JANT PHARMACAL CORPORATION

Cardio-Pulmonary Testing Panel - Genes and Diseases

Downloadable Panel Reference Guide

This sheet summarizes the genes covered in the Cardio-Pulmonary Testing Panel and the associated disease terms represented across the panel.

Total genes listed	Diseases covered
136	471

Genes Covered

ABCC9	ACTA2	ACTC1	ACTN2
ACVRL1	AKAP9	ALPK3	ANK2
ANKRD1	B3GAT3	BAG3	BGN
BMPR2	BRAF	CACNA1C	CACNA2D1
CACNB2	CALM1	CALM2	CALM3
CASQ2	CAV1	CAV3	CFTR
CHST14	COL12A1	COL1A2	COL2A1
COL3A1	COL5A1	CRYAB	CSRP3
CTNNA3	DES	DMD	DOLK
DSC2	DSE	DSG2	DSP
DTNA	EFEMP2	EIF2AK4	ELN
EMD	ENG	EYA4	F9
FBLN5	FBN1	FBN2	FHL1
FKRP	FKTN	FLNA	FLNC
GAA	GATA4	GATA5	GATA6
GATAD1	GDF2	GJA5	KCNJ8
KCNK3	KCNQ1	KRAS	LAMA4
LAMP2	LDB3	LMNA	LOX
LTBP4	MAP2K1	MAP2K2	MAT2A
MFAP5	MIB1	MYBPC3	MYH11

MYH6	MYH7	MYL2	MYL3
MYL4	MYLK	MYLK2	MYOZ2
MYPN	NEXN	NKX2-5	NOTCH1
NRAS	PKP2	PLN	PPA2
PRDM16	PRKAG2	PRKG1	PTPN11
RAF1	RANGRF	RASA1	RBM20
RIT1	RYR2	SCN10A	SCN1B
SCN2B	SCN3B	SCN4B	SCN5A
SGCD	SHOC2	SKI	SLC2A10
SMAD2	SMAD3	SMAD4	SMAD9
SNTA1	SOS1	TBX20	TCAP
TECRL	TGFB2	TGFB3	TGFBR1
TGFBR2	TMPO	TNNT2	TPM1
TRPM4	TTN	TXNRD2	VCL

Diseases Covered

#	Diseases
1	hypertrichotic osteochondrodysplasia Cantu type
2	dilated cardiomyopathy 1O
3	atrial fibrillation, familial, 12
4	Brugada syndrome
5	familial atrial fibrillation
6	familial isolated dilated cardiomyopathy
7	intellectual disability and myopathy syndrome
8	aortic aneurysm, familial thoracic 6
9	multisystemic smooth muscle dysfunction syndrome
10	Moyamoya disease 5
11	Moyamoya disease
12	familial thoracic aortic aneurysm and aortic dissection
13	hypertrophic cardiomyopathy 11
14	atrial septal defect 5
15	dilated cardiomyopathy 1R
16	left ventricular noncompaction
17	atrial septal defect, ostium secundum type
18	dilated cardiomyopathy 1AA
19	myopathy, congenital, with structured cores and z-line abnormalities
20	myopathy, distal, 6, adult-onset, autosomal dominant
21	telangiectasia, hereditary hemorrhagic, type 2

