Condition List (1200+ Panel)

NO.	Diseases	Genes
1	Laron dwarfism	*GHR*
2	Congenital Adrenal Hyperplasia due to 11-beta-Hydroxylase-Deficiency	*CYP11B1*
3	Congenital Adrenal Hyperplasia due to 17-alpha Hydroxylase Deficiency	*CYP17A1*
4	2,4-Dienoyl-CoA Reductase Deficiency	*NADK2*
5	2-Methylbutyryl Glycinuria	*ACADSB*
6	3MC Syndrome 1	*MASP1*
7	3MC Syndrome 2	*COLEC11*
8	3-M Syndrome 2	*OBSL1*
9	3-beta-Hydroxysteroid Dehydrogenase Deficiency	*HSD3B2*
10	3-Methylglutaconic Aciduria type 1	*AUH*
11	3-Methylglutaconic Aciduria type 3	*OPA3*
12	3-Methylglutaconic Aciduria type 5	*DNAJC19*
13	3-Methylglutaconic Aciduria type 8	*HTRA2*
14	3-Methylglutaconic Aciduria With Cataracts, Neurologic Involvement,And Neutropenia	*CLPB*
15	3-Methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	*SERAC1*
16	3-Hydroxy-3-Methylglutaryl-CoA Synthase 2 Deficiency	*HMGCS2*
17	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	*HMGCL*
18	3-Hydroxyisobutyryl-Coa Hydrolase Deficiency	*HIBCH*
19	Pyridoxal 5'-Phosphate-dependent Epilepsy	*PNPO*
20	GM2-gangliosidosis, AB variant	*GM2A*
21	Adams-Oliver Syndrome 2	*DOCK6*
22	Adams-Oliver Syndrome 4	*EOGT*
23	Aicardi-Goutieres Syndrome 1	*TREX1*
24	Aicardi-Goutieres Syndrome 2	*RNASEH2B*
25	Aicardi-Goutieres Syndrome 3	*RNASEH2C*
26	Aicardi-Goutieres Syndrome 4	*RNASEH2A*
27	Aicardi-Goutieres Syndrome 5	*SAMHD1*
28	Aicardi-Goutieres Syndrome 6	*ADAR*
29	Al Kaissi syndrome	*CDK10*
30	Alazami Syndrome	*LARP7*
31	Alkuraya-Kucinskas Syndrome	*BLTP1*
32	Allan-Herndon-Dudley syndrome	*SLC16A2*
33	Alstrom Syndrome	*ALMS1*
34	Anauxetic dysplasia 2	*POP1*

35	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis	*POR*
36	Arts Syndrome	*PRPS1*
37	Athabaskan Brain Stem Dysgenesis Syndrome	*HOXA1*
38	Baller-Gerold Syndrome	*RECQL4*
39	Bardet-Biedl syndrome 10	*BBS10*
40	Bardet-Biedl syndrome 12	*BBS12*
41	Bardet-Biedl syndrome 16	*SDCCAG8*
42	Bardet-Biedl syndrome 17	*LZTFL1*
43	Bardet-Biedl syndrome 1	*BBS1*
44	Bardet-Biedl syndrome 2	*BBS2*
45	Bardet-Biedl syndrome 3	*ARL6*
46	Bardet-Biedl syndrome 4	*BBS4*
47	Bardet-Biedl syndrome 5	*BBS5*
48	Bardet-Biedl syndrome 7	*BBS7*
49	Bardet-Biedl syndrome 8	*TTC8*
50	Bardet-Biedl syndrome 9	*BBS9*
51	Barth syndrome	*TAFAZZIN*
52	Bartter Syndrome 1	*SLC12A1*
53	Bartter Syndrome 2	*KCNJ1*
54	Basel-Vanagait-Smirin-Yosef syndrome	*MED25*
55	BEHR syndrome	*OPA1*
56	BH4-Deficient Hyperphenylalaninemia C	*QDPR*
57	Chondrodysplasia, Blomstrand Type	*PTH1R*
58	Bloom syndrome	*BLM*
59	Borjeson-Forssman-Lehmann syndrome	*PHF6*
60	Boucher-Neuhauser syndrome	*PNPLA6*
61	Brachyolmia Type 4	*PAPSS2*
62	Brown-Vialetto-Van Laere syndrome 1	*SLC52A3*
63	Brown-Vialetto-Van Laere syndrome 2	*SLC52A2*
64	Bruck Syndrome 1	*FKBP10*
65	Bruck Syndrome 2	*PLOD2*
66	Brunner syndrome	*MAOA*
67	Burn-Mckeown Syndrome	*TXNL4A*
68	Canavan Disease	*ASPA*
69	Carey-Fineman-Ziter syndrome	*MYMK*
70	Carpenter syndrome 1	*RAB23*
71	Carpenter Syndrome 2	*MEGF8*
72	Cerebellar Ataxia, Cayman Type	*ATCAY*
73	Cenani-Lenz Syndactyly Syndrome	*LRP4*
74	Chanarin-Dorfman syndrome	*ABHD5*
75	Charcot-Marie-Tooth disease, Axonal, Type 2A2B	*MFN2*
76	Charcot-Marie-Tooth disease type 3	*PRX*

77	Charcot-Marie-Tooth disease type 4B1	*MTMR2*
78	Charcot-Marie-Tooth disease type 4C	*SH3TC2*
79	Charcot-Marie-Tooth disease type 4D	*NDRG1*
80	Chediak-Higashi Syndrome	*LYST*
81	X-Linked Syndromic Mental Retardation, Christianson type	*SLC9A6*
82	Chudley-McCullough Syndrome	*GPSM2*
83	X-Linked Syndromic Mental Retardation, Claes-Jensen type	*KDM5C*
84	COACH syndrome	*TMEM67, CC2D2A*
85	Cockayne Syndrome A	*ERCC8*
86	Cockayne Syndrome B	*ERCC6*
87	CODAS Syndrome	*LONP1*
88	Coffin-Lowry Syndrome	*RPS6KA3*
89	Cohen Syndrome	*VPS13B*
90	Crigler-Najjar syndrome type 1	*UGT1A1*
91	D,L-2-hydroxyglutaric aciduria	*SLC25A1*
92	D-2-hydroxyglutaric aciduria 1	*D2HGDH*
93	DCLRE1C-Related Severe Combined Immunodeficiency	*DCLRE1C*
94	Desbuquois Dysplasia 1	*CANT1*
95	Desbuquois Dysplasia 2	*XYLT1*
96	Donnai-Barrow syndrome	*LRP2*
97	Dyggve-Melchior-Clausen Disease	*DYM*
98	D-Glyceric Aciduria	*GLYCTK*
99	Ehlers-Danlos Syndrome type VI	*PLOD1*
100	Ehlers-Danlos syndrome type VIIC	*ADAMTS2*
101	Ehlers-Danlos Syndrome with Progressive Kyphoscoliosis, Myopathy, and Hearing Loss	*FKBP14*
102	Ellis-van Creveld Syndrome	*EVC,EVC2*
103	Elsahy-Waters syndrome	*CDH11*
104	MULTIPLE PTERYGIUM SYNDROME, ESCOBAR VARIANT	*CHRNG*
105	Fabry Disease	*GLA*
106	Fanconi-Bickel Syndrome	*SLC2A2*
107	Farber Lipogranulomatosis	*ASAH1*
108	FG Syndrome Type 2	*FLNA*
109	FG Syndrome Type 4	*CASK*
110	Filippi Syndrome	*CKAP2L*
111	Frank-ter Haar Syndrome	*SH3PXD2B*
112	Fraser Syndrome 1	*FRAS1*
113	Fraser syndrome 2	*FREM2*
114	Fraser syndrome 3	*GRIP1*

115	X-linked Mental retardation, FRAXE type	*AFF2*
116	GABA-Transaminase Deficiency	*ABAT*
117	Galloway-Mowat Syndrome 1	*WDR73*
118	Galloway-Mowat syndrome 3	*OSGEP*
119	Geleophysic dysplasia 1	*ADAMTSL2*
120	Spondyloepimetaphyseal Dysplasia, Genevieve Type	*NANS*
121	Goldberg-Shprintzen syndrome	*KIFBP*
122	Gracile Syndrome	*BCS1L*
123	Greenberg dysplasia	*LBR*
124	Griscelli Syndrome 2	*RAB27A*
125	Dopa-Responsive Dystonia	*GCH1*
126	Hennekam Lymphangiectasia-Lymphedema Syndrome 1	*CCBE1*
127	Hennekam Lymphangiectasia-Lymphedema Syndrome 2	*FAT4*
128	Hermansky-Pudlak Syndrome 1	*HPS1*
129	Hermansky-Pudlak Syndrome 3	*HPS3*
130	Hermansky-Pudlak Syndrome 4	*HPS4*
131	Hermansky-Pudlak Syndrome 5	*HPS5*
132	Hermansky-Pudlak Syndrome 6	*HPS6*
133	Hurler-Scheie Syndrome	*IDUA*
134	Hurler Syndrome	*IDUA*
135	X-linked immunodysregulation, polyendocrinopathy, and enteropathy	*FOXP3*
136	Jalili Syndrome	*CNNM4*
137	Jervell and Lange-Nielsen syndrome 1	*KCNQ1*
138	Jervell and Lange-Nielsen syndrome 2	*KCNE1*
139	Johanson-Blizzard Syndrome	*UBR1*
140	Joubert Syndrome 10	*OFD1*
141	Joubert Syndrome 14	*TMEM237*
142	Joubert Syndrome 15	*CEP41*
143	Joubert Syndrome 16	*TMEM138*
144	Joubert Syndrome 17	*CPLANE1*
145	Joubert Syndrome 18	*TCTN3*
146	Joubert Syndrome 1	*INPP5E*
147	Joubert Syndrome 20	*TMEM231*
148	Joubert Syndrome 21	*CSPP1*
149	Joubert Syndrome 24	*TCTN2*
150	Joubert Syndrome 2	*TMEM216*
151	Joubert Syndrome 3	*AHI1*
152	Joubert Syndrome 4	*NPHP1*
153	Joubert Syndrome 5	*CEP290*
154	Joubert Syndrome 6	*TMEM67*
155	Joubert Syndrome 8	*ARL13B*
156	Joubert Syndrome 9	*CC2D2A*

