

	GENE SYMBOL	GENE NAME
1	AASS	Alpha-aminoadipic semialdehyde synthase, mitochondrial
2	ABAT	4-aminobutyrate aminotransferase, mitochondrial
3	ABCC8	ATP-binding cassette sub-family C member 8
4	ABCD1	ATP-binding cassette sub-family D member 1
5	ABHD5	1-acylglycerol-3-phosphate O-acyltransferase ABHD5
6	ACOX1	Peroxisomal acyl-coenzyme A oxidase 1
7	ACSL4	Long-chain-fatty-acid--CoA ligase 4
8	ADA	Adenosine deaminase
9	ADSL	Adenylosuccinate lyase
10	AFF2	AF4/FMR2 family member 2
11	AGA	N(4)-(beta-N-acetylglucosaminy)-L-asparaginase
12	AHDC1	AT-hook DNA-binding motif-containing protein 1
13	AK1	Adenylate kinase isoenzyme 1
14	ALDH18A1	Delta-1-pyrroline-5-carboxylate synthase
15	ALDH3A2	Fatty aldehyde dehydrogenase
16	ALDH4A1	Delta-1-pyrroline-5-carboxylate dehydrogenase, mitochondrial
17	ALDH5A1	Succinate-semialdehyde dehydrogenase, mitochondrial
18	ALDOA	Fructose-bisphosphate aldolase A
19	ALG12	Dol-P-Man:Man(7)GlcNAc(2)-PP-Dol alpha-1,6-mannosyltransferase
20	ALG2	Alpha-1,3/1,6-mannosyltransferase ALG2
21	ALG3	Dol-P-Man:Man(5)GlcNAc(2)-PP-Dol alpha-1,3-mannosyltransferase
22	ALG6	Dolichyl pyrophosphate Man9GlcNAc2 alpha-1,3-glucosyltransferase
23	AMT	Aminomethyltransferase, mitochondrial
24	ANKRD11	Ankyrin repeat domain-containing protein 11
25	AP1S2	AP-1 complex subunit sigma-2
26	AP1S3	AP-1 complex subunit sigma-3
27	ARG1	Arginase-1
28	ARHGEF9	Rho guanine nucleotide exchange factor 9
29	ARID1A	AT-rich interactive domain-containing protein 1A
30	ARID1B	AT-rich interactive domain-containing protein 1B
31	ARID2	AT-rich interactive domain-containing protein 2
32	ARSA	Arylsulfatase A
33	ARX	Homeobox protein ARX
34	ASAH1	Acid ceramidase
35	ASL	Argininosuccinate lyase
36	ASPA	Aspartoacylase
37	ASPM	Abnormal spindle-like microcephaly-associated protein
38	ASS1	Argininosuccinate synthase
39	ATN1	Atrophin-1
40	ATP2A2	Sarcoplasmic/endoplasmic reticulum calcium ATPase 2
41	ATP7A	Copper-transporting ATPase 1

42	ATR	Serine/threonine-protein kinase ATR
43	ATRX	Transcriptional regulator ATRX
44	AVPR2	Vasopressin V2 receptor
45	B4GALT1	Beta-1,4-galactosyltransferase 1
46	B4GALT7	Beta-1,4-galactosyltransferase 7
47	BBS1	Bardet-Biedl syndrome 1 protein
48	BBS2	Bardet-Biedl syndrome 2 protein
49	BBS4	Bardet-Biedl syndrome 4 protein
50	BCKDHA	2-oxoisovalerate dehydrogenase subunit alpha, mitochondrial
51	BCKDHB	2-oxoisovalerate dehydrogenase subunit beta, mitochondrial
52	BCOR	BCL-6 corepressor
53	BCS1L	Mitochondrial chaperone BCS1
54	BHLHE40	Class E basic helix-loop-helix protein 40
55	BRWD3	Bromodomain and WD repeat-containing protein 3
56	BSCL2	Seipin
57	BSND	Barttin
58	BTD	Biotinidase
59	C3ORF58	Deleted in autism protein 1
60	CA2	Carbonic anhydrase 2
61	CASK	Kappa-casein
62	CASK	Peripheral plasma membrane protein CASK
63	CBS	Cystathionine beta-synthase
64	CC2D1A	Coiled-coil and C2 domain-containing protein 1A