#	Disease Term
22	heritable pulmonary arterial hypertension
23	hereditary hemorrhagic telangiectasia
24	long QT syndrome 11
25	familial long QT syndrome
26	cardiomyopathy, familial hypertrophic 27
27	cardiac arrhythmia, ankyrin-B-related
28	Larsen-like syndrome, B3GAT3 type
29	myofibrillar myopathy 6
30	dilated cardiomyopathy 1HH
31	X-linked spondyloepimetaphyseal dysplasia
32	Meester-Loeys syndrome
33	X-linked severe syndromic thoracic aortic aneurysm and dissection
34	pulmonary venoocclusive disease
35	drug- or toxin-induced pulmonary arterial hypertension
36	pulmonary venoocclusive disease 1
37	pulmonary hypertension, primary, 1
38	colorectal cancer
39	cardiofaciocutaneous syndrome 1
40	Noonan syndrome with multiple lentigines
41	melanoma, cutaneous malignant, susceptibility to, 1
42	Noonan syndrome 1
43	lung cancer
44	Cushing disease due to pituitary adenoma
45	Noonan syndrome 7
46	LEOPARD syndrome 3
47	cardiofaciocutaneous syndrome
48	differentiated thyroid carcinoma
49	pilomyxoid astrocytoma
50	Langerhans cell histiocytosis
51	craniopharyngioma
52	hairy cell leukemia
53	syringocystadenoma papilliferum
54	large congenital melanocytic nevus
55	Timothy syndrome
56	Brugada syndrome 3
57	Timothy syndrome, atypical type
58	long qt syndrome 8
59	Timothy syndrome type 1
60	Timothy syndrome type 2
61	complex neurodevelopmental disorder
62	neurodevelopmental disorder with hypotonia, language delay, and skeletal defects with or without seizures
63	short QT syndrome

#	Disease Term
64	undetermined early-onset epileptic encephalopathy
65	developmental and epileptic encephalopathy 110
66	Brugada syndrome 4
67	catecholaminergic polymorphic ventricular tachycardia 4
68	long QT syndrome 14
69	catecholaminergic polymorphic ventricular tachycardia
70	long QT syndrome 15
71	long QT syndrome 16
72	catecholaminergic polymorphic ventricular tachycardia 1
73	catecholaminergic polymorphic ventricular tachycardia 2
74	partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome
75	congenital generalized lipodystrophy type 3
76	pulmonary hypertension, primary, 3
77	diffuse cutaneous systemic sclerosis
78	limited cutaneous systemic sclerosis
79	Berardinelli-Seip congenital lipodystrophy
80	creatine phosphokinase, elevated serum
81	hypertrophic cardiomyopathy 1
82	long QT syndrome 9
83	distal myopathy, Tateyama type
84	isolated asymptomatic elevation of creatine phosphokinase
85	rippling muscle disease 2
86	hereditary chronic pancreatitis
87	bronchiectasis with or without elevated sweat chloride 1
88	cystic fibrosis
89	congenital bilateral aplasia of vas deferens from CFTR mutation
90	congenital bilateral absence of vas deferens
91	idiopathic bronchiectasis
92	aquagenic palmoplantar keratoderma
93	Ehlers-Danlos syndrome, musculocontractural type
94	Ehlers-Danlos syndrome, musculocontractural type 1
95	Ullrich congenital muscular dystrophy
96	Bethlem myopathy
97	Ullrich congenital muscular dystrophy 2
98	Bethlem myopathy 2
99	osteoporosis
100	Ehlers-Danlos syndrome, arthrochalasia type
101	osteogenesis imperfecta type 1
102	osteogenesis imperfecta type 2
103	osteogenesis imperfecta type 4
104	Ehlers-Danlos syndrome, cardiac valvular type
105	osteogenesis imperfecta type 3

#	Disease Term
106	Ehlers-Danlos/osteogenesis imperfecta syndrome
107	high bone mass osteogenesis imperfecta
108	combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2
109	ehlers-danlos syndrome, arthrochalasia type, 2
110	Stickler syndrome type 1
111	multiple epiphyseal dysplasia, Beighton type
112	Legg-Calve-Perthes disease
113	platyspondylic dysplasia, Torrance type
114	Kniest dysplasia
115	spondyloepiphyseal dysplasia congenita
116	spondyloepimetaphyseal dysplasia, Strudwick type
117	spondylometaphyseal dysplasia, Schmidt type
118	spondylometaphyseal dysplasia, 'corner fracture' type
119	otospondylomegaepiphyseal dysplasia, autosomal dominant
120	achondrogenesis type II
121	spondyloperipheral dysplasia
122	mild spondyloepiphyseal dysplasia due to COL2A1 mutation with early-onset osteoarthritis
123	familial avascular necrosis of femoral head
124	spondyloepiphyseal dysplasia with metatarsal shortening
125	Stickler syndrome, type I, nonsyndromic ocular
126	spondyloepiphyseal dysplasia, Stanescu type
127	autosomal dominant rhegmatogenous retinal detachment
128	dysspondyloenchondromatosis
129	hypochondrogenesis
130	vitreoretinopathy with phalangeal epiphyseal dysplasia
131	avascular necrosis of femoral head, primary, 1
132	familial abdominal aortic aneurysm
133	autosomal dominant Ehlers-Danlos syndrome, vascular type
134	acrogeria
135	intracranial berry aneurysm
136	Ehlers-Danlos syndrome, vascular type
137	polymicrogyria with or without vascular-type Ehlers-Danlos syndrome
138	Ehlers-Danlos syndrome, classic type
139	Ehlers-Danlos syndrome, classic type, 1
140	fibromuscular dysplasia, multifocal
141	myofibrillar myopathy 2
142	cataract 16 multiple types
143	fatal infantile hypertonic myofibrillar myopathy
144	dilated cardiomyopathy 1II
145	early-onset lamellar cataract
146	early-onset nuclear cataract
147	early-onset posterior polar cataract