157	Kenny-Caffey Syndrome Type 1	*TBCE*
158	Keutel Syndrome	*MGP*
159	Kindler Syndrome	*FERMT1*
160	Knobloch Syndrome Type I	*COL18A1*
161	Kohlschutter-Tonz Syndrome	*ROGDI*
162	Krabbe Disease	*GALC*
163	L-2-hydroxyglutaric aciduria	*L2HGDH*
164	Lafora Disease	*EPM2A*
165	LAMA3-Related Junctional Epidermolysis Bullosa	*LAMA3*
166	LAMB3-Related Junctional Epidermolysis Bullosa	*LAMB3*
167	LAMC2-Related Junctional Epidermolysis Bullosa	*LAMC2*
168	Leber Congenital Amaurosis 12	*RD3*
169	Leber Congenital Amaurosis 13	*RDH12*
170	Leber congenital amaurosis 14	*LRAT*
171	Leber Congenital Amaurosis 1	*GUCY2D*
172	Leber Congenital Amaurosis 2	*RPE65*
173	Leber Congenital Amaurosis 3	*SPATA7*
174	Leber Congenital Amaurosis 5	*LCA5*
175	Leber Congenital Amaurosis 8	*CRB1*
176	Leber Congenital Amaurosis 9	*NMNAT1*
177	Lesch-Nyhan Syndrome	*HPRT1*
178	LIG4 syndrome	*LIG4*
179	Lowe Syndrome	*OCRL*
180	Lujan-Fryns syndrome	*MED12*
181	Majeed Syndrome	*LPIN2*
182	Marinesco-Sjogren Syndrome	*SIL1*
183	MASA syndrome	*L1CAM*
184	McKusick-Kaufman Syndrome	*MKKS*
185	Meckel syndrome 1	*MKS1*
186	Meckel Syndrome 2	*TMEM216*
187	Meckel Syndrome 3	*TMEM67*
188	Meckel Syndrome 4	*CEP290*
189	Meckel syndrome 5	*RPGRIP1L*
190	Neonatal Severe Encephalopathy Due To MECP2 Mutations	*MECP2*
191	Meester-Loeys syndrome	*BGN*
192	Congenital Muscular Dystrophy, Megaconial type	*CHKB*
193	MEHMO syndrome	*EIF2S3*
194	Meier-Gorlin Syndrome 1	*ORC1*
195	Meier-Gorlin Syndrome 3	*ORC6*
196	Meier-Gorlin Syndrome 4	*CDT1*
197	Meier-Gorlin syndrome 7	*CDC45*
198	Menkes Disease	*ATP7A*

199	Merosin-deficient congenital muscular dystrophy type 1A	*LAMA2*
200	Miller syndrome	*DHODH*
201	Mitchell-Riley syndrome	*RFX6*
202	Mohr-Tranebjaerg syndrome	*TIMM8A*
203	Recurrent Pyogenic Bacterial Infections due to MYD88 Deficiency	*MYD88*
204	X-Linked Syndromic Mental Retardation, Nascimento-type	*UBE2A*
205	Naxos Disease	*JUP*
206	Netherton syndrome	*SPINK5*
207	Neu-Laxova Syndrome 1	*PHGDH*
208	Neu-Laxova Syndrome 2	*PSAT1*
209	Niemann-Pick Disease Type A	*SMPD1*
210	Niemann-Pick Disease Type B	*SMPD1*
211	Niemann-Pick Disease Type C1	*NPC1*
212	Niemann-Pick Disease Type C2	*NPC2*
213	Norrie Disease	*NDP*
214	N-acetylglutamate synthase deficiency	*NAGS*
215	Ogden Syndrome	*NAA10*
216	Spondyloepiphyseal Dysplasia, Omani type	*CHST3*
217	Omenn syndrome	*RAG1, RAG2*
218	Opitz Gbbb Syndrome, Type I	*MID1*
219	Opsismodysplasia	*INPPL1*
220	PEHO syndrome	*ZNHIT3*
221	Perlman Syndrome	*DIS3L2*
222	Perrault Syndrome 3	*CLPP*
223	Perrault Syndrome 4	*LARS2*
224	Peters Plus Syndrome	*B3GLCT*
225	Pierson Syndrome	*LAMB2*
226	Pitt-Hopkins like syndrome 1	*CNTNAP2*
227	Poretti-Boltshauser syndrome	*LAMA1*
228	Raine Syndrome	*FAM20C*
229	X-Linked Syndromic Mental Retardation, Raymond type	*ZDHHC9*
230	Renpenning syndrome	*PQBP1*
231	Sandhoff Disease	*HEXB*
232	Metachromatic leukodystrophy due to Saposin B deficiency	*PSAP*
233	Schimke Immunoosseous Dysplasia	*SMARCAL1*
234	Schneckenbecken Dysplasia	*SLC35D1*
235	Schwartz-Jampel Syndrome, Type 1	*HSPG2*
236	Schwartz-Jampel Syndrome, Type 2	*LIFR*
237	SC Phocomelia Syndrome	*ESCO2*

238	Seckel Syndrome Type 1	*ATR*
239	Seckel Syndrome Type 2	*RBBP8*
240	Seckel Syndrome Type 5	*CEP152*
241	Tyrosine Hydroxylase Deficiency	*TH*
242	Sengers syndrome	*AGK*
243	Senior-Loken Syndrome 4	*NPHP4*
244	Senior-Loken syndrome 5	*IQCB1*
245	Senior-Loken Syndrome 8	*WDR19*
246	Shwachman-Diamond Syndrome	*SBDS*
247	X-linked Mental retardation syndrome, Siderius type	*PHF8*
248	Simpson-Golabi-Behmel Syndrome Type 1	*GPC3*
249	Sjögren-Larsson syndrome	*ALDH3A2*
250	Smith-Lemli-Opitz syndrome	*DHCR7*
251	Smith-McCort Dysplasia 2	*RAB33B*
252	Snyder-Robinson mental retardation syndrome	*SMS*
253	Steel Syndrome	*COL27A1*
254	TARP Syndrome	*RBM10*
255	Tay-Sachs Disease	*HEXA*
256	Temtamy Preaxial Brachydactyly Syndrome	*CHSY1*
257	Temtamy Syndrome	*C12orf57*
258	Tenascin-X deficiency type Ehlers-Danlos syndrome	*TNXB*
259	Ullrich congenital muscular dystrophy 1	*COL6A1,COL6A2,COL6A3*
260	Usher Syndrome Type IB	*MYO7A*
261	Usher Syndrome Type IC	*USH1C*
262	Usher syndrome, type 1D	*CDH23*
263	Usher Syndrome Type IF	*PCDH15*
264	Usher Syndrome Type IG	*USH1G*
265	Usher Syndrome Type IJ	*CIB2*
266	Usher Syndrome Type IIA	*USH2A*
267	Usher Syndrome Type IID	*WHRN*
268	Usher Syndrome Type IIIA	*CLRN1*
269	Van Den Ende-Gupta Syndrome	*SCARF2*
270	Van Maldergem Syndrome 1	*DCHS1*
271	Vici Syndrome	*EPG5*
272	Factor VII Deficiency	*F7*
273	Factor V deficiency	*F5*
274	Warburg Micro Syndrome 1	*RAB3GAP1*
275	Warburg Micro Syndrome 2	*RAB3GAP2*
276	Warburg Micro Syndrome 3	*RAB18*
277	Wieacker-Wolff Syndrome	*ZC4H2*
278	Wiskott-Aldrich Syndrome 1	*WAS*
279	Multiple Epiphyseal Dysplasia with Early-Onset Diabetes Mellitus	*EIF2AK3*

280	Wolfram Syndrome 1	*WFS1*
281	Wolfram Syndrome 2	*CISD2*
282	Woodhouse-Sakati syndrome	*DCAF17*
283	Alport syndrome 1, X-linked	*COL4A5*
284	X-linked Charcot-Marie-Tooth disease 4	*AIFM1*
285	X-linked Emery-Dreifuss Muscular Dystrophy 1	*EMD*
286	X-Linked Properdin Deficiency	*CFP*
287	X-Linked Hypophosphatemia	*PHEX*
288	Macular Degeneration, X-Linked Atrophic	*RPGR*
289	X-Linked Myopathy with Excessive Autophagy	*VMA21*
290	X-Linked Lymphoproliferative syndrome 1	*SH2D1A*
291	X-Linked Lymphoproliferative syndrome 2	*XIAP*
292	X-linked Chronic Granulomatous Disease	*CYBB*
293	X-Linked Juvenile Retinoschisis	*RS1*
294	X-Linked Hypohidrotic Ectodermal Dysplasia	*EDA*
295	X-Linked Adrenoleukodystrophy	*ABCD1*
296	Myopathy, X-linked, with postural muscle atrophy	*FHL1*
297	X-linked sideroblastic anemia and ataxia	*ABCB7*
298	X-linked Pigmentary disorder, reticulate, with systemic manifestations	*POLA1*
299	X-linked Lissencephaly 1	*DCX*
300	X-linked Lissencephaly 2	*ARX*
301	X-Linked Dyskeratosis Congenita	*DKC1*
302	X-Linked Adrenal Hypoplasia Congenita	*NR0B1*
303	X-Linked Ocular Albinism	*GPR143*
304	Spinal muscular atrophy, X-linked 2, infantile	*UBA1*
305	X-linked Mental Retardation	*NLGN4X*
306	X-Linked Mental Retardation 12	*THOC2*
307	X-Linked Mental Retardation 1	*IQSEC2*
308	X-Linked Mental Retardation 21	*IL1RAPL1*
309	X-Linked Mental Retardation 30	*PAK3*
310	X-Linked Mental Retardation 41	*GDI1*
311	X-Linked Mental Retardation 49	*CLCN4*
312	X-Linked Mental Retardation 58	*TSPAN7*
313	X-Linked Mental Retardation 61	*RLIM*
314	X-Linked Mental Retardation 72	*RAB39B*
315	X-Linked Mental Retardation 90	*DLG3*
316	X-Linked Mental Retardation 93	*BRWD3*
317	X-Linked Mental Retardation 96	*SYP*
318	X-Linked Mental Retardation 97	*ZNF711*
319	X-Linked Mental Retardation 98	*NEXMIF*
320	X-Linked Mental Retardation 99	*USP9X*
321	X-Linked Mental Retardation 9	*FTSJ1*

322	X-Linked Mental Retardation with Cerebellar Hypoplasia and Distinctive Facial Appearance	*OPHN1*
323	X-Linked Mental Retardation-Hypotonic Facies Syndrome 1	*ATRX*
324	X-Linked Centronuclear Myopathy	*MTM1*
325	X-Linked Severe Combined Immunodeficiency	*IL2RG*
326	X-Linked Syndromic Mental Retardation 14	*UPF3B*
327	X-Linked Syndromic Mental Retardation 15	*CUL4B*
328	X-Linked Syndromic Mental Retardation 35	*RPL10*
329	X-Linked Syndromic Mental Retardation 5	*AP1S2*
330	You-Hoover-Fong syndrome	*TELO2*
331	Yunis-Varon Syndrome	*FIG4*
332	Alpha1-Antitrypsin Deficiency	*SERPINA1*
333	Alpha-N-acetylgalactosaminidase deficiency	*NAGA*
334	Alpha-thalassemia	*HBA1, HBA2*
335	Alpha-Mannosidosis	*MAN2B1*
336	Beta-thalassemia	*HBB*
337	Beta-Mannosidosis	*MANBA*
338	Beta-Ureidopropionase Deficiency	*UPB1*
339	Beta-Ketothiolase Deficiency	*ACAT1*
340	Carbamoylphosphate Synthetase I Deficiency	*CPS1*
341	Prolidase deficiency	*PEPD*
342	Interleukin 1 Receptor Antagonist Deficiency	*IL1RN*
343	Cataracts, Growth Hormone Deficiency, Sensory Neuropathy,sensorineural hearing loss, and skeletal dysplasia	*IARS2*
344	Cataract 18	*FYCO1*
345	Cataract 40	*NHS*
346	Leukocyte Adhesion Deficiency type 1	*ITGB2*
347	Leukocyte Adhesion Deficiency type 3	*FERMT3*
348	Galactokinase Deficiency	*GALK1*
349	Galactosialidosis	*CTSA*
350	Galactosemia	*GALT*
351	Sudden Infant Death With Dysgenesis Of The Testes Syndrome	*TSPYL1*
352	Spondyloepimetaphyseal Dysplasia With Joint Laxity, Type 1, With Or Without Fractures	*B3GALT6*
353	Bowen-Conradi Syndrome	*EMG1*
354	Phenylketonuria	*PAH*
355	Bifid Nose With Or Without Anorectal And Renal Anomalies	*FREM1*
356	Pyridoxine-Refractory Sideroblastic Anemia 2	*SLC25A38*
357	Pyridoxine-Dependent Epilepsy	*ALDH7A1*

