JANT PHARMACAL CORPORATION

Hereditary Cancer Panel - Genes and Diseases

Downloadable Panel Reference Guide

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

Total genes listed	Diseases covered
53	224

Genes Covered

APC	ATM	AXIN2	BAP1
BARD1	BLM	BMPR1A	BRCA1
BRCA2	BRIP1	CDH1	CDKN1B
CDKN2A	CHEK2	EPCAM	FH
FLCN	GJB2	GREM1	HOXB13
MAX	MBD4	MEN1	MET
MLH1	MLH3	MSH2	MSH3
MSH6	MUTYH	NF1	NTHL1
PALB2	PMS2	POLD1	POLE
PTEN	RAD51C	RAD51D	RET
RNF43	SDHA	SDHAF2	SDHB
SDHC	SDHD	SMAD4	STK11
TMEM127	TP53	TSC1	TSC2
VHL			

Diseases Covered

#	Diseases
1	gastric cancer
2	colorectal cancer
3	hepatocellular carcinoma
4	desmoid tumor
5	Cenani-Lenz syndactyly syndrome
6	APC-related attenuated familial adenomatous polyposis

#	Disease Term
7	familial adenomatous polyposis due to 5q22.2 microdeletion
8	gastric adenocarcinoma and proximal polyposis of the stomach
9	Gardner syndrome
10	Turcot syndrome with polyposis
11	familial adenomatous polyposis 1
12	B-cell chronic lymphocytic leukemia
13	ataxia telangiectasia
14	hereditary breast carcinoma
15	ataxia - telangiectasia variant
16	familial colorectal cancer type X
17	mantle cell lymphoma
18	prostate cancer, hereditary
19	tooth agenesis
20	oligodontia-cancer predisposition syndrome
21	AXIN2-related attenuated familial adenomatous polyposis
22	malignant mesothelioma
23	uveal melanoma
24	melanoma, uveal, susceptibility to, 2
25	BAP1-related tumor predisposition syndrome
26	meningioma
27	familial melanoma
28	complex neurodevelopmental disorder
29	Kury-Isidor syndrome
30	hereditary breast ovarian cancer syndrome
31	Bloom syndrome
32	generalized juvenile polyposis/juvenile polyposis coli
33	hereditary mixed polyposis syndrome
34	polyposis syndrome, hereditary mixed, 2
35	juvenile polyposis syndrome
36	juvenile polyposis of infancy
37	breast-ovarian cancer, familial, susceptibility to, 1
38	pancreatic cancer, susceptibility to, 4
39	familial pancreatic carcinoma
40	primary peritoneal carcinoma
41	cholangiocarcinoma
42	Fanconi anemia
43	Fanconi anemia, complementation group S
44	medulloblastoma
45	Wilms tumor 1
46	chordoma
47	Fanconi anemia complementation group D1
48	breast-ovarian cancer, familial, susceptibility to, 2

#	Disease Term
49	glioma susceptibility 3
50	pancreatic cancer, susceptibility to, 2
51	kidney Wilms tumor
52	Fanconi anemia complementation group J
53	blepharocheilodontic syndrome
54	hereditary diffuse gastric adenocarcinoma
55	endometrial cancer
56	cleft lip/palate
57	blepharocheilodontic syndrome 1
58	CDH1-related diffuse gastric and lobular breast cancer syndrome
59	multiple endocrine neoplasia type 1
60	multiple endocrine neoplasia type 4
61	adrenal cortex carcinoma
62	melanoma, cutaneous malignant, susceptibility to, 2
63	melanoma and neural system tumor syndrome
64	melanoma-pancreatic cancer syndrome
65	familial atypical multiple mole melanoma syndrome
66	Li-Fraumeni syndrome
67	precursor T-cell acute lymphoblastic leukemia
68	B-lymphoblastic leukemia/lymphoma with t(9;22)(q34.1;q11.2)
69	bone osteosarcoma
70	Lynch syndrome
71	congenital diarrhea 5 with tufting enteropathy
72	Lynch syndrome 8
73	hereditary leiomyomatosis and renal cell cancer
74	fumaric aciduria
75	hereditary pheochromocytoma-paraganglioma
76	nonpapillary renal cell carcinoma
77	familial spontaneous pneumothorax
78	Potocki-Lupski syndrome
79	hereditary clear cell renal cell carcinoma
80	Birt-Hogg-Dube syndrome 1
81	keratoderma hereditarium mutilans
82	autosomal dominant keratitis-ichthyosis-hearing loss syndrome
83	palmoplantar keratoderma-deafness syndrome
84	Bart-Pumphrey syndrome
85	autosomal recessive nonsyndromic hearing loss 1A
86	X-linked mixed hearing loss with perilymphatic gusher
87	autosomal dominant nonsyndromic hearing loss 3A
88	ichthyosis, hystrix-like, with hearing loss
89	porokeratotic eccrine ostial and dermal duct nevus
90	KID syndrome