#	Disease Term
148	dilated cardiomyopathy 1M
149	hypertrophic cardiomyopathy 12
150	arrhythmogenic right ventricular dysplasia 13
151	familial isolated arrhythmogenic ventricular dysplasia, left dominant form
152	familial isolated arrhythmogenic ventricular dysplasia, biventricular form
153	familial isolated arrhythmogenic ventricular dysplasia, right dominant form
154	neurogenic scapuloperoneal syndrome, Kaeser type
155	myofibrillar myopathy 1
156	dilated cardiomyopathy 1I
157	Becker muscular dystrophy
158	dilated cardiomyopathy 3B
159	Duchenne muscular dystrophy
160	symptomatic form of muscular dystrophy of Duchenne and Becker in female carriers
161	non-syndromic X-linked intellectual disability
162	DK1-congenital disorder of glycosylation
163	arrhythmogenic right ventricular dysplasia 11
164	Ehlers-Danlos syndrome, musculocontractural type 2
165	arrhythmogenic right ventricular dysplasia 10
166	dilated cardiomyopathy 1BB
167	arrhythmogenic cardiomyopathy with wooly hair and keratoderma
168	arrhythmogenic right ventricular dysplasia 8
169	skin fragility-woolly hair-palmoplantar keratoderma syndrome
170	lethal acantholytic epidermolysis bullosa
171	keratosis palmoplantaris striata 2
172	severe dermatitis-multiple allergies-metabolic wasting syndrome
173	cardiomyopathy, dilated, with wooly hair, keratoderma, and tooth agenesis
174	striate palmoplantar keratoderma
175	interstitial lung disease 2
176	left ventricular noncompaction 1
177	cutis laxa, autosomal recessive, type 1B
178	lethal arteriopathy syndrome due to fibulin-4 deficiency
179	autosomal recessive cutis laxa type 1
180	pulmonary venoocclusive disease 2
181	cutis laxa, autosomal dominant 1
182	supravalvular aortic stenosis
183	Williams syndrome
184	autosomal dominant cutis laxa
185	X-linked Emery-Dreifuss muscular dystrophy
186	Emery-Dreifuss muscular dystrophy 1, X-linked
187	generalized juvenile polyposis/juvenile polyposis coli
188	telangiectasia, hereditary hemorrhagic, type 1
189	autosomal dominant nonsyndromic hearing loss 10

