Carrier Screening Condition List (1200+ panel)

No.	Diseases	Genes
1	Ichthyosis, congenital, autosomal recessive 4A	ABCA12
2	Ichthyosis, congenital, autosomal recessive 4B (harlequin)	ABCA12
3	Cholestasis, progressive familial intrahepatic 2	ABCB11
4	Cholestasis, progressive familial intrahepatic 3	ABCB4
5	Sitosterolemia 2	ABCG5
6	Sitosterolemia 1	ABCG8
7	Acyl-CoA dehydrogenase, medium chain, deficiency of	ACADM
8	Acyl-CoA dehydrogenase, short-chain, deficiency of	ACADS
9	VLCAD deficiency	ACADVL
10	Alpha-methylacetoacetic aciduria	ACAT1
11	Adenosine Deaminase Deficiency	ADA
12	Joubert Syndrome 3	AHI1
13	Sjogren-Larsson syndrome	ALDH3A2
14	Fructose intolerance, hereditary	ALDOB
15	Hypophosphatasia, childhood	ALPL
16	Hypophosphatasia, infantile	ALPL
17	Glycine encephalopathy 2	AMT
18	Metachromatic leukodystrophy	ARSA
19	Mucopolysaccharidosis type VI (Maroteaux-Lamy)	ARSB
20	Argininosuccinic aciduria	ASL
21	Canavan Disease	ASPA
22	Citrullinemia	ASS1
23	Wilson Disease	ATP7B
24	Maple syrup urine disease, type Ia	BCKDHA
25	Maple syrup urine disease, type Ib	BCKDHB
26	Biotinidase Deficiency	BTD
27	Albinism, oculocutaneous, type VII	LRMDA
28	Joubert Syndrome 17	CPLANE1
29	Muscular dystrophy, limb-girdle, autosomal recessive 1	CAPN3
30	Homocystinuria, B6-responsive and nonresponsive types	CBS
31	Joubert Syndrome 9	CC2D2A
32	COACH syndrome 2	CC2D2A
33	Joubert Syndrome 5	CEP290
34	Meckel Syndrome 4	CEP290
35	Cystic Fibrosis	CFTR
36	Ceroid lipofuscinosis, neuronal, 3	CLN3
37	Ceroid lipofuscinosis, neuronal, 5	CLN5
38	Ceroid lipofuscinosis, neuronal, 6B (Kufs type)	CLN6

39	Ceroid lipofuscinosis, neuronal, 6A	CLN6
40	Epidermolysis bullosa, junctional 4, intermediate	COL17A1
41	Alport syndrome 3B, autosomal recessive	COL4A3
42	Alport syndrome 2, autosomal recessive	COL4A4
43	Epidermolysis bullosa dystrophica, autosomal recessive	COL7A1
44	Carbamoylphosphate Synthetase I Deficiency	CPS1
45	Cystinosis, nephropathic	CTNS
46	Maple syrup urine disease, type II	DBT
47	Smith-Lemli-Opitz syndrome	DHCR7
48	Dihydrolipoamide dehydrogenase deficiency	DLD
49	Duchenne Muscular Dystrophy	DMD
50	Immunodeficiency-centromeric instability-facial anomalies syndrome 1	DNMT3B
51	Muscular dystrophy, limb-girdle, autosomal recessive 2	DYSF
52	Ectodermal dysplasia 1, hypohidrotic, X-linked	EDA
53	Glutaric acidemia IIA	ETFA
54	Glutaric acidemia IIB	ETFB
55	Glutaric acidemia IIC	ETFDH
56	Ellis-van Creveld Syndrome	EVC2, EVC
57	Hemophilia B	F9
58	Tyrosinemia, type I	FAH
59	Fanconi anemia, complementation group A	FANCA
60	Fanconi anemia, complementation group C	FANCC
61	Fanconi anemia, complementation group D2	FANCD2
62	Fanconi anemia, complementation group G	FANCG
63	Fanconi anemia, complementation group I	FANCI
64	Glycogen storage disease Ia	G6PC1
65	Glycogen storage disease II	GAA
66	Krabbe Disease	GALC
67	Mucopolysaccharidosis IVA	GALNS
68	Galactosemia	GALT
69	Glycogen storage disease IV	GBE1
70	Glutaricaciduria, type I	GCDH
71	Deafness, autosomal recessive 1A	GJB2
72	Fabry Disease	GLA
73	Mucopolysaccharidosis type IVB (Morquio)	GLB1
74	Glycine encephalopathy1	GLDC
75	Mucopolysaccharidosis type IIID	GNS
76	Ocular albinism, type I, Nettleship-Falls type	GPR143
77	Hyperinsulinemic hypoglycemia, familial, 4	HADH
78	Thalassemia, alpha-	HBA1, HBA2
79	Thalassemia, beta	HBB
80	Sickle cell disease	HBB

81	Tay-Sachs Disease	HEXA
82	Mucopolysaccharidosis type IIIC (Sanfilippo C)	HGSNAT
83	Holocarboxylase synthetase deficiency	HLCS
84	HMG-CoA synthase-2 deficiency	HMGCS2
85	Hermansky-Pudlak Syndrome 1	HPS1
86	Hermansky-Pudlak Syndrome 3	HPS3
87	Mucopolysaccharidosis II	IDS
88	Mucopolysaccharidosis Ih/s	IDUA
89	Mucopolysaccharidosis Is	IDUA
90	Mucopolysaccharidosis Ih	IDUA
91	Severe combined immunodeficiency, X-linked	IL2RG
92	Isovaleric Acidemia	IVD
93	Hyperinsulinemic hypoglycemia, familial, 2	KCNJ11
94	LAMA3-Related Junctional Epidermolysis Bullosa	LAMA3
95	LAMB3-Related Junctional Epidermolysis Bullosa	LAMB3
96	LAMC2-Related Junctional Epidermolysis Bullosa	LAMC2
97	Osteoporosis-pseudoglioma syndrome	LRP5
98	Mannosidosis, alpha-, types I and II	MAN2B1
99	3-Methylcrotonyl-CoA carboxylase 1 deficiency	MCCC1
100	3-Methylcrotonyl-CoA carboxylase 2 deficiency	MCCC2
101	Methylmalonyl-CoA epimerase deficiency	MCEE
102	Ceroid lipofuscinosis, neuronal, 7	MFSD8
103	Megalencephalic Leukoencephalopathy with Subcortical Cysts 1	MLC1
104	Methylmalonic aciduria, vitamin B12-responsive, cblA type	MMAA
105	Methylmalonic aciduria, vitamin B12-responsive, cblB type	MMAB
106	Methylmalonic aciduria and homocystinuria, cblC type	MMACHC
107	Methylmalonic aciduria and homocystinuria, cblD type	MMADHC
108	Molybdenum Cofactor Deficiency A	MOCS1
109	Myopathy, centronuclear, X-linked	MTM1
110	Homocystinuria-megaloblastic anemia, cblG complementation type	MTR
111	Homocystinuria-megaloblastic anemia, cbl E type	MTRR
112	Methylmalonic aciduria, mut(0) type	MMUT
113	Mucopolysaccharidosis type IIIB (Sanfilippo B)	NAGLU
114	Niemann-Pick disease, type C1	NPC1
115	Niemann-pick disease, type C2	NPC2
116	Nephronophthisis 3	NPHP3
117	Nephrotic syndrome, type 1	NPHS1
118	Adrenal hypoplasia, congenital	NR0B1
119	Albinism, oculocutaneous, type II	OCA2
120	Ornithine Transcarbamylase Deficiency	OTC
121	Phenylketonuria	PAH

122	Propionicacidemia	PCCA, PCCB
123	Peroxisome biogenesis disorder 1A (Zellweger)	PEX1
124	Congenital disorder of glycosylation, type Ia	PMM2
125	Ceroid lipofuscinosis, neuronal, 1	PPT1
126	Hemophagocytic lymphohistiocytosis, familial, 2	PRF1
127	Hyperphenylalaninemia, BH4-deficient, A	PTS
128	Omenn syndrome	RAG1, RAG2
129	Severe combined immunodeficiency, B cell-negative	RAG1, RAG2
130	Muscular dystrophy, limb-girdle, autosomal recessive 3	SGCA
131	Muscular dystrophy, limb-girdle, autosomal recessive 5	SGCG
132	Mucopolysaccharidosis type IIIA (Sanfilippo A)	SGSH
133	Carnitine deficiency, systemic primary	SLC22A5
134	Albinism, oculocutaneous, type VI	SLC24A5
135	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	SLC25A15
136	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	SLC26A4
137	Glycogen storage disease Ib	SLC37A4
138	Glycogen storage disease Ic	SLC37A4
139	Albinism, oculocutaneous, type IV	SLC45A2
140	Niemann-Pick disease, type A	SMPD1
141	Niemann-Pick disease, type B	SMPD1
142	Netherton syndrome	SPINK5
143	Hemophagocytic lymphohistiocytosis, familial, 4	STX11
144	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease	STXBP2
145	Osteopetrosis, autosomal recessive 1	TCIRG1
146	Ichthyosis, congenital, autosomal recessive 1	TGM1
147	Cholestasis, progressive familial intrahepatic 4	TJP2
148	Joubert Syndrome 2	TMEM216
149	Meckel Syndrome 2	TMEM216
150	Joubert Syndrome 6	TMEM67
151	Meckel Syndrome 3	TMEM67
152	COACH syndrome 1	TMEM67
153	Nephronophthisis 11	TMEM67
154	Ceroid lipofuscinosis, neuronal, 2	TPP1
155	Oculocutaneous Albinism Type 1	TYR
156	Albinism, oculocutaneous, type III	TYRP1
157	Hemophagocytic lymphohistiocytosis, familial, 3	UNC13D
158	Wolfram Syndrome 1	WFS1
159	Spinal Muscular Atrophy	SMN1
160	Aspartylglucosaminuria	AGA
161	Polycystic kidney disease 4, with or without hepatic disease	PKHD1

