JANT PHARMACAL CORPORATION

Hereditary Metabolic Panel - Genes and Diseases

Downloadable Panel Reference Guide

This sheet summarizes the genes covered in the Hereditary Metabolic Panel and the associated disease terms represented across the panel.

Total genes listed	Diseases covered
148	326

Genes Covered

ABCA1	ABCC8	ACADM	ACADVL
AGXT	ALAD	ALAS2	ALDOB
ALG1	ALG11	ALG12	ALG2
ALG3	ALG6	ALG8	ALG9
ANGPTL3	APOA1	APOA5	APOB
APOC2	APOE	APP	APPL1
ARG1	ARSB	ASL	ASS1
ATP7B	B4GALT1	BCKDHA	BCKDHB
BLK	BMP6	BTD	CACNA1S
CCDC115	COG1	COG4	COG5
COG6	COG7	COG8	CPOX
CPS1	CPT1A	CPT2	DBT
DDOST	DHCR7	DOLK	DPAGT1
DPM1	DPM3	DPYD	ETFA
ETFB	ETFDH	FAH	FBP1
FECH	G6PD	GAA	GALC
GALNS	GALT	GCDH	GCK
GLA	GLB1	GM2A	GPIHBP1
GRHPR	GUSB	HADHA	HADHB
HAMP	HEXA	HFE	HGSNAT
HJV	HMBS	HNF1A	HNF1B
HNF4A	HOGA1	HYAL1	IDS

IDUA	INS	IVD	KCNJ11
KLF11	LCAT	LCT	LDLR
LDLRAP1	LEP	LEPR	LIPC
LMF1	LPL	MC4R	MGAT2
MMUT	MOGS	MPDU1	MPI
MTHFR	MTTP	MVK	NAGLU
NAGS	NEUROD1	NGLY1	NPC1
NPC2	NTRK2	OTC	PAH
PAX4	PCCA	PCCB	PCSK1
PCSK9	PDX1	PGM1	PMM2
POMC	PPOX	RFT1	RYR1
SGSH	SH2B1	SIM1	SLC25A13
SLC25A15	SLC25A20	SLC35A1	SLC35A2
SLC35C1	SLC39A8	SLC40A1	SMPD1
TFR2	UGT1A1	UROD	UROS

Diseases Covered

#	Diseases
1	Tangier disease
2	hypoalphalipoproteinemia, primary, 1
3	type 2 diabetes mellitus
4	hypoglycemia, leucine-induced
5	hyperinsulinemic hypoglycemia, familial, 1
6	diabetes mellitus, transient neonatal, 1
7	diabetes mellitus, transient neonatal, 2
8	autosomal dominant hyperinsulinism due to SUR1 deficiency
9	diazoxide-resistant focal hyperinsulinism due to SUR1 deficiency
10	maturity-onset diabetes of the young
11	DEND syndrome
12	autosomal recessive hyperinsulinism due to SUR1 deficiency
13	diabetes mellitus, permanent neonatal 3
14	permanent neonatal diabetes mellitus
15	medium chain acyl-CoA dehydrogenase deficiency
16	very long chain acyl-CoA dehydrogenase deficiency
17	primary hyperoxaluria type 1
18	porphyria due to ALA dehydratase deficiency
19	X-linked erythropoietic protoporphyria
20	X-linked sideroblastic anemia 1

#	Disease Term
21	hereditary fructose intolerance
22	ALG1-congenital disorder of glycosylation
23	ALG11-congenital disorder of glycosylation
24	ALG12-congenital disorder of glycosylation
25	ALG2-congenital disorder of glycosylation
26	congenital myasthenic syndrome 14
27	ALG3-congenital disorder of glycosylation
28	ALG6-congenital disorder of glycosylation 1C
29	ALG8-congenital disorder of glycosylation
30	polycystic liver disease 3 with or without kidney cysts
31	autosomal dominant polycystic kidney disease
32	Gillessen-Kaesbach-Nishimura syndrome
33	ALG9-congenital disorder of glycosylation
34	familial hypobetalipoproteinemia 2
35	familial visceral amyloidosis
36	AApoAI amyloidosis
37	hypoalphalipoproteinemia, primary, 2
38	hypoalphalipoproteinemia, primary, 2, intermediate
39	hyperlipoproteinemia type V
40	hypertriglyceridemia 1
41	hypercholesterolemia, autosomal dominant, type B
42	familial hypobetalipoproteinemia 1
43	homozygous familial hypercholesterolemia
44	familial apolipoprotein C-II deficiency
45	Alzheimer disease 2
46	sea-blue histiocyte syndrome
47	age related macular degeneration 1
48	Alzheimer disease 4
49	Alzheimer disease 3
50	lipoprotein glomerulopathy
51	hyperlipoproteinemia type 3
52	Alzheimer disease type 1
53	cerebral amyloid angiopathy, APP-related
54	ABeta amyloidosis, dutch type
55	early-onset autosomal dominant Alzheimer disease
56	ABetaL34V amyloidosis
57	ABeta amyloidosis, Iowa type
58	ABeta amyloidosis, Italian type
59	ABetaA21G amyloidosis
60	ABeta amyloidosis, Arctic type
61	maturity-onset diabetes of the young type 14
62	hyperargininemia

