Genescreen Easy-Donor: gene list v.1

N.B. A question mark (?) indicates that the relationship between the condition and gene is provisional.

	Gene name	Condition
1	**AAAS**	Triple-A syndrome (achalasia-addisonianism-alacrimia)
2	**ABAT**	GABA-transaminase deficiency
3	**ABCA12**	Ichthyosis congenital autosomal recessive type 4A and 4B (harlequin)
4	**ABCA3**	Surfactant metabolism dysfunction, pulmonary, type 3
5	**ABCC8**	Hyperinsulinemic hypoglycemia, type 1 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)
6	**ABCD1**	Adrenoleukodystrophy
7	**ABHD12**	PHARC syndrome (polyneuropathy, hearing loss, ataxia, retinitis pigmentosa and cataract)
8	**ACAD9**	Acyl-CoA dehydrogenase 9 deficiency (mitochondrial complex I deficiency, nuclear, type 20)
9	**ACADM**	Medium-chain acyl-CoA dehydrogenase deficiency
10	**ACADS**	Short-chain acyl-CoA dehydrogenase deficiency
11	**ACADVL**	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
12	**ACAT1**	Alpha-methylacetoacetic aciduria (3-ketothiolase deficiency)
13	**ACE**	Renal tubular dysgenesis
14	**ACOX1**	Peroxisomal acyl-CoA oxidase deficiency
15	**ACTA1**	Nemaline myopathy 3; Congenital fiber-type disproportion myopathy 1
16	**ADA**	Severe combined immunodeficiency due to adenosine deaminase deficiency (ADA)
17	**ADAMTS10**	Weill-Marchesani syndrome, type 1 recessive
18	**ADAMTS13**	Thrombotic thrombocytopenic purpura, familial (Schulman-Upshaw syndrome)
19	**AFF2**	Intellectual developmental disorder, X-linked 109
20	**AGA**	Aspartylglucosaminuria (glycosylasparaginase deficiency)
21	**AGL**	Glycogen storage disease, type 3
22	**AGPAT2**	Congenital generalized lipodystrophy (Berardinelli-Seip syndrome)
23	**AGPS**	Rhizomelic chondrodysplasia punctata, type 3
24	**AGRN**	Myasthenic syndrome, congenital, type 8
25	**AGXT**	Hyperoxaluria, primary, type 1
26	**AHI1**	Joubert syndrome, type 3
27	**AIMP1**	Leukodystrophy, hypomyelinating, type 3
28	**AIRE**	Autoimmune polyendocrinopathy syndrome, type 1
29	**ALDH18A1**	Spastic paraplegia, type 9B, autosomal recessive; Cutis laxa, type 3A (De Barsy syndrome)
30	**ALDH3A2**	Sjogren-Larsson syndrome
31	**ALDH5A1**	Succinic semialdehyde dehydrogenase deficiency
32	**ALDH7A1**	Epilepsy, pyridoxine-dependent
33	**ALDOB**	Fructose intolerance, hereditary
34	**ALG12**	Congenital disorder of glycosylation, type 1G
35	**ALG6**	Congenital disorder of glycosylation, type 1C
36	**ALMS1**	Alström syndrome
37	**ALOX12B**	Ichthyosis, congenital, autosomal recessive, type 2
38	**ALPL**	Hypophosphatasia, infantile/childhood
39	**AMACR**	Bile acid synthesis defect, congenital, type 4; Alpha-methylacyl-CoA racemase deficiency
40	**AMT**	Glycine encephalopathy

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	Gene name	Condition
41	**ANO10**	Spinocerebellar ataxia, autosomal recessive, type 10
42	**ANTXR2**	Hyaline fibromatosis syndrome
43	**AP1S2**	Pettigrew syndrome
44	**APTX**	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia
45	**AQP2**	Diabetes insipidus, nephrogenic, type 2
46	**ARFGEF2**	Periventricular heterotopia with microcephaly
47	**ARL13B**	Joubert syndrome type 8
48	**ARL6**	Bardet-Biedl syndrome, type 3
49	**ARSA**	Metachromatic leukodystrophy
50	**ARSB**	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)
51	**ARSE**	Chondrodysplasia punctata, X-linked recessive
52	**ARX**	Epileptic encephalopathy, early infantile, type 1; ARX-related developmental disorders
53	**ASL**	Argininosuccinic aciduria
54	**ASPA**	Canavan disease
