JANT PHARMACAL CORPORATION

Immunodeficiency Genetics Disease Panel - Genes and Diseases

Downloadable Panel Reference Guide

This sheet summarizes the genes covered in the Immunodeficiency Genetics Disease Panel and the associated disease terms represented across the panel.

Total genes listed	Diseases covered
30	130

Genes Covered

ATM	**BTK**	**CFTR**	**CYBA**
CYBB	**G6PD**	**IFNGR1**	**IFNGR2**
ITGB2	**JAK2**	**MEFV**	**MPL**
MYD88	**NCF1**	**NFKB2**	**NRAS**
PIK3CD	**PLCG2**	**PTEN**	**PTPRC**
RAG1	**RAG2**	**RFXANK**	**SPINK5**
STAT1	**STAT3**	**STK4**	**TERT**
TNFRSF13B	**VPS13B**		

Diseases Covered

#	Diseases
1	B-cell chronic lymphocytic leukemia
2	ataxia telangiectasia
3	hereditary breast carcinoma
4	ataxia - telangiectasia variant
5	familial colorectal cancer type X
6	mantle cell lymphoma
7	prostate cancer, hereditary
8	Bruton-type agammaglobulinemia
9	isolated growth hormone deficiency type III
10	short stature due to isolated growth hormone deficiency with X-linked hypogammaglobulinemia
11	hereditary chronic pancreatitis
12	bronchiectasis with or without elevated sweat chloride 1

#	Disease Term
13	cystic fibrosis
14	congenital bilateral aplasia of vas deferens from CFTR mutation
15	congenital bilateral absence of vas deferens
16	idiopathic bronchiectasis
17	aquagenic palmoplantar keratoderma
18	granulomatous disease, chronic, autosomal recessive, cytochrome b-negative
19	chronic granulomatous disease
20	X-linked Mendelian susceptibility to mycobacterial diseases due to CYBB deficiency
21	granulomatous disease, chronic, X-linked
22	anemia, nonspherocytic hemolytic, due to G6PD deficiency
23	malaria, susceptibility to
24	Mycobacterium tuberculosis, susceptibility
25	Behcet disease
26	immunodeficiency 27A
27	Helicobacter pylori infection, susceptibility to
28	hepatitis B virus, susceptibility to
29	autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency
30	autosomal recessive Mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency
31	Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR1 deficiency
32	immunodeficiency 28
33	autosomal recessive Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR2 deficiency
34	autosomal recessive Mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR2 deficiency
35	autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR2 deficiency
36	leukocyte adhesion deficiency 1
37	essential thrombocythemia
38	primary familial polycythemia due to EPO receptor mutation
39	primary myelofibrosis
40	acquired polycythemia vera
41	Budd-Chiari syndrome
42	thrombocythemia 3
43	acute myeloid leukemia
44	familial thrombocytosis
45	breast implant-associated anaplastic large cell lymphoma
46	familial Mediterranean fever, autosomal dominant
47	autosomal recessive familial Mediterranean fever
48	sweet syndrome
49	intermittent hydrarthrosis
50	familial Mediterranean fever
51	thrombocythemia 2
52	congenital amegakaryocytic thrombocytopenia 1
53	pyogenic bacterial infections due to MyD88 deficiency
54	Waldenstrom macroglobulinemia

#	Disease Term
55	macroglobulinemia, Waldenstrom, 1
56	Williams syndrome
57	granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type 1
58	immunodeficiency, common variable, 10
59	common variable immunodeficiency
60	deficiency in anterior pituitary function - variable immunodeficiency syndrome
61	colorectal cancer
62	nevus, epidermal
63	linear nevus sebaceous syndrome
64	thyroid cancer, nonmedullary, 2
65	neurocutaneous melanocytosis
66	juvenile myelomonocytic leukemia
67	Noonan syndrome 6
68	autoimmune lymphoproliferative syndrome type 4
69	differentiated thyroid carcinoma
70	Langerhans cell histiocytosis
71	Noonan syndrome
72	large congenital melanocytic nevus
73	combined immunodeficiency with facio oculoskeletal anomalies
74	immunodeficiency 14
75	activated PI3K-delta syndrome
76	immunodeficiency 14b, autosomal recessive
77	familial cold autoinflammatory syndrome 3
78	autoinflammation-PLCG2-associated antibody deficiency-immune dysregulation
79	hereditary breast ovarian cancer syndrome
80	oral cavity squamous cell carcinoma
81	laryngeal squamous cell carcinoma
82	Bannayan-Riley-Ruvalcaba syndrome
83	Cowden syndrome 1
84	Proteus syndrome
85	macrocephaly-autism syndrome
86	bilateral frontoparietal polymicrogyria
87	familial meningioma
88	glioma susceptibility 2
89	segmental outgrowth-lipomatosis-arteriovenous malformation-epidermal nevus syndrome
90	Cowden disease
91	Proteus-like syndrome
92	Lhermitte-Duclos disease
93	juvenile polyposis of infancy
94	squamous cell carcinoma of lip
95	hypopharynx squamous cell carcinoma
96	oropharynx squamous cell carcinoma

#	Disease Term
97	paranasal sinus squamous cell carcinoma
98	T-B+ severe combined immunodeficiency due to CD45 deficiency
99	immunodeficiency 105
100	combined immunodeficiency with skin granulomas
101	severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-negative, NK cell-positive
102	Omenn syndrome
103	combined immunodeficiency due to partial RAG1 deficiency
104	MHC class II deficiency
105	Netherton syndrome
106	immunodeficiency 31B
107	autoimmune enteropathy and endocrinopathy - susceptibility to chronic infections syndrome
108	Mendelian susceptibility to mycobacterial diseases due to partial STAT1 deficiency
109	chronic lymphoproliferative disorder of NK-cells
110	hyper-IgE recurrent infection syndrome 1, autosomal dominant
111	acute promyelocytic leukemia
112	STAT3-related early-onset multisystem autoimmune disease
113	T-cell large granular lymphocyte leukemia
114	permanent neonatal diabetes mellitus
115	combined immunodeficiency due to STK4 deficiency
116	clear cell sarcoma of kidney
117	adrenal cortex carcinoma
118	dyskeratosis congenita, autosomal dominant 1
119	idiopathic aplastic anemia
120	dyskeratosis congenita, autosomal dominant 2
121	pulmonary fibrosis and/or bone marrow failure, Telomere-related, 1
122	melanoma, cutaneous malignant, susceptibility to, 9
123	dyskeratosis congenita
124	meningioma
125	Hoyeraal-Hreidarsson syndrome
126	familial melanoma
127	interstitial lung disease 2
128	immunodeficiency, common variable, 2
129	immunoglobulin A deficiency 2
130	Cohen syndrome