358	Epidermolytic hyperkeratosis	*KRT10*
359	Malonyl-Coa Decarboxylase Deficiency	*MLYCD*
360	Propionicacidemia	*PCCA, PCCB*
361	Pyruvate kinase deficiency	*PKLR*
362	Pyruvate Carboxylase Deficiency	*PC*
363	Pyruvate dehydrogenase E1-alpha deficiency	*PDHA1*
364	Pyruvate Dehydrogenase E1-Beta Deficiency	*PDHB*
365	Pyruvate Dehydrogenase Phosphatase Deficiency	*PDP1*
366	Pyruvate Dehydrogenase Lipoic Acid Synthetase Deficiency	*LIAS*
367	C1q deficiency	*C1QA,C1QB,C1QC*
368	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy	*CD55*
369	Complement Factor I Deficiency	*CFI*
370	Epimerase Deficiency Galactosemia	*GALE*
371	Hypomagnesemia 1, intestinal	*TRPM6*
372	Common Variable Immune Deficiency 1	*ICOS*
373	Common Variable Immune Deficiency 2	*TNFRSF13B*
374	Common Variable Immune Deficiency 6	*CD81*
375	Common Variable Immune Deficiency 8 with Autoimmunity	*LRBA*
376	Autosomal Spastic paraplegia 30	*KIF1A*
377	Autosomal Dyskeratosis Congenita 5/4	*RTEL1*
378	Alport syndrome 2, autosomal recessive	*COL4A3, COL4A4*
379	Autosomal Recessive Robinow Syndrome	*ROR2*
380	Autosomal Recessive T Cell-Negative、 B Cell-Positive、NK Cell-Negative Severe Combined Immunodeficiency	*JAK3*
381	Autosomal Recessive Persistent Hyperplastic Primary Vitreous	*ATOH7*
382	Autosomal Recessive Epidermolysis Bullosa Simplex 1	*KRT14,KRT5*
383	Autosomal recessive Thrombophilia due to protein C deficiency	*PROC*
384	Autosomal recessive Thrombophilia due to protein S deficiency	*PROS1*
385	Autosomal Recessive Deafness 1A	*GJB2*
386	Autosomal Recessive Deafness 3	*MYO15A*
387	Autosomal Recessive Deafness 4, with Enlarged Vestibular Aqueduct	*SLC26A4*
388	Autosomal Recessive Deafness 7	*TMC1*
389	Autosomal Recessive Deafness 8/10	*TMPRSS3*

390	Autosomal Recessive Deafness 9	*OTOF*
391	Autosomal Recessive Osteopetrosis 1	*TCIRG1*
392	Autosomal Recessive Osteopetrosis 2	*TNFSF11*
393	Autosomal Recessive Osteopetrosis 3	*CA2*
394	Autosomal Recessive Osteopetrosis 4	*CLCN7*
395	Autosomal Recessive Osteopetrosis 5	*OSTM1*
396	Autosomal Recessive Osteopetrosis 7	*TNFRSF11A*
397	Autosomal Recessive Spinocerebellar Ataxia 10	*ANO10*
398	Autosomal Recessive Spinocerebellar Ataxia 13	*GRM1*
399	Autosomal Recessive Spinocerebellar Ataxia 16	*STUB1*
400	Autosomal Recessive Spinocerebellar Ataxia 1	*SETX*
401	Autosomal Recessive Spinocerebellar ataxia 20	*SNX14*
402	Autosomal Recessive Spinocerebellar ataxia 21	*SCYL1*
403	Autosomal Recessive Spinocerebellar Ataxia 2	*PMPCA*
404	Autosomal Recessive Spastic Paraplegia type 11	*SPG11*
405	Autosomal Recessive Spastic paraplegia 15	*ZFYVE26*
406	Autosomal Recessive Spastic paraplegia 23	*DSTYK*
407	Autosomal Recessive Spastic paraplegia 26	*B4GALNT1*
408	Autosomal Recessive Spastic paraplegia 35	*FA2H*
409	Autosomal Recessive Spastic paraplegia 45	*NT5C2*
410	Autosomal Recessive Spastic paraplegia 46	*GBA2*
411	Autosomal Recessive Spastic paraplegia 47	*AP4B1*
412	Autosomal Recessive Spastic paraplegia 50	*AP4M1*
413	Autosomal Recessive Spastic paraplegia 52	*AP4S1*
414	Autosomal Recessive Spastic paraplegia 53	*VPS37A*
415	Autosomal Recessive Spastic paraplegia 54	*DDHD2*
416	Autosomal Recessive Spastic paraplegia 56	*CYP2U1*
417	Autosomal Recessive Spastic Paraplegia 9B	*ALDH18A1*
418	Autosomal Recessive Cutis Laxa type 1A	*FBLN5*
419	Autosomal Recessive Cutis Laxa type 1B	*EFEMP2*
420	Autosomal Recessive Cutis Laxa type 1C	*LTBP4*
421	Autosomal Recessive Cutis Laxa type 2A	*ATP6V0A2*
422	Autosomal Recessive Cutis Laxa type 2B	*PYCR1*
423	Autosomal Recessive Cytochrome B-Positive Chronic Granulomatous Disease Type I	*NCF1*
424	Autosomal Recessive Cytochrome B-Positive Chronic Granulomatous Disease Type II	*NCF2*
425	Autosomal Recessive Cytochrome B-Negative Chronic Granulomatous Disease	*CYBA*
426	Autosomal Recessive Myotonia Congenita	*CLCN1*
427	Autosomal Recessive Dyskeratosis Congenita 3	*WRAP53*
428	Autosomal Recessive Microcephaly And Chorioretinopathy 1	*TUBGCP6*

429	Autosomal Recessive Microcephaly And Chorioretinopathy 3	*TUBGCP4*
430	Autosomal Recessive Spastic Ataxia 8 with Hypomyelinating Leukodystrophy	*NKX6-2*
431	Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	*SACS*
432	Autosomal Recessive Congenital Ichthyosis 1	*TGM1*
433	Autosomal Recessive Congenital Ichthyosis 4A	*ABCA12*
434	Autosomal Recessive Congenital Ichthyosis 4B	*ABCA12*
435	Autosomal Recessive Congenital Ichthyosis 5	*CYP4F22*
436	Autosomal Recessive Congenital Ichthyosis 6	*NIPAL4*
437	Autosomal Recessive Congenital Ichthyosis 9	*CERS3*
438	Autosomal Recessive Epidermolysis Bullosa Dystrophica	*COL7A1*
439	Hypertrophic Osteoarthropathy, Primary, Autosomal Recessive, 1	*HPGD*
440	Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy	*MED23*
441	Intellectual developmental disorder, autosomal recessive 27	*LINS1*
442	Intellectual developmental disorder, autosomal recessive 38	*HERC2*
443	Intellectual developmental disorder, autosomal recessive 57	*MBOAT7*
444	Autosomal Recessive Mental Retardation 13	*TRAPPC9*
445	Autosomal Recessive Mental Retardation 15	*MAN1B1*
446	Autosomal Recessive Mental Retardation 36	*ADAT3*
447	Autosomal Recessive Mental Retardation 39	*TTI2*
448	Autosomal Recessive Mental Retardation 3	*CC2D1A*
449	Autosomal Recessive Mental Retardation 41	*KPTN*
450	Autosomal Recessive Mental Retardation 42	*PGAP1*
451	Autosomal Recessive Mental Retardation 44	*METTL23*
452	Autosomal Recessive Mental Retardation 49	*GPT2*
453	Autosomal Recessive Mental Retardation 58	*ELP2*
454	Autosomal Recessive Mental Retardation 5	*NSUN2*
455	Autosomal Recessive Mental Retardation 7	*TUSC3*
456	Severe combined immunodeficiency, B cell-negative	*RAG1, RAG2*
457	Osteogenesis Imperfecta type XV	*WNT1*
458	Osteogenesis Imperfecta type VI	*SERPINF1*
459	Osteogenesis Imperfecta type VIII	*P3H1*
460	Osteogenesis imperfecta, type X	*SERPINH1*
461	Achalasia-Addisonianism-Alacrima Syndrome	*AAAS*

462	Postnatal Progressive Microcephaly With Seizures And Brain Atrophy	*MED17*
463	Hemorrhagic Destruction of the Brain, Subependymal Calcification and Cataracts	*JAM3*
464	Adrenocorticotropic hormone Deficiency	*TBX19*
465	Neurodegeneration due to Cerebral Folate Transport Deficiency	*FOLR1*
466	Epidermolysis Bullosa with Pyloric Atresia	*ITGB4,ITGA6*
467	Macrocephaly, Alopecia, Cutis Laxa, and Scoliosis(MACS syndrome)	*RIN2*
468	Band-Like Calcification with Simplified Gyration and Polymicrogyria	*OCLN*
469	Band heterotopia	*EML1*
470	Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects	*B3GAT3*
471	Leukodystrophy, hypomyelinating, 2	*GJC2*
472	HSD10 mitochondrial disease	*HSD17B10*
473	D-bifunctional protein deficiency	*HSD17B4*
474	Autosomal Recessive Distal Spinal Muscular Atrophy 1	*IGHMBP2*
475	Donohue Syndrome	*INSR*
476	Gillespie syndrome, Autosomal recessive	*ITPR1*
477	PERCHING syndrome	*KLHL7*
478	Familial Lecithin cholesterol acyltransferase deficiency	*LCAT*
479	Lysosomal acid lipase deficiency	*LIPA*
480	Congenital Hydrocephalus 2 with or without brain or eye anomalies	*MPDZ*
481	Hypertrophic Neuropathy of Dejerine Sottas	*MPZ*
482	Cleft lip/palate-ectodermal dysplasia syndrome	*NECTIN1*
483	myoclonic epilepsy of Lafora	*NHLRC1*
484	Insensitivity to pain, congenital, with anhidrosis	*NTRK1*
485	Coloboma, Congenital Heart Disease, Ichthyosiform Dermatosis, Mental Retardation, and Ear Anomalies Syndrome	*PIGL*
486	Plasminogen deficiency, type I	*PLG*
487	Microcephaly, seizures, and developmental delay	*PNKP*
488	OBESITY, EARLY-ONSET, WITH ADRENAL INSUFFICIENCY AND RED HAIR	*POMC*
489	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)	*SLC19A3*
490	Autosomal Recessive Spastic paraplegia 20	*SPART*
491	Salt and pepper developmental regression syndrome, Autosomal recessive	*ST3GAL5*
492	Gastrointestinal defects and immunodeficiency	*TTC7A*

