecomastia	264300	<i>HSD17B3</i>
603	Pseudohypoaldosteronism type 1, autosomal recessive (gene SCNN1A)	264350	<i>SCNN1A</i>
604	Pseudohypoaldosteronism type 1, autosomal recessive (gene SCNN1B)	264350	<i>SCNN1B</i>
605	Pseudohypoaldosteronism type 1, autosomal recessive (gene SCNN1G)	264350	<i>SCNN1G</i>
606	Pseudovaginal perineoscrotal hypospadias	264600	<i>SRD5A2</i>
607	Pycnodysostosis	265800	<i>CTSK</i>
608	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency	612260	<i>MYD88</i>
609	Pyridoxal phosphate-responsive seizures	610090	<i>PNPO</i>
610	Pyruvate carboxylase deficiency	266150	<i>PC</i>
611	Pyruvate dehydrogenase phosphatase deficiency	608782	<i>PDPI</i>
612	Renal-hepatic-pancreatic dysplasia	208540	<i>NPHP3</i>
613	Renpenning syndrome	309500	<i>PQBP1</i>
614	Rett syndrome, congenital variant	613454	<i>FOXG1</i>



615	Rhizomelic chondrodysplasia punctata type 1	215100	<i>PEX7</i>
616	Rhizomelic chondrodysplasia punctata type 3	600121	<i>AGPS</i>
617	Rigid spine syndrome	602771	<i>SEPNI</i>
618	Roberts syndrome	269000	<i>ESCO2</i>
619	Roussy-Levy syndrome	180800	<i>MPZ</i>
620	Roussy-Levy syndrome	180800	<i>PMP22</i>
621	Sandhoff disease	268800	<i>HEXB</i>
622	Sanfilippo syndrome type C	252930	<i>HGSNAT</i>
623	Schneckenbecken dysplasia	269250	<i>SLC35D1</i>
624	Schwartz-Jampel syndrome	255800	<i>HSPG2</i>
625	Seckel syndrome	210600	<i>ATR</i>
626	Senior-Loken syndrome	610189	<i>CEP290</i>
627	Senior-Loken syndrome	606996	<i>NPHP4</i>
628	Senior-Loken syndrome 1	266900	<i>NPHP3</i>
629	Senior-Loken syndrome 5	609254	<i>IQCB1</i>
630	Sensory ataxic neuropathy - dysarthria - ophthalmoparesis	607459	<i>POLG</i>
631	Severe combined immunodeficiency due to adenosine deaminase deficiency	102700	<i>ADA</i>
632	Severe combined immunodeficiency due to complete RAG1/2 deficiency	601457	<i>RAG1</i>
633	Severe combined immunodeficiency due to complete RAG1/2 deficiency	601457	<i>RAG2</i>
634	Severe combined immunodeficiency due to DCLRE1C deficiency	602450	<i>DCLRE1C</i>
635	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	611291	<i>NHEJ1</i>
636	Severe combined immunodeficiency with sensitivity to ionizing radiation	602450	<i>LIG4</i>
637	Severe generalized recessive dystrophic epidermolysis bullosa	226600	<i>COL7A1</i>
638	Severe neonatal-onset encephalopathy with microcephaly	300673	<i>MECP2</i>
639	Severe T-cell immunodeficiency - congenital alopecia - nail dystrophy	601705	<i>FOXN1</i>
640	Short-rib thoracic dysplasia 3 with or without polydactyly	613091	<i>DYNC2H1</i>
641	Shwachman-Diamond syndrome	260400	<i>SBDS</i>
642	Sialidosis, type I	256550	<i>NEU1</i>
643	Sialidosis, type II	256550	<i>NEU1</i>
644	Sickle cell anemia	603903	<i>HBB</i>
645	Simpson-Golabi-Behmel syndrome type 2	300209	<i>OFD1</i>
646	Simpson-Golabi-Behmel syndrome, type 1	312870	<i>GPC3</i>
647	Síndrome de Dursun	612541	<i>G6PC3</i>
648	Sjogren-Larsson syndrome	270200	<i>ALDH3A2</i>
649	Smith-Lemli-Opitz syndrome	270400	<i>DHCR7</i>
650	Sotos syndrome 1	117550	<i>NSD1</i>
651	Spastic paralysis, infantile onset ascending	607225	<i>ALS2</i>
652	Spastic paraplegia type 2, X-linked	312920	<i>PLP1</i>
653	Spinal muscular atrophy with respiratory distress	604320	<i>IGHMBP2</i>