65	CDK5RAP2	CDK5 regulatory subunit-associated protein 2
66	CDKL5	Cyclin-dependent kinase-like 5
67	CENPJ	Centromere protein J
68	CFC1B	Cryptic family protein 1B
69	CHD7	Chromodomain-helicase-DNA-binding protein 7
70	CHD8	Chromodomain-helicase-DNA-binding protein 8
71	CLCNKB	Chloride channel protein CIC-Kb
72	CLN3	Battenin
73	CLN5	Ceroid-lipofuscinosis neuronal protein 5
74	CLN6	Ceroid-lipofuscinosis neuronal protein 6
75	CLN8	Protein CLN8
76	COX10	Protoheme IX farnesyltransferase, mitochondrial
77	CPS1	Carbamoyl-phosphate synthase [ammonia], mitochondrial
78	CRBN	Protein cereblon
79	CREBBP	CREB-binding protein
80	CTCF	Transcriptional repressor CTCF
81	CTSA	Lysosomal protective protein
82	CUL4B	Cullin-4B
83	CYP27A1	Sterol 26-hydroxylase, mitochondrial

84	DBT	Lipoamide acyltransferase of branched-chain alpha-keto acid dehydrogenase complex, mitochondrial
85	DCX	Neuronal migration protein doublecortin
86	DHCR7	7-dehydrocholesterol reductase
87	DIAPH1	Protein diaphanous homolog 1
88	DKC1	H/ACA ribonucleoprotein complex subunit 4
89	DLD	Dihydrolipoyl dehydrogenase, mitochondrial
90	DLG3	Disks large homolog 3
91	DMD	Dystrophin
92	DMPK	Myotonin-protein kinase
93	DNMT3B	DNA (cytosine-5)-methyltransferase 3B
94	DOCK8	Dedicator of cytokinesis protein 8
95	DPAGT1	UDP-N-acetylglucosamine--dolichyl-phosphate N-acetylglucosaminophosphotransferase
96	DPM1	Dolichol-phosphate mannosyltransferase subunit 1
97	DPYD	Dihydropyrimidine dehydrogenase [NADP(+)]
98	DPYS	Dihydropyrimidinase
99	DUOX2	Dual oxidase 2
100	DYM	Dymeclin
101	EDEM1	ER degradation-enhancing alpha-mannosidase-like protein 1
102	EHMT1	Histone-lysine N-methyltransferase EHMT1
103	EIF2AK3	Eukaryotic translation initiation factor 2-alpha kinase 3
104	EMX2	Homeobox protein EMX2
105	EP300	Histone acetyltransferase p300
106	ERCC2	TFIIH basal transcription factor complex helicase XPD subunit
107	ERCC3	TFIIH basal transcription factor complex helicase XPB subunit
108	ERCC5	DNA repair protein complementing XP-G cells
109	ERCC6	DNA excision repair protein ERCC-6
110	ERCC8	DNA excision repair protein ERCC-8
111	EZH2	Histone-lysine N-methyltransferase EZH2
112	FANCA	Fanconi anemia group A protein
113	FANCB	Fanconi anemia group B protein
114	FANCC	Fanconi anemia group C protein
115	FBN1	Fibrillin-1
116	FGD1	FYVE, RhoGEF and PH domain-containing protein 1
117	FGFR1	Fibroblast growth factor receptor 1
118	FGFR2	Fibroblast growth factor receptor 2
119	FGFR3	Fibroblast growth factor receptor 3
120	FH	Fumarate hydratase, mitochondrial
121	FKRP	Fukutin-related protein
122	FKTN	Fukutin
123	FLNA	Filamin-A
124	FMR1	Fragile X mental retardation protein 1
125	FOXE1	Forkhead box protein E1

126	FOXP2	Forkhead box protein P2
127	FRAS1	Extracellular matrix protein FRAS1
128	FTSJ1	Putative tRNA (cytidine(32)/guanosine(34)-2'-O)-methyltransferase
129	FUCA1	Tissue alpha-L-fucosidase
130	FXR1	Fragile X mental retardation syndrome-related protein 1