#	Disease Term
91	autosomal dominant nonsyndromic hearing loss
92	hearing loss, autosomal recessive
93	prostate cancer, hereditary, 9
94	pheochromocytoma
95	polydactyly-macrocephaly syndrome
96	melanoma, uveal, susceptibility to, 1
97	tumor predisposition syndrome 2
98	pancreatic insulin-producing neuroendocrine tumor
99	prolactin-producing pituitary gland adenoma
100	familial isolated hyperparathyroidism
101	silent pituitary adenoma
102	null pituitary adenoma
103	pituitary gigantism
104	hereditary papillary renal cell carcinoma
105	osteofibrous dysplasia
106	autosomal recessive nonsyndromic hearing loss 97
107	papillary renal cell carcinoma
108	pediatric hepatocellular carcinoma
109	arthrogryposis, distal, IIa 11
110	Muir-Torre syndrome
111	mismatch repair cancer syndrome 1
112	Lynch syndrome 2
113	mismatch repair cancer syndrome
114	colorectal cancer, hereditary nonpolyposis, type 7
115	Lynch syndrome 1
116	mismatch repair cancer syndrome 2
117	familial adenomatous polyposis 4
118	Lynch syndrome 5
119	mismatch repair cancer syndrome 3
120	familial adenomatous polyposis 2
121	neurofibromatosis, familial spinal
122	Watson syndrome
123	embryonal rhabdomyosarcoma
124	alveolar rhabdomyosarcoma
125	neurofibromatosis-Noonan syndrome
126	juvenile myelomonocytic leukemia
127	chromosome 17q11.2 deletion syndrome, 1.4Mb
128	17q11.2 microduplication syndrome
129	pleomorphic rhabdomyosarcoma
130	neurofibromatosis type 1 due to NF1 mutation or intragenic deletion
131	neurofibromatosis type 1
132	familial adenomatous polyposis 3

#	Disease Term
133	Fanconi anemia complementation group N
134	pancreatic cancer, susceptibility to, 3
135	breast-ovarian cancer, familial, susceptibility to, 5
136	Lynch syndrome 4
137	mismatch repair cancer syndrome 4
138	colorectal cancer, susceptibility to, 10
139	mandibular hypoplasia-deafness-progeroid syndrome
140	Polymerase proofreading-related adenomatous polyposis
141	IMAGe syndrome
142	colorectal cancer, susceptibility to, 12
143	facial dysmorphism-immunodeficiency-livedo-short stature syndrome
144	intrauterine growth retardation, metaphyseal dysplasia, adrenal hypoplasia congenita, genital anomalies, and immunodeficiency
145	oral cavity squamous cell carcinoma
146	laryngeal squamous cell carcinoma
147	Bannayan-Riley-Ruvalcaba syndrome
148	Cowden syndrome 1
149	Proteus syndrome
150	macrocephaly-autism syndrome
151	bilateral frontoparietal polymicrogyria
152	familial meningioma
153	glioma susceptibility 2
154	segmental outgrowth-lipomatosis-arteriovenous malformation-epidermal nevus syndrome
155	Cowden disease
156	Proteus-like syndrome
157	activated PI3K-delta syndrome
158	Lhermitte-Duclos disease
159	squamous cell carcinoma of lip
160	hypopharynx squamous cell carcinoma
161	oropharynx squamous cell carcinoma
162	paranasal sinus squamous cell carcinoma
163	Fanconi anemia complementation group O
164	breast-ovarian cancer, familial, susceptibility to, 3
165	breast-ovarian cancer, familial, susceptibility to, 4
166	Hirschsprung disease, susceptibility to, 1
167	familial medullary thyroid carcinoma
168	multiple endocrine neoplasia type 2B
169	multiple endocrine neoplasia type 2A
170	differentiated thyroid carcinoma
171	bilateral renal agenesis
172	sporadic pheochromocytoma/secreting paraganglioma
173	Hirschsprung disease
174	renal agenesis, unilateral

#	Disease Term
175	Haddad syndrome
176	sessile serrated polyposis cancer syndrome
177	hyperplastic polyposis syndrome
178	gastrointestinal stromal tumor
179	dilated cardiomyopathy 1GG
180	paragangliomas 5
181	neurodegeneration with ataxia and late-onset optic atrophy
182	mitochondrial complex II deficiency, nuclear type 1
183	familial isolated dilated cardiomyopathy
184	paragangliomas 2
185	paragangliomas 4
186	Carney-Stratakis syndrome
187	mitochondrial complex 2 deficiency, nuclear type 4
188	paragangliomas 3
189	paragangliomas 1
190	mitochondrial complex 2 deficiency, nuclear type 3
191	carcinoid syndrome
192	Myhre syndrome
193	juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome
194	hereditary hemorrhagic telangiectasia
195	familial thoracic aortic aneurysm and aortic dissection
196	melanoma, cutaneous malignant, susceptibility to, 1
197	Peutz-Jeghers syndrome
198	testicular germ cell tumor
199	clear cell papillary renal cell carcinoma
200	essential thrombocythemia
201	small cell lung carcinoma
202	adrenocortical carcinoma, hereditary
203	Cushing disease due to pituitary adenoma
204	choroid plexus papilloma
205	nasopharyngeal carcinoma, susceptibility to, 1
206	basal cell carcinoma, susceptibility to, 7
207	adult hepatocellular carcinoma
208	gliosarcoma
209	giant cell glioblastoma
210	choroid plexus carcinoma
211	myelodysplastic syndrome
212	glioma susceptibility 1
213	bone marrow failure syndrome 5
214	tuberous sclerosis
215	lung lymphangioleiomyomatosis
216	tuberous sclerosis 1

#	Disease Term
217	lymphangioleiomyomatosis
218	isolated focal cortical dysplasia type II
219	isolated focal cortical dysplasia type IIb
220	autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis
221	tuberous sclerosis 2
222	isolated focal cortical dysplasia type IIa
223	von Hippel-Lindau disease
224	Chuvash polycythemia