JANT PHARMACAL CORPORATION

Hereditary Eye Disorder Panel - Genes and Diseases

Downloadable Panel Reference Guide

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

Total genes listed	Diseases covered
51	202

Genes Covered

ATXN7	BEST1	BFSP1	BFSP2
CAV1	CDH23	CDKL5	CFH
CLRN1	CNGA1	CRYAA	CRYAB
CRYGC	CYP1B1	EYS	FOXC1
FOXE3	FTL	GALK1	GJB2
GJB6	HSF4	LTBP2	MYO15A
MYO7A	NR2E3	NRL	OTOF
PAX6	PCDH15	PDE6A	PDE6B
PITX2	POLG	PRPF31	RHO
RP1	RP2	RPE65	RPGR
SIX1	SIX6	SLC26A4	TCF4
TGFBI	TMC1	TMPRSS3	USH1C
USH1G	USH2A	WFS1	

Diseases Covered

#	Diseases
1	autosomal dominant cerebellar ataxia type II
2	nanophthalmia
3	vitelliform macular dystrophy 2
4	autosomal dominant vitreoretinochoroidopathy
5	adult-onset foveomacular vitelliform dystrophy
6	autosomal recessive bestrophinopathy

#	Disease Term
7	retinitis pigmentosa 50
8	MRCS syndrome
9	retinitis pigmentosa
10	cataract 33
11	early-onset nuclear cataract
12	pulverulent cataract
13	cataract 12 multiple types
14	early-onset lamellar cataract
15	early-onset sutural cataract
16	partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome
17	congenital generalized lipodystrophy type 3
18	pulmonary hypertension, primary, 3
19	diffuse cutaneous systemic sclerosis
20	limited cutaneous systemic sclerosis
21	heritable pulmonary arterial hypertension
22	Berardinelli-Seip congenital lipodystrophy
23	Cushing disease due to pituitary adenoma
24	Usher syndrome type 1
25	prolactin-producing pituitary gland adenoma
26	Usher syndrome type 1D
27	autosomal recessive nonsyndromic hearing loss 12
28	familial isolated pituitary adenoma
29	hearing loss, autosomal recessive
30	TSH-secreting pituitary adenoma
31	pituitary adenoma 5, multiple types
32	developmental and epileptic encephalopathy, 2
33	atypical Rett syndrome
34	infantile spasms
35	developmental and epileptic encephalopathy
36	Doyne honeycomb retinal dystrophy
37	basal laminar drusen
38	HELLP syndrome
39	hemolytic uremic syndrome, atypical, susceptibility to, 1
40	complement factor H deficiency
41	age related macular degeneration 4
42	immunoglobulin-mediated membranoproliferative glomerulonephritis
43	immunodeficiency with factor H anomaly
44	dense deposit disease
45	atypical hemolytic uremic syndrome with complement gene abnormality
46	Usher syndrome type 3A
47	retinitis pigmentosa 61
48	Usher syndrome type 3

#	Disease Term
49	retinitis pigmentosa 49
50	cataract 9 multiple types
51	cataract - microcornea syndrome
52	early-onset anterior polar cataract
53	total early-onset cataract
54	myofibrillar myopathy 2
55	cataract 16 multiple types
56	fatal infantile hypertonic myofibrillar myopathy
57	dilated cardiomyopathy 1II
58	early-onset posterior polar cataract
59	familial isolated dilated cardiomyopathy
60	cataract 2, multiple types
61	glaucoma 3A
62	glaucoma 3, primary infantile, B
63	Peters anomaly
64	anterior segment dysgenesis 6
65	juvenile open angle glaucoma
66	retinitis pigmentosa 25
67	isolated aniridia
68	Axenfeld-Rieger syndrome type 3
69	Axenfeld-Rieger syndrome
70	Rieger anomaly
71	Axenfeld anomaly
72	anterior segment dysgenesis 3
73	congenital primary aphakia
74	cataract 34 multiple types
75	familial thoracic aortic aneurysm and aortic dissection
76	aortic aneurysm, familial thoracic 11, susceptibility to
77	hereditary hyperferritinemia with congenital cataracts
78	neuroferritinopathy
79	L-ferritin deficiency
80	galactokinase deficiency
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	Clouston syndrome
93	autosomal dominant nonsyndromic hearing loss 3B
94	autosomal recessive nonsyndromic hearing loss 1B
95	cataract 5 multiple types
96	microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma
97	glaucoma 3, primary congenital, D
98	Weill-Marchesani syndrome 3
99	glaucoma secondary to spherophakia/ectopia lentis and megalocornea
100	Weill-Marchesani syndrome
101	autosomal recessive nonsyndromic hearing loss 3
102	autosomal recessive nonsyndromic hearing loss 2
103	autosomal dominant nonsyndromic hearing loss 11
104	Usher syndrome type 2
105	Usher syndrome type 1B
106	retinitis pigmentosa 37
107	enhanced S-cone syndrome
108	Goldmann-Favre syndrome
109	retinitis pigmentosa 27
110	autosomal recessive nonsyndromic hearing loss 9
111	coloboma, ocular, autosomal dominant
112	coloboma of macula
113	coloboma of optic nerve
114	foveal hypoplasia 1
115	autosomal dominant keratitis
116	isolated optic nerve hypoplasia
117	WAGR syndrome
118	aniridia-cerebellar ataxia-intellectual disability syndrome
119	foveal hypoplasia-presenile cataract syndrome
120	morning glory syndrome
121	coloboma of choroid and retina
122	coloboma of eye lens
123	coloboma of iris
124	coloboma of eyelid
125	aniridia 1
126	Usher syndrome type 1F
127	autosomal recessive nonsyndromic hearing loss 23
128	retinitis pigmentosa 43
129	congenital stationary night blindness autosomal dominant 2
130	retinitis pigmentosa 40
131	congenital stationary night blindness
132	anterior segment dysgenesis 4