#	Disease Term
63	mucopolysaccharidosis type 6
64	mucopolysaccharidosis type 6, rapidly progressing
65	mucopolysaccharidosis type 6, slowly progressing
66	argininosuccinic aciduria
67	citrullinemia type I
68	acute neonatal citrullinemia type I
69	adult-onset citrullinemia type I
70	Wilson disease
71	B4GALT1-congenital disorder of glycosylation
72	combined low LDL and fibrinogen
73	classic maple syrup urine disease
74	intermediate maple syrup urine disease
75	intermittent maple syrup urine disease
76	thiamine-responsive maple syrup urine disease
77	maple syrup urine disease type 1A
78	maple syrup urine disease type 1B
79	systemic lupus erythematosus
80	maturity-onset diabetes of the young type 11
81	iron overload, susceptibility to
82	biotinidase deficiency
83	hypokalemic periodic paralysis
84	thyrotoxic periodic paralysis, susceptibility to, 1
85	malignant hyperthermia, susceptibility to, 5
86	malignant hyperthermia of anesthesia
87	thyrotoxic periodic paralysis
88	hypokalemic periodic paralysis, type 1
89	congenital myopathy 18
90	CCDC115-CDG
91	COG1-congenital disorder of glycosylation
92	COG4-congenital disorder of glycosylation
93	microcephalic osteodysplastic dysplasia, Saul-Wilson type
94	COG5-congenital disorder of glycosylation
95	COG6-congenital disorder of glycosylation
96	hypohidrosis-enamel hypoplasia-palmoplantar keratoderma-intellectual disability syndrome
97	COG7-congenital disorder of glycosylation
98	COG8-congenital disorder of glycosylation
99	hereditary coproporphyria
100	harderoporphyria
101	carbamoyl phosphate synthetase I deficiency disease
102	pulmonary hypertension, neonatal, susceptibility to
103	carnitine palmitoyl transferase 1A deficiency
104	carnitine palmitoyl transferase II deficiency, myopathic form

#	Disease Term
105	carnitine palmitoyl transferase II deficiency, severe infantile form
106	carnitine palmitoyl transferase II deficiency, neonatal form
107	encephalopathy, acute, infection-induced, susceptibility to, 4
108	acute necrotizing encephalopathy of childhood
109	maple syrup urine disease type 2
110	DDOST-congenital disorder of glycosylation
111	Smith-Lemli-Opitz syndrome
112	DK1-congenital disorder of glycosylation
113	familial isolated dilated cardiomyopathy
114	DPAGT1-congenital disorder of glycosylation
115	congenital myasthenic syndrome 13
116	congenital disorder of glycosylation type 1E
117	DPM3-congenital disorder of glycosylation
118	muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15
119	dihydropyrimidine dehydrogenase deficiency
120	1p21.3 microdeletion syndrome
121	multiple acyl-CoA dehydrogenase deficiency
122	multiple acyl-CoA dehydrogenase deficiency, severe neonatal type
123	multiple acyl-CoA dehydrogenase deficiency, mild type
124	tyrosinemia type I
125	fructose-1,6-bisphosphatase deficiency
126	protoporphyria, erythropoietic, 1
127	autosomal erythropoietic protoporphyria
128	anemia, nonspherocytic hemolytic, due to G6PD deficiency
129	malaria, susceptibility to
130	glycogen storage disease II
131	glycogen storage disease due to acid maltase deficiency, infantile onset
132	glycogen storage disease due to acid maltase deficiency, late-onset
133	Krabbe disease
134	infantile Krabbe disease
135	late-infantile/juvenile Krabbe disease
136	adult Krabbe disease
137	mucopolysaccharidosis type 4A
138	classic galactosemia
139	glutaryl-CoA dehydrogenase deficiency
140	maturity-onset diabetes of the young type 2
141	hyperinsulinism due to glucokinase deficiency
142	permanent neonatal diabetes mellitus 1
143	Fabry disease
144	GM1 gangliosidosis type 1
145	GM1 gangliosidosis type 2
146	GM1 gangliosidosis type 3