55	**ASPM**	Primary microcephaly type 5 autosomal recessive
56	**ASS1**	Citrullinemia, type 1
57	**ATM**	Ataxia-telangiectasia
58	**ATP6V0A2**	Cutis laxa, autosomal recessive, type 2A; Wrinkly skin syndrome
59	**ATP7A**	Menkes disease; Occipital horn syndrome
60	**ATP7B**	Wilson disease
61	**ATR**	Seckel syndrome, type 1
62	**ATRX**	Intellectual disability-hypotonic facies syndrome, X-linked; Alpha-thalassemia/intellectual developmental disorder syndrome
63	**AUH**	3-methylglutaconic aciduria, type 1
64	**BBS1**	Bardet-Biedl syndrome, type 1
65	**BBS10**	Bardet-Biedl syndrome, type 10
66	**BBS12**	Bardet-Biedl syndrome, type 12
67	**BBS2**	Bardet-Biedl syndrome, type 2
68	**BBS7**	Bardet-Biedl syndrome, type 7
69	**BBS9**	Bardet-Biedl syndrome, type 9
70	**BCKDHA**	Maple syrup urine disease, type 1A
71	**BCKDHB**	Maple syrup urine disease, type 1B
72	**BCS1L**	Mitochondrial complex III deficiency nuclear type 1; GRACILE syndrome; Bjornstad syndrome
73	**BEST1**	Bestrophinopathy, AR
74	**BLM**	Bloom syndrome
75	**BRWD3**	Intellectual developmental disorder, X-linked 93
76	**BSCL2**	Congenital generalized lipodystrophy, type 2; Encephalopathy, progressive, with or withou lipodystrophy
77	**BTD**	Biotinidase deficiency
78	**BUB1B**	Mosaic variegated aneuploidy syndrome 1
79	**CBS**	Homocystinuria due to cystathionine beta-synthase
80	**CC2D2A**	Joubert syndrome, type 9; Meckel syndrome, type 6; COACH syndrome, 2
81	**CCDC88C**	Hydrocephalus, congenital, type 1
82	**CD40LG**	Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)
83	**CDAN1**	Dyserythropoietic anemia, congenital, type 1A
84	**CDH23**	Deafness, autosomal recessive, type 12; Usher syndrome, type 1D

	Gene name	Condition
85	**CDK5RAP2**	Primary microcephaly type 3 autosomal recessive
86	**CENPJ**	Primary microcephaly type 6 autosomal recessive
87	**CEP152**	Primary microcephaly type 9 autosomal recessive
88	**CEP290**	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10
89	**CFL2**	Nemaline myopathy, type 7 autosomal recessive
90	**CFTR**	Cystic fibrosis
91	**CHAT**	Myasthenic syndrome, congenital, type 6 presynaptic
92	**CHM**	Choroideremia
93	**CHRND**	Myasthenic syndrome, congenital, type 3B, fast-channel; Multiple pterygium syndrome, lethal type
94	**CHRNE**	Myasthenic syndrome, congenital, type 4B, fast-channel; Myasthenic syndrome, congenital, type 4C, associated with acetylcholine receptor deficiency
95	**CHRNG**	Multiple pterygium syndrome (MPS), Escobar type; MPS, lethal type
96	**CHST14**	Ehlers-Danlos syndrome, musculocontractural, type 1
97	**CHST3**	Spondyloepiphyseal dysplasia with congenital joint dislocations
98	**CLCN1**	Myotonia congenita, recessive
99	**CLN3**	Ceroid lipofuscinosis, neuronal, type 3
100	**CLN5**	Ceroid lipofuscinosis, neuronal, type 5
101	**CLN6**	Ceroid lipofuscinosis, neuronal, type 6
102	**CLN8**	Ceroid lipofuscinosis, neuronal, type 8
103	**CLRN1**	Usher syndrome, type 3A
104	**CNGB3**	Achromatopsia, type 3
105	**COG4**	Congenital disorder of glycosylation, type 2J
106	**COL11A2**	Otospondylomegaepiphyseal dysplasia, autosomal recessive
107	**COL17A1**	Epidermolysis bullosa, junctional, non-Herlitz type
108	**COL4A3**	Alport syndrome, autosomal recessive, type 2
109	**COL4A5**	Alport syndrome, X-linked
110	**COL7A1**	Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial
111	**COLQ**	Myasthenic syndrome, congenital, type 5
112	**COQ2**	Primary coenzyme Q10 deficiency, type 1
113	**CORO1A**	Immunodeficiency, type 8
