GeneScreen®: Elenco dei geni analizzati e malattie genetiche investigate

	DISEASE NAME	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	Achalasia-addisonianism-alacrimia syndrome	231550	*AAAS*
12	Achondrogenesis type 1B	600972	*SLC26A2*
13	Acyl-CoA dehydrogenase 9 deficiency	611126	*ACAD9*
14	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	202010	*CYP11B1*
15	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	613743	*CYP11A1*
16	Adrenocortical insufficiency	612965	*NR5A1*
17	Adrenoleukodystrophy	300100	*ABCD1*
18	Adult neuronal ceroid lipofuscinosis	256730	*PPT1*
19	Adult neuronal ceroid lipofuscinosis 10	610127	*CTSD*
20	Adult neuronal ceroid lipofuscinosis 4A	204300	*CLN6*
21	Aicardi-Goutières syndrome	225750	*TREX1*
22	Aicardi-Goutieres syndrome 2	610181	*RNASEH2B*
23	Aicardi-Goutieres syndrome 3	610329	*RNASEH2C*
24	Aicardi-Goutieres syndrome 4	610333	*RNASEH2A*
25	Aicardi-Goutieres syndrome 5	612952	*SAMHD1*
26	Aldosteronism, glucocorticoid-remediable	103900	*CYP11B1*
27	Allan-Herndon-Dudley syndrome	300523	*SLC16A2*
28	Alpers syndrome	203700	*POLG*
29	Alpha-methylacyl-Coa Racemase deficiency	614307	*AMACR*
30	Alpha-thalassemia	604131	*HBA1*
31	Alpha-thalassemia myelodysplasia syndrome, somatic	300448	*ATRX*
32	Alpha-thalassemia/mental retardation syndrome	301040	*ATRX*
33	Alport syndrome	301050	*COL4A5*
34	Alport syndrome autosomal recessive (gene COL4A3)	203780	*COL4A3*
35	Alport syndrome autosomal recessive (gene COL4A4)	203780	*COL4A4*
36	Alström syndrome	203800	*ALMS1*

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CHIAMATA GRATUITA
NUMEROVERDE
800-501651

37	Amish infantile epilepsy syndrome	609056	*ST3GAL5*
38	Amyotrophic lateral sclerosis 2, juvenile	205100	*ALS2*
39	Anauxetic dysplasia	607095	*RMRP*
40	Angelman syndrome	105830	*UBE3A*
41	Antenatal Bartter syndrome	241200	*KCNJ1*
42	Antenatal Bartter syndrome type 1	601678	*SLC12A1*
43	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	201750	*POR*
44	Aplasia/hypoplasia of limbs and pelvis	276820	*WNT7A*
45	Aplastic anemia	609135	*NBN*
46	Apparent mineralocorticoid excess	218030	*HSD11B2*
47	Argininosuccinic aciduria	207900	*ASL*
48	Aromatic L-amino acid decarboxylase deficiency	608643	*DDC*
49	Arthrogryposis - renal dysfunction - cholestasis	208085	*VPS33B*
50	Arthrogryposis, renal dysfunction, and cholestasis 2	613404	*VIPAR*
51	Ataxia - oculomotor apraxia type 1	208920	*APTX*
52	Ataxia with vitamin E deficiency	277460	*TTPA*
53	Ataxia-telangiectasia	208900	*ATM*
54	Atelosteogenesis type II	256050	*SLC26A2*
55	Autism, susceptibility to, X-linked 5	300847	*RPL10*
56	Autoimmune lymphoproliferative syndrome, type IA	601859	*FAS*
57	Autoimmune lymphoproliferative syndrome, type IB	601859	*FASLG*
58	Autoimmune lymphoproliferative syndrome, type II	603909	*CASP10*
59	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia	240300	*AIRE*
60	Autosomal dominant Charcot-Marie-Tooth disease type 2K	607831	*GDAP1*
61	Autosomal recessive ataxia due to ubiquinone deficiency	612016	*ADCK3*
62	Autosomal recessive Charcot-Marie-Tooth disease with hoarseness	607706	*GDAP1*
63	Autosomal recessive distal spinal muscular atrophy type 4	611067	*PLEKHG5*
64	Autosomal recessive dopa-responsive dystonia	605407	*TH*
65	Autosomal recessive hypophosphatemic rickets 1	241520	*DMP1*
66	Autosomal recessive hypophosphatemic rickets 2	613312	*ENPP1*
67	Autosomal recessive intermediate Charcot-Marie-Tooth disease type A	608340	*GDAP1*
68	Autosomal recessive limb-girdle muscular dystrophy type 2I	607155	*FKRP*
69	Autosomal recessive limb-girdle muscular dystrophy type 2M	611588	*FKTN*
70	Autosomal recessive limb-girdle muscular dystrophy type C	613157	*POMGNT1*
71	Autosomal recessive limb-girdle muscular dystrophy type C	609308	*POMT1*
72	Autosomal recessive limb-girdle muscular dystrophy type C	613158	*POMT2*
73	Autosomal recessive malignant osteopetrosis 1	259700	*TCIRG1*
74	Autosomal recessive malignant osteopetrosis 4	611490	*CLCN7*