162	Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	SACS
163	Bloom syndrome	BLM
164	Combined pituitary hormone deficiency 2	PROP1
165	Dysautonomia, familial	ELP1
166	GRACILE Syndrome	BCS1L
167	Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum	SLC12A6
168	Usher Syndrome Type 1F	PCDH15
169	Usher Syndrome Type 3A	CLRN1
170	Argininemia	ARG1
171	LCHAD deficiency	HADHA
172	Mitochondrial trifunctional protein deficiency 1	HADHA
173	Mitochondrial trifunctional protein deficiency 2	HADHB
174	Ataxia-telangiectasia	ATM
175	Hemolytic anemia, G6PD deficient (favism)	G6PD
176	Galactokinase deficiency with cataracts	GALK1
177	Mucolipidosis IV	MCOLN1
178	Familial Mediterranean fever, AR	MEFV
179	Nemaline myopathy 2, autosomal recessive	NEB
180	Alpha- 1 antitrypsin deficiency	SERPINA1
181	Gitelman syndrome	SLC12A3
182	Spastic paraplegia 11, autosomal recessive	SPG11
183	Segawa syndrome, recessive	TH
184	Laron dwarfism	GHR
185	Congenital Adrenal Hyperplasia due to 11-beta-Hydroxylase-Deficiency	CYP11B1
186	Congenital Adrenal Hyperplasia due to 17-alpha Hydroxylase Deficiency	CYP17A1
187	2,4-Dienoyl-CoA Reductase Deficiency	NADK2
188	2-Methylbutyryl Glycinuria	ACADSB
189	3MC Syndrome 1	MASP1
190	3MC Syndrome 2	COLEC11
191	3-M Syndrome 2	OBSL1
192	3-beta-Hydroxysteroid Dehydrogenase Deficiency	HSD3B2
193	3-Methylglutaconic Aciduria type 1	AUH
194	3-Methylglutaconic Aciduria type 3	OPA3
195	3-Methylglutaconic Aciduria type 5	DNAJC19
196	3-Methylglutaconic Aciduria type 8	HTRA2
197	3-Methylglutaconic Aciduria With Cataracts, Neurologic Involvement,And Neutropenia	CLPB
198	3-Methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	SERAC1
199	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	HMGCL

200	3-Hydroxyisobutyryl-Coa Hydrolase Deficiency	HIBCH
201	Pyridoxal 5'-Phosphate-dependent Epilepsy	PNPO
202	GM2-gangliosidosis, AB variant	GM2A
203	Adams-Oliver Syndrome 2	DOCK6
204	Adams-Oliver Syndrome 4	EOGT
205	Aicardi-Goutieres Syndrome 1	TREX1
206	Aicardi-Goutieres Syndrome 2	RNASEH2B
207	Aicardi-Goutieres Syndrome 3	RNASEH2C
208	Aicardi-Goutieres Syndrome 4	RNASEH2A
209	Aicardi-Goutieres Syndrome 5	SAMHD1
210	Aicardi-Goutieres Syndrome 6	ADAR
211	Al Kaissi syndrome	CDK10
212	Alazami Syndrome	LARP7
213	Alkuraya-Kucinskas Syndrome	BLTP1
214	Allan-Herndon-Dudley syndrome	SLC16A2
215	Alstrom Syndrome	ALMS1
216	Anauxetic dysplasia 2	POP1
217	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis	POR
218	Arts Syndrome	PRPS1
219	Athabaskan Brain Stem Dysgenesis Syndrome	HOXA1
220	Baller-Gerold Syndrome	RECQL4
221	Bardet-Biedl syndrome 10	BBS10
222	Bardet-Biedl syndrome 12	BBS12
223	Bardet-Biedl syndrome 16	SDCCAG8
224	Bardet-Biedl syndrome 17	LZTFL1
225	Bardet-Biedl syndrome 1	BBS1
226	Bardet-Biedl syndrome 2	BBS2
227	Bardet-Biedl syndrome 3	ARL6
228	Bardet-Biedl syndrome 4	BBS4
229	Bardet-Biedl syndrome 5	BBS5
230	Bardet-Biedl syndrome 7	BBS7
231	Bardet-Biedl syndrome 8	TTC8
232	Bardet-Biedl syndrome 9	BBS9
233	Barth syndrome	TAFAZZIN
234	Bartter Syndrome 1	SLC12A1
235	Bartter Syndrome 2	KCNJ1
236	Basel-Vanagait-Smirin-Yosef syndrome	MED25
237	BEHR syndrome	OPA1
238	BH4-Deficient Hyperphenylalaninemia C	QDPR
239	Chondrodysplasia, Blomstrand Type	PTH1R
240	Borjeson-Forssman-Lehmann syndrome	PHF6
241	Boucher-Neuhauser syndrome	PNPLA6

242	Brachyolmia Type 4	PAPSS2
243	Brown-Vialetto-Van Laere syndrome 1	SLC52A3
244	Brown-Vialetto-Van Laere syndrome 2	SLC52A2
245	Bruck Syndrome 1	FKBP10
246	Bruck Syndrome 2	PLOD2
247	Brunner syndrome	MAOA
248	Burn-Mckeown Syndrome	TXNL4A
249	Carey-Fineman-Ziter syndrome	MYMK
250	Carpenter syndrome 1	RAB23
251	Carpenter Syndrome 2	MEGF8
252	Cerebellar Ataxia, Cayman Type	ATCAY
253	Cenani-Lenz Syndactyly Syndrome	LRP4
254	Chanarin-Dorfman syndrome	ABHD5
255	Charcot-Marie-Tooth disease, Axonal, Type 2A2B	MFN2
256	Charcot-Marie-Tooth disease type 3	PRX
257	Charcot-Marie-Tooth disease type 4B1	MTMR2
258	Charcot-Marie-Tooth disease type 4C	SH3TC2
259	Charcot-Marie-Tooth disease type 4D	NDRG1
260	Chediak-Higashi Syndrome	LYST
261	X-Linked Syndromic Mental Retardation, Christianson type	SLC9A6
262	Chudley-McCullough Syndrome	GPSM2
263	X-Linked Syndromic Mental Retardation, Claes-Jensen type	KDM5C
264	Cockayne Syndrome A	ERCC8
265	Cockayne Syndrome B	ERCC6
266	CODAS Syndrome	LONP1
267	Coffin-Lowry Syndrome	RPS6KA3
268	Cohen Syndrome	VPS13B
269	Crigler-Najjar syndrome type 1	UGT1A1
270	D,L-2-hydroxyglutaric aciduria	SLC25A1
271	D-2-hydroxyglutaric aciduria 1	D2HGDH
272	DCLRE1C-Related Severe Combined Immunodeficiency	DCLRE1C
273	Desbuquois Dysplasia 1	CANT1
274	Desbuquois Dysplasia 2	XYLT1
275	Donnai-Barrow syndrome	LRP2
276	Dyggve-Melchior-Clausen Disease	DYM
277	D-Glyceric Aciduria	GLYCTK
278	Ehlers-Danlos Syndrome type VI	PLOD1
279	Ehlers-Danlos syndrome type VIIC	ADAMTS2
280	Ehlers-Danlos Syndrome with Progressive Kyphoscoliosis, Myopathy, and Hearing Loss	FKBP14
281	Elsahy-Waters syndrome	CDH11
282	MULTIPLE PTERYGIUM SYNDROME, ESCOBAR VARIANT	CHRNG

283	Fanconi-Bickel Syndrome	SLC2A2
284	Farber Lipogranulomatosis	ASAH1
285	FG Syndrome Type 2	FLNA
286	FG Syndrome Type 4	CASK
287	Filippi Syndrome	CKAP2L
288	Frank-ter Haar Syndrome	SH3PXD2B
289	Fraser Syndrome 1	FRAS1
290	Fraser syndrome 2	FREM2
291	Fraser syndrome 3	GRIP1
292	X-linked Mental retardation, FRAXE type	AFF2
293	GABA-Transaminase Deficiency	ABAT
294	Galloway-Mowat Syndrome 1	WDR73
295	Galloway-Mowat syndrome 3	OSGEP
296	Geleophysic dysplasia 1	ADAMTSL2
297	Spondyloepimetaphyseal Dysplasia, Genevieve Type	NANS
298	Goldberg-Shprintzen syndrome	KIFBP
299	Greenberg dysplasia	LBR
300	Griscelli Syndrome 2	RAB27A
301	Dopa-Responsive Dystonia	GCH1
302	Hennekam Lymphangiectasia-Lymphedema Syndrome 1	CCBE1
303	Hennekam Lymphangiectasia-Lymphedema Syndrome 2	FAT4
304	Hermansky-Pudlak Syndrome 4	HPS4
305	Hermansky-Pudlak Syndrome 5	HPS5
306	Hermansky-Pudlak Syndrome 6	HPS6
307	X-linked immunodysregulation, polyendocrinopathy, and enteropathy	FOXP3
308	Jalili Syndrome	CNNM4
309	Jervell and Lange-Nielsen syndrome 1	KCNQ1
310	Jervell and Lange-Nielsen syndrome 2	KCNE1
311	Johanson-Blizzard Syndrome	UBR1
312	Joubert Syndrome 10	OFD1
313	Joubert Syndrome 14	TMEM237
314	Joubert Syndrome 15	CEP41
315	Joubert Syndrome 16	TMEM138
316	Joubert Syndrome 18	TCTN3
317	Joubert Syndrome 1	INPP5E
318	Joubert Syndrome 20	TMEM231
319	Joubert Syndrome 21	CSPP1
320	Joubert Syndrome 24	TCTN2
321	Joubert Syndrome 4	NPHP1
322	Joubert Syndrome 8	ARL13B
323	Kenny-Caffey Syndrome Type 1	TBCE
324	Keutel Syndrome	MGP