114	**CPS1**	Carbamoylphosphate synthetase 1 deficiency
115	**CPT1A**	Carnitine palmitoyltransferase type 1A deficiency, hepatic
116	**CPT2**	Carnitine palmitoyltransferase type 2 deficiency, lethal neonatal; Carnitine palmitoyltransferase type 2 deficiency, infantile
117	**CRLF1**	Cold-induced sweating syndrome type 1
118	**CRTAP**	Osteogenesis imperfecta, type 7
119	**CSTB**	Epilepsy, progressive myoclonic type 1A (Unverricht and Lundborg)
120	**CTC1**	Cerebroretinal microangiopathy with calcifications and cysts
121	**CTNS**	Nephropathic cystinosis
122	**CTSD**	Ceroid lipofuscinosis, neuronal, type 10
123	**CTSK**	Pycnodysostosis
124	**CUL4B**	Intellectual developmental disorder, X-linked syndromic, Cabezas type
125	**CUL7**	3M syndrome 1
126	**CYBB**	Chronic granulomatous disease, X-linked
127	**CYP11A1**	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency

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	Gene name	Condition
128	**CYP1B1**	Glaucoma, primary congenital, type 3A
129	**CYP21A2**	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency
130	**CYP27A1**	Cerebrotendinous xanthomatosis
131	**CYP27B1**	Vitamin D-dependent rickets, type 1
132	**D2HGDH**	D-2-hydroxyglutaric aciduria
133	**DBH**	Dopamine beta-hydroxylase deficiency
134	**DBT**	Maple syrup urine disease, type 2
135	**DCLRE1C**	Omenn syndrome; Severe combined immunodeficiency, Athabascan type
136	**DCX**	Lissencephaly, X-linked, type 1
137	**DDC**	Aromatic L-amino acid decarboxylase deficiency
138	**DGUOK**	DGUOK-related mitochondrial DNA depletion syndrome
139	**DHCR7**	Smith-Lemli-Opitz syndrome
140	**DHDDS**	Retinitis pigmentosa, type 59
141	**DHH**	46,XY complete gonadal dysgenesis
142	**DLAT**	Pyruvate dehydrogenase E2 deficiency
143	**DLG3**	Intellectual developmental disorder, X-linked 90
144	**DMD**	Duchenne/Becker muscular dystrophy
145	**DMD**	Duchenne/Becker muscular dystrophy
146	**DMP1**	Hypophosphatemic rickets, autosomal recessive
147	**DNAH11**	Ciliary dyskinesia, primary, type 7 with or without situs inversus
148	**DNAH5**	Ciliary dyskinesia, primary, type 3 with or without situs inversus
149	**DNAL1**	Ciliary dyskinesia, primary, type 16
150	**DOCK8**	Hyper-IgE recurrent infection syndrome, autosomal recessive
151	**DOK7**	Fetal akinesia deformation sequence, type 3; Myasthenic syndrome, congenital, type 10
152	**DPAGT1**	Congenital disorder of glycosylation, type 1J; Myasthenic syndrome, congenital, type 13
153	**DPYD**	Dihydropyrimidine dehydrogenase deficiency
154	**DYNC2H1**	Short-rib thoracic dysplasia, type 3 with or without polydactyly
155	**EDA**	Ectodermal dysplasia, type 1 hypohidrotic, X-linked
156	**EDNRB**	ABCD syndrome
157	**EIF2AK3**	Wolcott-Rallison syndrome
158	**EIF2B2**	Leukoencephalopathy with vanishing white matter (VWM)
159	**EIF2B3**	Leukoencephalopathy with vanishing white matter (VWM)
160	**EIF2B4**	Leukoencephalopathy with vanishing white matter (VWM)
161	**EIF2B5**	Leukoencephalopathy with vanishing white matter (VWM)
162	**EMD**	Emery-Dreifuss muscular dystrophy, type 1 X-linked
163	**ENPP1**	Arterial calcification, generalized, of infancy, type 1
164	**EPG5**	Vici syndrome
165	**EPM2A**	Epilepsy, progressive myoclonic, type 2A (Lafora)
166	**ERCC2**	Trichothiodystrophy, type 1
167	**ERCC4**	Fanconi anemia, complementation group Q
168	**ERCC5**	Cerebrooculofacioskeletal syndrome, type 3
169	**ERCC6**	Cockayne syndrome, type B; Cerebrooculofacioskeletal syndrome, type 1
170	**ERCC8**	Cockayne syndrome, type A