	syndrome	
493	Kaufman oculocerebrofacial syndrome	*UBE3B*
494	Epidermolysis bullosa simplex with pyloric atresia	*PLEC*
495	Isolated Microphthalmia 8	*ALDH1A3*
496	Monocarboxylate Transporter 1 Deficiency	*SLC16A1*
497	Proteasome-Associated Autoinflammatory Syndrome 1	*PSMB8*
498	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1)	*ANOS1*
499	Hypomyelinating Leukodystrophy 10	*PYCR2*
500	Hypomyelinating Leukodystrophy 12	*VPS11*
501	Hypomyelinating Leukodystrophy 14	*UFM1*
502	Hypomyelinating Leukodystrophy 3	*AIMP1*
503	Hypomyelinating Leukodystrophy 4	*HSPD1*
504	Hypomyelinating Leukodystrophy 5	*HYCC1*
505	Hypomyelinating Leukodystrophy 7 with or without oligodontia and/or hypogonadotropic hypogonadism	*POLR3A*
506	Hypomyelinating Leukodystrophy 8	*POLR3B*
507	Epilepsy, Hearing Loss, And Mental Retardation Syndrome	*AFG2A*
508	Epilepsy with Variable Learning Disabilities and Behavior Disorders	*SYN1*
509	Seizures, Sensorineural Deafness, Ataxia, Mental Retardation, and Electrolyte Imbalance Syndrome	*KCNJ10*
510	Arterial tortuosity syndrome	*SLC2A10*
511	Short-Rib Thoracic Dysplasia 14 With Polydactyly	*KIAA0586*
512	Short-rib throacic dysplasia 15 with polydactyly	*DYNC2LI1*
513	Short-rib thoracic dysplasia 10 with or without polydactyly	*IFT172*
514	Short-rib thoracic dysplasia 11 with or without polydactyly	*DYNC2I2*
515	Short-rib thoracic dysplasia 13 with or without polydactyly	*CEP120*
516	Short-rib thoracic dysplasia 2 with or without polydactyly	*IFT80*
517	Short-rib thoracic dysplasia 3 with or without polydactyly	*DYNC2H1*
518	Short-rib thoracic dysplasia 4 with or without polydactyly	*TTC21B*
519	Short-rib thoracic dysplasia 6 with or without polydactyly	*NEK1*
520	Short-rib thoracic dysplasia 7 with or without polydactyly	*WDR35*
521	Short-rib thoracic dysplasia 8 with or without	*DYNC2I1*

	polydactyly	
522	Short-rib thoracic dysplasia 9 with or without polydactyly	*IFT140*
523	Short Stature, Microcephaly, And Endocrine Dysfunction	*XRCC4*
524	Multicentric Osteolysis, Nodulosis, and Arthropathy	*MMP2*
525	Multiple Sulfatase Deficiency	*SUMF1*
526	Multiple Congenital Anomalies-Hypotonia-Seizures Syndrome 1	*PIGN*
527	Multiple Congenital Anomalies-Hypotonia-Seizures Syndrome 3	*PIGT*
528	Multiple Pterygium Syndrome,lethal type	*CHRNA1*
529	Polycystic Kidney Disease 4 with or without Polycystic Liver Disease	*PKHD1*
530	Multiminicore disease	*RYR1*
531	Multisystem Autoimmune Disease With Facial Dysmorphism	*ITCH*
532	Multiple mitochondrial dysfunctions syndrome 1	*NFU1*
533	Multiple mitochondrial dysfunctions syndrome 2	*BOLA3*
534	Multiple mitochondrial dysfunctions syndrome 3	*IBA57*
535	Multiple mitochondrial dysfunctions syndrome 4	*ISCA2*
536	Polymicrogyria with Seizures	*RTTN*
537	Catecholaminergic Polymorphic Ventricular Tachycardia 2	*CASQ2*
538	Catecholaminergic Polymorphic Ventricular Tachycardia 5, with or without muscle weakness	*TRDN*
539	Childhood-Onset Polyarteritis Nodosa	*ADA2*
540	Hypophosphatasia, childhood	*ALPL*
541	Childhood-onset neurodegeneration with ataxia, dystonia, and gaze palsy	*SQSTM1*
542	Developmental Delay With Short Stature, Dysmorphic Features, And Sparse Hair	*DPH1*
543	GAPO Syndrome	*ANTXR1*
544	Leigh Syndrome, French-Canadian Type	*LRPPRC*
545	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration	*TANGO2*
546	Fanconi anemia, complementation group A	*FANCA*
547	Fanconi anemia, complementation group B	*FANCB*
548	Fanconi anemia, complementation group C	*FANCC*
549	Fanconi anemia, complementation group D2	*FANCD2*
550	Fanconi anemia, complementation group E	*FANCE*
551	Fanconi anemia, complementation group F	*FANCF*

552	Fanconi anemia, complementation group G	*FANCG*
553	Fanconi anemia, complementation group I	*FANCI*
554	Fanconi anemia, complementation group L	*FANCL*
555	Fanconi anemia, complementation group Q	*ERCC4*
556	Fanconi anemia, complementation group T	*UBE2T*
557	Non-Herlitz type Junctional Epidermolysis Bullosa	*COL17A1*
558	Nonphotosensitive Trichothiodystrophy 4	*MPLKIP*
559	Pulmonary Surfactant Metabolism Dysfunction 1	*SFTPB*
560	Pulmonary Surfactant Metabolism Dysfunction 3	*ABCA3*
561	Pulmonary Venoocclusive Disease 2	*EIF2AK4*
562	Maple Syrup Urine Disease Type 1A	*BCKDHA*
563	Maple Syrup Urine Disease Type 1B	*BCKDHB*
564	Maple Syrup Urine Disease, type 2	*DBT*
565	Maple Syrup Urine Disease Type 3	*DLD*
566	Combined Oxidative Phosphorylation Deficiency 10	*MTO1*
567	Combined Oxidative Phosphorylation Deficiency 11	*RMND1*
568	Combined Oxidative Phosphorylation Deficiency 12	*EARS2*
569	Combined Oxidative Phosphorylation Deficiency 14	*FARS2*
570	Combined Oxidative Phosphorylation Deficiency 15	*MTFMT*
571	Combined Oxidative Phosphorylation Deficiency 17	*ELAC2*
572	Combined Oxidative Phosphorylation Deficiency 1	*GFM1*
573	Combined Oxidative Phosphorylation Deficiency 20	*VARS2*
574	Combined Oxidative Phosphorylation Deficiency 23	*GTPBP3*
575	Combined Oxidative Phosphorylation Deficiency 24	*NARS2*
576	Combined Oxidative Phosphorylation Deficiency 27	*CARS2*
577	Combined Oxidative Phosphorylation Deficiency 35	*TRIT1*
578	Combined Oxidative Phosphorylation Deficiency 3	*TSFM*
579	Combined Oxidative Phosphorylation Deficiency 4	*TUFM*
580	Combined Oxidative Phosphorylation Deficiency 7	*MTRFR*
581	Combined Oxidative Phosphorylation Deficiency 8	*AARS2*
582	Diarrhea 5 With Congenital Tufting Enteropathy	*EPCAM*
583	Diarrhea 7	*DGAT1*
584	Diarrhea with Microvillus Atrophy 2	*MYO5B*
585	Glycine encephalopathy	*AMT, GLDC*
586	Glycerol Kinase Deficiency	*GK*
587	Nemaline Myopathy 10	*LMOD3*
588	Nemaline Myopathy 1	*TPM3*
589	Nemaline Myopathy 2	*NEB*
590	Nemaline Myopathy 5	*TNNT1*
591	Nemaline Myopathy 7	*CFL2*
592	Nemaline Myopathy 8	*KLHL40*
593	Nemaline Myopathy 9	*KLHL41*
594	Wilson Disease	*ATP7B*

595	Hepatic Veno-Occlusive Disease with Immunodeficiency	*SP110*
596	Hyper IgE Syndrome	*DOCK8*
597	Homocystinuria-Megaloblastic Anemia cblG type	*MTR*
598	Hyperphosphatasia with Mental Retardation Syndrome 1	*PIGV*
599	Hyperphosphatasia with Mental Retardation Syndrome 2	*PIGO*
600	Hyperphosphatasia with Mental Retardation Syndrome 3	*PGAP2*
601	Hyperphosphatasia with Mental Retardation Syndrome 4	*PGAP3*
602	Hypermanganesemia With Dystonia 1	*SLC30A10*
603	Hypermanganesemia with dystonia 2	*SLC39A14*
604	Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	*SLC25A15*
605	Hyperuricemia, Pulmonary Hypertension, Renal Failure, And Alkalosis syndrome	*SARS2*
606	Hyperprolinemia type I	*PRODH*
607	Methemoglobinemia Due to Deficiency of Methemoglobin Reductase	*CYB5R3*
608	Ataxia with oculomotor apraxia type 1	*APTX*
609	Ataxia with vitamin E deficiency	*TTPA*
610	Ataxia-Telangiectasia-Like Disorder 1	*MRE11*
611	Ataxia-telangiectasia	*ATM*
612	Microphthalmia, isolated 3	*RAX*
613	Glutamate Formiminotransferase Deficiency	*FTCD*
614	Sitosterolemia	*ABCG5, ABCG8*
615	Glutathione synthetase deficiency	*GSS*
616	Bone marrow failure syndrome 2	*ERCC6L2*
617	Bone marrow failure syndrome 3	*DNAJC21*
618	Sclerosteosis 1	*SOST*
619	Osteoporosis-pseudoglioma syndrome	*LRP5*
620	Citrullinemia	*ASS1*
621	Cerebral creatine deficiency syndrome 2	*GAMT*
622	Arthrogryposis, Renal Dysfunction, and Cholestasis Syndrome 1	*VPS33B*
623	Arthrogryposis, Renal Dysfunction, and Cholestasis Syndrome 2	*VIPAS39*
624	Arthrogryposis, Mental Retardation and Seizures	*SLC35A3*
625	Photosensitive Trichothiodystrophy 3	*GTF2H5*
626	Homocystinuria Due to Cystathionine Beta-Synthase Deficiency	*CBS*
627	Fructose 1,6 Bisphosphatase Deficiency	*FBP1*
628	Peroxisome biogenesis disorder 10A	*PEX3*
629	Peroxisome biogenesis disorder 11A	*PEX13*
630	Peroxisome Biogenesis Disorder 14B	*PEX11B*