#	Disease Term
133	Axenfeld-Rieger syndrome type 1
134	ring dermoid of cornea
135	familial atrial fibrillation
136	autosomal dominant progressive external ophthalmoplegia
137	mitochondrial DNA depletion syndrome 4a
138	progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 1
139	mitochondrial DNA depletion syndrome 1
140	sensory ataxic neuropathy, dysarthria, and ophthalmoparesis
141	mitochondrial DNA depletion syndrome 4b
142	spinocerebellar ataxia with epilepsy
143	autosomal recessive progressive external ophthalmoplegia
144	mitochondrial neurogastrointestinal encephalomyopathy
145	recessive mitochondrial ataxia syndrome
146	progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 1
147	retinitis pigmentosa 11
148	fundus albipunctatus
149	congenital stationary night blindness autosomal dominant 1
150	retinitis pigmentosa 4
151	retinitis punctata albescens
152	retinitis pigmentosa 1
153	retinitis pigmentosa 2
154	Leber congenital amaurosis 2
155	severe early-childhood-onset retinal dystrophy
156	retinitis pigmentosa 20
157	Leber congenital amaurosis
158	retinitis pigmentosa 87 with choroidal involvement
159	retinitis pigmentosa 3
160	macular degeneration, X-linked atrophic
161	X-linked cone-rod dystrophy 1
162	cone-rod dystrophy
163	primary ciliary dyskinesia
164	achromatopsia
165	branchio-oto-renal syndrome
166	branchiootorenal syndrome 1
167	autosomal dominant nonsyndromic hearing loss 23
168	branchiootic syndrome 3
169	branchiootic syndrome
170	microphthalmia, isolated, with coloboma
171	anophthalmia/microphthalmia-esophageal atresia syndrome
172	colobomatous optic disc-macular atrophy-chorioretinopathy syndrome
173	Pendred syndrome
174	autosomal recessive nonsyndromic hearing loss 4

#	Disease Term
175	athyreosis
176	thyroid hypoplasia
177	Fuchs' endothelial dystrophy
178	Pitt-Hopkins syndrome
179	corneal dystrophy, Fuchs endothelial, 3
180	primary sclerosing cholangitis
181	autosomal dominant non-syndromic intellectual disability
182	epithelial basement membrane dystrophy
183	granular corneal dystrophy type I
184	lattice corneal dystrophy type I
185	Thiel-Behnke corneal dystrophy
186	granular corneal dystrophy type II
187	Reis-Bucklers corneal dystrophy
188	corneal dystrophy, lattice type 3A
189	autosomal recessive nonsyndromic hearing loss 7
190	autosomal dominant nonsyndromic hearing loss 36
191	autosomal recessive nonsyndromic hearing loss 8
192	Usher syndrome type 1C
193	autosomal recessive nonsyndromic hearing loss 18A
194	Usher syndrome type 1G
195	Usher syndrome type 2A
196	retinitis pigmentosa 39
197	type 2 diabetes mellitus
198	cataract 41
199	Wolfram syndrome 1
200	autosomal dominant nonsyndromic hearing loss 6
201	Wolfram-like syndrome
202	Wolfram syndrome