131	FXR2	Fragile X mental retardation syndrome-related protein 2
132	G6PD	Glucose-6-phosphate 1-dehydrogenase
133	GALE	UDP-glucose 4-epimerase
134	GALT	Galactose-1-phosphate uridylyltransferase
135	GAMT	Guanidinoacetate N-methyltransferase
136	GATM	Glycine amidinotransferase, mitochondrial
137	GCDH	Glutaryl-CoA dehydrogenase, mitochondrial
138	GCH1	GTP cyclohydrolase 1
139	GCSH	Glycine cleavage system H protein, mitochondrial
140	GDI1	Rab GDP dissociation inhibitor alpha
141	GFAP	Glial fibrillary acidic protein
142	GK	Glycerol kinase
143	GLB1	Beta-galactosidase
144	GLDC	Glycine dehydrogenase (decarboxylating), mitochondrial
145	GLI3	Transcriptional activator GLI3
146	GM2A	Ganglioside GM2 activator
147	GNAS	Guanine nucleotide-binding protein G(s) subunit alpha isoforms XLas
148	GNPAT	Dihydroxyacetone phosphate acyltransferase
149	GNS	N-acetylglucosamine-6-sulfatase
150	GPC3	Glypican-3
151	GPI	Glucose-6-phosphate isomerase
152	GRIA3	Glutamate receptor 3
153	GSS	Glutathione synthetase
154	GTF2I	General transcription factor II-I
155	GTF2IRD1	General transcription factor II-I repeat domain-containing protein 1
156	GUSB	Beta-glucuronidase
157	HCCS	Cytochrome c-type heme lyase
158	HCFC1	Host cell factor 1
159	HDAC8	Histone deacetylase 8
160	HERC2	E3 ubiquitin-protein ligase HERC2
161	HESX1	Homeobox expressed in ES cells 1
162	HEXA	Beta-hexosaminidase subunit alpha
163	HEXB	Beta-hexosaminidase subunit beta
164	HLCS	Biotin--protein ligase
165	HMGCL	Hydroxymethylglutaryl-CoA lyase, mitochondrial
166	HPD	4-hydroxyphenylpyruvate dioxygenase
167	HPRT1	Hypoxanthine-guanine phosphoribosyltransferase

168	HSD17B10	3-hydroxyacyl-CoA dehydrogenase type-2
169	HUWE1	E3 ubiquitin-protein ligase HUWE1
170	IDS	Iduronate 2-sulfatase
171	IDUA	Alpha-L-iduronidase
172	IGF1	Insulin-like growth factor I
173	IKBKG	NF-kappa-B essential modulator
174	IL1RAPL1	Interleukin-1 receptor accessory protein-like 1
175	IL5RA	Interleukin-5 receptor subunit alpha
176	IQSEC2	IQ motif and SEC7 domain-containing protein 2
177	ITGA7	Integrin alpha-7
178	ITPR1	Inositol 1,4,5-trisphosphate receptor type 1
179	JAG1	Protein jagged-1
180	JARID2	Protein Jumonji
181	KANSL1	KAT8 regulatory NSL complex subunit 1
182	KAT6A	Histone acetyltransferase KAT6A
183	KAT6B	Histone acetyltransferase KAT6B
184	KCNJ1	ATP-sensitive inward rectifier potassium channel 1
185	KCNJ11	ATP-sensitive inward rectifier potassium channel 11
186	KDM5C	Lysine-specific demethylase 5C
187	KDM6A	Lysine-specific demethylase 6A
188	KDM6B	Lysine-specific demethylase 6B
189	KIRREL3	Kin of IRRE-like protein 3
190	KMT2A	Histone-lysine N-methyltransferase 2A
191	KMT2D	Histone-lysine N-methyltransferase 2D
192	L1CAM	Neural cell adhesion molecule L1
193	LAMA2	Laminin subunit alpha-2
194	LAMP2	Lysosome-associated membrane glycoprotein 2
195	LARGE	Glycosyltransferase-like protein LARGE1