#	Disease Term
190	dilated cardiomyopathy 1J
191	autosomal dominant nonsyndromic hearing loss
192	thrombophilia, X-linked, due to factor 9 defect
193	hemophilia B
194	severe hemophilia B
195	moderately severe hemophilia B
196	mild hemophilia B
197	symptomatic form of hemophilia B in female carriers
198	cutis laxa, autosomal recessive, type 1A
199	macular degeneration, age-related, 3
200	cutis laxa, autosomal dominant 2
201	hereditary sensorimotor neuropathy with hyperelastic skin
202	Charcot-Marie-Tooth disease, demyelinating, IIA 1H
203	geleophysic dysplasia
204	Acromicric dysplasia
205	ectopia lentis 1, isolated, autosomal dominant
206	Marfan syndrome
207	Shprintzen-Goldberg syndrome
208	stiff skin syndrome
209	MASS syndrome
210	Weill-Marchesani syndrome 2, dominant
211	geleophysic dysplasia 2
212	progeroid and marfanoid aspect-lipodystrophy syndrome
213	isolated ectopia lentis
214	neonatal Marfan syndrome
215	Weill-Marchesani syndrome
216	congenital contractural arachnodactyly
217	macular degeneration, early-onset
218	Uruguay Faciocardiomusculoskeletal syndrome
219	X-linked scapuloperoneal muscular dystrophy
220	X-linked myopathy with postural muscle atrophy
221	myopathy, reducing body, X-linked, early-onset, severe
222	myopathy, reducing body, X-linked, childhood-onset
223	reducing body myopathy
224	muscular dystrophy-dystroglycanopathy, type A
225	muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A1
226	muscular dystrophy-dystroglycanopathy type B5
227	autosomal recessive limb-girdle muscular dystrophy type 2I
228	muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A5
229	congenital muscular dystrophy with intellectual disability
230	muscle-eye-brain disease
231	congenital muscular dystrophy without intellectual disability

#	Disease Term
232	muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4
233	autosomal recessive limb-girdle muscular dystrophy type 2M
234	dilated cardiomyopathy 1X
235	muscular dystrophy-dystroglycanopathy (congenital without intellectual disability), type B4
236	intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked
237	heterotopia, periventricular, X-linked dominant
238	terminal osseous dysplasia-pigmentary defects syndrome
239	FG syndrome 2
240	otopalatodigital syndrome type 2
241	X-linked Ehlers-Danlos syndrome
242	Melnick-Needles syndrome
243	otopalatodigital syndrome type 1
244	cardiac valvular dysplasia, X-linked
245	congenital short bowel syndrome
246	frontometaphyseal dysplasia
247	periventricular nodular heterotopia
248	frontometaphyseal dysplasia 1
249	X-linked keloid scarring-reduced joint mobility-increased optic cup-to-disc ratio syndrome
250	visceral neuropathy, familial, 1, autosomal recessive
251	myofibrillar myopathy 5
252	distal myopathy with posterior leg and anterior hand involvement
253	hypertrophic cardiomyopathy 26
254	glycogen storage disease II
255	glycogen storage disease due to acid maltase deficiency, infantile onset
256	glycogen storage disease due to acid maltase deficiency, late-onset
257	tetralogy of fallot
258	atrial septal defect 2
259	ventricular septal defect 1
260	atrioventricular septal defect 4
261	testicular anomalies with or without congenital heart disease
262	8p23.1 microdeletion syndrome
263	46,XY partial gonadal dysgenesis
264	complete atrioventricular canal-ventricle hypoplasia syndrome
265	complete atrioventricular canal-tetralogy of fallot syndrome
266	familial bicuspid aortic valve
267	congenital heart defects, multiple types, 5
268	congenital diaphragmatic hernia
269	pancreatic hypoplasia-diabetes-congenital heart disease syndrome
270	atrioventricular septal defect 5
271	atrial septal defect 9
272	conotruncal heart malformations
273	dilated cardiomyopathy 2B