#	Disease Term
147	mucopolysaccharidosis type 4B
148	Tay-Sachs disease AB variant
149	hyperlipoproteinemia, type 1D
150	primary hyperoxaluria type 2
151	mucopolysaccharidosis type 7
152	mitochondrial trifunctional protein deficiency
153	long chain 3-hydroxyacyl-CoA dehydrogenase deficiency
154	acute fatty liver of pregnancy
155	mitochondrial trifunctional protein deficiency 1
156	mitochondrial trifunctional protein deficiency 2
157	hemochromatosis type 2B
158	hemochromatosis type 2
159	digenic hemochromatosis
160	Tay-Sachs disease
161	Tay-Sachs disease, b variant, infantile form
162	Tay-Sachs disease, b variant, juvenile form
163	Tay-Sachs disease, B variant, adult form
164	sporadic porphyria cutanea tarda
165	familial porphyria cutanea tarda
166	cystic fibrosis
167	hemochromatosis type 1
168	mucopolysaccharidosis type 3C
169	retinitis pigmentosa 73
170	retinitis pigmentosa
171	hemochromatosis type 2A
172	acute intermittent porphyria
173	encephalopathy, porphyria-related
174	leukoencephalopathy, porphyria-related
175	type 1 diabetes mellitus
176	hepatic adenomas, familial
177	nonpapillary renal cell carcinoma
178	maturity-onset diabetes of the young type 3
179	type 1 diabetes mellitus 20
180	chromophobe renal cell carcinoma
181	hyperinsulinism due to HNF1A deficiency
182	clear cell papillary renal cell carcinoma
183	renal cysts and diabetes syndrome
184	Mayer-Rokitansky-Küster-Hauser syndrome type 2
185	chromosome 17q12 deletion syndrome
186	medullary sponge kidney
187	renal dysplasia, unilateral
188	renal dysplasia, bilateral

#	Disease Term
189	unilateral multicystic dysplastic kidney
190	bilateral multicystic dysplastic kidney
191	prostate cancer, hereditary
192	maturity-onset diabetes of the young type 1
193	Fanconi renotubular syndrome 4 with maturity-onset diabetes of the young
194	hyperinsulinism due to HNF4A deficiency
195	atypical Fanconi syndrome-neonatal hyperinsulinism syndrome
196	primary hyperoxaluria type 3
197	mucopolysaccharidosis type 9
198	mucopolysaccharidosis type 2
199	mucopolysaccharidosis type 2, severe form
200	mucopolysaccharidosis type 2, attenuated form
201	Hurler syndrome
202	Hurler-Scheie syndrome
203	Scheie syndrome
204	type 1 diabetes mellitus 2
205	maturity-onset diabetes of the young type 10
206	hyperproinsulinemia
207	diabetes mellitus, permanent neonatal 4
208	isovaleric acidemia
209	hyperinsulinemic hypoglycemia, familial, 2
210	diabetes mellitus, transient neonatal, 3
211	maturity-onset diabetes of the young type 13
212	autosomal dominant hyperinsulinism due to Kir6.2 deficiency
213	diazoxide-resistant focal hyperinsulinism due to Kir6.2 deficiency
214	autosomal recessive hyperinsulinism due to Kir6.2 deficiency
215	intermediate DEND syndrome
216	diabetes mellitus, permanent neonatal 2
217	maturity-onset diabetes of the young type 7
218	fish eye disease
219	Norum disease
220	congenital lactase deficiency
221	hypercholesterolemia, familial, 1
222	hypercholesterolemia, familial, 4
223	obesity due to congenital leptin deficiency
224	obesity due to leptin receptor gene deficiency
225	hyperlipidemia due to hepatic triglyceride lipase deficiency
226	lipase deficiency, combined
227	hyperlipidemia, familial combined, LPL related
228	familial lipoprotein lipase deficiency
229	obesity due to melanocortin 4 receptor deficiency
230	MGAT2-congenital disorder of glycosylation