171	**ESCO2**	Roberts syndrome; Juberg-Hayward syndrome
172	**ETFDH**	Glutaric acidemia, type 2C
173	**ETHE1**	Ethylmalonic encephalopathy

	Gene name	Condition
174	**EVC**	Ellis-van Creveld syndrome
175	**EVC2**	Ellis-van Creveld syndrome
176	**EXOSC3**	Pontocerebellar hypoplasia, type 1B
177	**F11**	Factor XI deficiency
178	**F8**	Hemophilia A
179	**F9**	Hemophilia B
180	**FA2H**	Spastic paraplegia, type 35 autosomal recessive
181	**FAH**	Tyrosinemia, type 1
182	**FAM20C**	Raine syndrome
183	**FANCA**	Fanconi anemia, complementation group A
184	**FANCC**	Fanconi anemia, complementation group C
185	**FANCF**	Fanconi anemia, complementation group F
186	**FANCG**	Fanconi anemia, complementation group G
187	**FANCI**	Fanconi anemia, complementation group I
188	**FGD1**	Aarskog-Scott syndrome; intellectual developmental disorder, X-linked syndromic, type 16
189	**FKRP**	Muscular dystrophy-dystroglycanopathy, type 5A (Walker-Warburg syndrome); Type 5B; Type 5C (limb-girdle muscular dystrophy, type 9 [LGMDR9])
190	**FKTN**	Muscular dystrophy-dystroglycanopathy, type 4A (Walker-Warburg syndrome); Type 4B; Type 4C (limb-girdle muscular dystrophy, type 13 [LGMD R13])
191	**FMO3**	Trimethylaminuria
192	**FMR1**	Fragile X syndrome
193	**FRAS1**	Fraser syndrome, type 1
194	**FTSJ1**	Intellectual developmental disorder, X-linked 9
195	**FUCA1**	Fucosidosis
196	**G6PC**	Glycogen storage disease, type 1A
197	**G6PD**	Hemolytic anemia, G6PD deficient (favism)
198	**GAA**	Glycogen storage disease, type 2
199	**GALC**	Krabbe disease
200	**GALT**	Galactosemia
201	**GAMT**	Cerebral creatine deficiency syndrome, type 2
202	**GAN**	Giant axonal neuropathy, type 1
203	**GATM**	Cerebral creatine deficiency syndrome, type 3
204	**GBA**	Gaucher Disease
205	**GBE1**	Glycogen storage disease, type 4
206	**GCDH**	Glutaricaciduria, type 1
207	**GFM1**	Combined oxidative phosphorylation deficiency, type 1
208	**GFPT1**	Myasthenia, congenital, type 12 with tubular aggregates
209	**GJB1**	Charcot-Marie-Tooth neuropathy, X-linked dominant, type 1
210	**GJB2**	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6
211	**GJB6**	Deafness, autosomal recessive, type 1B; Deafness, digenic GJB2/GJB6
212	**GLA**	Fabry disease
213	**GLB1**	GM1-gangliosidosis, types 1-3; Mucopolysaccharidosis, type 4B (Morquio)
214	**GLDC**	Glycine encephalopathy
215	**GLRA1**	Hyperekplexia, type 1
216	**GNPTAB**	Mucolipidosis 2 alpha/beta; Mucolipidosis 3 alpha/beta
217	**GNRHR**	Hypogonadotropic hypogonadism, type 7 without anosmia

	Gene name	Condition
218	**GOSR2**	Epilepsy, progressive myoclonic, type 6
219	**GP1BA**	Bernard-Soulier syndrome, type A1
220	**GP1BB**	Bernard-Soulier syndrome, type B
221	**GPHN**	Molybdenum cofactor deficiency C
222	**GPR143**	Ocular albinism, type 1 (Nettleship-Falls type)
223	**GRHPR**	Hyperoxaluria, primary, type 2
224	**GRIP1**	Fraser syndrome 3
225	**HADH**	3-hydroxyacyl-CoA dehydrogenase deficiency
226	**HADHA**	Long-chain 3-hydroxyl-CoA dehydrogenase (LCHAD) deficiency; Mitochondrial trifunctional protein deficiency
227	**HBA1**	Alpha-thalassemia
228	**HBA2**	Alpha-thalassemia
229	**HBB**	Beta-thalassemia, Sickle cell anemia and other HBB-related hemoglobinopathies
230	**HEXA**	Tay-Sachs disease
231	**HEXB**	Sandhoff disease, infantile, juvenile, and adult forms
232	**HGSNAT**	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)
233	**HIBCH**	3-hydroxyisobutryl-CoA hydrolase deficiency
234	**HLCS**	Holocarboxylase synthetase deficiency
235	**HPS3**	Hermansky-Pudlak syndrome, type 3