196	LRPPRC	Leucine-rich PPR motif-containing protein, mitochondrial
197	LRRN1	Leucine-rich repeat neuronal protein 1
198	MAN2B1	Lysosomal alpha-mannosidase
199	MANBA	Beta-mannosidase
200	MAOA	Amine oxidase [flavin-containing] A
201	MAP2K2	Dual specificity mitogen-activated protein kinase kinase 2
202	MAT1A	S-adenosylmethionine synthase isoform type-1
203	MBD5	Methyl-CpG-binding domain protein 5
204	MBTPS2	Membrane-bound transcription factor site-2 protease
205	MCCC1	Methylcrotonoyl-CoA carboxylase subunit alpha, mitochondrial
206	MCCC2	Methylcrotonoyl-CoA carboxylase beta chain, mitochondrial
207	MCOLN1	Mucolipin-1
208	MCPH1	Microcephalin
209	MECP2	Methyl-CpG-binding protein 2

210	MED12	Mediator of RNA polymerase II transcription subunit 12
211	MGAT2	Alpha-1,6-mannosyl-glycoprotein 2-beta-N-acetylglucosaminyltransferase
212	MID1	E3 ubiquitin-protein ligase Midline-1
213	MKKS	McKusick-Kaufman/Bardet-Biedl syndromes putative chaperonin
214	MLYCD	Malonyl-CoA decarboxylase, mitochondrial
215	MOCS1	Molybdenum cofactor biosynthesis protein 1
216	MOCS2	Molybdopterin synthase catalytic subunit
217	MOGS	Mannosyl-oligosaccharide glucosidase
218	MPDU1	Mannose-P-dolichol utilization defect 1 protein
219	MTHFR	Methylenetetrahydrofolate reductase
220	MTR	Methionine synthase
221	MTRR	Methionine synthase reductase
222	MYO5A	Unconventional myosin-Va
223	NAGLU	Alpha-N-acetylglucosaminidase
224	NBN	Nibrin
225	NDP	Norrin
226	NDUFA1	NADH dehydrogenase [ubiquinone] 1 alpha subcomplex subunit 1
227	NDUFS1	NADH-ubiquinone oxidoreductase 75 kDa subunit, mitochondrial
228	NDUFS2	NADH dehydrogenase [ubiquinone] iron-sulfur protein 2, mitochondrial
229	NDUFS4	NADH dehydrogenase [ubiquinone] iron-sulfur protein 4, mitochondrial
230	NDUFS7	NADH dehydrogenase [ubiquinone] iron-sulfur protein 7, mitochondrial
231	NDUFS8	NADH dehydrogenase [ubiquinone] iron-sulfur protein 8, mitochondrial
232	NDUFV1	NADH dehydrogenase [ubiquinone] flavoprotein 1, mitochondrial
233	NEU1	Oxytocin-neurophysin 1
234	NEU1	Sialidase-1
235	NF1	Neurofibromin
236	NHS	Nance-Horan syndrome protein
237	NIPBL	Nipped-B-like protein
238	NKX2-1	Homeobox protein Nkx-2.1
239	NLGN4X	Neuroigin-4, X-linked
240	NLRP3	NACHT, LRR and PYD domains-containing protein 3
241	NPC1	Niemann-Pick C1 protein
242	NPC2	Epididymal secretory protein E1
243	NSD1	Histone-lysine N-methyltransferase, H3 lysine-36 and H4 lysine-20 specific
244	NSDHL	Sterol-4-alpha-carboxylate 3-dehydrogenase, decarboxylating
245	NTRK1	High affinity nerve growth factor receptor
246	NUFIP1	Nuclear fragile X mental retardation-interacting protein 1
247	OAT	Ornithine aminotransferase, mitochondrial
248	OCRL	Inositol polyphosphate 5-phosphatase OCRL-1
249	OFD1	Oral-facial-digital syndrome 1 protein
250	OPHN1	Oligophrenin-1
251	OTC	Ornithine carbamoyltransferase, mitochondrial

252	PAFAH1B1	Platelet-activating factor acetylhydrolase IB subunit alpha