#	Disease Term
231	methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
232	vitamin B12-unresponsive methylmalonic acidemia type mut0
233	vitamin B12-unresponsive methylmalonic acidemia type mut-
234	MOGS-congenital disorder of glycosylation
235	MPDU1-congenital disorder of glycosylation
236	MPI-congenital disorder of glycosylation
237	schizophrenia
238	thrombophilia due to thrombin defect
239	homocystinuria due to methylene tetrahydrofolate reductase deficiency
240	neural tube defects, folate-sensitive
241	isolated anencephaly
242	isolated exencephaly
243	abetalipoproteinemia
244	porokeratosis 3, disseminated superficial actinic type
245	hyperimmunoglobulinemia D with periodic fever
246	mevalonic aciduria
247	porokeratosis of Mibelli
248	disseminated superficial actinic porokeratosis
249	mucopolysaccharidosis type 3B
250	Charcot-Marie-Tooth disease axonal type 2V
251	hyperammonemia due to N-acetylglutamate synthase deficiency
252	maturity-onset diabetes of the young type 6
253	congenital disorder of deglycosylation 1
254	Niemann-Pick disease, type C1
255	Niemann-Pick disease type C, severe perinatal form
256	Niemann-Pick disease type C, severe early infantile neurologic onset
257	Niemann-Pick disease type C, late infantile neurologic onset
258	Niemann-Pick disease type C, juvenile neurologic onset
259	Niemann-Pick disease type C, adult neurologic onset
260	Niemann-Pick disease, type C2
261	obesity, hyperphagia, and developmental delay
262	pilomyxoid astrocytoma
263	infantile spasms
264	undetermined early-onset epileptic encephalopathy
265	developmental and epileptic encephalopathy, 58
266	early-onset obesity-hyperphagia-severe developmental delay syndrome
267	ornithine carbamoyltransferase deficiency
268	phenylketonuria
269	maternal phenylketonuria
270	tetrahydrobiopterin-responsive hyperphenylalaninemia/phenylketonuria
271	mild phenylketonuria
272	classic phenylketonuria

#	Disease Term
273	mild hyperphenylalaninemia
274	maturity-onset diabetes of the young type 9
275	diabetes mellitus, ketosis-prone
276	propionic acidemia
277	obesity due to prohormone convertase I deficiency
278	hypercholesterolemia, autosomal dominant, 3
279	pancreatic agenesis
280	maturity-onset diabetes of the young type 4
281	pancreatic agenesis 1
282	PGM1-congenital disorder of glycosylation
283	PMM2-congenital disorder of glycosylation
284	obesity due to pro-opiomelanocortin deficiency
285	inherited obesity
286	variegate porphyria
287	variegate porphyria, childhood-onset
288	RFT1-congenital disorder of glycosylation
289	central core myopathy
290	malignant hyperthermia, susceptibility to, 1
291	autosomal dominant centronuclear myopathy
292	lethal multiple pterygium syndrome
293	congenital multicore myopathy with external ophthalmoplegia
294	autosomal recessive centronuclear myopathy
295	moderate multiminicore disease with hand involvement
296	benign Samaritan congenital myopathy
297	congenital myopathy with myasthenic-like onset
298	exercise-induced malignant hyperthermia
299	King-Denborough syndrome
300	mucopolysaccharidosis type 3A
301	proximal 16p11.2 microdeletion syndrome
302	distal 16p11.2 microdeletion syndrome
303	severe early-onset obesity-insulin resistance syndrome due to SH2B1 deficiency
304	6q16 deletion syndrome
305	obesity due to SIM1 deficiency
306	SIM1-related Prader-Willi-like syndrome
307	citrullinemia, type II, adult-onset
308	neonatal intrahepatic cholestasis due to citrin deficiency
309	citrullinemia type II
310	ornithine translocase deficiency
311	carnitine-acylcarnitine translocase deficiency
312	SLC35A1-congenital disorder of glycosylation
313	SLC35A2-congenital disorder of glycosylation
314	isolated focal cortical dysplasia type Ia

#	Disease Term
315	leukocyte adhesion deficiency type II
316	SLC39A8-CDG
317	hemochromatosis type 4
318	Niemann-Pick disease type A
319	Niemann-Pick disease type B
320	hemochromatosis type 3
321	Gilbert syndrome
322	transient familial neonatal hyperbilirubinemia
323	Crigler-Najjar syndrome type 2
324	Crigler-Najjar syndrome type 1
325	hepatoerythropoietic porphyria
326	cutaneous porphyria