325	Kindler Syndrome	FERMT1
326	Knobloch Syndrome Type I	COL18A1
327	Kohlschutter-Tonz Syndrome	ROGDI
328	L-2-hydroxyglutaric aciduria	L2HGDH
329	Lafora Disease	EPM2A
330	Leber Congenital Amaurosis 12	RD3
331	Leber Congenital Amaurosis 13	RDH12
332	Leber congenital amaurosis 14	LRAT
333	Leber Congenital Amaurosis 1	GUCY2D
334	Leber Congenital Amaurosis 2	RPE65
335	Leber Congenital Amaurosis 3	SPATA7
336	Leber Congenital Amaurosis 5	LCA5
337	Leber Congenital Amaurosis 8	CRB1
338	Leber Congenital Amaurosis 9	NMNAT1
339	Lesch-Nyhan Syndrome	HPRT1
340	LIG4 syndrome	LIG4
341	Lowe Syndrome	OCRL
342	Lujan-Fryns syndrome	MED12
343	Majeed Syndrome	LPIN2
344	Marinesco-Sjogren Syndrome	SIL1
345	MASA syndrome	L1CAM
346	McKusick-Kaufman Syndrome	MKKS
347	Meckel syndrome 1	MKS1
348	Meckel syndrome 5	RPGRIP1L
349	Neonatal Severe Encephalopathy Due To MECP2 Mutations	MECP2
350	Meester-Loeys syndrome	BGN
351	Congenital Muscular Dystrophy, Megaconial type	CHKB
352	MEHMO syndrome	EIF2S3
353	Meier-Gorlin Syndrome 1	ORC1
354	Meier-Gorlin Syndrome 3	ORC6
355	Meier-Gorlin Syndrome 4	CDT1
356	Meier-Gorlin syndrome 7	CDC45
357	Menkes Disease	ATP7A
358	Merosin-deficient congenital muscular dystrophy type 1A	LAMA2
359	Miller syndrome	DHODH
360	Mitchell-Riley syndrome	RFX6
361	Mohr-Tranebjaerg syndrome	TIMM8A
362	Recurrent Pyogenic Bacterial Infections due to MYD88 Deficiency	MYD88
363	X-Linked Syndromic Mental Retardation, Nascimento-type	UBE2A
364	Naxos Disease	JUP
365	Neu-Laxova Syndrome 1	PHGDH
366	Neu-Laxova Syndrome 2	PSAT1

367	Norrie Disease	NDP
368	N-acetylglutamate synthase deficiency	NAGS
369	Ogden Syndrome	NAA10
370	Spondyloepiphyseal Dysplasia, Omani type	CHST3
371	Opitz Gbbb Syndrome, Type I	MID1
372	Opsismodysplasia	INPPL1
373	PEHO syndrome	ZNHIT3
374	Perlman Syndrome	DIS3L2
375	Perrault Syndrome 3	CLPP
376	Perrault Syndrome 4	LARS2
377	Peters Plus Syndrome	B3GLCT
378	Pierson Syndrome	LAMB2
379	Pitt-Hopkins like syndrome 1	CNTNAP2
380	Poretti-Boltshauser syndrome	LAMA1
381	Raine Syndrome	FAM20C
382	X-Linked Syndromic Mental Retardation, Raymond type	ZDHHC9
383	Renpenning syndrome	PQBP1
384	Sandhoff Disease	HEXB
385	Metachromatic leukodystrophy due to Saposin B deficiency	PSAP
386	Schimke Immunoosseous Dysplasia	SMARCAL1
387	Schneckenbecken Dysplasia	SLC35D1
388	Schwartz-Jampel Syndrome, Type 1	HSPG2
389	Schwartz-Jampel Syndrome, Type 2	LIFR
390	SC Phocomelia Syndrome	ESCO2
391	Seckel Syndrome Type 1	ATR
392	Seckel Syndrome Type 2	RBBP8
393	Seckel Syndrome Type 5	CEP152
394	Sengers syndrome	AGK
395	Senior-Loken Syndrome 4	NPHP4
396	Senior-Loken syndrome 5	IQCB1
397	Senior-Loken Syndrome 8	WDR19
398	Shwachman-Diamond Syndrome	SBDS
399	X-linked Mental retardation syndrome, Siderius type	PHF8
400	Simpson-Golabi-Behmel Syndrome Type 1	GPC3
401	Smith-McCort Dysplasia 2	RAB33B
402	Snyder-Robinson mental retardation syndrome	SMS
403	Steel Syndrome	COL27A1
404	TARP Syndrome	RBM10
405	Temtamy Preaxial Brachydactyly Syndrome	CHSY1
406	Temtamy Syndrome	C12orf57
407	Tenascin-X deficiency type Ehlers-Danlos syndrome	TNXB
408	Ullrich congenital muscular dystrophy 1	COL6A1, COL6A2, COL6A3
409	Usher Syndrome Type IB	MYO7A

410	Usher Syndrome Type IC	USH1C
411	Usher syndrome, type 1D	CDH23
412	Usher Syndrome Type IG	USH1G
413	Usher Syndrome Type IJ	CIB2
414	Usher Syndrome Type IIA	USH2A
415	Usher Syndrome Type IID	WHRN
416	Van Den Ende-Gupta Syndrome	SCARF2
417	Van Maldergem Syndrome 1	DCHS1
418	Vici Syndrome	EPG5
419	Factor VII Deficiency	F7
420	Factor V deficiency	F5
421	Warburg Micro Syndrome 1	RAB3GAP1
422	Warburg Micro Syndrome 2	RAB3GAP2
423	Warburg Micro Syndrome 3	RAB18
424	Wieacker-Wolff Syndrome	ZC4H2
425	Wiskott-Aldrich Syndrome 1	WAS
426	Multiple Epiphyseal Dysplasia with Early-Onset Diabetes Mellitus	EIF2AK3
427	Wolfram Syndrome 2	CISD2
428	Woodhouse-Sakati syndrome	DCAF17
429	Alport syndrome 1, X-linked	COL4A5
430	X-linked Charcot-Marie-Tooth disease 4	AIFM1
431	X-linked Emery-Dreifuss Muscular Dystrophy 1	EMD
432	X-Linked Properdin Deficiency	CFP
433	X-Linked Hypophosphatemia	PHEX
434	Macular Degeneration, X-Linked Atrophic	RPGR
435	X-Linked Myopathy with Excessive Autophagy	VMA21
436	X-Linked Lymphoproliferative syndrome 1	SH2D1A
437	X-Linked Lymphoproliferative syndrome 2	XIAP
438	X-linked Chronic Granulomatous Disease	CYBB
439	X-Linked Juvenile Retinoschisis	RS1
440	X-Linked Adrenoleukodystrophy	ABCD1
441	Myopathy, X-linked, with postural muscle atrophy	FHL1
442	X-linked sideroblastic anemia and ataxia	ABCB7
443	X-linked Pigmentary disorder, reticulate, with systemic manifestations	POLA1
444	X-linked Lissencephaly 1	DCX
445	X-linked Lissencephaly 2	ARX
446	X-Linked Dyskeratosis Congenita	DKC1
447	Spinal muscular atrophy, X-linked 2, infantile	UBA1
448	X-linked Mental Retardation	NLGN4X
449	X-Linked Mental Retardation 12	THOC2
450	X-Linked Mental Retardation 1	IQSEC2

451	X-Linked Mental Retardation 21	IL1RAPL1
452	X-Linked Mental Retardation 30	PAK3
453	X-Linked Mental Retardation 41	GDI1
454	X-Linked Mental Retardation 49	CLCN4
455	X-Linked Mental Retardation 58	TSPAN7
456	X-Linked Mental Retardation 61	RLIM
457	X-Linked Mental Retardation 72	RAB39B
458	X-Linked Mental Retardation 90	DLG3
459	X-Linked Mental Retardation 93	BRWD3
460	X-Linked Mental Retardation 96	SYP
461	X-Linked Mental Retardation 97	ZNF711
462	X-Linked Mental Retardation 98	NEXMIF
463	X-Linked Mental Retardation 99	USP9X
464	X-Linked Mental Retardation 9	FTSJ1
465	X-Linked Mental Retardation with Cerebellar Hypoplasia and Distinctive Facial Appearance	OPHN1
466	X-Linked Mental Retardation-Hypotonic Facies Syndrome 1	ATRX
467	X-Linked Syndromic Mental Retardation 14	UPF3B
468	X-Linked Syndromic Mental Retardation 15	CUL4B
469	X-Linked Syndromic Mental Retardation 35	RPL10
470	X-Linked Syndromic Mental Retardation 5	AP1S2
471	You-Hoover-Fong syndrome	TELO2
472	Yunis-Varon Syndrome	FIG4
473	Alpha-N-acetylgalactosaminidase deficiency	NAGA
474	Beta-Mannosidosis	MANBA
475	Beta-Ureidopropionase Deficiency	UPB1
476	Prolidase deficiency	PEPD
477	Interleukin 1 Receptor Antagonist Deficiency	IL1RN
478	Cataracts, Growth Hormone Deficiency, Sensory Neuropathy,sensorineural hearing loss, and skeletal dysplasia	IARS2
479	Cataract 18	FYCO1
480	Cataract 40	NHS
481	Leukocyte Adhesion Deficiency type 1	ITGB2
482	Leukocyte Adhesion Deficiency type 3	FERMT3
483	Galactosialidosis	CTSA
484	Sudden Infant Death With Dysgenesis Of The Testes Syndrome	TSPYL1
485	Spondyloepimetaphyseal Dysplasia With Joint Laxity, Type 1, With Or Without Fractures	B3GALT6
486	Bowen-Conradi Syndrome	EMG1
487	Bifid Nose With Or Without Anorectal And Renal Anomalies	FREM1
488	Pyridoxine-Refractory Sideroblastic Anemia 2	SLC25A38
489	Pyridoxine-Dependent Epilepsy	ALDH7A1