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

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

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151	Combined oxidative phosphorylation defect type 5	611719	*MRPS22*
152	Combined oxidative phosphorylation deficiency 4	610678	*TUFM*
153	Combined pituitary hormone deficiencies, genetic forms	182230	*HESX1*
154	Combined pituitary hormone deficiencies, genetic forms	613038	*POU1F1*
155	Combined pituitary hormone deficiencies, genetic forms	262600	*PROP1*
156	Combined pituitary hormone deficiency with spine abnormalities	221750	*LHX3*
157	Complete androgen insensitivity syndrome	300068	*AR*
158	Complex I, mitochondrial respiratory chain, deficiency of	252010	*NDUFS6*
159	Congenital bile acid synthesis defect type 4	214950	*AMACR*
160	Congenital disorder of glycosylation type 1a	212065	*PMM2*
161	Congenital disorder of glycosylation type 1b	602579	*MPI*
162	Congenital disorder of glycosylation type 1e	608799	*DPM1*
163	Congenital disorder of glycosylation type 1j	608093	*DPAGT1*
164	Congenital disorder of glycosylation type 2a	212066	*MGAT2*
165	Congenital disorder of glycosylation type 2c	266265	*SLC35C1*
166	Congenital disorder of glycosylation type 2d	607091	*B4GALT1*
167	Congenital disorder of glycosylation type 2f	603585	*SLC35A1*
168	Congenital disorder of glycosylation type Ic	603147	*ALG6*
169	Congenital disorder of glycosylation type Ik	608540	*ALG1*
170	Congenital disorder of glycosylation, type Id	601110	*ALG3*
171	Congenital disorder of glycosylation, type If	609180	*MPDU1*
172	Congenital disorder of glycosylation, type Ig	607143	*ALG12*
173	Congenital disorder of glycosylation, type Ih	608104	*ALG8*
174	Congenital disorder of glycosylation, type Ii	607906	*ALG2*
175	Congenital disorder of glycosylation, type IIb	606056	*MOGS*
176	Congenital disorder of glycosylation, type IIe	608779	*COG7*
177	Congenital disorder of glycosylation, type IIg	611209	*COG1*
178	Congenital disorder of glycosylation, type IIh	611182	*COG8*
179	Congenital disorder of glycosylation, type Il	608776	*ALG9*
180	Congenital disorder of glycosylation, type Im	610768	*DOLK*
181	Congenital disorder of glycosylation, type In	612015	*RFT1*
182	Congenital disorder of glycosylation, type Iq	612379	*SRD5A3*
183	Congenital fibrinogen deficiency (gene FGA)	202400	*FGA*
184	Congenital heart defects, nonsyndromic, 1, X-linked	306955	*ZIC3*
185	Congenital hereditary endothelial dystrophy type II	217700	*SLC4A11*
186	Congenital lipoid adrenal hyperplasia	201710	*STAR*
187	Congenital malabsorptive diarrhea due to paucity of enteroendocrine cells	610370	*NEUROG3*
188	Congenital muscular dystrophy type 1A	607855	*LAMA2*
189	Congenital muscular dystrophy type 1D	608840	*LARGE*