253	PAH	Phenylalanine-4-hydroxylase
254	PAK3	Serine/threonine-protein kinase PAK 3
255	PANK2	Pantothenate kinase 2, mitochondrial
256	PAX8	Paired box protein Pax-8
257	PC	Pyruvate carboxylase, mitochondrial
258	PCCA	Propionyl-CoA carboxylase alpha chain, mitochondrial
259	PCCB	Propionyl-CoA carboxylase beta chain, mitochondrial
260	PCDH19	Protocadherin-19
261	PDHA1	Pyruvate dehydrogenase E1 component subunit alpha, somatic form, mitochondrial
262	PEPD	Xaa-Pro dipeptidase
263	PEX1	Peroxisome biogenesis factor 1
264	PEX10	Peroxisome biogenesis factor 10
265	PEX12	Peroxisome assembly protein 12
266	PEX13	Peroxisomal membrane protein PEX13
267	PEX16	Peroxisomal membrane protein PEX16
268	PEX19	Peroxisomal biogenesis factor 19
269	PEX2	Peroxisome biogenesis factor 2
270	PEX3	Peroxisomal biogenesis factor 3
271	PEX5	Peroxisomal targeting signal 1 receptor
272	PEX6	Peroxisome assembly factor 2
273	PEX7	Peroxisomal targeting signal 2 receptor
274	PGK1	Phosphoglycerate kinase 1
275	PHF6	PHD finger protein 6
276	PHF8	Histone lysine demethylase PHF8
277	PLP1	Myelin proteolipid protein
278	PMM2	Phosphomannomutase 2
279	PNP	Purine nucleoside phosphorylase
280	POMGNT1	Protein O-linked-mannose beta-1,2-N-acetylglucosaminyltransferase 1
281	POMT1	Protein O-mannosyl-transferase 1
282	POMT2	Protein O-mannosyl-transferase 2
283	PORCN	Protein-cysteine N-palmitoyltransferase porcupine
284	POU1F1	Pituitary-specific positive transcription factor 1
285	PPOX	Protoporphyrinogen oxidase
286	PPT1	Palmitoyl-protein thioesterase 1
287	PQBP1	Polyglutamine-binding protein 1
288	PRPS1	Ribose-phosphate pyrophosphokinase 1
289	PRR12	Proline-rich protein 12
290	PRSS12	Neurotrypsin
291	PSAP	Prosaposin
292	PTCH1	Protein patched homolog 1
293	PTCH2	Protein patched homolog 2

294	PTCHD1	Patched domain-containing protein 1
295	PTEN	Phosphatidylinositol 3,4,5-trisphosphate 3-phosphatase and dual-specificity protein phosphatase PTEN
296	PTPN11	Tyrosine-protein phosphatase non-receptor type 11
297	PTPN23	Tyrosine-protein phosphatase non-receptor type 23
298	PTS	6-pyruvoyl tetrahydrobiopterin synthase
299	PVRL1	Poliovirus receptor-related protein 1
300	PYCR1	Pyrroline-5-carboxylate reductase 1, mitochondrial
301	QDPR	Dihydropteridine reductase
302	RAB39B	Ras-related protein Rab-39B
303	RAI1	Retinoic acid-induced protein 1
304	RBM10	RNA-binding protein 10
305	RELN	Reelin
306	RPS6KA3	Ribosomal protein S6 kinase alpha-3
307	SAR1B	GTP-binding protein SAR1b
308	SATB2	DNA-binding protein SATB2
309	SCN8a	Sodium channel protein type 8 subunit alpha
310	SCO2	Protein SCO2 homolog, mitochondrial
311	SDHA	Succinate dehydrogenase [ubiquinone] flavoprotein subunit, mitochondrial
312	SETD2	Histone-lysine N-methyltransferase SETD2
313	SETD5	SET domain-containing protein 5
314	SETMAR	Histone-lysine N-methyltransferase SETMAR
315	SGSH	N-sulphoglucosamine sulphohydrolase