#	Disease Term
274	telangiectasia, hereditary hemorrhagic, type 5
275	atrial standstill 1
276	chromosome 1q21.1 deletion syndrome
277	atrial fibrillation, familial, 11
278	pulmonary hypertension, primary, 4
279	Jervell and Lange-Nielsen syndrome
280	Beckwith-Wiedemann syndrome
281	atrial fibrillation, familial, 3
282	short QT syndrome type 2
283	Jervell and Lange-Nielsen syndrome 1
284	long QT syndrome 1
285	gastric cancer
286	urinary bladder cancer
287	Lynch syndrome
288	arteriovenous malformations of the brain
289	linear nevus sebaceous syndrome
290	Toriello-Lacassie-Droste syndrome
291	juvenile myelomonocytic leukemia
292	Noonan syndrome 3
293	encephalocraniocutaneous lipomatosis
294	autoimmune lymphoproliferative syndrome type 4
295	cardiofaciocutaneous syndrome 2
296	familial pancreatic carcinoma
297	hereditary breast carcinoma
298	acute myeloid leukemia
299	Noonan syndrome
300	dilated cardiomyopathy 1JJ
301	Danon disease
302	dilated cardiomyopathy 1C
303	myofibrillar myopathy 4
304	dilated cardiomyopathy 1A
305	familial partial lipodystrophy, Dunnigan type
306	Hutchinson-Gilford progeria syndrome
307	dilated cardiomyopathy-hypergonadotropic hypogonadism syndrome
308	mandibuloacral dysplasia with type A lipodystrophy
309	Charcot-Marie-Tooth disease type 2B1
310	familial partial lipodystrophy, Kobberling type
311	heart-hand syndrome, Slovenian type
312	congenital muscular dystrophy due to LMNA mutation
313	Emery-Dreifuss muscular dystrophy 3, autosomal recessive
314	autosomal semi-dominant severe lipodystrophic laminopathy
315	LMNA-related cardiocutaneous progeria syndrome

#	Disease Term
316	atypical Werner syndrome
317	autosomal dominant Emery-Dreifuss muscular dystrophy
318	Emery-Dreifuss muscular dystrophy 2, autosomal dominant
319	restrictive dermopathy 2
320	restrictive dermopathy
321	aortic aneurysm, familial thoracic 10
322	cutis laxa with severe pulmonary, gastrointestinal and urinary anomalies
323	melorheostosis
324	cardiofaciocutaneous syndrome 3
325	neurofibromatosis-Noonan syndrome
326	cardiofaciocutaneous syndrome 4
327	aortic aneurysm, familial thoracic 9
328	left ventricular noncompaction 7
329	hypertrophic cardiomyopathy 4
330	left ventricular noncompaction 10
331	aortic aneurysm, familial thoracic 4
332	acute myeloid leukemia with abnormal bone marrow eosinophils inv(16)(p13q22) or t(16;16)(p13;q22)
333	aortic aneurysm, familial thoracic 1
334	megacystis-microcolon-intestinal hypoperistalsis syndrome 2
335	megacystis-microcolon-intestinal hypoperistalsis syndrome 1
336	visceral myopathy 2
337	familial sick sinus syndrome
338	hypertrophic cardiomyopathy 14
339	dilated cardiomyopathy 1EE
340	atrial septal defect 3
341	sick sinus syndrome 3, susceptibility to
342	MYH7-related skeletal myopathy
343	congenital myopathy 7A, myosin storage, autosomal dominant
344	Ebstein anomaly
345	myopathy, myosin storage, autosomal recessive
346	dilated cardiomyopathy 1S
347	classic multiminicore myopathy
348	congenital fiber-type disproportion myopathy
349	hypertrophic cardiomyopathy 10
350	myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy
351	hypertrophic cardiomyopathy 8
352	atrial fibrillation, familial, 18
353	aortic aneurysm, familial thoracic 7
354	hypertrophic cardiomyopathy 16
355	dilated cardiomyopathy 1KK
356	MYPN-related myopathy
357	childhood-onset nemaline myopathy

#	Disease Term
358	cap myopathy
359	dilated cardiomyopathy 1CC
360	hypertrophic cardiomyopathy 20
361	hypoplastic left heart syndrome
362	atrial septal defect 7
363	hypothyroidism, congenital, nongoitrous, 5
364	familial isolated congenital asplenia
365	ventricular septal defect 3
366	hypoplastic left heart syndrome 2
367	deletion 5q35
368	progressive familial heart block
369	thyroid ectopia
370	athyreosis
371	Adams-Oliver syndrome
372	Adams-Oliver syndrome 5
373	aortic valve disease 1
374	nevus, epidermal
375	thyroid cancer, nonmedullary, 2
376	neurocutaneous melanocytosis
377	Noonan syndrome 6
378	arrhythmogenic right ventricular dysplasia 9
379	dilated cardiomyopathy 1P
380	hypertrophic cardiomyopathy 18
381	sudden cardiac failure, infantile
382	sudden cardiac failure, alcohol-induced
383	chromosome 1p36 deletion syndrome
384	left ventricular noncompaction 8
385	Wolff-Parkinson-White syndrome
386	lethal congenital glycogen storage disease of heart
387	hypertrophic cardiomyopathy 6
388	aortic aneurysm, familial thoracic 8
389	metachondromatosis
390	LEOPARD syndrome 1
391	Noonan syndrome 5
392	LEOPARD syndrome 2
393	dilated cardiomyopathy 1NN
394	basal cell carcinoma, susceptibility to, 1
395	capillary malformation-arteriovenous malformation syndrome
396	capillary malformation-arteriovenous malformation 1
397	dilated cardiomyopathy 1DD
398	Noonan syndrome 8
399	ventricular arrhythmias due to cardiac ryanodine receptor calcium release deficiency syndrome