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190	Congenital muscular dystrophy type 4B	613152	*FKTN*
191	Congenital muscular dystrophy type 5B	606612	*FKRP*
192	Congenital muscular dystrophy with cerebellar involvement	613151	*POMGNT1*
193	Congenital muscular dystrophy with cerebellar involvement	613155	*POMT1*
194	Congenital muscular dystrophy with cerebellar involvement	613156	*POMT2*
195	Corneal dystrophy - perceptive deafness	217400	*SLC4A11*
196	Corpus callosum agenesis - neuronopathy	218000	*SLC12A6*
197	Corpus callosum hypoplasia-retardation-adducted thumbs-spasticity-hydrocephalus syndrome	307000	*L1CAM*
198	Cowden syndrome 1	158350	*PTEN*
199	Craniofrontonasal dysplasia	304110	*EFNB1*
200	Cutis laxa, autosomal dominant 2	614434	*FBLN5*
201	Cutis laxa, autosomal recessive, type IA	219100	*FBLN5*
202	Cutis laxa, autosomal recessive, type IB	614437	*EFEMP2*
203	Cutis laxa, autosomal recessive, type IIA	219200	*ATP6V0A2*
204	Cystic fibrosis; mucoviscidosis	219700	*CFTR*
205	Cystinosis	219800	*CTNS*
206	Deafness - encephaloneuropathy - obesity - valvulopathy	614651	*PDSS1*
207	Dejerine-Sottas disease	145900	*MPZ*
208	Dejerine-Sottas disease	145900	*PMP22*
209	Dent disease	300009	*CLCN5*
210	Dent disease 2	300555	*OCRL*
211	Desmosterolosis	602398	*DHCR24*
212	Diabetes mellitus, noninsulin-dependent	125853	*ABCC8*
213	Diabetes mellitus, permanent neonatal	606176	*ABCC8*
214	Diabetes mellitus, transient neonatal 2	610374	*ABCC8*
215	Diastrophic dwarfism	222600	*SLC26A2*
216	Dihydropyrimidine dehydrogenase deficiency	274270	*DPYD*
217	Dilated cardiomyopathy with ataxia	610198	*DNAJC19*
218	Donnai-Barrow syndrome	222448	*LRP2*
219	Duchenne muscular dystrophy	310200	*DMD*
220	Dyskeratosis congenita X-linked	305000	*DKC1*
221	Dystrophic epidermolysis bullosa pruriginosa	604129	*COL7A1*
222	Early infantile epileptic encephalopathy	308350	*ARX*
223	Early infantile epileptic encephalopathy	609304	*SLC25A22*
224	Ectodermal dysplasia 1, hypohidrotic, X-linked	305100	*EDA*
225	Ectodermal dysplasia, hypohidrotic, with immune deficiency	300291	*IKBKG*
226	Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency	300301	*IKBKG*
227	Ehlers-Danlos syndrome type 6	225400	*PLOD1*
228	Ehlers-Danlos syndrome, cardiac valvular type	225320	*COL1A2*

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

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CHIAMATA GRATUITA
NUMEROVERDE
800-501651

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

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306	Hemophilia A	306700	*F8*
307	Hemophilia B	306900	*F9*
308	Hepatic venoocclusive disease with immunodeficiency	235550	*SP110*
309	Hepatoencephalopathy due to combined oxidative phosphorylation deficiency type 1	609060	*GFM1*
310	Hereditary fructose intolerance	229600	*ALDOB*
311	Hereditary sensory and autonomic neuropathy type 4	256800	*NTRK1*
312	Hermansky-Pudlak syndrome 2	608233	*AP3B1*
313	Hermansky-pudlak syndrome 9	614171	*PLDN*
314	Heterotaxy, visceral, 1, X-linked	306955	*ZIC3*
315	Histidinemia	235800	*HAMP*
316	Holocarboxylase synthetase deficiency	253270	*HLCS*
317	Hoyeraal-Hreidarsson syndrome	300240	*DKC1*
318	Hyaline fibromatosis syndrome	228600	*ANTXR2*
319	Hyperammonemia due to N-acetylglutamate synthetase deficiency	237310	*NAGS*
320	Hyper-IgE recurrent infection syndrome, autosomal recessive	243700	*DOCK8*
321	Hyperinsulinemic hypoglycemia, familial, 1	256450	*ABCC8*
322	Hyperornithinemia-hyperammonemia-homocitrullinuria	238970	*SLC25A15*
323	Hypoglycemia of infancy, leucine-sensitive	240800	*ABCC8*
324	Hypogonadotropic hypogonadism 7 without anosmia	146110	*GNRHR*
325	Hypomyelination - congenital cataract	610532	*FAM126A*
326	Hypoparathyroidism - intellectual deficit - dysmorphism syndrome	241410	*TBCE*
327	Hypophosphatemic rickets	300554	*CLCN5*
328	Ichthyosis follicularis - alopecia - photophobia	308205	*MBTPS2*
329	Ichthyosis, autosomal recessive 4B (harlequin)	242500	*ABCA12*
330	Ichthyosis, congenital, autosomal recessive 1	242300	*TGM1*
331	Ichthyosis, congenital, autosomal recessive 4A	601277	*ABCA12*
332	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis 607626		*CLDN1*
333	Immunodeficiency 10	612783	*STIM1*
334	Immunodeficiency 17, CD3 gamma deficient	615607	*CD3G*
335	Immunodeficiency 18, SCID variant	615615	*CD3E*
336	Immunodeficiency 19	615617	*CD3D*
337	Immunodeficiency 27A, mycobacteriosis, AR	209950	*IFNGR1*
338	Immunodeficiency 28, mycobacteriosis	614889	*IFNGR2*
339	Immunodeficiency 29, mycobacteriosis	614890	*IL12B*
340	Immunodeficiency 30	614891	*IL12RB1*
341	Immunodeficiency 31A, mycobacteriosis, autosomal dominant	614892	*STAT1*
342	Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive	613796	*STAT1*
343	Immunodeficiency 31C, autosomal dominant	614162	*STAT1*