654	Stocco dos Santos X-linked mental retardation syndrome	300434	<i>SHROOM4</i>
655	Stormorken syndrome	185070	<i>STIMI</i>
656	Stüve-Wiedemann syndrome	601559	<i>LIFR</i>
657	Subcortical laminal heteropia, X-linked	300067	<i>DCX</i>
658	Succinyl CoA:3-oxoacid CoA transferase deficiency	245050	<i>OXCT1</i>
659	Sudden infant death with dysgenesis of the testes syndrome	608800	<i>TSPYL1</i>
660	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A (gene MOCS1)	252150	<i>MOCS1</i>
661	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A (gene MOCS2)	252150	<i>MOCS2</i>
662	Sulfocysteinuria	272300	<i>SUOX</i>
663	Surfactant metabolism dysfunction, pulmonary, 1	265120	<i>SFTPB</i>
664	Surfactant metabolism dysfunction, pulmonary, 2	610913	<i>SFTPC</i>
665	Surfactant metabolism dysfunction, pulmonary, 3	610921	<i>ABCA3</i>
666	Syndromic microphthalmia type 9	601186	<i>STRA6</i>
667	Tay-Sachs disease	272800	<i>HEXA</i>
668	T-B+ severe combined immunodeficiency due to gamma chain deficiency	300400	<i>IL2RG</i>
669	T-B+ severe combined immunodeficiency due to JAK3 deficiency	600802	<i>JAK3</i>
670	T-B+ severe combined immunodeficiency, X-linked	312863	<i>IL2RG</i>
671	Tetra-amelia, autosomal recessive	273395	<i>WNT3</i>
672	Thrombocythemia 2	601977	<i>MPL</i>
673	Thrombocytopenia, congenital amegakaryocytic	604498	<i>MPL</i>
674	Thrombotic thrombocytopenic purpura, familial	274150	<i>ADAMTS13</i>
675	Tooth agenesis, selective, X-linked 1	313500	<i>EDA</i>
676	Trichothiodystrophy, complementation group A	601675	<i>GTF2H5</i>
677	Tyrosinemia type 1	276700	<i>FAH</i>
678	Tyrosinemia type 2	276600	<i>TAT</i>
679	Tyrosinemia type 3	276710	<i>HPD</i>
680	Ullrich congenital muscular dystrophy	254090	<i>COL6A1</i>
681	Ullrich congenital muscular dystrophy	254090	<i>COL6A2</i>
682	Ullrich congenital muscular dystrophy	254090	<i>COL6A3</i>
683	Unverricht-Lundborg disease	254800	<i>CSTB</i>
684	Usher syndrome type 1	276900	<i>MYO7A</i>
685	Usher syndrome type 1C	276904	<i>USH1C</i>
686	Usher syndrome type 1G	606943	<i>USH1G</i>
687	Usher syndrome type 2A	276901	<i>USH2A</i>
688	Usher syndrome type 2C	605472	<i>GPR98</i>
689	Usher syndrome type 3A	276902	<i>CLRN1</i>
690	Very long chain acyl-CoA dehydrogenase deficiency	201475	<i>ACADVL</i>
691	Vitamin B12-responsive methylmalonic acidemia type cblA	251100	<i>MMAA</i>

692	Vitamin B12-responsive methylmalonic acidemia type cblB	251110	<i>MMAB</i>
693	Vitamin B12-unresponsive methylmalonic acidemia type mut-	251000	<i>MUT</i>
694	Vitamin D-dependent rickets type 2A	277440	<i>VDR</i>
695	Vitamin D-dependent rickets, type I	264700	<i>CYP27B1</i>
696	Waardenburg-Shah syndrome 4A	277580	<i>EDNRB</i>
697	Waardenburg-Shah syndrome 4B	613265	<i>EDN3</i>
698	Walker-Warburg syndrome (gene POMGNT1)	253280	<i>POMGNT1</i>
699	Walker-Warburg syndrome (gene POMT1)	236670	<i>POMT1</i>
700	Walker-Warburg syndrome (gene POMT2)	613150	<i>POMT2</i>
701	Weyers acrodistal dysostosis	193530	<i>EVC</i>
702	Wilson disease	277900	<i>ATP7B</i>
703	Wiskott-Aldrich syndrome	301000	<i>WAS</i>
704	Wolcott-Rallison syndrome	226980	<i>EIF2AK3</i>
705	Wrinkly skin syndrome	278250	<i>ATP6V0A2</i>
706	Xeroderma pigmentosum complementation group A	278700	<i>XPA</i>
707	Xeroderma pigmentosum complementation group E	278740	<i>DDB2</i>
708	Xeroderma pigmentosum, group C	278720	<i>XPC</i>
709	Xeroderma pigmentosum/Cockayne syndrome complex complementation group B	610651	<i>ERCC3</i>
710	Xeroderma pigmentosum/Cockayne syndrome complex complementation group D	278730	<i>ERCC2</i>
711	Xeroderma pigmentosum/Cockayne syndrome complex complementation group F	278760	<i>ERCC4</i>
712	Xeroderma pigmentosum/Cockayne syndrome complex complementation group G	278780	<i>ERCC5</i>
713	X-linked agammaglobulinemia	300755	<i>BTK</i>
714	X-linked centronuclear myopathy	310400	<i>MTM1</i>
715	X-linked Charcot-Marie-Tooth disease type 5	311070	<i>PRPS1</i>
716	X-linked creatine transporter deficiency	300352	<i>SLC6A8</i>
717	X-linked distal spinal muscular atrophy	300489	<i>ATP7A</i>
718	X-linked hyper-IgM syndrome	308230	<i>CD40LG</i>
719	X-linked intellectual deficit with marfanoid habitus	309520	<i>MED12</i>
720	X-linked lymphoproliferative disease	308240	<i>SH2D1A</i>
721	X-linked severe congenital neutropenia	300299	<i>WNT10A</i>
722	X-linked spinal muscular atrophy type 2	301830	<i>UBA1</i>
723	Zellweger syndrome 1A	214100	<i>PEX1</i>
724	Zellweger syndrome 7A	614872	<i>PEX26</i>