490	Epidermolytic hyperkeratosis	KRT10
491	Malonyl-Coa Decarboxylase Deficiency	MLYCD
492	Pyruvate kinase deficiency	PKLR
493	Pyruvate Carboxylase Deficiency	PC
494	Pyruvate dehydrogenase E1-alpha deficiency	PDHA1
495	Pyruvate Dehydrogenase E1-Beta Deficiency	PDHB
496	Pyruvate Dehydrogenase Phosphatase Deficiency	PDP1
497	Pyruvate Dehydrogenase Lipoic Acid Synthetase Deficiency	LIAS
498	C1q deficiency	C1QA, C1QB, C1QC
499	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy	CD55
500	Complement Factor I Deficiency	CFI
501	Epimerase Deficiency Galactosemia	GALE
502	Hypomagnesemia 1, intestinal	TRPM6
503	Common Variable Immune Deficiency 1	ICOS
504	Common Variable Immune Deficiency 2	TNFRSF13B
505	Common Variable Immune Deficiency 6	CD81
506	Common Variable Immune Deficiency 8 with Autoimmunity	LRBA
507	Autosomal Spastic paraplegia 30	KIF1A
508	Autosomal Dyskeratosis Congenita 5/4	RTEL1
509	Autosomal Recessive Robinow Syndrome	ROR2
510	Autosomal Recessive T Cell-Negative、 B Cell-Positive、NK Cell-Negative Severe Combined Immunodeficiency	JAK3
511	Autosomal Recessive Persistent Hyperplastic Primary Vitreous	ATOH7
512	Autosomal Recessive Epidermolysis Bullosa Simplex 1	KRT14, KRT5
513	Autosomal recessive Thrombophilia due to protein C deficiency	PROC
514	Autosomal recessive Thrombophilia due to protein S deficiency	PROS1
515	Autosomal Recessive Deafness 3	MYO15A
516	Autosomal Recessive Deafness 7	TMC1
517	Autosomal Recessive Deafness 8/10	TMPRSS3
518	Autosomal Recessive Deafness 9	OTOF
519	Autosomal Recessive Osteopetrosis 2	TNFSF11
520	Autosomal Recessive Osteopetrosis 3	CA2
521	Autosomal Recessive Osteopetrosis 4	CLCN7
522	Autosomal Recessive Osteopetrosis 5	OSTM1
523	Autosomal Recessive Osteopetrosis 7	TNFRSF11A
524	Autosomal Recessive Spinocerebellar Ataxia 10	ANO10
525	Autosomal Recessive Spinocerebellar Ataxia 13	GRM1
526	Autosomal Recessive Spinocerebellar Ataxia 16	STUB1
527	Autosomal Recessive Spinocerebellar Ataxia 1	SETX

528	Autosomal Recessive Spinocerebellar ataxia 20	SNX14
529	Autosomal Recessive Spinocerebellar ataxia 21	SCYL1
530	Autosomal Recessive Spinocerebellar Ataxia 2	PMPCA
531	Autosomal Recessive Spastic paraplegia 15	ZFYVE26
532	Autosomal Recessive Spastic paraplegia 23	DSTYK
533	Autosomal Recessive Spastic paraplegia 26	B4GALNT1
534	Autosomal Recessive Spastic paraplegia 35	FA2H
535	Autosomal Recessive Spastic paraplegia 45	NT5C2
536	Autosomal Recessive Spastic paraplegia 46	GBA2
537	Autosomal Recessive Spastic paraplegia 47	AP4B1
538	Autosomal Recessive Spastic paraplegia 50	AP4M1
539	Autosomal Recessive Spastic paraplegia 52	AP4S1
540	Autosomal Recessive Spastic paraplegia 53	VPS37A
541	Autosomal Recessive Spastic paraplegia 54	DDHD2
542	Autosomal Recessive Spastic paraplegia 56	CYP2U1
543	Autosomal Recessive Spastic Paraplegia 9B	ALDH18A1
544	Autosomal Recessive Cutis Laxa type 1A	FBLN5
545	Autosomal Recessive Cutis Laxa type 1B	EFEMP2
546	Autosomal Recessive Cutis Laxa type 1C	LTBP4
547	Autosomal Recessive Cutis Laxa type 2A	ATP6V0A2
548	Autosomal Recessive Cutis Laxa type 2B	PYCR1
549	Autosomal Recessive Cytochrome B-Positive Chronic Granulomatous Disease Type I	NCF1
550	Autosomal Recessive Cytochrome B-Positive Chronic Granulomatous Disease Type II	NCF2
551	Autosomal Recessive Cytochrome B-Negative Chronic Granulomatous Disease	CYBA
552	Autosomal Recessive Myotonia Congenita	CLCN1
553	Autosomal Recessive Dyskeratosis Congenita 3	WRAP53
554	Autosomal Recessive Microcephaly And Chorioretinopathy 1	TUBGCP6
555	Autosomal Recessive Microcephaly And Chorioretinopathy 3	TUBGCP4
556	Autosomal Recessive Spastic Ataxia 8 with Hypomyelinating Leukodystrophy	NKX6-2
557	Autosomal Recessive Congenital Ichthyosis 5	CYP4F22
558	Autosomal Recessive Congenital Ichthyosis 6	NIPAL4
559	Autosomal Recessive Congenital Ichthyosis 9	CERS3
560	Hypertrophic Osteoarthropathy, Primary, Autosomal Recessive, 1	HPGD
561	Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy	MED23
562	Intellectual developmental disorder, autosomal recessive 27	LINS1
563	Intellectual developmental disorder, autosomal recessive 38	HERC2
564	Intellectual developmental disorder, autosomal recessive 57	MBOAT7

565	Autosomal Recessive Mental Retardation 13	TRAPPC9
566	Autosomal Recessive Mental Retardation 15	MAN1B1
567	Autosomal Recessive Mental Retardation 36	ADAT3
568	Autosomal Recessive Mental Retardation 39	TTI2
569	Autosomal Recessive Mental Retardation 3	CC2D1A
570	Autosomal Recessive Mental Retardation 41	KPTN
571	Autosomal Recessive Mental Retardation 42	PGAP1
572	Autosomal Recessive Mental Retardation 44	METTL23
573	Autosomal Recessive Mental Retardation 49	GPT2
574	Autosomal Recessive Mental Retardation 58	ELP2
575	Autosomal Recessive Mental Retardation 5	NSUN2
576	Autosomal Recessive Mental Retardation 7	TUSC3
577	Osteogenesis Imperfecta type XV	WNT1
578	Osteogenesis Imperfecta type VI	SERPINF1
579	Osteogenesis Imperfecta type VIII	P3H1
580	Osteogenesis imperfecta, type X	SERPINH1
581	Achalasia-Addisonianism-Alacrima Syndrome	AAAS
582	Postnatal Progressive Microcephaly With Seizures And Brain Atrophy	MED17
583	Hemorrhagic Destruction of the Brain, Subependymal Calcification and Cataracts	JAM3
584	Adrenocorticotropic hormone Deficiency	TBX19
585	Neurodegeneration due to Cerebral Folate Transport Deficiency	FOLR1
586	Epidermolysis Bullosa with Pyloric Atresia	ITGB4, ITGA6
587	Macrocephaly, Alopecia, Cutis Laxa, and Scoliosis(MACS syndrome)	RIN2
588	Band-Like Calcification with Simplified Gyration and Polymicrogyria	OCLN
589	Band heterotopia	EML1
590	Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects	B3GAT3
591	Leukodystrophy, hypomyelinating, 2	GJC2
592	HSD10 mitochondrial disease	HSD17B10
593	D-bifunctional protein deficiency	HSD17B4
594	Autosomal Recessive Distal Spinal Muscular Atrophy 1	IGHMBP2
595	Donohue Syndrome	INSR
596	Gillespie syndrome, Autosomal recessive	ITPR1
597	PERCHING syndrome	KLHL7
598	Familial Lecithin cholesterol acyltransferase deficiency	LCAT
599	Lysosomal acid lipase deficiency	LIPA
600	Congenital Hydrocephalus 2 with or without brain or eye anomalies	MPDZ

601	Hypertrophic Neuropathy of Dejerine Sottas	MPZ
602	Cleft lip/palate-ectodermal dysplasia syndrome	NECTIN1
603	myoclonic epilepsy of Lafora	NHLRC1
604	Insensitivity to pain, congenital, with anhidrosis	NTRK1
605	Coloboma, Congenital Heart Disease, Ichthyosiform Dermatosis, Mental Retardation, and Ear Anomalies Syndrome	PIGL
606	Plasminogen deficiency, type I	PLG
607	Microcephaly, seizures, and developmental delay	PNKP
608	OBESITY, EARLY-ONSET, WITH ADRENAL INSUFFICIENCY AND RED HAIR	POMC
609	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)	SLC19A3
610	Autosomal Recessive Spastic paraplegia 20	SPART
611	Salt and pepper developmental regression syndrome, Autosomal recessive	ST3GAL5
612	Gastrointestinal defects and immunodeficiency syndrome	TTC7A
613	Kaufman oculocerebrofacial syndrome	UBE3B
614	Epidermolysis bullosa simplex with pyloric atresia	PLEC
615	Isolated Microphthalmia 8	ALDH1A3
616	Monocarboxylate Transporter 1 Deficiency	SLC16A1
617	Proteasome-Associated Autoinflammatory Syndrome 1	PSMB8
618	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1)	ANOS1
619	Hypomyelinating Leukodystrophy 10	PYCR2
620	Hypomyelinating Leukodystrophy 12	VPS11
621	Hypomyelinating Leukodystrophy 14	UFM1
622	Hypomyelinating Leukodystrophy 3	AIMP1
623	Hypomyelinating Leukodystrophy 4	HSPD1
624	Hypomyelinating Leukodystrophy 5	HYCC1
625	Hypomyelinating Leukodystrophy 7 with or without oligodontia and/or hypogonadotropic hypogonadism	POLR3A
626	Hypomyelinating Leukodystrophy 8	POLR3B
627	Epilepsy, Hearing Loss, And Mental Retardation Syndrome	AFG2A
628	Epilepsy with Variable Learning Disabilities and Behavior Disorders	SYN1
629	Seizures, Sensorineural Deafness, Ataxia, Mental Retardation, and Electrolyte Imbalance Syndrome	KCNJ10
630	Arterial tortuosity syndrome	SLC2A10
631	Short-Rib Thoracic Dysplasia 14 With Polydactyly	KIAA0586
632	Short-rib throacic dysplasia 15 with polydactyly	DYNC2LI1
633	Short-rib thoracic dysplasia 10 with or without polydactyly	IFT172
634	Short-rib thoracic dysplasia 11 with or without polydactyly	DYNC2I2