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344	Immunodeficiency 33	300636	*IKBKG*
345	Immunodeficiency 35	611521	*TYK2*
346	Immunodeficiency 9	612782	*ORAI1*
347	Immunodeficiency, common variable, 1	607594	*ICOS*
348	Immunodeficiency, common variable, 3	613493	*CD19*
349	Immunodeficiency-centromeric instability-facial anomalies syndrome 1	242860	*DNMT3B*
350	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	304790	*FOXP3*
351	Incontinentia pigmenti, type II	308300	*IKBKG*
352	Infantile bilateral striatal necrosis	271930	*NUP62*
353	Infantile hypophosphatasia	241500	*ALPL*
354	Infantile neuroaxonal dystrophy 2A	256600	*PLA2G6*
355	Infantile neuroaxonal dystrophy 2B	610217	*PLA2G6*
356	Infantile onset spinocerebellar ataxia	271245	*C10orf2*
357	Interleukin 1 receptor antagonist deficiency	612852	*IL1RN*
358	Isolated CoQ-cytochrome C reductase deficiency	124000	*BCS1L*
359	Isolated growth hormone deficiency type III	307200	*BTK*
360	Isolated thyroid-stimulating hormone deficiency	275100	*TSHB*
361	Isovaleric acidemia	243500	*IVD*
362	Jeune syndrome	611263	*IFT80*
363	Johanson-Blizzard syndrome	243800	*UBR1*
364	Joubert syndrome 4	609583	*NPHP1*
365	Joubert syndrome 6	610688	*TMEM67*
366	Joubert syndrome with hepatic defect	216360	*RPGRIP1L*
367	Joubert syndrome with ocular defect	608629	*AHI1*
368	Joubert syndrome with oculorenal defect 5	610188	*CEP290*
369	Junctional epidermolysis bullosa - pyloric atresia	226730	*ITGA6*
370	Junctional epidermolysis bullosa with piloric atresia	226730	*ITGB4*
371	Junctional epidermolysis bullosa, Herlitz type (gene LAMA3)	226700	*LAMA3*
372	Junctional epidermolysis bullosa, Herlitz type (gene LAMB3)	226700	*LAMA3*
373	Junctional epidermolysis bullosa, Herlitz type (gene LAMC2)	226700	*LAMC2*
374	Junctional epidermolysis bullosa, non-Herlitz type	226650	*ITGB4*
375	Junctional epidermolysis bullosa, non-Herlitz type (gene LAMA3)	226650	*LAMA3*
376	Junctional epidermolysis bullosa, non-Herlitz type (gene LAMB3)	226650	*LAMB3*
377	Junctional epidermolysis bullosa, non-Herlitz type (gene LAMC2)	226650	*LAMC2*
378	Juvenile neuronal ceroid lipofuscinosis 3	204200	*CLN3*
379	Kahrizi syndrome	612713	*SRD5A3*
380	Kelley-Seegmiller syndrome	300323	*HPRT1*
381	Kennedy disease	313200	*AR*
382	Ketoacidosis due to beta-ketothiolase deficiency	203750	*ACAT1*

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

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422	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	609016	*HADH*
423	Luteinizing hormone resistance, female	238320	*LHCGR*
424	Lymphoproliferative syndrome, X-linked, 2	300635	*XIAP*
425	Macrocephaly/autism syndrome	605309	*PTEN*
426	Macroglobulinemia, Waldenstrom	153600	*MYD88*
427	Macular degeneration, age-related, 3	608895	*FBLN5*
428	Mandibuloacral dysplasia with type A lipodystrophy	248370	*LMNA*
429	Mandibuloacral dysplasia with type B lipodystrophy	608612	*ZMPSTE24*
430	Mannosidosis, alpha-, types I and II	248500	*MAN2B1*
431	Maple syrup urine disease	248600	*DLD*
432	Maple syrup urine disease (gene BCKDHA)	248600	*BCKDHA*
433	Maple syrup urine disease (gene BCKDHB)	248600	*BCKDHB*
434	Marinesco-Sjögren syndrome	248800	*SIL1*
435	Masa syndrome	303350	*L1CAM*
436	Meckel syndrome type 1	249000	*MKS1*
437	Meckel syndrome, type 5	611561	*RPGRIP1L*
438	Medium chain acyl-CoA dehydrogenase deficiency	201450	*ACADM*
439	Megalencephalic leukoencephalopathy with subcortical cysts	604004	*MLC1*
440	Menkes disease	309400	*ATP7A*
441	Mental retardation and microcephaly with pontine and cerebellar hypoplasia	300749	*CASK*
442	Mental retardation, autosomal recessive 1	249500	*PRSS12*
443	Mental retardation, autosomal recessive 12	611090	*ST3GAL3*
444	Mental retardation, autosomal recessive 13	613192	*TRAPPC9*
445	Mental retardation, autosomal recessive 5	611091	*NSUN2*
446	Mental retardation, autosomal recessive, 6	611092	*GRIK2*
447	Mental retardation, with or without nystagmus	300422	*CASK*
448	Mental retardation, X-linked	300495	*NLGN4X*
449	Mental retardation, X-linked 19	300844	*RPS6KA3*
450	Mental retardation, X-linked 21/34	300143	*IL1RAPL1*
451	Mental retardation, X-linked 30/47	300558	*PAK3*
452	Mental retardation, X-linked 41	300849	*GDI1*
453	Mental retardation, X-linked 46	300436	*ARHGEF6*
454	Mental retardation, X-linked 63	300387	*ACSL4*
455	Mental retardation, X-linked 72	300271	*RAB39B*
456	Mental retardation, X-linked 9	309549	*FTSJ1*
457	Mental retardation, X-linked 90	300850	*DLG3*
458	Mental retardation, X-linked 93	300659	*BRWD3*
459	Mental retardation, X-linked 96	300802	*SYP*
460	Mental retardation, X-linked 97	300803	*ZNF711*

