Rheumatology

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Single Gene

Code	Diseases	Gene
sCe5/0007	Acne inversa familial type 3	PSEN1
sCe5/0057	Atypical Mycobacterial infection	IFNGR2
sCe5/0058	Atypical Mycobacterial infection	IKBKG
sCe5/0059	Atypical Mycobacterial infection	IL12RB1
sCe5/0060	Atypical Mycobacterial infection	STAT1
sCe5/0061	Atypical Mycobacterial infection, IL12RB2 related	IL12RB2
sCe5/0062	Autoimmune lymphoproliferative syndrome type 1A	FAS
sCe5/0063	Autoimmune lymphoproliferative syndrome type 1B	FASLG
sCe5/0064	Autoimmune lymphoproliferative syndrome type 2A	CASP10
sCe5/0065	Autoimmune lymphoproliferative syndrome type 2B	CASP8
sCe5/0066	Autoimmune lymphoproliferative syndrome type 3	PRKCD
sCe5/0067	Autoimmune polyendocrinopathy syndrome type 1	AIRE
sCe5/0068	Autoinflammation, lipodystroph and dermatosis syndrome	PSMB8
sCe5/0070	Bare lymphocyte syndrome, type 2	RFXANK
sCe5/0071	Bare lymphocyte syndrome, type 2, complementation group A	CIITA
sCe5/0072	B-cell expansion with NFKB and T-cell anergy	CARD11
sCe5/0077	C1q deficiency	C1QA
sCe5/0078	C2 deficiency	C2
sCe5/0079	C3 deficiency	C3
sCe5/0080	C5 deficiency	C5
sCe5/0081	C7 deficiency	C7
sCe5/0082	Chediak-Higashi syndrome	LYST
sCe5/0083	Choanal atresia and lymphedema	PTPN14
sCe5/0089	Cold autoinflammatory syndrome type 2	NLRP12
sCe5/0091	Combined cellular and humoral immune defects with granulomas	RAG2

sCe5/0092	Combined immunodeficiency, B cell-negative, T cell-negative, NK cell positive	RAG2
sCe5/0093	Combined immunodeficiency, X-linked, moderate	IL2RG
sCe5/0116	Dermatitis, atopic type 2	FLG
sCe5/0119	Diarrhea type 2 with microvillus atrophy	MYO5B
sCe5/0120	Diarrhea type 6	GUCY2C
sCe5/0154	Emberger syndrome	GATA2
sCe5/0181	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis and hyper IgE	DSG1
sCe5/0184	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis	COX4I2
sCe5/0187	Fanconi anemia, complementation group Q	ERCC4
sCe5/0201	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type 1	NCF1
sCe5/0202	Granulomatous disease, chronic, autosomal recessive, cytochrome b-negative	CYBA
sCe5/0203	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type 2	NCF2
sCe5/0204	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type 3	NCF4
sCe5/0205	Granulomatous disease, chronic, X-linked	CYBB
sCe5/0211	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy	CD59
sCe5/0212	Hemophagocytic lymphohistiocytosis type 2	PRF1
sCe5/0213	Hemophagocytic lymphohistiocytosis type 3	UNC13D
sCe5/0214	Hemophagocytic lymphohistiocytosis type 4	STX11
sCe5/0215	Hemophagocytic lymphohistiocytosis type 5	STXBP2
sCe5/0216	Hennekam lymphangiectasia-lymphedema syndrome type 1	CCBE1
sCe5/0217	Hepatic venoocclusive disease with immunodeficiency	SP110
sCe5/0218	Herpes simplex encephalitis type 2, susceptibility to	TLR3
sCe5/0219	Histiocytosis-lymphadenopathy plus syndrome	SLC29A3
sCe5/0221	Hyper-IgE recurrent infection syndrome	STAT3
sCe5/0222	Hyper-IgE recurrent infection syndrome, autosomal recessive	DOCK8

sCe5/0241	Hypotrichosis-lymphedema-telangiectasia syndrome	SOX18
sCe5/0260	Immunodeficiency common variable type 1	ICOS
sCe5/0261	Immunodeficiency common variable type 10	NFKB2
sCe5/0262	Immunodeficiency common variable type 2	TNFRSF13B
sCe5/0263	Immunodeficiency common variable type 3	CD19
sCe5/0264	Immunodeficiency common variable type 4	TNFRSF13C
sCe5/0265	Immunodeficiency common variable type 6	CD81
sCe5/0266	Immunodeficiency common variable type 8	LRBA
sCe5/0267	Immunodeficiency due to defect in MAPBP-interacting protein	LAMTOR2
sCe5/0268	Immunodeficiency due to purine nucleoside phosphorylase deficiency	PNP
sCe5/0269	Immunodeficiency type 10	STIM1
sCe5/0270	Immunodeficiency type 11	CARD11
sCe5/0271	Immunodeficiency type 12	MALT1
sCe5/0272	Immunodeficiency type 14	PIK3CD
sCe5/0273	Immunodeficiency type 15	IKBKB
sCe5/0274	Immunodeficiency type 18	CD3E
sCe5/0275	Immunodeficiency type 19	CD3D
sCe5/0276	Immunodeficiency type 2, with hyper-IgM	AICDA
sCe5/0277	Immunodeficiency type 21	GATA2
sCe5/0278	Immunodeficiency type 22	LCK
sCe5/0279	Immunodeficiency type 24	CTPS1
sCe5/0280	Immunodeficiency type 25	CD247
sCe5/0281	Immunodeficiency type 26, with or without neurologic abnormalities	PRKDC
sCe5/0282	Immunodeficiency type 3, with hyper-IgM	CD40
sCe5/0283	Immunodeficiency type 32A, mycobacteriosis, autosomal dominant	IRF8
sCe5/0284	Immunodeficiency type 32B, monocyte and dendritic cell deficiency, autosomal recessive	IRF8
sCe5/0285	Immunodeficiency type 34	CYBB
sCe5/0286	Immunodeficiency type 35	TYK2
sCe5/0287	Immunodeficiency type 36	PIK3R1