236	**HPS4**	Hermansky-Pudlak syndrome, type 4
237	**HPS6**	Hermansky-Pudlak syndrome, type 6
238	**HSD17B3**	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency
239	**HSD17B4**	D-bifunctional protein deficiency
240	**HSD3B2**	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency
241	**HSPG2**	Dyssegmental dysplasia, Silverman-Handmaker type
242	**IDS**	Mucopolysaccharidosis, type 2
243	**IDUA**	Mucopolysaccharidosis type 1
244	**IGHMBP2**	Charcot-Marie-Tooth disease, axonal, type 2S
245	**IL1RAPL1**	Intellectual developmental disorder, X-linked 21
246	**IL2RG**	Severe combined immunodeficiency, X-linked
247	**IL7R**	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type
248	**INPP5E**	Joubert syndrome, type 1
249	**INSR**	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, type A
250	**ITGB4**	Epidermolysis bullosa, junctional, with pyloric atresia
251	**IVD**	Isovaleric acidemia
252	**JAK3**	Severe Combined Immunodeficiency, autosomal recessive, T-negative/B-positive type
253	**KCNJ1**	Bartter syndrome, type 2
254	**KCNJ13**	Leber congenital amaurosis, type 16
255	**KCTD7**	Epilepsy, progressive myoclonic, type 3 with or without intracellular inclusions
256	**KDM5C**	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type
257	**KIF7**	Acrocallosal syndrome; Joubert syndrome, type 12
258	**L1CAM**	L1 Syndrome
259	**LAMA2**	LAMA2-related muscular dystrophy
260	**LAMA3**	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
261	**LAMB2**	Pierson syndrome; Nephrotic syndrome, type 5 with or without ocular abnormalities

	Gene name	Condition
262	**LAMB3**	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
263	**LAMC2**	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
264	**LIFR**	Stuve-Wiedemann syndrome / Schwartz-Jampel type 2 syndrome
265	**LMNA**	Cardiomyopathy, dilated, 1A
266	**LPL**	Lipoprotein lipase deficiency
267	**LRP2**	Donnai-Barrow syndrome
268	**LRPPRC**	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian)
269	**LTBP4**	Cutis laxa, autosomal recessive, type 1C
270	**MAN2B1**	Alpha-mannosidosis
271	**MCCC2**	3-Methylcrotonyl-CoA carboxylase deficiency, type 2
272	**MCOLN1**	Mucolipidosis type 4
273	**MCPH1**	Microcephaly type 1 primary, autosomal recessive
274	**MFN2**	Charcot-Marie-Tooth disease, axonal, type 2A2B
275	**MFSD8**	Ceroid lipofuscinosis, neuronal, type 7
276	**MID1**	Opitz GBBB syndrome, type 1
277	**MKS1**	Bardet-Biedl syndrome type 13; Meckel syndrome, type 1; Joubert syndrome, type 28
278	**MLC1**	Megalencephalic leukoencephalopathy with subcortical cysts
279	**MMAB**	Methylmalonic aciduria, vitamin B12-responsive, type cblB
280	**MMACHC**	Methylmalonic aciduria and homocystinuria, cblC type
281	**MMADHC**	Homocystinuria, cblD type, variant 1
282	**MMUT**	Methylmalonic aciduria, mut(0) type
283	**MPDU1**	Congenital disorder of glycosylation, type 1F
284	**MPI**	Congenital disorder of glycosylation, type 1B
285	**MPV17**	Mitochondrial DNA depletion syndrome type 6 (hepatocerebral); Charcot-Marie-Tooth disease, axonal, type 2EE
286	**MPZ**	Dejerine-Sottas disease
287	**MTHFR**	Homocystinuria due to MTHFR deficiency
288	**MTM1**	Myotubular myopathy, X-linked
289	**MTO1**	Combined oxidative phosphorylation deficiency 10
290	**MTR**	Homocystinuria-megaloblastic anemia, cblG complementation type
291	**MUSK**	Fetal akinesia deformation sequence, type 1; Myasthenic syndrome, congenital, type 9 associated with acetylcholine receptor deficiency
292	**MVK**	Mevalonic aciduria