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461	Mental retardation, X-linked syndromic 16	305400	*FGD1*
462	Mental retardation, X-linked syndromic 5	304340	*AP1S2*
463	Mental retardation, X-linked syndromic, Christianson type	300243	*SLC9A6*
464	Mental retardation, X-linked syndromic, Nascimento-type	300860	*UBE2A*
465	Mental retardation, X-linked syndromic, Raymond type	300799	*ZDHHC9*
466	Mental retardation, X-linked syndromic, Turner type	300706	*HUWE1*
467	Mental retardation, X-linked, FRAXE type	309548	*AFF2*
468	Mental retardation, X-linked, Snyder-Robinson type	309583	*SMS*
469	Mental retardation, X-linked, syndromic 14	300676	*UPF3B*
470	Mental retardation, X-linked, syndromic 15 (Cabezas type)	300354	*CUL4B*
471	Mental retardation, X-linked, syndromic, Claes-Jensen type	300534	*KDM5C*
472	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	300486	*OPHN1*
473	Mental retardation, X-linked, with isolated growth hormone deficiency	300123	*SOX3*
474	Mental retardation-hypotonic facies syndrome, X-linked	309580	*ATRX*
475	Metachromatic leukodystrophy	250100	*ARSA*
476	Metachromatic leukodystrophy	249900	*PSAP*
477	Metaphyseal chondrodysplasia, Murk Jansen type	156400	*PTH1R*
478	Metaphyseal dysplasia without hypotrichosis	250460	*RMRP*
479	Methylmalonic acidemia with homocystinuria, type cblC	277400	*MMACHC*
480	Methylmalonic acidemia with homocystinuria, type cblD	277410	*MMACHC*
481	Mevalonic aciduria	610377	*MVK*
482	Micro syndrome	600118	*RAB3GAP1*
483	Microphthalmia, syndromic 2	300166	*BCOR*
484	Mitochondrial complex I deficiency	252010	*NDUFA1*
485	Mitochondrial complex I deficiency	252010	*NDUFAF2*
486	Mitochondrial complex I deficiency	252010	*NDUFAF4*
487	Mitochondrial complex I deficiency	252010	*NDUFS3*
488	Mitochondrial complex I deficiency	252010	*NDUFS4*
489	Mitochondrial complex I deficiency	252010	*NDUFV1*
490	Mitochondrial complex IV deficiency	220110	*COX10*
491	Mitochondrial complex IV deficiency	220110	*COX6B1*
492	Mitochondrial complex IV deficiency	220110	*FASTKD2*
493	Mitochondrial complex IV deficiency		*SCO1*
494	Mitochondrial DNA depletion syndrome 1 (MNGIE type)	603041	*TYMP*
495	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	612073	*SUCLA2*
496	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)	612075	*RRM2B*
497	Mitochondrial DNA depletion syndrome 8B (MNGIE type)	612075	*RRM2B*
498	Mitochondrial DNA depletion syndrome, hepatocerebral form due to	251880	*DGUOK*