631	Peroxisome biogenesis disorder 1A(Zellweger)	*PEX1*
632	Peroxisome biogenesis disorder 2A	*PEX5*
633	Peroxisome biogenesis disorder 3A(Zellweger)	*PEX12*
634	Peroxisome biogenesis disorder 4A	*PEX6*
635	Peroxisome biogenesis disorder 5A	*PEX2*
636	Peroxisome biogenesis disorder 6A(Zellweger)	*PEX10*
637	Peroxisome biogenesis disorder 7A	*PEX26*
638	Peroxisome biogenesis disorder 8A(Zellweger)	*PEX16*
639	Peroxisomal Acyl-CoA oxidase deficiency	*ACOX1*
640	Treacher Collins Syndrome 3	*POLR1C*
641	Ataxia, posterior column, with retinitis pigmentosa	*FLVCR1*
642	Succinic Semialdehyde Dehydrogenase Deficiency	*ALDH5A1*
643	Warsaw Breakage Syndrome	*DDX11*
644	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency	*FLAD1*
645	Myopathy, lactic acidosis, and sideroblastic anemia 1	*PUS1*
646	Myopathy, Lactic acidosis, and Sideroblastic anemia 2	*YARS2*
647	Myopathy With Extrapyramidal Signs	*MICU1*
648	Mulibrey nanism	*TRIM37*
649	Ehlers-Danlos Syndrome, Musculocontractural type 1	*CHST14*
650	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7	*CRPPA*
651	Myofibrillar Myopathy 7	*KY*
652	Myofibrillar Myopathy 8	*PYROXD1*
653	Acyl-CoA Dehydrogenase Deficiency,Very Long-Chain	*ACADVL*
654	Acute Recurrent Myoglobinuria	*LPIN1*
655	Spinal Muscular Atrophy	*SMN1*
656	Spondylo-Megaepiphyseal-Metaphyseal Dysplasia	*NKX3-2*
657	Spondylocarpotarsal Synostosis Syndrome	*FLNB*
658	Spondyloocular syndrome	*XYLT2*
659	Spondylometaphyseal Dysplasia with Cone-Rod Dystrophy	*PCYT1A*
660	Spondylocostal dysostosis 1	*DLL3*
661	Spondylocostal dysostosis 2	*MESP2*
662	Spondylocostal dysostosis 4	*HES7*
663	Familial Mediterranean Fever	*MEFV*
664	Familial Cirrhosis	*KRT8*
665	Hypercholesterolemia, familial,1	*LDLR*
666	Familial Hyperinsulinemic Hypoglycemia 1	*ABCC8*
667	Familial Hyperinsulinemic Hypoglycemia 2	*KCNJ11*
668	Familial Hyperinsulinemic Hypoglycemia 4	*HADH*
669	Familial Chloride Diarrhea	*SLC26A3*
670	Familial Candidiasis 2	*CARD9*

671	Hemophagocytic lymphohistiocytosis, familial, 2	*PRF1*
672	Hemophagocytic lymphohistiocytosis, familial, 3	*UNC13D*
673	Hemophagocytic lymphohistiocytosis, familial, 4	*STX11*
674	Hemophagocytic lymphohistiocytosis, familial, 5	*STXBP2*
675	Familial Normophosphatemic Tumoral Calcinosis	*SAMD9*
676	MCEE-Related Methylmalonic Acidemia	*MCEE*
677	Methylmalonic aciduria and homocystinuria CblF type	*LMBRD1*
678	Methylmalonic aciduria and homocysteinemia, cblX type	*HCFC1*
679	MMAA-Related Methylmalonic Acidemia	*MMAA*
680	MMAB-Related Methylmalonic Acidemia	*MMAB*
681	MUT-Related Methylmalonic Acidemia	*MMUT*
682	Methylmalonic Aciduria and Homocystinuria cblC type	*MMACHC*
683	Methylmalonic Aciduria and Homocystinuria cblD type	*MMADHC*
684	Mevalonic Aciduria	*MVK*
685	Thyroid dyshormonogenesis 5	*DUOXA2*
686	Thyroid dyshormonogenesis 6	*DUOX2*
687	Pseudohypoaldosteronism, type I	*SCNN1A,SCNN1B*
688	Interstitial lung and liver disease	*MARS1*
689	Omodysplasia 1	*GPC6*
690	Brittle Cornea Syndrome 1	*ZNF469*
691	Brittle Cornea Syndrome 2	*PRDM5*
692	Progressive Myoclonic Epilepsy 1A	*CSTB*
693	Progressive Myoclonic Epilepsy 1B	*PRICKLE1*
694	Progressive Myoclonic Epilepsy 3	*KCTD7*
695	Progressive Myoclonic Epilepsy 4	*SCARB2*
696	Progressive Myoclonic Epilepsy 6	*GOSR2*
697	Progressive Familial Intrahepatic Cholestasis 1	*ATP8B1*
698	Progressive Familial Intrahepatic Cholestasis 2	*ABCB11*
699	Progressive Familial Intrahepatic Cholestasis 3	*ABCB4*
700	Progressive Familial Intrahepatic Cholestasis 4	*TJP2*
701	Duchenne Muscular Dystrophy	*DMD*
702	Progressive Pseudorheumatoid Dysplasia	*CCN6*
703	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy	*NAXE*
704	Proximal Renal Tubular Acidosis with Ocular Abnormalities	*SLC4A4*
705	Argininosuccinic aciduria	*ASL*
706	Cerebral creatine deficiency syndrome 3	*GATM*
707	Argininemia	*ARG1*
708	Spastic Ataxia 2	*KIF1C*
709	Spastic Ataxia 3	*MARS2*

710	Spastic Paraplegia And Psychomotor Retardation With Or Without Seizures	*HACE1*
711	Megalencephalic Leukoencephalopathy with Subcortical Cysts 1	*MLC1*
712	Megalencephalic Leukoencephalopathy with Subcortical Cysts 2A	*HEPACAM*
713	Giant Axonal Neuropathy-1	*GAN*
714	Orofaciodigital syndrome XVI	*TMEM107*
715	Orofaciodigital Syndrome V	*DDX59*
716	Dilated Cardiomyopathy With Woolly Hair And Keratoderma	*DSP*
717	Lysinuric Protein Intolerance	*SLC7A7*
718	Familial Dysautonomia	*ELP1*
719	Geroderma Osteodysplasticum	*GORAB*
720	Tyrosinemia Type 1	*FAH*
721	Tyrosinemia Type II	*TAT*
722	Tyrosinemia Type III	*HPD*
723	CK Syndrome	*NSDHL*
724	Refsum disease	*PHYH*
725	Ehlers-Danlos syndrome, spondylodysplastic type, 1	*B4GALT7*
726	Cold-induced Sweating Syndrome 1	*CRLF1*
727	Cold-induced Sweating Syndrome 2	*CLCF1*
728	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia	*MTHFD1*
729	Combined pituitary hormone deficiency 1	*POU1F1*
730	Combined pituitary hormone deficiency 2	*PROP1*
731	Combined pituitary hormone deficiency 3	*LHX3*
732	Sickle Cell Anemia	*HBB*
733	Desmosterolosis	*DHCR24*
734	Split-Hand/Foot Malformation 6	*WNT10B*
735	Lymphoproliferative Syndrome 1	*ITK*
736	Lymphoproliferative Syndrome 2	*CD27*
737	Triosephosphate Isomerase Deficiency	*TPI1*
738	Phosphoglycerate Kinase Deficiency	*PGK1*
739	Phosphoserine Phosphatase Deficiency	*PSPH*
740	Thiamine-Responsive Megaloblastic Anemia Syndrome	*SLC19A2*
741	Thiamine Metabolism Dysfunction Syndrome 4 (Bilateral Striatal Degeneration and Progressive Polyneuropathy Type)	*SLC25A19*
742	Thiamine Metabolism Dysfunction Syndrome 5 (Episodic Encephalopathy type)	*TPK1*
743	Sulfocysteinuria	*SUOX*
744	Craniolenticulosutural Dysplasia	*SEC23A*

745	Craniofrontonasal syndrome	*EFNB1*
746	Craniosynostosis and Dental Anomalies	*IL11RA*
747	Cranioectodermal Dysplasia 1	*IFT122*
748	Craniofacial Dysmorphism, Skeletal Anomalies, And Mental Retardation syndrome	*TMCO1*
749	Bare lymphocyte syndrome, type II, complementation group A	*CIITA*
750	Bare lymphocyte syndrome, type II, complementation group B	*RFXANK*
751	Bare lymphocyte syndrome, type II, complementation group D	*RFXAP*
752	Chronic Atrial And Intestinal Dysrhythmia	*SGO1*
753	Trichohepatoenteric syndrome 1	*SKIC3*
754	Trichohepatoenteric Syndrome 2	*SKIC2*
755	Ichthyosis Follicularis-Atrichia-Photophobia Syndrome	*MBTPS2*
756	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	*QARS1*
757	Immunoskeletal dysplasia with neurodevelopmental abnormalities	*EXTL3*
758	Immunodeficiency with Hyper-IgM, type 1	*CD40LG*
759	Immunodeficiency with Hyper-IgM, type 3	*CD40*
760	Immunodeficiency-centromeric instability-facial anomalies syndrome 1	*DNMT3B*
761	Immunodeficiency-Centromeric Instability-Facial Anomalies Syndrome 2	*ZBTB24*
762	Immunodeficiency 10	*STIM1*
763	Immunodeficiency 11A	*CARD11*
764	Immunodeficiency 12	*MALT1*
765	Immunodeficiency 15B	*IKBKB*
766	Immunodeficiency 19	*CD3D*
767	Immunodeficiency 23	*PGM3*
768	Immunodeficiency 24	*CTPS1*
769	Immunodeficiency 27A	*IFNGR1*
770	Immunodeficiency 28	*IFNGR2*
771	Immunodeficiency 31B	*STAT1*
772	Immunodeficiency 35	*TYK2*
773	Immunodeficiency 40	*DOCK2*
774	Immunodeficiency 42	*RORC*
775	Immunodeficiency 47	*ATP6AP1*
776	Immunodeficiency 48	*ZAP70*
777	Immunodeficiency 51	*IL17RA*
778	Immunodeficiency 52	*LAT*
779	Immunodeficiency 54	*MCM4*