635	Short-rib thoracic dysplasia 13 with or without polydactyly	CEP120
636	Short-rib thoracic dysplasia 2 with or without polydactyly	IFT80
637	Short-rib thoracic dysplasia 3 with or without polydactyly	DYNC2H1
638	Short-rib thoracic dysplasia 4 with or without polydactyly	TTC21B
639	Short-rib thoracic dysplasia 6 with or without polydactyly	NEK1
640	Short-rib thoracic dysplasia 7 with or without polydactyly	WDR35
641	Short-rib thoracic dysplasia 8 with or without polydactyly	DYNC2I1
642	Short-rib thoracic dysplasia 9 with or without polydactyly	IFT140
643	Short Stature, Microcephaly, And Endocrine Dysfunction	XRCC4
644	Multicentric Osteolysis, Nodulosis, and Arthropathy	MMP2
645	Multiple Sulfatase Deficiency	SUMF1
646	Multiple Congenital Anomalies-Hypotonia-Seizures Syndrome 1	PIGN
647	Multiple Congenital Anomalies-Hypotonia-Seizures Syndrome 3	PIGT
648	Multiple Pterygium Syndrome,lethal type	CHRNA1
649	Multiminicore disease	RYR1
650	Multisystem Autoimmune Disease With Facial Dysmorphism	ITCH
651	Multiple mitochondrial dysfunctions syndrome 1	NFU1
652	Multiple mitochondrial dysfunctions syndrome 2	BOLA3
653	Multiple mitochondrial dysfunctions syndrome 3	IBA57
654	Multiple mitochondrial dysfunctions syndrome 4	ISCA2
655	Polymicrogyria with Seizures	RTTN
656	Catecholaminergic Polymorphic Ventricular Tachycardia 2	CASQ2
657	Catecholaminergic Polymorphic Ventricular Tachycardia 5, with or without muscle weakness	TRDN
658	Childhood-Onset Polyarteritis Nodosa	ADA2
659	Childhood-onset neurodegeneration with ataxia, dystonia, and gaze palsy	SQSTM1
660	Developmental Delay With Short Stature, Dysmorphic Features, And Sparse Hair	DPH1
661	GAPO Syndrome	ANTXR1
662	Leigh Syndrome, French-Canadian Type	LRPPRC
663	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration	TANGO2
664	Fanconi anemia, complementation group B	FANCB
665	Fanconi anemia, complementation group E	FANCE
666	Fanconi anemia, complementation group F	FANCF
667	Fanconi anemia, complementation group L	FANCL
668	Fanconi anemia, complementation group Q	ERCC4
669	Fanconi anemia, complementation group T	UBE2T
670	Nonphotosensitive Trichothiodystrophy 4	MPLKIP
671	Pulmonary Surfactant Metabolism Dysfunction 1	SFTPB

672	Pulmonary Surfactant Metabolism Dysfunction 3	ABCA3
673	Pulmonary Venoocclusive Disease 2	EIF2AK4
674	Combined Oxidative Phosphorylation Deficiency 10	MTO1
675	Combined Oxidative Phosphorylation Deficiency 11	RMND1
676	Combined Oxidative Phosphorylation Deficiency 12	EARS2
677	Combined Oxidative Phosphorylation Deficiency 14	FARS2
678	Combined Oxidative Phosphorylation Deficiency 15	MTFMT
679	Combined Oxidative Phosphorylation Deficiency 17	ELAC2
680	Combined Oxidative Phosphorylation Deficiency 1	GFM1
681	Combined Oxidative Phosphorylation Deficiency 20	VARS2
682	Combined Oxidative Phosphorylation Deficiency 23	GTPBP3
683	Combined Oxidative Phosphorylation Deficiency 24	NARS2
684	Combined Oxidative Phosphorylation Deficiency 27	CARS2
685	Combined Oxidative Phosphorylation Deficiency 35	TRIT1
686	Combined Oxidative Phosphorylation Deficiency 3	TSFM
687	Combined Oxidative Phosphorylation Deficiency 4	TUFM
688	Combined Oxidative Phosphorylation Deficiency 7	MTRFR
689	Combined Oxidative Phosphorylation Deficiency 8	AARS2
690	Diarrhea 5 With Congenital Tufting Enteropathy	EPCAM
691	Diarrhea 7	DGAT1
692	Diarrhea with Microvillus Atrophy 2	MYO5B
693	Glycerol Kinase Deficiency	GK
694	Nemaline Myopathy 10	LMOD3
695	Nemaline Myopathy 1	TPM3
696	Nemaline Myopathy 5	TNNT1
697	Nemaline Myopathy 7	CFL2
698	Nemaline Myopathy 8	KLHL40
699	Nemaline Myopathy 9	KLHL41
700	Hepatic Veno-Occlusive Disease with Immunodeficiency	SP110
701	Hyper IgE Syndrome	DOCK8
702	Hyperphosphatasia with Mental Retardation Syndrome 1	PIGV
703	Hyperphosphatasia with Mental Retardation Syndrome 2	PIGO
704	Hyperphosphatasia with Mental Retardation Syndrome 3	PGAP2
705	Hyperphosphatasia with Mental Retardation Syndrome 4	PGAP3
706	Hypermanganesemia With Dystonia 1	SLC30A10
707	Hypermanganesemia with dystonia 2	SLC39A14
708	Hyperuricemia, Pulmonary Hypertension, Renal Failure, And Alkalosis syndrome	SARS2
709	Hyperprolinemia type I	PRODH
710	Methemoglobinemia Due to Deficiency of Methemoglobin Reductase	CYB5R3
711	Ataxia with oculomotor apraxia type 1	APTX
712	Ataxia with vitamin E deficiency	TTPA

713	Ataxia-Telangiectasia-Like Disorder 1	MRE11
714	Microphthalmia, isolated 3	RAX
715	Glutamate Formiminotransferase Deficiency	FTCD
716	Glutathione synthetase deficiency	GSS
717	Bone marrow failure syndrome 2	ERCC6L2
718	Bone marrow failure syndrome 3	DNAJC21
719	Sclerosteosis 1	SOST
720	Cerebral creatine deficiency syndrome 2	GAMT
721	Arthrogryposis, Renal Dysfunction, and Cholestasis Syndrome 1	VPS33B
722	Arthrogryposis, Renal Dysfunction, and Cholestasis Syndrome 2	VIPAS39
723	Arthrogryposis, Mental Retardation and Seizures	SLC35A3
724	Photosensitive Trichothiodystrophy 3	GTF2H5
725	Fructose 1,6 Bisphosphatase Deficiency	FBP1
726	Peroxisome biogenesis disorder 10A	PEX3
727	Peroxisome biogenesis disorder 11A	PEX13
728	Peroxisome Biogenesis Disorder 14B	PEX11B
729	Peroxisome biogenesis disorder 2A	PEX5
730	Peroxisome biogenesis disorder 3A(Zellweger)	PEX12
731	Peroxisome biogenesis disorder 4A	PEX6
732	Peroxisome biogenesis disorder 5A	PEX2
733	Peroxisome biogenesis disorder 6A(Zellweger)	PEX10
734	Peroxisome biogenesis disorder 7A	PEX26
735	Peroxisome biogenesis disorder 8A(Zellweger)	PEX16
736	Peroxisomal Acyl-CoA oxidase deficiency	ACOX1
737	Treacher Collins Syndrome 3	POLR1C
738	Ataxia, posterior column, with retinitis pigmentosa	FLVCR1
739	Succinic Semialdehyde Dehydrogenase Deficiency	ALDH5A1
740	Warsaw Breakage Syndrome	DDX11
741	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency	FLAD1
742	Myopathy, lactic acidosis, and sideroblastic anemia 1	PUS1
743	Myopathy, Lactic acidosis, and Sideroblastic anemia 2	YARS2
744	Myopathy With Extrapyramidal Signs	MICU1
745	Mulibrey nanism	TRIM37
746	Ehlers-Danlos Syndrome, Musculocontractural type 1	CHST14
747	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7	CRPPA
748	Myofibrillar Myopathy 7	KY
749	Myofibrillar Myopathy 8	PYROXD1
750	Acute Recurrent Myoglobinuria	LPIN1
751	Spondylo-Megaepiphyseal-Metaphyseal Dysplasia	NKX3-2

752	Spondylocarpotarsal Synostosis Syndrome	FLNB
753	Spondyloocular syndrome	XYLT2
754	Spondylometaphyseal Dysplasia with Cone-Rod Dystrophy	PCYT1A
755	Spondylocostal dysostosis 1	DLL3
756	Spondylocostal dysostosis 2	MESP2
757	Spondylocostal dysostosis 4	HES7
758	Familial Cirrhosis	KRT8
759	Hypercholesterolemia, familial,1	LDLR
760	Familial Hyperinsulinemic Hypoglycemia 1	ABCC8
761	Familial Chloride Diarrhea	SLC26A3
762	Familial Candidiasis 2	CARD9
763	Familial Normophosphatemic Tumoral Calcinosis	SAMD9
764	Methylmalonic aciduria and homocystinuria CblF type	LMBRD1
765	Methylmalonic aciduria and homocysteinemia, cblX type	HCFC1
766	Mevalonic Aciduria	MVK
767	Thyroid dyshormonogenesis 5	DUOXA2
768	Thyroid dyshormonogenesis 6	DUOX2
769	Pseudohypoaldosteronism, type I	SCNN1A, SCNN1B
770	Interstitial lung and liver disease	MARS1
771	Omodysplasia 1	GPC6
772	Brittle Cornea Syndrome 1	ZNF469
773	Brittle Cornea Syndrome 2	PRDM5
774	Progressive Myoclonic Epilepsy 1A	CSTB
775	Progressive Myoclonic Epilepsy 1B	PRICKLE1
776	Progressive Myoclonic Epilepsy 3	KCTD7
777	Progressive Myoclonic Epilepsy 4	SCARB2
778	Progressive Myoclonic Epilepsy 6	GOSR2
779	Progressive Familial Intrahepatic Cholestasis 1	ATP8B1
780	Progressive Pseudorheumatoid Dysplasia	CCN6
781	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy	NAXE
782	Proximal Renal Tubular Acidosis with Ocular Abnormalities	SLC4A4
783	Cerebral creatine deficiency syndrome 3	GATM
784	Spastic Ataxia 2	KIF1C
785	Spastic Ataxia 3	MARS2
786	Spastic Paraplegia And Psychomotor Retardation With Or Without Seizures	HACE1
787	Megalencephalic Leukoencephalopathy with Subcortical Cysts 2A	HEPACAM
788	Giant Axonal Neuropathy-1	GAN
789	Orofaciodigital syndrome XVI	TMEM107
790	Orofaciodigital Syndrome V	DDX59
791	Dilated Cardiomyopathy With Woolly Hair And Keratoderma	DSP