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	DGUOK deficiency 3		
499	**Mitochondrial DNA depletion syndrome, myopathic form**	**609560**	***TK2***
500	**Mitochondrial neurogastrointestinal encephalomyopathy**	**613662**	***POLG***
501	**Mitochondrial respiratory chain complex III deficiency**	**124000**	***UQCRB***
502	**Mitochondrial respiratory chain complex III deficiency**	**124000**	***UQCRQ***
503	**Mitochondrial trifunctional protein deficiency**	**609015**	***HADHA***
504	**Mitochondrial trifunctional protein deficiency**	**609015**	***HADHB***
505	**Mohr-Tranebjaerg syndrome**	**304700**	***TIMM8A***
506	**Mowat-Wilson syndrome**	**235730**	***ZEB2***
507	**Mucolipidosis type 2**	**252500**	***GNPTAB***
508	**Mucolipidosis type 3**	**252600**	***GNPTAB***
509	**Mucolipidosis type 4**	**252650**	***MCOLN1***
510	**Mucopolysaccharidosis Ih**	**607014**	***IDUA***
511	**Mucopolysaccharidosis Ih/s**	**607015**	***IDUA***
512	**Mucopolysaccharidosis Is**	**607016**	***IDUA***
513	**Mucopolysaccharidosis type 2**	**309900**	***IDS***
514	**Mucopolysaccharidosis type 3A (Sanfilippo syndrome type A)**	**252900**	***SGSH***
515	**Mucopolysaccharidosis type 4B**	**253010**	***GLB1***
516	**Mucopolysaccharidosis type 6**	**253200**	***ARSB***
517	**Mucopolysaccharidosis type 7**	**253220**	***GUSB***
518	**Mucopolysaccharidosis type IIIB (Sanfilippo B)**	**252920**	***NAGLU***
519	**MULIBREY nanism**	**253250**	***TRIM37***
520	**Multiple epiphyseal dysplasia type 4**	**226900**	***SLC26A2***
521	**Multiple pterygium syndrome, lethal type**	**253290**	***CHRNA1***
522	**Multiple pterygium syndrome, lethal type**	**253290**	***CHRND***
523	**Multiple pterygium syndrome, lethal type**	**253290**	***CHRNG***
524	**Muscle-eye-brain disease**	**613153**	***FKRP***
525	**Muscle-eye-brain disease**	**613154**	***LARGE***
526	**Myasthenia gravis, neonatal transient**	**100730**	***CHRNG***
527	**Myasthenia, limb-girdle, familial**	**254300**	***DOK7***
528	**Myasthenic syndrome, fast-channel congenital**	**608930**	***CHRNA1***
529	**Myasthenic syndrome, fast-channel congenital**	**608930**	***CHRND***
530	**Myasthenic syndrome, slow-channel congenital**	**601462**	***CHRNA1***
531	**Myasthenic syndrome, slow-channel congenital**	**601462**	***CHRND***
532	**Myopathy, tubular aggregate, 1**	**160565**	***STIM1***
533	**Myopathy, tubular aggregate, 2**	**615883**	***ORAI1***
534	**Nance-Horan syndrome**	**302350**	***NHS***
535	**Navajo neurohepatopathy**	**256810**	***MPV17***
536	**Nemaline myopathy 2**	**256030**	***NEB***

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CHIAMATA GRATUITA
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537	Neonatal adrenoleukodystrophy (gene PEX12)	266510	*PEX12*
538	Neonatal adrenoleukodystrophy (gene PEX26)	614873	*PEX26*
539	Neonatal adrenoleukodystrophy (gene PEX5)	202370	*PEX5*
540	Nephrolithiasis, type I	310468	*CLCN5*
541	Nephronophthisis 2, infantile	602088	*INVS*
542	Nephrotic syndrome, tupe 3	610725	*PLCE1*
543	Nephrotic syndrome, type 1	256300	*NPHS1*
544	Nephrotic syndrome, type 2	600995	*NPHS2*
545	Nephrotic syndrome, type 5, with or without ocular abnormalities	614199	*LAMB2*
546	Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency	250620	*HIBCH*
547	Neurodegeneration due to cerebral folate transport deficiency	613068	*FOLR1*
548	Neuronal ceroid lipofuscinosis 2	204500	*TPP1*
549	Neuropathy, congenital hypomyelinating	605253	*MPZ*
550	Neutropenia, severe congenital 3, autosomal recessive	610738	*HAX1*
551	Niemann-Pick disease type A	257200	*SMPD1*
552	Niemann-Pick disease type B	607616	*SMPD1*
553	Niemann-Pick disease type C1	257220	*NPC1*
554	Niemann-Pick disease type C2	607625	*NPC2*
555	Nijmegen breakage syndrome	251260	*NBN*
556	Norrie disease	310600	*NDP*
557	ntal retardation, autosomal recessive 7	611093	*TUSC3*
558	Occipital horn syndrome	304150	*ATP7A*
559	Oculocerebrorenal syndrome	309000	*OCRL*
560	Omenn syndrome	603554	*DCLRE1C*
561	Omenn syndrome (gene RAG1)	603554	*RAG1*
562	Omenn syndrome (gene RAG2)	603554	*RAG2*
563	Opitz GBBB syndrome, type I	300000	*MID1*
564	Ornithine transcarbamylase deficiency	311250	*OTC*
565	Osteogenesis imperfecta type 8	610915	*LEPRE1*
566	Osteogenesis imperfecta type VII	610682	*CRTAP*
567	Osteogenesis imperfecta, type I	166200	*COL1A1*
568	Osteogenesis imperfecta, type II	166210	*COL1A1*
569	Osteogenesis imperfecta, type III	259420	*COL1A1*
570	Osteogenesis imperfecta, type IV	166220	*COL1A1*
571	Osteopetrosis with renal tubular acidosis	259730	*CA2*
572	Osteopetrosis, autosomal recessive 5	259720	*OSTM1*
573	Paget disease, juvenile	239000	*TNFRSF11B*
574	Panhypopituitarism, X-linked	312000	*SOX3*
575	Pantothenate kinase-associated neurodegeneration	234200	*PANK2*