780	Immunodeficiency 9	*ORAI1*
781	Arthrogryposis, distal, with impaired proprioception and touch	*PIEZO2*
782	Dopa-responsive dystonia due to sepiapterin reductase deficiency	*SPR*
783	Molybdenum cofactor deficiency C	*GPHN*
784	Molybdenum Cofactor Deficiency A	*MOCS1*
785	Molybdenum Cofactor Deficiency Complementation Group B	*MOCS2*
786	Nijmegen breakage syndrome,	*NBN*
787	Nijmegen Breakage Syndrome-like Disorder	*RAD50*
788	Cystic Leukoencephalopathy without Megalencephaly	*RNASET2*
789	Cystic Fibrosis	*CFTR*
790	Cerebral Dysgenesis, Neuropathy, Ichthyosis, And Palmoplantar Keratoderma Syndrome	*SNAP29*
791	Leukoencephalopathy with Brain Stem and Spinal Cord Involvement and Lactate Elevation	*DARS2*
792	Hypomyelination with brainstem and spinal cord involvement and leg spasticity	*DARS1*
793	Cerebral Creatine Deficiency Syndrome 1	*SLC6A8*
794	Hydrolethalus Syndrome 1	*HYLS1*
795	Hydrolethalus Syndrome 2	*KIF7*
796	Cerebrotendinous xanthomatosis	*CYP27A1*
797	Pontocerebellar hypoplasia type 10	*CLP1*
798	Pontocerebellar hypoplasia type 11	*TBC1D23*
799	Pontocerebellar hypoplasia type 1A	*VRK1*
800	Pontocerebellar Hypoplasia type 1B	*EXOSC3*
801	Pontocerebellar Hypoplasia, Type 1C	*EXOSC8*
802	Pontocerebellar hypoplasia type 2A	*TSEN54*
803	Pontocerebellar hypoplasia type 2B	*TSEN2*
804	Pontocerebellar Hypoplasia type 2D	*SEPSECS*
805	Pontocerebellar Hypoplasia, Type 2E	*VPS53*
806	Pontocerebellar hypoplasia type 6	*RARS2*
807	Pontocerebellar hypoplasia, type 7	*TOE1*
808	Pontocerebellar hypoplasia type 9	*AMPD2*
809	Cerebroretinal Microangiopathy With Calcifications And Cysts	*CTC1*
810	Ventriculomegaly With Cystic Kidney Disease	*CRB2*
811	Periventricular Nodular Heterotopia 2	*ARFGEF2*
812	Cerebrooculofacioskeletal Syndrome 2	*ERCC2*
813	Visceral Heterotaxy 1	*ZIC3*
814	Visceral Heterotaxy 7	*MMP21*
815	Mucopolysaccharidosis type VII	*GUSB*

816	Mucopolysaccharidosis Type IIIA	*SGSH*
817	Mucopolysaccharidosis Type IIIB	*NAGLU*
818	Mucopolysaccharidosis type IIIC	*HGSNAT*
819	Mucopolysaccharidosis type IIID	*GNS*
820	Mucopolysaccharidosis II	*IDS*
821	Mucopolysaccharidosis type IVA	*GALNS*
822	Mucopolysaccharidosis type IVB	*GLB1*
823	Mucopolysaccharidosis type VI	*ARSB*
824	Mucopolysaccharidosis type V	*IDUA*
825	Ornithine Transcarbamylase Deficiency	*OTC*
826	Urofacial Syndrome 1	*HPSE2*
827	Urofacial Syndrome 2	*LRIG2*
828	Parkinson Disease 15	*FBXO7*
829	Parkinson Disease 19	*DNAJC6*
830	Pelizaeus-Merzbacher disease	*PLP1*
831	Poikiloderma with Neutropenia	*USB1*
832	Purine Nucleoside Phosphorylase Deficiency	*PNP*
833	Horizontal gaze palsy with progressive scoliosis 1	*ROBO3*
834	Polyglucosan Body Myopathy 1 With Or Without Immunodeficiency	*RBCK1*
835	Mosaic variegated aneuploidy syndrome 1	*BUB1B*
836	Rigid Spine Muscular Dystrophy 1	*SELENON*
837	Juvenile Paget Disease	*TNFRSF11B*
838	Primary Lateral Sclerosis, Juvenile	*ALS2*
839	Mild non-BH4-deficient Hyperphenylalaninemia	*DNAJC12*
840	Achromatopsia 2	*CNGA3*
841	Achromatopsia 4	*GNAT2*
842	Achromatopsia 7	*ATF6*
843	Hyaline fibromatosis syndrome	*ANTXR2*
844	Holocarboxylase synthetase deficiency	*HLCS*
845	Carnitine Palmitoyltransferase I Deficiency	*CPT1A*
846	Carnitine Palmitoyltransferase II Deficiency	*CPT2*
847	Carnitine-Acylcarnitine Translocase Deficiency	*SLC25A20*
848	Chylomicron Retention Disease	*SAR1B*
849	Cartilage-hair hypoplasia	*RMRP*
850	Achondrogenesis type 1A	*TRIP11*
851	Achondrogenesis type 1B	*SLC26A2*
852	Trifunctional Protein Deficiency	*HADHA,HADHB*
853	Trimethylaminuria	*FMO3*
854	Short Stature, Optic Nerve Atrophy, And Pelger-Huet Anomaly	*NBAS*
855	Short Stature, Onychodysplasia, Facial Dysmorphism, And Hypotrichosis	*POC1A*

856	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies	*PLAA*
857	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies	*PRUNE1*
858	Neurodevelopmental Disorder with Spastic Quadriplegia and Brain Abnormalities with or without Seizures	*WDR45B*
859	Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy	*PIGG*
860	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy	*VARS1*
861	Neurodegeneration with brain iron accumulation 1	*PANK2*
862	Neurodegeneration with brain iron accumulation 2B	*PLA2G6*
863	Neurodegeneration with brain iron accumulation 4(Mitochondrial Membrane Protein-Associated Neurodegeneration)	*C19orf12*
864	Neuronal Ceroid-Lipofuscinoses 1	*PPT1*
865	Neuronal Ceroid-Lipofuscinoses 2	*TPP1*
866	Neuronal Ceroid-Lipofuscinoses 3	*CLN3*
867	Neuronal Ceroid-Lipofuscinoses 4A	*CLN6*
868	Neuronal Ceroid-Lipofuscinoses 5	*CLN5*
869	Neuronal Ceroid-Lipofuscinoses 6	*CLN6*
870	Neuronal Ceroid-Lipofuscinoses 7	*MFSD8*
871	Neuronal Ceroid-Lipofuscinoses 10	*CTSD*
872	Neuronal Ceroid Lipofuscinosis 8	*CLN8*
873	Nephropathic Cystinosis	*CTNS*
874	Nephrotic Syndrome Type 11	*NUP107*
875	Nephrotic Syndrome Type 12	*NUP93*
876	Nephrotic Syndrome Type 14	*SGPL1*
877	Nephrotic syndrome, type 1	*NPHS1*
878	Nephrotic Syndrome Type 2	*NPHS2*
879	Nephrotic Syndrome Type 3	*PLCE1*
880	Nephrotic Syndrome Type 7	*DGKE*
881	Nephrotic Syndrome Type 9	*COQ8B*
882	Nephronophthisis 11	*TMEM67*
883	Nephronophthisis 16	*ANKS6*
884	Nephronophthisis 19	*DCDC2*
885	Nephronophthisis 20	*MAPKBP1*
886	Nephronophthisis 2	*INVS*
887	Nephronophthisis 3	*NPHP3*
888	Nephronophthisis-Like Nephropathy 1	*XPNPEP3*
889	Renal-Hepatic-Pancreatic Dysplasia 2	*NEK8*
890	Renal tubular dysgenesis	*ACE,AGT,REN*
891	Diabetes insipidus, nephrogenic, 2	*AQP2*

892	Hypomagnesemia 5, renal, with ocular involvement	*CLDN19*
893	Biotinidase Deficiency	*BTD*
894	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy	*IARS1*
895	Growth Retardation, Developmental Delay, Facial Dysmorphism	*FTO*
896	Septooptic Dysplasia	*HESX1*
897	Optic Atrophy 10 With Or Without Ataxia, Mental Retardation, And Seizures	*RTN4IP1*
898	Retinal Arterial Macroaneurysm With Supravalvular Pulmonic Stenosis	*IGFBP7*
899	Retinitis pigmentosa 14	*TULP1*
900	Retinitis pigmentosa 59	*DHDDS*
901	Retinitis pigmentosa 77	*REEP6*
902	Retinitis pigmentosa with or without skeletal anomalies	*CWC27*
903	Retinal dystrophy with macular staphyloma	*CFAP410*
904	Cone-rod dystrophy	*AIPL1*
905	Cone-Rod Dystrophy 10	*SEMA4A*
906	Cone-Rod Dystrophy 3	*ABCA4*
907	Spondylometaepiphyseal Dysplasia, Short Limb-Hand type	*DDR2*
908	bilateral frontoparietal polymicrogyria	*ADGRG1*
909	Hyperphenylalaninemia, BH4-deficient, A	*PTS*
910	Limb pelvis hypoplasia aplasia syndrome	*WNT7A*
911	Spastic Tetraplegia, Thin Corpus Callosum, And Progressive Microcephaly	*SLC1A4*
912	Leukodystrophy, hypomyelinating, 9	*RARS1*
913	Fetal akinesia deformation sequence 2	*RAPSN*
914	Meconium Ileus	*GUCY2C*
915	Glycosylphosphatidylinositol Biosynthesis Defect 15	*GPAA1*
916	Glucocorticoid Deficiency 1	*MC2R*
917	Glucocorticoid Deficiency 2	*MRAP*
918	Glucocorticoid Deficiency 4	*NNT*
919	Glycogen Storage Disease Type Ia	*G6PC*
920	Glycogen Storage Disease Type Ib	*SLC37A4*
921	Glycogen Storage Disease Type Ic	*SLC37A4*
922	Glycogen Storage Disease Type II	*GAA*
923	Glycogen Storage Disease type IV	*GBE1*
924	Glycogen Storage Disease type III	*AGL*
925	Glycogen storage disease type IXa1/IXa2	*PHKA2*
926	Glycogen storage disease type IXb	*PHKB*
927	Glycogen storage disease type IXc	*PHKG2*
928	Glycogen storage disease type IXd	*PHKA1*