293	**MYO15A**	Deafness, autosomal recessive, type 3
294	**MYO5A**	Griscelli syndrome, type 1
295	**MYO5B**	Microvillus inclusion disease
296	**MYO7A**	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2
297	**NAGA**	Schindler disease, type I
298	**NBN**	Nijmegen breakage syndrome
299	**NDP**	Norrie disease
300	**NEB**	Nemaline myopathy type 2
301	**NEUROG3**	Diarrhea 4 malabsorptive, congenital
302	**NHLRC1**	Epilepsy, progressive myoclonic, type 2B (Lafora)
303	**NPC1**	Niemann-Pick disease, type C1
304	**NPC2**	Niemann-pick disease, type C2
305	**NPHP1**	Joubert syndrome type 4

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306	**NPHP3**	Meckel syndrome type 7
307	**NPHP4**	Nephronophthisis type 4
308	**NPHS1**	Nephrotic syndrome, type 1
309	**NPHS2**	Nephrotic syndrome, type 2
310	**NR0B1**	Adrenal hypoplasia, congenital
311	**NTRK1**	Insensitivity to pain, congenital, with anhidrosis
312	**NUP62**	Striatonigral degeneration, infantile
313	**OBSL1**	3M syndrome 2
314	**OCA2**	Oculocutaneous albinism type 2
315	**OCRL**	Lowe Syndrome; Dent disease type 2
316	**OPHN1**	Intellectual developmental disorder, X-linked syndromic, Billuart type
317	**OTC**	Ornithine transcarbamylase deficiency
318	**PAH**	Phenylketonuria
319	**PAK3**	Intellectual developmental disorder, X-linked 30
320	**PC**	Pyruvate carboxylase deficiency
321	**PCCA**	Propionic acidemia
322	**PCDH15**	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic
323	**PDHA1**	Pyruvate dehydrogenase E1-alpha deficiency
324	**PDHB**	Pyruvate dehydrogenase E1-beta deficiency
325	**PDHX**	Lacticacidemia due to PDX1 deficiency
326	**PEX1**	Heimler syndrome type 1
327	**PEX10**	Peroxisome biogenesis disorder, type 6A (Zellweger syndrome); Peroxisome biogenesis disorder, type 6B
328	**PEX16**	Peroxisome biogenesis disorder, type 8A (Zellweger syndrome); Peroxisome biogenesis disorder, type 8B
329	**PEX7**	Rhizomelic chondrodysplasia punctata, type 1
330	**PGK1**	Phosphoglycerate kinase 1 deficiency
331	**PHF8**	Intellectual developmental disorder, X-linked syndromic, Siderius type
332	**PKHD1**	Polycystic kidney disease type 4
333	**PKP1**	Ectodermal dysplasia/skin fragility syndrome
334	**PLA2G6**	Infantile neuroaxonal dystrophy type 1
335	**PLEC**	Epidermolysis bullosa simplex with muscular dystrophy
336	**PLOD1**	Ehlers-Danlos syndrome, kyphoscoliotic type, 1
337	**PLP1**	Pelizaeus-Merzbacher disease
338	**PMM2**	Congenital disorder of glycosylation, type 1A
339	**PNPLA1**	Ichthyosis, congenital, autosomal recessive, type 10
340	**PNPO**	Pyridoxamine 5'-phosphate oxidase deficiency
341	**POLG**	POLG-related disorders
342	**POLR1C**	Leukodystrophy, hypomyelinating, type 11; Treacher Collins syndrome 3
343	**POMGNT1**	Muscular dystrophy-dystroglycanopathy, type 3A (Walker-Warburg syndrome); Type 3B; Type 3C (limb-girdle muscular dystrophy, type 15 [LGMDR15])
344	**POMT1**	Muscular dystrophy-dystroglycanopathy, type 1A (Walker-Warburg syndrome); Type 1B; Type 1C (limb-girdle muscular dystrophy, type 11 [LGMD R11]) muscular dystrophy, type 1 [LGMD R11])
345	**POMT2**	Muscular dystrophy-dystroglycanopathy, type 2A (Walker-Warburg syndrome); Type 2B; Type 2C (limb-girdle muscular dystrophy, type 14 [LGMD R14]) muscular dystrophy, type 14 [LGMD R14])

	Gene name	Condition
346	**POU1F1**	Pituitary hormone deficiency, combined, type 1
347	**POU3F4**	Deafness, X-linked, type 2
348	**PPT1**	Ceroid lipofuscinosis, neuronal, type 1
349	**PQBP1**	Renpenning syndrome