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576	Partial androgen insensitivity syndrome	312300	*AR*
577	Pelizaeus-Merzbacher-like due to GJC2 mutation	608804	*GJC2*
578	Peroxisomal acyl-CoA oxidase deficiency	264470	*ACOX1*
579	Peroxisome biogenesis disorder 11A (Zellweger)	614883	*PEX13*
580	Peroxisome biogenesis disorder 11B	614885	*PEX13*
581	Peroxisome biogenesis disorder 6A (Zellweger)	614870	*PEX10*
582	Peroxisome biogenesis disorder 6B	614871	*PEX10*
583	Perrault syndrome	233400	*HSD17B4*
584	Phenylketonuria	261600	*PAH*
585	Pierson syndrome	609049	*LAMB2*
586	Pitt-Hopkins syndrome	610954	*TCF4*
587	Plasminogen deficiency type 1	217090	*PLG*
588	Pontocerebellar hypoplasia type 2A	277470	*TSEN54*
589	Pontocerebellar hypoplasia type 4	225753	*TSEN54*
590	Porphyria, congenital erythropoietic	263700	*UROS*
591	Precocious puberty, male	176410	*LHCGR*
592	Primary lateral sclerosis, juvenile	606353	*ALS2*
593	Progressive epilepsy - intellectual deficit, Finnish type	610003	*CLN8*
594	Properdin deficiency, X-linked	312060	*CFP*
595	Propionic acidemia (gene PCCA)	606054	*PCCA*
596	Propionic acidemia (gene PCCB)	606054	*PCCB*
597	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis 308990		*CLCN5*
598	Proximal spinal muscular atrophy type 1	253300	*SMN1*
599	Proximal spinal muscular atrophy type 2	253550	*SMN1*
600	Proximal spinal muscular atrophy type 3	253400	*SMN1*
601	Proximal spinal muscular atrophy type 4	271150	*SMN1*
602	Pseudohermaphroditism, male, with gynecomastia	264300	*HSD17B3*
603	Pseudohypoaldosteronism type 1, autosomal recessive (gene SCNN1A)	264350	*SCNN1A*
604	Pseudohypoaldosteronism type 1, autosomal recessive (gene SCNN1B)	264350	*SCNN1B*
605	Pseudohypoaldosteronism type 1, autosomal recessive (gene SCNN1G)	264350	*SCNN1G*
606	Pseudovaginal perineoscrotal hypospadias	264600	*SRD5A2*
607	Pycnodysostosis	265800	*CTSK*
608	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency	612260	*MYD88*
609	Pyridoxal phosphate-responsive seizures	610090	*PNPO*
610	Pyruvate carboxylase deficiency	266150	*PC*
611	Pyruvate dehydrogenase phosphatase deficiency	608782	*PDP1*
612	Renal-hepatic-pancreatic dysplasia	208540	*NPHP3*
613	Renpenning syndrome	309500	*PQBP1*
614	Rett syndrome, congenital variant	613454	*FOXG1*