#	Disease Term
400	primary erythermalgia
401	paroxysmal extreme pain disorder
402	channelopathy-associated congenital insensitivity to pain, autosomal recessive
403	episodic pain syndrome, familial, 2
404	generalized epilepsy with febrile seizures plus, type 1
405	Brugada syndrome 5
406	atrial fibrillation, familial, 13
407	generalized epilepsy with febrile seizures plus
408	developmental and epileptic encephalopathy, 52
409	developmental and epileptic encephalopathy
410	atrial fibrillation, familial, 14
411	Brugada syndrome 7
412	long QT syndrome 10
413	progressive familial heart block, type 1A
414	Brugada syndrome 1
415	dilated cardiomyopathy 1E
416	ventricular fibrillation, paroxysmal familial, type 1
417	long QT syndrome 3
418	atrial fibrillation, familial, 10
419	atrial standstill
420	sick sinus syndrome 1
421	paroxysmal familial ventricular fibrillation
422	autosomal recessive limb-girdle muscular dystrophy type 2F
423	dilated cardiomyopathy 1L
424	Noonan syndrome-like disorder with loose anagen hair
425	Noonan syndrome-like disorder with loose anagen hair 1
426	arterial tortuosity syndrome
427	Loeys-Dietz syndrome
428	Loeys-Dietz syndrome 6
429	congenital heart defects, multiple types, 8, with or without heterotaxy
430	aneurysm-osteoarthritis syndrome
431	Myhre syndrome
432	juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome
433	juvenile polyposis syndrome
434	pulmonary hypertension, primary, 2
435	long QT syndrome 12
436	fibromatosis, gingival, 1
437	Noonan syndrome 4
438	hereditary gingival fibromatosis
439	atrial septal defect 4
440	autosomal recessive limb-girdle muscular dystrophy type 2G
441	hypertrophic cardiomyopathy 25

#	Disease Term
442	catecholaminergic polymorphic ventricular tachycardia 3
443	Loeys-Dietz syndrome 4
444	arrhythmogenic right ventricular dysplasia 1
445	Rienhoff syndrome
446	multiple self-healing squamous epithelioma
447	Loeys-Dietz syndrome 1
448	esophageal squamous cell carcinoma
449	esophageal cancer
450	Loeys-Dietz syndrome 2
451	colorectal cancer, hereditary nonpolyposis, type 6
452	hypertrophic cardiomyopathy 2
453	dilated cardiomyopathy 1D
454	cardiomyopathy, familial restrictive, 3
455	hypertrophic cardiomyopathy 3
456	dilated cardiomyopathy 1Y
457	progressive familial heart block type IB
458	erythrokeratodermia variabilis
459	erythrokeratodermia variabilis et progressiva 6
460	tibial muscular dystrophy
461	myopathy, myofibrillar, 9, with early respiratory failure
462	dilated cardiomyopathy 1G
463	autosomal recessive limb-girdle muscular dystrophy type 2J
464	early-onset myopathy with fatal cardiomyopathy
465	hypertrophic cardiomyopathy 9
466	autosomal recessive centronuclear myopathy
467	childhood-onset progressive contractures-limb-girdle weakness-muscle dystrophy syndrome
468	familial glucocorticoid deficiency
469	glucocorticoid deficiency 5
470	dilated cardiomyopathy 1W
471	hypertrophic cardiomyopathy 15