792	Lysinuric Protein Intolerance	SLC7A7
793	Geroderma Osteodysplasticum	GORAB
794	Tyrosinemia Type II	TAT
795	Tyrosinemia Type III	HPD
796	CK Syndrome	NSDHL
797	Refsum disease	PHYH
798	Ehlers-Danlos syndrome, spondylodysplastic type, 1	B4GALT7
799	Cold-induced Sweating Syndrome 1	CRLF1
800	Cold-induced Sweating Syndrome 2	CLCF1
801	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia	MTHFD1
802	Combined pituitary hormone deficiency 1	POU1F1
803	Combined pituitary hormone deficiency 3	LHX3
804	Desmosterolosis	DHCR24
805	Split-Hand/Foot Malformation 6	WNT10B
806	Lymphoproliferative Syndrome 1	ITK
807	Lymphoproliferative Syndrome 2	CD27
808	Triosephosphate Isomerase Deficiency	TPI1
809	Phosphoglycerate Kinase Deficiency	PGK1
810	Phosphoserine Phosphatase Deficiency	PSPH
811	Thiamine-Responsive Megaloblastic Anemia Syndrome	SLC19A2
812	Thiamine Metabolism Dysfunction Syndrome 4 (Bilateral Striatal Degeneration and Progressive Polyneuropathy Type)	SLC25A19
813	Thiamine Metabolism Dysfunction Syndrome 5 (Episodic Encephalopathy type)	TPK1
814	Sulfocysteinuria	SUOX
815	Craniolenticulosutural Dysplasia	SEC23A
816	Craniofrontonasal syndrome	EFNB1
817	Craniosynostosis and Dental Anomalies	IL11RA
818	Cranioectodermal Dysplasia 1	IFT122
819	Craniofacial Dysmorphism, Skeletal Anomalies, And Mental Retardation syndrome	TMCO1
820	Bare lymphocyte syndrome, type II, complementation group A	CIITA
821	Bare lymphocyte syndrome, type II, complementation group B	RFXANK
822	Bare lymphocyte syndrome, type II, complementation group D	RFXAP
823	Chronic Atrial And Intestinal Dysrhythmia	SGO1
824	Trichohepatoenteric syndrome 1	SKIC3
825	Trichohepatoenteric Syndrome 2	SKIC2
826	Ichthyosis Follicularis-Atrichia-Photophobia Syndrome	MBTPS2

827	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	QARS1
828	Immunoskeletal dysplasia with neurodevelopmental abnormalities	EXTL3
829	Immunodeficiency with Hyper-IgM, type 1	CD40LG
830	Immunodeficiency with Hyper-IgM, type 3	CD40
831	Immunodeficiency-Centromeric Instability-Facial Anomalies Syndrome 2	ZBTB24
832	Immunodeficiency 10	STIM1
833	Immunodeficiency 11A	CARD11
834	Immunodeficiency 12	MALT1
835	Immunodeficiency 15B	IKBKB
836	Immunodeficiency 19	CD3D
837	Immunodeficiency 23	PGM3
838	Immunodeficiency 24	CTPS1
839	Immunodeficiency 27A	IFNGR1
840	Immunodeficiency 28	IFNGR2
841	Immunodeficiency 31B	STAT1
842	Immunodeficiency 35	TYK2
843	Immunodeficiency 40	DOCK2
844	Immunodeficiency 42	RORC
845	Immunodeficiency 47	ATP6AP1
846	Immunodeficiency 48	ZAP70
847	Immunodeficiency 51	IL17RA
848	Immunodeficiency 52	LAT
849	Immunodeficiency 54	MCM4
850	Immunodeficiency 9	ORAI1
851	Arthrogryposis, distal, with impaired proprioception and touch	PIEZO2
852	Dopa-responsive dystonia due to sepiapterin reductase deficiency	SPR
853	Molybdenum cofactor deficiency C	GPHN
854	Molybdenum Cofactor Deficiency Complementation Group B	MOCS2
855	Nijmegen breakage syndrome,	NBN
856	Nijmegen Breakage Syndrome-like Disorder	RAD50
857	Cystic Leukoencephalopathy without Megalencephaly	RNASET2
858	Cerebral Dysgenesis, Neuropathy, Ichthyosis, And Palmoplantar Keratoderma Syndrome	SNAP29
859	Leukoencephalopathy with Brain Stem and Spinal Cord Involvement and Lactate Elevation	DARS2
860	Hypomyelination with brainstem and spinal cord involvement and leg spasticity	DARS1
861	Cerebral Creatine Deficiency Syndrome 1	SLC6A8

862	Hydrolethalus Syndrome 1	HYLS1
863	Hydrolethalus Syndrome 2	KIF7
864	Cerebrotendinous xanthomatosis	CYP27A1
865	Pontocerebellar hypoplasia type 10	CLP1
866	Pontocerebellar hypoplasia type 11	TBC1D23
867	Pontocerebellar hypoplasia type 1A	VRK1
868	Pontocerebellar Hypoplasia type 1B	EXOSC3
869	Pontocerebellar Hypoplasia, Type 1C	EXOSC8
870	Pontocerebellar hypoplasia type 2A	TSEN54
871	Pontocerebellar hypoplasia type 2B	TSEN2
872	Pontocerebellar Hypoplasia type 2D	SEPSECS
873	Pontocerebellar Hypoplasia, Type 2E	VPS53
874	Pontocerebellar hypoplasia type 6	RARS2
875	Pontocerebellar hypoplasia, type 7	TOE1
876	Pontocerebellar hypoplasia type 9	AMPD2
877	Cerebroretinal Microangiopathy With Calcifications And Cysts	CTC1
878	Ventriculomegaly With Cystic Kidney Disease	CRB2
879	Periventricular Nodular Heterotopia 2	ARFGEF2
880	Cerebrooculofacioskeletal Syndrome 2	ERCC2
881	Visceral Heterotaxy 1	ZIC3
882	Visceral Heterotaxy 7	MMP21
883	Mucopolysaccharidosis type VII	GUSB
884	Urofacial Syndrome 1	HPSE2
885	Urofacial Syndrome 2	LRIG2
886	Parkinson Disease 15	FBXO7
887	Parkinson Disease 19	DNAJC6
888	Pelizaeus-Merzbacher disease	PLP1
889	Poikiloderma with Neutropenia	USB1
890	Purine Nucleoside Phosphorylase Deficiency	PNP
891	Horizontal gaze palsy with progressive scoliosis 1	ROBO3
892	Polyglucosan Body Myopathy 1 With Or Without Immunodeficiency	RBCK1
893	Mosaic variegated aneuploidy syndrome 1	BUB1B
894	Rigid Spine Muscular Dystrophy 1	SELENON
895	Juvenile Paget Disease	TNFRSF11B
896	Primary Lateral Sclerosis, Juvenile	ALS2
897	Mild non-BH4-deficient Hyperphenylalaninemia	DNAJC12
898	Achromatopsia 2	CNGA3
899	Achromatopsia 4	GNAT2
900	Achromatopsia 7	ATF6
901	Hyaline fibromatosis syndrome	ANTXR2
902	Carnitine Palmitoyltransferase I Deficiency	CPT1A

903	Carnitine Palmitoyltransferase II Deficiency	CPT2
904	Carnitine-Acylcarnitine Translocase Deficiency	SLC25A20
905	Chylomicron Retention Disease	SAR1B
906	Cartilage-hair hypoplasia	RMRP
907	Achondrogenesis type 1A	TRIP11
908	Achondrogenesis type 1B	SLC26A2
909	Trimethylaminuria	FMO3
910	Short Stature, Optic Nerve Atrophy, And Pelger-Huet Anomaly	NBAS
911	Short Stature, Onychodysplasia, Facial Dysmorphism, And Hypotrichosis	POC1A
912	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies	PLAA
913	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies	PRUNE1
914	Neurodevelopmental Disorder with Spastic Quadriplegia and Brain Abnormalities with or without Seizures	WDR45B
915	Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy	PIGG
916	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy	VARS1
917	Neurodegeneration with brain iron accumulation 1	PANK2
918	Neurodegeneration with brain iron accumulation 2B	PLA2G6
919	Neurodegeneration with brain iron accumulation 4(Mitochondrial Membrane Protein-Associated Neurodegeneration)	C19orf12
920	Neuronal Ceroid-Lipofuscinoses 10	CTSD
921	Neuronal Ceroid Lipofuscinosis 8	CLN8
922	Nephrotic Syndrome Type 11	NUP107
923	Nephrotic Syndrome Type 12	NUP93
924	Nephrotic Syndrome Type 14	SGPL1
925	Nephrotic Syndrome Type 2	NPHS2
926	Nephrotic Syndrome Type 3	PLCE1
927	Nephrotic Syndrome Type 7	DGKE
928	Nephrotic Syndrome Type 9	COQ8B
929	Nephronophthisis 16	ANKS6
930	Nephronophthisis 19	DCDC2
931	Nephronophthisis 20	MAPKBP1
932	Nephronophthisis 2	INVS
933	Nephronophthisis-Like Nephropathy 1	XPNPEP3
934	Renal-Hepatic-Pancreatic Dysplasia 2	NEK8
935	Renal tubular dysgenesis	ACE, AGT, REN
936	Diabetes insipidus, nephrogenic, 2	AQP2