sCe5/0288	Immunodeficiency type 38	ISG15
sCe5/0289	Immunodeficiency type 5, with hyper IgM	UNG
sCe5/0290	Immunodeficiency type 8	CORO1A
sCe5/0291	Immunodeficiency type 9	ORAI1
sCe5/0292	Immunodeficiency, isolated	IKBKG
sCe5/0293	Immunodeficiency, primary, autosomal recessive, IL21R-related	IL21R
sCe5/0294	Immunodeficiency, X-linked with hyper-IgM	CD40LG
sCe5/0295	Immunodeficiency-centromeric instability-facial anomalies syndrome type 1	DNMT3B
sCe5/0296	Immunodeficiency-centromeric instability-facial anomalies syndrome type 2	ZBTB24
sCe5/0297	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	FOXP3
sCe5/0298	Immunological disorder, PECAM1 related	PECAM1
sCe5/0299	Immunological disorder, PICALM related	PICALM
sCe5/0301	Inflammatory bowel disease type 13	ABCB1
sCe5/0302	Inflammatory skin and bowel disease, neonatal, type 1	ADAM17
sCe5/0303	Interleukin 12A deficiency	IL12A
sCe5/0304	Interleukin 2 receptor deficiency	IL2RA
sCe5/0305	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital	ITGA3
sCe5/0306	Invasive pneumococcal disease, recurrent isolated type 1	IRAK4
sCe5/0307	Invasive pneumococcal disease, recurrent isolated type 2	IKBKG
sCe5/0308	IRAK4 deficiency	IRAK4
sCe5/0319	Leukocyte adhesion deficiency type 3	FERMT3
sCe5/0320	LIG4 syndrome	LIG4
sCe5/0326	Lymphedema, hereditary, type 1A	FLT4
sCe5/0327	Lymphedema, hereditary, type IC	GJC2
sCe5/0328	Lymphoproliferative syndrome type 2	CD27
sCe5/0329	Majeed syndrome	LPIN2
sCe5/0330	Major histocompatibility comples 1 deficiency	MR1
sCe5/0336	Meconium ileus	GUCY2C
sCe5/0347	Mycobacterial infection, atypical, familial disseminated	IFNGR1

sCe5/0355	Neutropenia, nonimmune chronic idiopathic, of adults	GFI1
sCe5/0356	Neutropenia, severe congenital type 2, autosomal dominant	GFI1
sCe5/0357	Neutropenia, severe congenital type 4, autosomal recessive	G6PC3
sCe5/0358	Neutropenia, severe congenital type 5, autosomal recessive	VPS45
sCe5/0359	Neutropenia, severe congenital type 6, autosomal recessive	JAGN1
sCe5/0360	Neutrophil immunodeficiency syndrome	RAC2
sCe5/0363	Omenn syndrome	RAG2
sCe5/0364	Omenn syndrome	DCLRE1C
sCe5/0413	Poikiloderma with neutropenia	USB1
sCe5/0424	Pulmonary fibrosis and/or bone marrow failure, telomere-related, type 1	TERT
sCe5/0425	Pulmonary fibrosis and/or bone marrow failure, telomere-related, type 4	PARN
sCe5/0427	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency	MYD88
sCe5/0428	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne	PSTPIP1
sCe5/0433	Reticular dysgenesis	AK2
sCe5/0434	Rheumatoid arthritis, susceptibility to	AFF3
sCe5/0435	Rheumatoid arthritis, TNFAIP3 related	TNFAIP3
sCe5/0440	Selective T-cell defect	ZAP70
sCe5/0441	Severe combined immunodeficiency due to ADA deficiency	ADA
sCe5/0442	Severe combined immunodeficiency due to IL2 deficiency	IL2
sCe5/0443	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	NHEJ1
sCe5/0444	Severe combined immunodeficiency, Athabascan type	DCLRE1C
sCe5/0445	Severe combined immunodeficiency, B cell-negative	RAG1
sCe5/0446	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive	PTPRC
sCe5/0447	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type	IL7R
sCe5/0463	Systemic lupus erythematosus	DNASE1
sCe5/0464	Systemic lupus erythematosus type 16	DNASE1L3
sCe5/0465	Systemic lupus erythematosus, susceptibility to	ITGAM

sCe5/0466	T-cell immunodeficiency, congenital alopecia, and nail dystrophy	FOXN1
sCe5/0485	Vitiligo-associated multiple autoimmune disease	NLRP1
sCe5/0490	WHIM syndrome	CXCR4

Panel

Code	product name	TAT-day
	Clinical Exome Whole exome Solo	
pCe5/0002	Autoimmune lymphoproliferative syndrome panel	25
pCe5/0003	B-negative SCID panel	25
pCe5/0004	B-positive SCID panel	25
pCe5/0005	Chronic granulomatous disease panel	25
pCe5/0006	Common variable immune deficiency (CVID) panel	25
pCe5/0012	Fanconi anemia panel	25
pCe5/0013	Hemophagocytic Lymphohistiocytosis panel	25
pCe5/0020	Periodic fever syndrome panel	25
pCe5/0021	Primary antibody deficiency panel	25
pCe5/0022	Susceptibility to atypical mycobacterium disease panel	25