350	**PRF1**	Hemophagocytic lymphohistiocytosis, familial, type 2
351	**PRICKLE1**	Epilepsy, progressive myoclonic, type 1B
352	**PROP1**	Pituitary hormone deficiency, combined, type 2
353	**PRPS1**	Phosphoribosylpyrophosphate synthetase (PRS) deficiency
354	**PSAP**	Combined SAP deficiency
355	**PYGL**	Glycogen storage disease, type 6
356	**RAB3GAP1**	Warburg micro syndrome; Martsolf syndrome
357	**RAB3GAP2**	Warburg micro syndrome; Martsolf syndrome
358	**RAD51C**	Fanconi anemia, complementation group O
359	**RAG1**	Omenn syndrome; Severe combined immunodeficiency, B cell-negative
360	**RAG2**	Omenn syndrome; Severe combined immunodeficiency, B cell-negative
361	**RAPSN**	Fetal akinesia deformation sequence, type 2; Myasthenic syndrome, congenital, type 11 associated with AChR deficiency
362	**RARS2**	Pontocerebellar hypoplasia, type 6
363	**RELN**	Lissencephaly 2 (Norman-Roberts type)
364	**RMRP**	Anauxetic dysplasia 1
365	**RNASEH2B**	Aicardi-Goutieres syndrome, type 2
366	**ROR2**	Robinow syndrome, autosomal recessive
367	**RP2**	Retinitis pigmentosa, type 2 X-linked
368	**RPGR**	Retinitis pigmentosa, type 3 X-linked; Cone-rod dystrophy, X-linked, 1
369	**RPGRIP1**	Leber congenital amaurosis, type 6
370	**RPGRIP1L**	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome
371	**RS1**	Retinoschisis
372	**RTEL1**	Dyskeratosis congenita, autosomal recessive type 5
373	**RYR1**	RYR1 related congenitalmyopathy
374	**SACS**	Spastic ataxia, Charlevoix-Saguenay, type
375	**SBDS**	Shwachman-Diamond syndrome
376	**SCN4A**	Myasthenic syndrome, congenital, type 16
377	**SCN9A**	Indifference to pain and autosomal recessive hereditary sensory neuropathy type 2D
378	**SCNN1B**	Pseudohypoaldosteronism, type 1
379	**SCO2**	Mitochondrial complex IV deficiency, nuclear type 2
380	**SGCA**	Limb-girdle muscular dystrophy, type 3 (LGMD R3)
381	**SGCB**	Limb-girdle muscular dystrophy, type 4 (LGMD R4)
382	**SH3TC2**	Charcot-Marie-Tooth disease, type 4C
383	**SIL1**	Marinesco-Sjogren syndrome
384	**SLC12A1**	Bartter syndrome, type 1
385	**SLC12A6**	Agenesis of the corpus callosum with peripheral neuropathy
386	**SLC16A2**	Allan-Herndon-Dudley syndrome
387	**SLC17A5**	Salla disease
388	**SLC19A2**	Thiamine-responsive megaloblastic anemia syndrome
389	**SLC19A3**	Thiamine metabolism dysfunction syndrome, type 2 (biotin- or thiamine-responsive encephalopathy type)

	Gene name	Condition
390	**SLC25A19**	Microcephaly, Amish type; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type)
391	**SLC25A22**	Developmental and epileptic encephalopathy 3
392	**SLC26A2**	Achondrogenesis, type 1B (diastrophic dysplasia)
393	**SLC26A4**	Deafness, autosomal recessive, type 4; Pendred syndrome
394	**SLC2A2**	Fanconi-Bickel syndrome
395	**SLC35A1**	Congenital disorder of glycosylation, type 2F
396	**SLC35C1**	Congenital disorder of glycosylation, type 2C
397	**SLC37A4**	Glycogen storage disease, type 1B
398	**SLC3A1**	Cystinuria
399	**SLC45A2**	Albinism, oculocutaneous, type 4
400	**SLC4A11**	Corneal endothelial dystrophy, autosomal recessive
401	**SLC6A8**	Cerebral creatine deficiency syndrome, type 1
402	**SMN1**	Spinal muscular atrophy
403	**SMPD1**	Niemann-Pick disease, type A; Niemann-Pick disease, type B
404	**SNAP29**	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome
405	**SP110**	Hepatic venoocclusive disease with immunodeficiency
406	**SPATA7**	Leber congenital amaurosis, type 3
407	**SRD5A2**	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency (pseudovaginal perineoscrotal hypospadias)