937	Hypomagnesemia 5, renal, with ocular involvement	CLDN19
938	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy	IARS1
939	Growth Retardation, Developmental Delay, Facial Dysmorphism	FTO
940	Septooptic Dysplasia	HESX1
941	Optic Atrophy 10 With Or Without Ataxia, Mental Retardation, And Seizures	RTN4IP1
942	Retinal Arterial Macroaneurysm With Supravalvular Pulmonic Stenosis	IGFBP7
943	Retinitis pigmentosa 14	TULP1
944	Retinitis pigmentosa 59	DHDDS
945	Retinitis pigmentosa 77	REEP6
946	Retinitis pigmentosa with or without skeletal anomalies	CWC27
947	Retinal dystrophy with macular staphyloma	CFAP410
948	Cone-rod dystrophy	AIPL1
949	Cone-Rod Dystrophy 10	SEMA4A
950	Cone-Rod Dystrophy 3	ABCA4
951	Spondylometaepiphyseal Dysplasia, Short Limb-Hand type	DDR2
952	bilateral frontoparietal polymicrogyria	ADGRG1
953	Limb pelvis hypoplasia aplasia syndrome	WNT7A
954	Spastic Tetraplegia, Thin Corpus Callosum, And Progressive Microcephaly	SLC1A4
955	Leukodystrophy, hypomyelinating, 9	RARS1
956	Fetal akinesia deformation sequence 2	RAPSN
957	Meconium Ileus	GUCY2C
958	Glycosylphosphatidylinositol Biosynthesis Defect 15	GPAA1
959	Glucocorticoid Deficiency 1	MC2R
960	Glucocorticoid Deficiency 2	MRAP
961	Glucocorticoid Deficiency 4	NNT
962	Glycogen Storage Disease type III	AGL
963	Glycogen storage disease type IXa1/IXa2	PHKA2
964	Glycogen storage disease type IXb	PHKB
965	Glycogen storage disease type IXc	PHKG2
966	Glycogen storage disease type IXd	PHKA1
967	Glycogen Storage Disease type VII	PFKM
968	Glycogen Storage Disease type VI	PYGL
969	Glycogen Storage Disease type V	PYGM
970	Glycogen Storage Disease type XIV	PGM1
971	Isolated growth hormone deficiency type III	BTK
972	Intellectual developmental disorder, X-linked, Turner type	HUWE1
973	Asparagine Synthetase Deficiency	ASNS

974	Sideroblastic Anemia With B-Cell Immunodeficiency, Periodic Fevers,And Developmental Delay	TRNT1
975	Diaphanospondylodysostosis	BMPER
976	Encephalopathy, Progressive, With Or Without Lipodystrophy	BSCL2
977	Sialidosis	NEU1
978	Ectodermal dysplasia, Ectrodactyly, and macular dystrophy Syndrome	CDH3
979	Ectodermal Dysplasia 10b, Hypohidrotic/Hair/Tooth type	EDAR
980	Ectodermal dysplasia and immunodeficiency 1	IKBKG
981	Lathosterolosis	SC5D
982	Reticular Dysgenesis	AK2
983	Epilepsy, early-onset, vitamin B6-dependent	PLPBP
984	Vitamin D-dependent rickets Type IA	CYP27B1
985	Vitamin K-dependent clotting factors, combined deficiency of, 2	VKORC1
986	Abetalipoproteinemia	MTTP
987	Alacrima, Achalasia, And Mental Retardation Syndrome	GMPPA
988	Lissencephaly 4	NDE1
989	Lissencephaly 5	LAMB1
990	Lissencephaly 6	KATNB1
991	Lissencephaly 8	TMTC3
992	Acheiropody	LMBR1
993	Atransferrinemia	TF
994	Chorea-Acanthocytosis	VPS13A
995	Fatal Infantile Cardioencephalomyopathy due to Cytochrome c Oxidase Deficiency 1	SCO2
996	Fatal Infantile Cardioencephalomyopathy due to Cytochrome c Oxidase Deficiency 2	COX15
997	Congenital Diarrhea 8, Secretory Sodium	SLC9A3
998	Congenital Cataracts, Hearing Loss, And Neurodegeneration	SLC33A1
999	Congenital Bile Acid Synthesis Defect 1	HSD3B7
1000	Congenital Bile Acid Synthesis Defect 2	AKR1D1
1001	Congenital Bile Acid Synthesis Defect 3	CYP7B1
1002	Congenital short bowel syndrome(CLMP)	CLMP
1003	Multiple congenital anomalies-hypotonia-seizures syndrome 2	PIGA
1004	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect	LGI4
1005	Hypothyroidism Congenital Nongoitrous 1	TSHR
1006	Hypothyroidism Congenital Nongoitrous 4	TSHB
1007	Congenital Dyserythropoietic Anemia Type II	SEC23B
1008	Congenital Myasthenic Syndrome 10	DOK7
1009	Congenital Myasthenic Syndrome 13	DPAGT1

1010	Congenital Myasthenic Syndrome 14	ALG2
1011	Congenital Myasthenic Syndrome 20	SLC5A7
1012	Congenital Myasthenic Syndrome 3B, fast-channel	CHRND
1013	Congenital Myasthenic Syndrome 4A, slow-channel	CHRNE
1014	Congenital Myasthenic Syndrome 5	COLQ
1015	Congenital Myasthenic Syndrome 6	CHAT
1016	Congenital Myasthenic Syndrome 9	MUSK
1017	Muscular dystrophy, congenital, with cataracts and intellectual disability	INPP5K
1018	Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies type A 10	RXYLT1
1019	Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies type A 11	B3GALNT2
1020	Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies type A 12	POMK
1021	Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies type A1	POMT1
1022	Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies type A 2	POMT2
1023	Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies type A 3	POMGNT1
1024	Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies type A 4	FKTN
1025	Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies type A 5	FKRP
1026	Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies type A 6	LARGE1
1027	Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies type A 8	POMGNT2
1028	Congenital Stationary Night Blindness, Type 1A	NYX
1029	Congenital Stationary Night Blindness, Type 1E	GPR179
1030	Congenital Stationary Night Blindness, Type 2B	CABP4
1031	Congenital Sodium Diarrhea	SPINT2
1032	Congenital Hydrocephalus 1	CCDC88C
1033	Congenital Prothrombin Deficiency	F2
1034	Lipodystrophy, congenital generalized, type 4	CAVIN1
1035	Adrenal Insufficiency, Congenital, with 46XY Sex Reversal, Partial or Complete	CYP11A1
1036	Congenital Disorders of Glycosylation Ib	MPI
1037	Congenital Disorders of Glycosylation Ic	ALG6
1038	Congenital Disorders of Glycosylation Id	ALG3
1039	Congenital Disorders of Glycosylation Ig	ALG12
1040	Congenital Disorders of Glycosylation Ih	ALG8

1041	Congenital Disorders of Glycosylation Ik	ALG1
1042	Congenital Disorders of Glycosylation Il	ALG9
1043	Congenital Disorders of Glycosylation Im	DOLK
1044	Congenital Disorders of Glycosylation In	RFT1
1045	Congenital Disorders of Glycosylation Ip	ALG11
1046	Congenital Disorders of Glycosylation Iq	SRD5A3
1047	Congenital Disorders of Glycosylation Iv	NGLY1
1048	Congenital Disorders of Glycosylation Iy	SSR4
1049	Congenital Disorders of Glycosylation IIa	MGAT2
1050	Congenital Disorders of Glycosylation IIe	COG7
1051	Congenital Disorders of Glycosylation IIk	TMEM165
1052	Congenital Disorders of Glycosylation III	COG6
1053	Congenital Disorders of Glycosylation Iin	SLC39A8
1054	Congenital Disorders of Glycosylation Iio	CCDC115
1055	Congenital Insensitivity to Pain	SCN9A
1056	Congenital alopecia and T-Cell Immunodeficiency and nail dystrophy	FOXN1
1057	Congenital Amegakaryocytic Thrombocytopenia	MPL
1058	Congenital Afibrinogenemia	FGA, FGB, FGG
1059	Congenital fiber-type disproportion myopathy	ACTA1
1060	Congenital Thrombotic thrombocytopenic purpura	ADAMTS13
1061	Camptodactyly-Arthropathy-Coxa Vara-Pericarditis Syndrome	PRG4
1062	Fibrochondrogenesis 1	COL11A1
1063	Fibrochondrogenesis 2	COL11A2
1064	Mitochondrial DNA depletion syndrome 11	MGME1
1065	Mitochondrial DNA depletion syndrome 13	FBXL4
1066	Mitochondrial DNA depletion syndrome 1	TYMP
1067	Mitochondrial DNA depletion syndrome 2	TK2
1068	Mitochondrial DNA depletion syndrome 3	DGUOK
1069	Mitochondrial DNA depletion syndrome 4A	POLG
1070	Mitochondrial DNA depletion syndrome 5	SUCLA2
1071	Mitochondrial DNA depletion syndrome 6	MPV17
1072	Mitochondrial DNA depletion syndrome 7	TWNK
1073	Mitochondrial DNA depletion syndrome 8A/8B	RRM2B
1074	Mitochondrial DNA depletion syndrome 9	SUCLG1
1075	Mitochondrial Short-Chain Enoyl-CoA Hydratase 1 Deficiency	ECHS1
1076	Mitochondrial complex I deficiency, nuclear type 1	NDUFS4
1077	Mitochondrial complex I deficiency, nuclear type 10	NDUFAF2
1078	Mitochondrial complex I deficiency, nuclear type 12	NDUFA1
1079	Mitochondrial complex I deficiency, nuclear type 14	NDUFA11
1080	Mitochondrial complex I deficiency, nuclear type 16	NDUFAF5

1081	Mitochondrial complex I deficiency, nuclear type 17	NDUFAF6
1082	Mitochondrial complex I deficiency, nuclear type 19	FOXRED1
1083	Mitochondrial complex I deficiency, nuclear type 21	NUBPL
1084	Mitochondrial complex I deficiency, nuclear type 22	NDUFA10
1085	Mitochondrial complex I deficiency, nuclear type 4	NDUFV1
1086	Mitochondrial complex I deficiency, nuclear type 5	NDUFS1
1087	Mitochondrial complex I deficiency, nuclear type 6	NDUFS2
1088	Mitochondrial complex I deficiency, nuclear type 7	NDUFV2
1089	Mitochondrial complex I deficiency, nuclear type 9	NDUFS6
1090	Mitochondrial complex I deficiency, nuclear type 2	NDUFS8
1091	Mitochondrial complex I deficiency, nuclear type 3	NDUFS7
1092	Mitochondrial complex II deficiency, nuclear type 2	SDHAF1
1093	Mitochondrial complex IV deficiency, nuclear type 1	SURF1
1094	Mitochondrial complex IV deficiency, nuclear type 3	COX10
1095	Mitochondrial complex III deficiency nuclear type 2	TTC19
1096	Mitochondrial complex III deficiency nuclear type 4	UQCRQ
1097	Mitochondrial complex III deficiency nuclear type 5	UQCRC2
1098	Mitochondrial complex III deficiency nuclear type 8	LYRM7
1099	Mitochondrial complex IV deficiency, nuclear type 11	COX20
1100	Mitochondrial complex IV deficiency, nuclear type 12	PET100
1101	Mitochondrial complex IV deficiency, nuclear type 17	COA8
1102	Mitochondrial complex I deficiency, nuclear type 20	ACAD9
1103	Mitochondrial Complex V (ATP Synthase) Deficiency, Nuclear Type 2	TMEM70
1104	Mitochondrial Neurodevelopmental disorder with abnormal movements and lactic acidosis with or without seizures	WARS2
1105	Adenylosuccinase Deficiency	ADSL
1106	Infantile Transient Liver Failure	TRMU
1107	Cerebellofaciodental Syndrome	BRF1
1108	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 1	VLDLR
1109	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2	WDR81
1110	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4	ATP8A2
1111	Microcephalic Osteodysplastic Primordial Dwarfism, Type I	RNU4ATAC
1112	Microcephalic Osteodysplastic Primordial Dwarfism, Type II	PCNT
1113	Microcephaly,Short Stature, And Limb Abnormalities	DONSON
1114	Microcephaly, Seizures, Spasticity, And Brain Calcifications	PCDH12
1115	Microcephaly, Epilepsy, and Diabetes Syndrome	IER3IP1
1116	Microcephaly, Short Stature, And Impaired Glucose Metabolism 1	TRMT10A
1117	Microcephaly-Capillary Malformation Syndrome	STAMBP