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615	Rhizomelic chondrodysplasia punctata type 1	215100	*PEX7*
616	Rhizomelic chondrodysplasia punctata type 3	600121	*AGPS*
617	Rigid spine syndrome	602771	*SEPN1*
618	Roberts syndrome	269000	*ESCO2*
619	Roussy-Levy syndrome	180800	*MPZ*
620	Roussy-Levy syndrome	180800	*PMP22*
621	Sandhoff disease	268800	*HEXB*
622	Sanfilippo syndrome type C	252930	*HGSNAT*
623	Schneckenbecken dysplasia	269250	*SLC35D1*
624	Schwartz-Jampel syndrome	255800	*HSPG2*
625	Seckel syndrome	210600	*ATR*
626	Senior-Loken syndrome	610189	*CEP290*
627	Senior-Loken syndrome	606996	*NPHP4*
628	Senior-Loken syndrome 1	266900	*NPHP3*
629	Senior-Loken syndrome 5	609254	*IQCB1*
630	Sensory ataxic neuropathy - dysarthria - ophthalmoparesis	607459	*POLG*
631	Severe combined immunodeficiency due to adenosine deaminase deficiency	102700	*ADA*
632	Severe combined immunodeficiency due to complete RAG1/2 deficiency	601457	*RAG1*
633	Severe combined immunodeficiency due to complete RAG1/2 deficiency	601457	*RAG2*
634	Severe combined immunodeficiency due to DCLRE1C deficiency	602450	*DCLRE1C*
635	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	611291	*NHEJ1*
636	Severe combined immunodeficiency with sensitivity to ionizing radiation	602450	*LIG4*
637	Severe generalized recessive dystrophic epidermolysis bullosa	226600	*COL7A1*
638	Severe neonatal-onset encephalopathy with microcephaly	300673	*MECP2*
639	Severe T-cell immunodeficiency - congenital alopecia - nail dystrophy	601705	*FOXN1*
640	Short-rib thoracic dysplasia 3 with or without polydactyly	613091	*DYNC2H1*
641	Shwachman-Diamond syndrome	260400	*SBDS*
642	Sialidosis, type I	256550	*NEU1*
643	Sialidosis, type II	256550	*NEU1*
644	Sickle cell anemia	603903	*HBB*
645	Simpson-Golabi-Behmel syndrome type 2	300209	*OFD1*
646	Simpson-Golabi-Behmel syndrome, type 1	312870	*GPC3*
647	Síndrome de Dursun	612541	*G6PC3*
648	Sjogren-Larsson syndrome	270200	*ALDH3A2*
649	Smith-Lemli-Opitz syndrome	270400	*DHCR7*
650	Sotos syndrome 1	117550	*NSD1*
651	Spastic paralysis, infantile onset ascending	607225	*ALS2*
652	Spastic paraplegia type 2, X-linked	312920	*PLP1*
653	Spinal muscular atrophy with respiratory distress	604320	*IGHMBP2*

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654	Stocco dos Santos X-linked mental retardation syndrome	300434	*SHROOM4*
655	Stormorken syndrome	185070	*STIM1*
656	Stüve-Wiedemann syndrome	601559	*LIFR*
657	Subcortical laminal heteropia, X-linked	300067	*DCX*
658	Succinyl CoA:3-oxoacid CoA transferase deficiency	245050	*OXCT1*
659	Sudden infant death with dysgenesis of the testes syndrome	608800	*TSPYL1*
660	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A (gene MOCS1)	252150	*MOCS1*
661	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A (gene MOCS2)	252150	*MOCS2*
662	Sulfocysteinuria	272300	*SUOX*
663	Surfactant metabolism dysfunction, pulmonary, 1	265120	*SFTPB*
664	Surfactant metabolism dysfunction, pulmonary, 2	610913	*SFTPC*
665	Surfactant metabolism dysfunction, pulmonary, 3	610921	*ABCA3*
666	Syndromic microphthalmia type 9	601186	*STRA6*
667	Tay-Sachs disease	272800	*HEXA*
668	T-B+ severe combined immunodeficiency due to gamma chain deficiency	300400	*IL2RG*
669	T-B+ severe combined immunodeficiency due to JAK3 deficiency	600802	*JAK3*
670	T-B+ severe combined immunodeficiency, X-linked	312863	*IL2RG*
671	Tetra-amelia, autosomal recessive	273395	*WNT3*
672	Thrombocythemia 2	601977	*MPL*
673	Thrombocytopenia, congenital amegakaryocytic	604498	*MPL*
674	Thrombotic thrombocytopenic purpura, familial	274150	*ADAMTS13*
675	Tooth agenesis, selective, X-linked 1	313500	*EDA*
676	Trichothiodystrophy, complementation group A	601675	*GTF2H5*
677	Tyrosinemia type 1	276700	*FAH*
678	Tyrosinemia type 2	276600	*TAT*
679	Tyrosinemia type 3	276710	*HPD*
680	Ullrich congenital muscular dystrophy	254090	*COL6A1*
681	Ullrich congenital muscular dystrophy	254090	*COL6A2*
682	Ullrich congenital muscular dystrophy	254090	*COL6A3*
683	Unverricht-Lundborg disease	254800	*CSTB*
684	Usher syndrome type 1	276900	*MYO7A*
685	Usher syndrome type 1C	276904	*USH1C*
686	Usher syndrome type 1G	606943	*USH1G*
687	Usher syndrome type 2A	276901	*USH2A*
688	Usher syndrome type 2C	605472	*GPR98*
689	Usher syndrome type 3A	276902	*CLRN1*
690	Very long chain acyl-CoA dehydrogenase deficiency	201475	*ACADVL*
691	Vitamin B12-responsive methylmalonic acidemia type cblA	251100	*MMAA*

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

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