316	SHH	Sonic hedgehog protein
317	SIX3	Homeobox protein SIX3
318	SLC12A1	Solute carrier family 12 member 1
319	SLC12A6	Solute carrier family 12 member 6
320	SLC16A2	Monocarboxylate transporter 8
321	SLC17A5	Sialin
322	SLC25A15	Mitochondrial ornithine transporter 1
323	SLC35C1	GDP-fucose transporter 1
324	SLC4A10	Sodium-driven chloride bicarbonate exchanger
325	SLC4A4	Electrogenic sodium bicarbonate cotransporter 1
326	SLC5A5	Sodium/iodide cotransporter
327	SLC6A8	Sodium- and chloride-dependent creatine transporter 1
328	SLC7A7	Y+L amino acid transporter 1
329	SLC9A6	Sodium/hydrogen exchanger 6
330	SMARCA2	Probable global transcription activator SNF2L2
331	SMARCA4	Transcription activator BRG1
332	SMARCB1	SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily B member 1
333	SMARCE1	SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily E member 1
334	SMC1A	Structural maintenance of chromosomes protein 1A
335	SMC3	Structural maintenance of chromosomes protein 3

336	SMPD1	Sphingomyelin phosphodiesterase
337	SMS	Spermine synthase
338	SMS	Somatostatin
339	SNRPN	Small nuclear ribonucleoprotein-associated protein N
340	SOX10	Transcription factor SOX-10
341	SOX3	Transcription factor SOX-3
342	SRCAP	Helicase SRCAP
343	SRGAP3	SLIT-ROBO Rho GTPase-activating protein 3
344	STS	Steryl-sulfatase
345	SUMF1	Sulfatase-modifying factor 1
346	SUOX	Sulfite oxidase, mitochondrial
347	SURF1	Surfeit locus protein 1
348	SYN1	Synapsin-1
349	SYNGAP1	Ras/Rap GTPase-activating protein SynGAP
350	SYP	Synaptophysin
351	TAF1	Transcription initiation factor TFIID subunit 1
352	TAT	Tyrosine aminotransferase
353	TBCE	Tubulin-specific chaperone E
354	TCF4	Transcription factor 4
355	TCN2	Transcobalamin-2
356	TCOF1	Treacle protein
357	TDGF1	Teratocarcinoma-derived growth factor 1
358	TG	Thyroglobulin
359	TGIF1	Homeobox protein TGIF1
360	THRB	Thyroid hormone receptor beta
361	THRB	Prothrombin
362	TIMM8A	Mitochondrial import inner membrane translocase subunit Tim8 A
363	TPI1	Triosephosphate isomerase
364	TPO	Thrombopoietin
365	TPO	Thyroid peroxidase
366	TPP1	Tripeptidyl-peptidase 1
367	TRNT1	CCA tRNA nucleotidyltransferase 1, mitochondrial
368	TSC1	Hamartin
369	TSC2	Tuberin
370	TSHB	Thyrotropin subunit beta
371	TSHR	Thyrotropin receptor
372	TSPAN7	Tetraspanin-7
373	TTF1	Transcription termination factor 1
374	TTF2	Transcription termination factor 2
375	UBE2A	Ubiquitin-conjugating enzyme E2 A
376	UBE3A	Ubiquitin-protein ligase E3A
377	UPF3B	Regulator of nonsense transcripts 3B

378	VLDLR	Very low-density lipoprotein receptor
379	VPS53	Vacuolar protein sorting-associated protein 53 homolog
380	XPA	DNA repair protein complementing XP-A cells
381	ZBTB24	Zinc finger and BTB domain-containing protein 24
382	ZDHHC9	Palmitoyltransferase ZDHHC9
383	ZEB2	Zinc finger E-box-binding homeobox 2
384	ZIC2	Zinc finger protein ZIC 2
385	ZNF711	Zinc finger protein 711