1118	Microphthalmia with coloboma 3	VSX2
1119	Acrodermatitis Enteropathica, Zinc-Deficiency Type	SLC39A4
1120	Neonatal Bartter syndrome type 4A with sensorineural deafness	BSND
1121	Citrullinemia, Type II, Neonatal-Onset	SLC25A13
1122	Neonatal Diabetes Mellitus with Congenital Hypothyroidism	GLIS3
1123	Neonatal Severe Hyperparathyroidism	CASR
1124	Lethal Neonatal Rigidity and Multifocal Seizure Syndrome	BRAT1
1125	Hemochromatosis, type 2A	HJV
1126	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease	ARPC1B
1127	Homocystinuria due to MTHFR deficiency	MTHFR
1128	Moyamoya disease 6 with or without achalasia	GUCY1A1
1129	Fumarase Deficiency	FH
1130	Severe Congenital Neutropenia, Autosomal Recessive,3	HAX1
1131	Severe Congenital Neutropenia, Autosomal Recessive,4	G6PC3
1132	Severe Congenital Neutropenia, Autosomal Recessive,5	VPS45
1133	Severe Congenital Neutropenia, Autosomal Recessive,6	JAGN1
1134	Fucosidosis	FUCA1
1135	Inflammatory Bowel Disease 28	IL10RA
1136	Anterior segement dysgenesis 2	FOXE3
1137	Anterior segement dysgenesis 7	PXDN
1138	Polyhydramnios, Megalencephaly, And Symptomatic Epilepsy	STRADA
1139	Insulin-Like Growth Factor I, Resistance to	IGF1R
1140	Hereditary Sensory and Autonomic Neuropathy type IIB	RETREG1
1141	Hereditary Sensory and Autonomic Neuropathy type II	WNK1
1142	Hereditary Sensory and Autonomic Neuropathy type V	NGF
1143	Hereditary Sensory and Autonomic Neuropathy type VIII	PRDM12
1144	Hereditary Hyperekplexia 3	SLC6A5
1145	Hereditary 4	ATAD1
1146	Hemochromatosis type 2B	HAMP
1147	Hereditary Folate Malabsorption	SLC46A1
1148	Hereditary Motor And Sensory Neuropathy type VIB	SLC25A46
1149	Ethylmalonic Encephalopathy	ETHE1
1150	Isobutyryl-CoA dehydrogenase deficiency	ACAD8
1151	Infantile liver failure syndrome 1	LARS1
1152	Striatonigral Degeneration, Infantile	NUP62
1153	Generalized Arterial Calcification of Infancy 2	ABCC6
1154	Hypotonia, Infantile, With Psychomotor Retardation And Characteristic facies 1	NALCN
1155	Hypotonia, Infantile, With Psychomotor Retardation And Characteristic facies 2	UNC80

1156	Hypotonia, Infantile, With Psychomotor Retardation And Characteristic facies 3	TBCK
1157	Infantile Parkinsonism-Dystonia	SLC6A3
1158	Infantile Sudden cardiac failure	PPA2
1159	Free sialic acid storage disease, infantile form	SLC17A5
1160	Infantile Cerebellar-Retinal Degeneration	ACO2
1161	Right Atrial Isomerism	GDF1
1162	Primary Autosomal Recessive Microcephaly 10	ZNF335
1163	Primary Autosomal Recessive Microcephaly 15	MFSD2A
1164	Primary Autosomal Recessive Microcephaly 17	CIT
1165	Primary Autosomal Recessive Microcephaly 1	MCPH1
1166	Primary Autosomal Recessive Microcephaly 20	KIF14
1167	Primary Autosomal Recessive Microcephaly 2, With Or Without Corticalmalformations	WDR62
1168	Primary Autosomal Recessive Microcephaly 3	CDK5RAP2
1169	Primary Autosomal Recessive Microcephaly 4	KNL1
1170	Primary Autosomal Recessive Microcephaly 5	ASPM
1171	Primary Autosomal Recessive Microcephaly 6	CENPJ
1172	Primary Autosomal Recessive Microcephaly 7	STIL
1173	Primary Coenzyme Q10 deficiency 1	COQ2
1174	Primary Coenzyme Q10 deficiency 4	COQ8A
1175	Primary Coenzyme Q10 deficiency 6	COQ6
1176	Primary Coenzyme Q10 deficiency 7	COQ4
1177	Primary Hyperoxaluria Type I	AGXT
1178	Primary Open Angle Glaucoma 3A	CYP1B1
1179	Distal Arthrogryposis type 5D	ECEL1
1180	Distal Renal Tubular Acidosis with Hemolytic Anemia	SLC4A1
1181	Early-Onset Myopathy, Areflexia, Respiratory Distress, and Dysphagia	MEGF10
1182	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum	TBCD
1183	Early Infantile Epileptic Encephalopathy 16	TBC1D24
1184	Early Infantile Epileptic Encephalopathy 25	SLC13A5
1185	Early Infantile Epileptic Encephalopathy 28	WWOX
1186	Early Infantile Epileptic Encephalopathy 34	SLC12A5
1187	Early Infantile Epileptic Encephalopathy 37	FRRS1L
1188	Early Infantile Epileptic Encephalopathy 38	ARV1
1189	Early Infantile Epileptic Encephalopathy 3	SLC25A22
1190	Early Infantile Epileptic Encephalopathy 44	UBA5
1191	Early Infantile Epileptic Encephalopathy 48	AP3B2
1192	Early Infantile Epileptic Encephalopathy 49	DENND5A
1193	Early Infantile Epileptic Encephalopathy 8	ARHGEF9
1194	Early Infantile Epileptic Encephalopathy 9	PCDH19

1195	Proliferative Vasculopathy And Hydranencephaly-Hydrocephaly Syndrome	FLVCR2
1196	Mucolipidosis III Alpha&Beta	GNPTAB
1197	Mucolipidosis III Gamma	GNPTG
1198	Xeroderma Pigmentosum Group A	XPA
1199	Xeroderma Pigmentosum Group C	XPC
1200	Xeroderma Pigmentosum Group G	ERCC5
1201	Cortical Malformations, Occipital	LAMC3
1202	Branched-chain Ketoacid Dehydrogenase Kinase Deficiency	BCKDK
1203	Limb-Girdle Muscular Dystrophy type 2E	SGCB
1204	Limb-Girdle Muscular Dystrophy type 2F	SGCD
1205	Limb-Girdle Muscular Dystrophy type 2G	TCAP
1206	Limb-Girdle Muscular Dystrophy type 2H	TRIM32
1207	Limb-Girdle Muscular Dystrophy type 2S	TRAPPC11
1208	Limb-Girdle Muscular Dystrophy type 2T	GMPPB
1209	Acromesomelic dysplasia 1	NPR2
1210	Acromesomelic dysplasia 2A	GDF5
1211	Acromesomelic dysplasia 3	BMPR1B
1212	Rhizomelic Chondrodysplasia Punctata type 1	PEX7
1213	Rhizomelic Chondrodysplasia Punctata type 2	GNPAT
1214	Rhizomelic chondrodysplasia punctata, type 3	AGPS
1215	Lipoyltransferase 1 deficiency	LIPT1
1216	Lipoid Congenital Adrenal Hyperplasia	STAR
1217	Pycnodysostosis	CTSK
1218	Lethal Arthrogryposis With Anterior Horn Cell Disease	GLE1
1219	Popliteal Pterygium Syndrome, Lethal Type	RIPK4
1220	Lethal congenital contracture syndrome 11	GLDN
1221	Lethal Congenital Contracture Syndrome 2	ERBB3
1222	Lethal Congenital Contracture Syndrome 3	PIP5K1C
1223	Lethal Congenital Contracture Syndrome 7	CNTNAP1
1224	Lethal Restrictive Dermopathy	LMNA, ZMPSTE24
1225	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies	OTUD6B
1226	Intellectual developmental disorder with cardiac arrhythmia	GNB5
1227	Foveal Hypoplasia 2	SLC38A8
1228	Centronuclear Myopathy 2	BIN1
1229	Severe Combined Immunodeficiency with Microcephaly, Growth Retardation, and Sensitivity to Ionizing Radiation	NHEJ1
1230	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive	IL7R
1231	Transcobalamin II Deficiency	TCN2
1232	Transaldolase Deficiency	TALDO1
1233	Autoimmune Polyendocrine Syndrome Type 1	AIRE

1234	Syndromic Microphthalmia 12	RARB
1235	Microphthalmia, syndromic 9	STRA6
1236	Histiocytosis-lymphadenopathy plus syndrome	SLC29A3
1237	Hypoplastic Left Heart Syndrome 1	GJA1
1238	Aromatic L-Amino Acid Decarboxylase Deficiency	DDC
1239	Orofaciodigital Syndrome XIV	C2CD3
1240	Cone-Rod Dystrophy 16	CFAP418
1241	Achromatopsia 3	CNGB3