929	Glycogen Storage Disease type VII	*PFKM*
930	Glycogen Storage Disease type VI	*PYGL*
931	Glycogen Storage Disease type V	*PYGM*
932	Glycogen Storage Disease type XIV	*PGM1*
933	Isolated growth hormone deficiency type III	*BTK*
934	Intellectual developmental disorder, X-linked, Turner type	*HUWE1*
935	Aspartylglucosaminuria	*AGA*
936	Asparagine Synthetase Deficiency	*ASNS*
937	Sideroblastic Anemia With B-Cell Immunodeficiency, Periodic Fevers,And Developmental Delay	*TRNT1*
938	Homocystinuria-megaloblastic anemia cblE type	*MTRR*
939	Diaphanospondylodysostosis	*BMPER*
940	Encephalopathy, Progressive, With Or Without Lipodystrophy	*BSCL2*
941	Sialidosis	*NEU1*
942	Ectodermal dysplasia, Ectrodactyly, and macular dystrophy Syndrome	*CDH3*
943	Ectodermal Dysplasia 10b, Hypohidrotic/Hair/Tooth type	*EDAR*
944	Ectodermal dysplasia and immunodeficiency 1	*IKBKG*
945	Lathosterolosis	*SC5D*
946	Reticular Dysgenesis	*AK2*
947	Epilepsy, early-onset, vitamin B6-dependent	*PLPBP*
948	Vitamin D-dependent rickets Type IA	*CYP27B1*
949	Vitamin K-dependent clotting factors, combined deficiency of, 2	*VKORC1*
950	Abetalipoproteinemia	*MTTP*
951	Alacrima, Achalasia, And Mental Retardation Syndrome	*GMPPA*
952	Lissencephaly 4	*NDE1*
953	Lissencephaly 5	*LAMB1*
954	Lissencephaly 6	*KATNB1*
955	Lissencephaly 8	*TMTC3*
956	Acheiropody	*LMBR1*
957	Atransferrinemia	*TF*
958	Chorea-Acanthocytosis	*VPS13A*
959	Glutaric acidemia IIA	*ETFA*
960	Glutaric acidemia IIB	*ETFB*
961	Glutaric acidemia IIC	*ETFDH*
962	Glutaric Acidemia I	*GCDH*
963	Fatal Infantile Cardioencephalomyopathy due to Cytochrome c Oxidase Deficiency 1	*SCO2*

964	Fatal Infantile Cardioencephalomyopathy due to Cytochrome c Oxidase Deficiency 2	*COX15*
965	Congenital Diarrhea 8, Secretory Sodium	*SLC9A3*
966	Congenital Cataracts, Hearing Loss, And Neurodegeneration	*SLC33A1*
967	Congenital Bile Acid Synthesis Defect 1	*HSD3B7*
968	Congenital Bile Acid Synthesis Defect 2	*AKR1D1*
969	Congenital Bile Acid Synthesis Defect 3	*CYP7B1*
970	Congenital short bowel syndrome(CLMP)	*CLMP*
971	Multiple congenital anomalies-hypotonia-seizures syndrome 2	*PIGA*
972	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect	*LGI4*
973	Hypothyroidism Congenital Nongoitrous 1	*TSHR*
974	Hypothyroidism Congenital Nongoitrous 4	*TSHB*
975	Congenital Dyserythropoietic Anemia Type II	*SEC23B*
976	Congenital Myasthenic Syndrome 10	*DOK7*
977	Congenital Myasthenic Syndrome 13	*DPAGT1*
978	Congenital Myasthenic Syndrome 14	*ALG2*
979	Congenital Myasthenic Syndrome 20	*SLC5A7*
980	Congenital Myasthenic Syndrome 3B, fast-channel	*CHRND*
981	Congenital Myasthenic Syndrome 4A, slow-channel	*CHRNE*
982	Congenital Myasthenic Syndrome 5	*COLQ*
983	Congenital Myasthenic Syndrome 6	*CHAT*
984	Congenital Myasthenic Syndrome 9	*MUSK*
985	Muscular dystrophy, congenital, with cataracts and intellectual disability	*INPP5K*
986	Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies type A 10	*RXYLT1*
987	Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies type A 11	*B3GALNT2*
988	Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies type A 12	*POMK*
989	Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies type A1	*POMT1*
990	Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies type A 2	*POMT2*
991	Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies type A 3	*POMGNT1*
992	Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies type A 4	*FKTN*
993	Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies type A 5	*FKRP*

994	Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies type A 6	*LARGE1*
995	Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies type A 8	*POMGNT2*
996	Congenital Stationary Night Blindness, Type 1A	*NYX*
997	Congenital Stationary Night Blindness, Type 1E	*GPR179*
998	Congenital Stationary Night Blindness, Type 2B	*CABP4*
999	Congenital Sodium Diarrhea	*SPINT2*
1000	Congenital Hydrocephalus 1	*CCDC88C*
1001	Congenital Prothrombin Deficiency	*F2*
1002	Lipodystrophy, congenital generalized, type 4	*CAVIN1*
1003	Adrenal Insufficiency, Congenital, with 46XY Sex Reversal, Partial or Complete	*CYP11A1*
1004	Congenital Disorders of Glycosylation Ia	*PMM2*
1005	Congenital Disorders of Glycosylation Ib	*MPI*
1006	Congenital Disorders of Glycosylation Ic	*ALG6*
1007	Congenital Disorders of Glycosylation Id	*ALG3*
1008	Congenital Disorders of Glycosylation Ig	*ALG12*
1009	Congenital Disorders of Glycosylation Ih	*ALG8*
1010	Congenital Disorders of Glycosylation Ik	*ALG1*
1011	Congenital Disorders of Glycosylation Il	*ALG9*
1012	Congenital Disorders of Glycosylation Im	*DOLK*
1013	Congenital Disorders of Glycosylation In	*RFT1*
1014	Congenital Disorders of Glycosylation Ip	*ALG11*
1015	Congenital Disorders of Glycosylation Iq	*SRD5A3*
1016	Congenital Disorders of Glycosylation Iv	*NGLY1*
1017	Congenital Disorders of Glycosylation Iy	*SSR4*
1018	Congenital Disorders of Glycosylation IIa	*MGAT2*
1019	Congenital Disorders of Glycosylation IIe	*COG7*
1020	Congenital Disorders of Glycosylation IIk	*TMEM165*
1021	Congenital Disorders of Glycosylation III	*COG6*
1022	Congenital Disorders of Glycosylation Iin	*SLC39A8*
1023	Congenital Disorders of Glycosylation Iio	*CCDC115*
1024	Congenital Insensitivity to Pain	*SCN9A*
1025	Congenital alopecia and T-Cell Immunodeficiency and nail dystrophy	*FOXN1*
1026	Congenital Amegakaryocytic Thrombocytopenia	*MPL*
1027	Congenital Afibrinogenemia	*FGA,FGB,FGG*
1028	Congenital fiber-type disproportion myopathy	*ACTA1*
1029	Congenital Thrombotic thrombocytopenic purpura	*ADAMTS13*
1030	Camptodactyly-Arthropathy-Coxa Vara-Pericarditis Syndrome	*PRG4*
1031	Fibrochondrogenesis 1	*COL11A1*

1032	Fibrochondrogenesis 2	*COL11A2*
1033	Mitochondrial DNA depletion syndrome 11	*MGME1*
1034	Mitochondrial DNA depletion syndrome 13	*FBXL4*
1035	Mitochondrial DNA depletion syndrome 1	*TYMP*
1036	Mitochondrial DNA depletion syndrome 2	*TK2*
1037	Mitochondrial DNA depletion syndrome 3	*DGUOK*
1038	Mitochondrial DNA depletion syndrome 4A	*POLG*
1039	Mitochondrial DNA depletion syndrome 5	*SUCLA2*
1040	Mitochondrial DNA depletion syndrome 6	*MPV17*
1041	Mitochondrial DNA depletion syndrome 7	*TWNK*
1042	Mitochondrial DNA depletion syndrome 8A/8B	*RRM2B*
1043	Mitochondrial DNA depletion syndrome 9	*SUCLG1*
1044	Mitochondrial Short-Chain Enoyl-CoA Hydratase 1 Deficiency	*ECHS1*
1045	Mitochondrial complex I deficiency, nuclear type 1	*NDUFS4*
1046	Mitochondrial complex I deficiency, nuclear type 10	*NDUFAF2*
1047	Mitochondrial complex I deficiency, nuclear type 12	*NDUFA1*
1048	Mitochondrial complex I deficiency, nuclear type 14	*NDUFA11*
1049	Mitochondrial complex I deficiency, nuclear type 16	*NDUFAF5*
1050	Mitochondrial complex I deficiency, nuclear type 17	*NDUFAF6*
1051	Mitochondrial complex I deficiency, nuclear type 19	*FOXRED1*
1052	Mitochondrial complex I deficiency, nuclear type 21	*NUBPL*
1053	Mitochondrial complex I deficiency, nuclear type 22	*NDUFA10*
1054	Mitochondrial complex I deficiency, nuclear type 4	*NDUFV1*
1055	Mitochondrial complex I deficiency, nuclear type 5	*NDUFS1*
1056	Mitochondrial complex I deficiency, nuclear type 6	*NDUFS2*
1057	Mitochondrial complex I deficiency, nuclear type 7	*NDUFV2*
1058	Mitochondrial complex I deficiency, nuclear type 9	*NDUFS6*
1059	Mitochondrial complex I deficiency, nuclear type 2	*NDUFS8*
1060	Mitochondrial complex I deficiency, nuclear type 3	*NDUFS7*
1061	Mitochondrial complex II deficiency, nuclear type 2	*SDHAF1*
1062	Mitochondrial complex IV deficiency, nuclear type 1	*SURF1*
1063	Mitochondrial complex IV deficiency, nuclear type 3	*COX10*
1064	Mitochondrial complex III deficiency nuclear type 2	*TTC19*
1065	Mitochondrial complex III deficiency nuclear type 4	*UQCRQ*
1066	Mitochondrial complex III deficiency nuclear type 5	*UQCRC2*
1067	Mitochondrial complex III deficiency nuclear type 8	*LYRM7*
1068	Mitochondrial complex IV deficiency, nuclear type 11	*COX20*
1069	Mitochondrial complex IV deficiency, nuclear type 12	*PET100*
1070	Mitochondrial complex IV deficiency, nuclear type 17	*COA8*
1071	Mitochondrial complex I deficiency, nuclear type 20	*ACAD9*
1072	Mitochondrial Complex V (ATP Synthase) Deficiency, Nuclear Type 2	*TMEM70*

1073	Mitochondrial Neurodevelopmental disorder with abnormal movements and lactic acidosis with or without seizures	*WARS2*
1074	Adenylosuccinase Deficiency	*ADSL*
1075	Adenosine Deaminase Deficiency	*ADA*
1076	Infantile Transient Liver Failure	*TRMU*
1077	Cerebellofaciodental Syndrome	*BRF1*
1078	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 1	*VLDLR*
1079	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2	*WDR81*
1080	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4	*ATP8A2*
1081	Microcephalic Osteodysplastic Primordial Dwarfism, Type I	*RNU4ATAC*
1082	Microcephalic Osteodysplastic Primordial Dwarfism, Type II	*PCNT*
1083	Microcephaly,Short Stature, And Limb Abnormalities	*DONSON*
1084	Microcephaly, Seizures, Spasticity, And Brain Calcifications	*PCDH12*
1085	Microcephaly, Epilepsy, and Diabetes Syndrome	*IER3IP1*
1086	Microcephaly, Short Stature, And Impaired Glucose Metabolism 1	*TRMT10A*
1087	Microcephaly-Capillary Malformation Syndrome	*STAMBP*
1088	Microphthalmia with coloboma 3	*VSX2*
1089	Acrodermatitis Enteropathica, Zinc-Deficiency Type	*SLC39A4*
1090	Neonatal Bartter syndrome type 4A with sensorineural deafness	*BSND*
1091	Citrullinemia, Type II, Neonatal-Onset	*SLC25A13*
1092	Neonatal Diabetes Mellitus with Congenital Hypothyroidism	*GLIS3*
1093	Neonatal Severe Hyperparathyroidism	*CASR*
1094	Lethal Neonatal Rigidity and Multifocal Seizure Syndrome	*BRAT1*
1095	Hemochromatosis, type 2A	*HJV*
1096	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease	*ARPC1B*
1097	Homocystinuria due to MTHFR deficiency	*MTHFR*
1098	Moyamoya disease 6 with or without achalasia	*GUCY1A1*
1099	Fumarase Deficiency	*FH*
1100	Severe Congenital Neutropenia, Autosomal Recessive,3	*HAX1*
1101	Severe Congenital Neutropenia, Autosomal Recessive,4	*G6PC3*
1102	Severe Congenital Neutropenia, Autosomal Recessive,5	*VPS45*