408	**SRD5A3**	Congenital disorder of glycosylation, type 1Q; Kahrizi syndrome
409	**ST3GAL5**	Salt and pepper developmental regression syndrome
410	**STAR**	Lipoid adrenal hyperplasia
411	**STIL**	Microcephaly, type 7 primary, autosomal recessive
412	**STRA6**	Microphthalmia, isolated, with coloboma, type 8
413	**SUOX**	Sulfite oxidase deficiency
414	**SURF1**	Charcot-Marie-Tooth disease, type 4K; Leigh syndrome, due to COX IV deficiency
415	**SYN1**	Epilepsy, X-linked, with variable learning disabilities and behavior disorders
416	**TCTN1**	Joubert syndrome, type 13
417	**TCTN2**	Joubert syndrome, type 24; ?Meckel syndrome, type 8
418	**TF**	Atransferrinemia
419	**TGM1**	Ichthyosis, congenital, autosomal recessive, type 1
420	**TH**	Segawa syndrome, recessive
421	**THOC2**	intellectual developmental disorder, X-linked 12
422	**TJP2**	Cholestasis, progressive familial intrahepatic, type 4
423	**TMEM138**	Joubert syndrome 16
424	**TMEM216**	Joubert syndrome, type 2; Meckel syndrome, type 2
425	**TMEM237**	Joubert syndrome, type 14
426	**TMEM67**	Joubert syndrome, type 6; Meckel syndrome, type 3; COACH syndrome
427	**TNNT1**	Nemaline myopathy , type 5 Amish type
428	**TNXB**	Ehlers-Danlos syndrome, classic-like
429	**TPK1**	Episodic encephalopathy due to thiamine pyrophosphokinase deficiency
430	**TPP1**	Ceroid lipofuscinosis, neuronal, type 2; Spinocerebellar ataxia, autosomal recessive, type 7
431	**TREX1**	Aicardi-Goutieres syndrome, type 1
432	**TRIM32**	Limb-girdle muscular dystrophy, type 8 (LGMD R8)
433	**TSEN2**	Pontocerebellar hypoplasia, type 2B

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	Gene name	Condition
434	**TSEN34**	Pontocerebellar hypoplasia type 2C
435	**TSEN54**	Pontocerebellar hypoplasia, type 2A; Pontocerebellar hypoplasia, type 4
436	**TSFM**	Combined oxidative phosphorylation deficiency, type 3
437	**TTC21B**	Short-rib thoracic dysplasia, type 4 with or without polydactyly
438	**TTC8**	Bardet-Biedl syndrome, type 8
439	**TTN**	Limb-girdle muscular dystrophy type 10 (LGMDR10); Early-onset myopathy with fatal cardiomyopathy (Salih myopathy)
440	**TTPA**	Ataxia with isolated vitamin E deficiency
441	**TYR**	Oculocutaneous albinism (OCA) type 1A; OCA type 1B
442	**UBR1**	Johanson-Blizzard syndrome
443	**UNC13D**	Hemophagocytic lymphohistiocytosis, familial, type 3
444	**UPF3B**	Intellectual developmental disorder, X-linked syndromic 14
445	**USH1C**	Usher syndrome, type 1C; Deafness, autosomal recessive, type 18A
446	**USH1G**	Usher syndrome, type 1G
447	**USH2A**	Usher syndrome, type 2A
448	**VLDLR**	Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1
449	**VPS13B**	Cohen syndrome
450	**VPS33B**	Arthrogryposis, renal dysfunction and cholestasis, type 1
451	**WAS**	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked
452	**WDR62**	Microcephaly, type 2 primary, autosomal recessive, with or without cortical malformations
453	**WFS1**	Wolfram syndrome, type 1
454	**WNT7A**	Fuhrmann syndrome
455	**WRN**	Werner syndrome
456	**XPA**	Xeroderma pigmentosum, group A
457	**XPC**	Xeroderma pigmentosum, group C
458	**ZAP70**	Autoimmune disease, multisystem, infantile-onset, type 2; Immunodeficiency, type 48
459	**ZDHHC9**	Intellectual developmental disorder, X-linked syndromic, Raymond type
460	**ZMPSTE24**	Mandibuloacral dysplasia with, type B lipodystrophy
461	**ZNF711**	Intellectual developmental disorder, X-linked 97

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