1103	Severe Congenital Neutropenia, Autosomal Recessive,6	*JAGN1*
1104	Fucosidosis	*FUCA1*
1105	Inflammatory Bowel Disease 28	*IL10RA*
1106	Oculocutaneous Albinism Type 1	*TYR*
1107	Oculocutaneous Albinism Type 2	*OCA2*
1108	Oculocutaneous Albinism Type 3	*TYRP1*
1109	Oculocutaneous Albinism Type 4	*SLC45A2*
1110	Oculocutaneous Albinism Type 6	*SLC24A5*
1111	Oculocutaneous Albinism Type 7	*LRMDA*
1112	Anterior segement dysgenesis 2	*FOXE3*
1113	Anterior segement dysgenesis 7	*PXDN*
1114	Polyhydramnios, Megalencephaly, And Symptomatic Epilepsy	*STRADA*
1115	Insulin-Like Growth Factor I, Resistance to	*IGF1R*
1116	Hereditary Sensory and Autonomic Neuropathy type IIB	*RETREG1*
1117	Hereditary Sensory and Autonomic Neuropathy type II	*WNK1*
1118	Hereditary Sensory and Autonomic Neuropathy type V	*NGF*
1119	Hereditary Sensory and Autonomic Neuropathy type VIII	*PRDM12*
1120	Hereditary Fructose Intolerance	*ALDOB*
1121	Hereditary Hyperekplexia 3	*SLC6A5*
1122	Hereditary 4	*ATAD1*
1123	Hemochromatosis type 2B	*HAMP*
1124	Hereditary Folate Malabsorption	*SLC46A1*
1125	Hereditary Motor And Sensory Neuropathy type VIB	*SLC25A46*
1126	Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum	*SLC12A6*
1127	Ethylmalonic Encephalopathy	*ETHE1*
1128	Hemophilia B	*F9*
1129	Isobutyryl-CoA dehydrogenase deficiency	*ACAD8*
1130	Metachromatic Leukodystrophy due to Arylsulfatase A	*ARSA*
1131	Isovaleric Acidemia	*IVD*
1132	Infantile liver failure syndrome 1	*LARS1*
1133	Striatonigral Degeneration, Infantile	*NUP62*
1134	Hypophosphatasia, infantile	*ALPL*
1135	Generalized Arterial Calcification of Infancy 2	*ABCC6*
1136	Hypotonia, Infantile, With Psychomotor Retardation And Characteristic facies 1	*NALCN*
1137	Hypotonia, Infantile, With Psychomotor Retardation And Characteristic facies 2	*UNC80*
1138	Hypotonia, Infantile, With Psychomotor Retardation And Characteristic facies 3	*TBCK*
1139	Infantile Parkinsonism-Dystonia	*SLC6A3*

1140	Infantile Sudden cardiac failure	*PPA2*
1141	Free sialic acid storage disease, infantile form	*SLC17A5*
1142	Infantile Cerebellar-Retinal Degeneration	*ACO2*
1143	Right Atrial Isomerism	*GDF1*
1144	Primary Autosomal Recessive Microcephaly 10	*ZNF335*
1145	Primary Autosomal Recessive Microcephaly 15	*MFSD2A*
1146	Primary Autosomal Recessive Microcephaly 17	*CIT*
1147	Primary Autosomal Recessive Microcephaly 1	*MCPH1*
1148	Primary Autosomal Recessive Microcephaly 20	*KIF14*
1149	Primary Autosomal Recessive Microcephaly 2, With Or Without Corticalmalformations	*WDR62*
1150	Primary Autosomal Recessive Microcephaly 3	*CDK5RAP2*
1151	Primary Autosomal Recessive Microcephaly 4	*KNL1*
1152	Primary Autosomal Recessive Microcephaly 5	*ASPM*
1153	Primary Autosomal Recessive Microcephaly 6	*CENPJ*
1154	Primary Autosomal Recessive Microcephaly 7	*STIL*
1155	Primary Coenzyme Q10 deficiency 1	*COQ2*
1156	Primary Coenzyme Q10 deficiency 4	*COQ8A*
1157	Primary Coenzyme Q10 deficiency 6	*COQ6*
1158	Primary Coenzyme Q10 deficiency 7	*COQ4*
1159	Primary Hyperoxaluria Type I	*AGXT*
1160	Primary Open Angle Glaucoma 3A	*CYP1B1*
1161	Primary Carnitine Deficiency	*SLC22A5*
1162	Distal Arthrogryposis type 5D	*ECEL1*
1163	Distal Renal Tubular Acidosis with Hemolytic Anemia	*SLC4A1*
1164	Early-Onset Myopathy, Areflexia, Respiratory Distress, and Dysphagia	*MEGF10*
1165	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum	*TBCD*
1166	Early Infantile Epileptic Encephalopathy 16	*TBC1D24*
1167	Early Infantile Epileptic Encephalopathy 25	*SLC13A5*
1168	Early Infantile Epileptic Encephalopathy 28	*WWOX*
1169	Early Infantile Epileptic Encephalopathy 34	*SLC12A5*
1170	Early Infantile Epileptic Encephalopathy 37	*FRRS1L*
1171	Early Infantile Epileptic Encephalopathy 38	*ARV1*
1172	Early Infantile Epileptic Encephalopathy 3	*SLC25A22*
1173	Early Infantile Epileptic Encephalopathy 44	*UBA5*
1174	Early Infantile Epileptic Encephalopathy 48	*AP3B2*
1175	Early Infantile Epileptic Encephalopathy 49	*DENND5A*
1176	Early Infantile Epileptic Encephalopathy 8	*ARHGEF9*
1177	Early Infantile Epileptic Encephalopathy 9	*PCDH19*
1178	Proliferative Vasculopathy And Hydranencephaly-Hydrocephaly Syndrome	*FLVCR2*

1179	Mucolipidosis III Alpha&Beta	*GNPTAB*
1180	Mucolipidosis III Gamma	*GNPTG*
1181	Mucolipidosis IV	*MCOLN1*
1182	Long-Chain 3-Hydroxyacyl-Coa Dehydrogenase Deficiency	*HADHA*
1183	Xeroderma Pigmentosum Group A	*XPA*
1184	Xeroderma Pigmentosum Group C	*XPC*
1185	Xeroderma Pigmentosum Group G	*ERCC5*
1186	Cortical Malformations, Occipital	*LAMC3*
1187	Branched-chain Ketoacid Dehydrogenase Kinase Deficiency	*BCKDK*
1188	Limb-Girdle Muscular Dystrophy type 2A	*CAPN3*
1189	Limb-Girdle Muscular Dystrophy type 2B	*DYSF*
1190	Limb-Girdle Muscular Dystrophy type 2C	*SGCG*
1191	Limb-Girdle Muscular Dystrophy type 2D	*SGCA*
1192	Limb-Girdle Muscular Dystrophy type 2E	*SGCB*
1193	Limb-Girdle Muscular Dystrophy type 2F	*SGCD*
1194	Limb-Girdle Muscular Dystrophy type 2G	*TCAP*
1195	Limb-Girdle Muscular Dystrophy type 2H	*TRIM32*
1196	Limb-Girdle Muscular Dystrophy type 2S	*TRAPPC11*
1197	Limb-Girdle Muscular Dystrophy type 2T	*GMPPB*
1198	Acromesomelic dysplasia 1	*NPR2*
1199	Acromesomelic dysplasia 2A	*GDF5*
1200	Acromesomelic dysplasia 3	*BMPR1B*
1201	Rhizomelic Chondrodysplasia Punctata type 1	*PEX7*
1202	Rhizomelic Chondrodysplasia Punctata type 2	*GNPAT*
1203	Rhizomelic chondrodysplasia punctata, type 3	*AGPS*
1204	Lipoyltransferase 1 deficiency	*LIPT1*
1205	Lipoid Congenital Adrenal Hyperplasia	*STAR*
1206	Pycnodysostosis	*CTSK*
1207	Lethal Arthrogryposis With Anterior Horn Cell Disease	*GLE1*
1208	Popliteal Pterygium Syndrome, Lethal Type	*RIPK4*
1209	Lethal congenital contracture syndrome 11	*GLDN*
1210	Lethal Congenital Contracture Syndrome 2	*ERBB3*
1211	Lethal Congenital Contracture Syndrome 3	*PIP5K1C*
1212	Lethal Congenital Contracture Syndrome 7	*CNTNAP1*
1213	Lethal Restrictive Dermopathy	*LMNA,ZMPSTE24*
1214	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies	*OTUD6B*
1215	Intellectual developmental disorder with cardiac arrhythmia	*GNB5*
1216	Acyl-CoA Dehydrogenase Deficiency,Medium-Chain	*ACADM*
1217	Foveal Hypoplasia 2	*SLC38A8*

1218	Centronuclear Myopathy 2	*BIN1*
1219	Severe Combined Immunodeficiency with Microcephaly, Growth Retardation, and Sensitivity to Ionizing Radiation	*NHEJ1*
1220	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive	*IL7R*
1221	Transcobalamin II Deficiency	*TCN2*
1222	Transaldolase Deficiency	*TALDO1*
1223	Autoimmune Polyendocrine Syndrome Type 1	*AIRE*
1224	Syndromic Microphthalmia 12	*RARB*
1225	Microphthalmia, syndromic 9	*STRA6*
1226	Histiocytosis-lymphadenopathy plus syndrome	*SLC29A3*
1227	Hypoplastic Left Heart Syndrome 1	*GJA1*
1228	Aromatic L-Amino Acid Decarboxylase Deficiency	*DDC*
1229	Orofaciodigital Syndrome XIV	*C2CD3*
1230	Cone-Rod Dystrophy 16	*CFAP418*
1231	3-Methylcrotonyl-CoA carboxylase 1 deficiency	*MCCC1*
1232	3-Methylcrotonyl-CoA carboxylase 2 deficiency	*MCCC2*
1233	Gitelman syndrome	*SLC12A3*
1234	Glucose-6-Phosphate Dehydrogenase Deficiency	*G6PD*
1235	Short Chain Acyl-CoA Dehydrogenase Deficiency	*ACADS*
1236	Achromatopsia 3	*CNGB3*