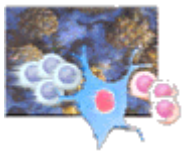
PRIMARY IMMUNODEFICIENCIES



Affected cells	*Name*	*Gene's anomaly/locus*	*Clinical symptoms*
T cells and B cells	X-Linked *Severe Combined Immunode-ficiency (SCID)*	IL2RG on Xq13.1, which encodes γ chain (CD132) receptor for IL-2 as well as IL-4, IL-7, IL-9, IL-12, IL-13, and IL-15	Very severe infections from the birth ("Bubble Boy Disease")
T cells and B cells	*SCID*, Reticular dysgenesis	AK2 (Mitochondrial adenylate kinase-2) on 1p35.1 that leads to disability to haemopoiesis	Very severe infections from the birth, cytopenias
T cells and B cells	*SCID*, Adenosine deaminase deficiency	ADA (Adenosine deaminase) on 20q13.12 that leads to disorder of lymphopoiesis	Very severe infections from the birth or later
T cells and B cells	*SCID*, Type I Bare Lymphocyte Syndrome	TAP1/TAP2 on 6p21.32	Severe infections of respiratory tract in babies, toddlers and older children
T cells and B cells	*SCID*, Type II Bare Lymphocyte Syndrome	Different regulatory genes on 1q21.3, 13q13.3, 16p13.13 and 19p13.11, which are required for transcription of HLA II genes	Very severe infections from the birth or later
T cells	*SCID*, T cell-negative, B cell-positive, NK cell-positive	IL7RA on 5p13.2 that results in disorder of signaling through IL-7Rα (CD127) and blockade in T lymphopoieses	Very severe infections from the birth
T cells	*SCID*, T cell-negative, B cell-positive, NK cell-negative	JAK3 on 19p13.11 that leads to blockade in T-cell and NK-cell development	Very severe infections from the birth
T cells	*Immunodeficiency 17/19*	CD3G/ CD3D on 11q23.3 that leads to disorder of T lymphopoiesis and decrease in CD3γδ	Recurrent infections in babies, toddlers and later
T cells	*Immunodeficiency 48*	ZAP70 on 2q11.2 that leads to selective disorder of CD8+T-cell lymphopoiesis	Recurrent infections in babies, toddlers and later
B cells	*SCID*, Omenn Syndrome	RAG1/RAG2 on 11p12 that leads to disorder of B lymphopoiesis and partially T lymphopoiesis	Early infections, dwarfism, reticuloendotheliosis with eosinophilia, predisposition to tumors
B cells	X-Linked *Agamma-globulinaemia (Bruton's Syndrome)*	BTK (Bruton's tyrosine kinase) on Xq21.3-q22	Recurrent pyogenic infections in male babies, underdeveloped secondary lymphoid organs
B cells	*Common Variable Immunodeficiency (CVID)*	ICOS on 2q33.2 (CVID1), TNFRSF13B on 17p11.2 (CVID2), CD19 on 16p11.2	Recurrent sinopulmonary infections at any age, increased incidence of

	(heterogenous group)	(CVID3), TNFRSF13C on 22q13.2 (CVID4), CD20 (MS4A1) on 11q12.2 (CVID5), CD81 on 11p15.5 (CVID6), CD21 (CR2) on 1q32.2 (CVID7), etc	autoimmune disorders and tumors
B cells	*Hyper-IgM Syndrome*, types 1-5	Different mutations: TNFSF5 on Xq26.3 (type 1), AICDA on 12p13.31 (type 2), CD40 on 20q13.12 (type 3), unknown (type 4) and UNG on 12q24.11 (type 5), which lead to inability of B cells to undergo antibody isotype switching	Recurrent pyogenic infections in babies, Underdeveloped secondary lymphoid organs, autoimmune cytopenias
Combined	*Wiskott-Aldrich Syndrome*	WASP (Wiskott-Aldrich Syndrome Protein) on Xp11.22-p11.23 that leads to cytoskeleton defect	Clinical triad: (1) increased susceptibility to infections in babies and toddlers, (2) thrombocytopenic purpura, (3) mild eczema as well as increased incidence of tumors
DNA defects	*DiGeorge Syndrome*	1.5- to 3.0-Mb deletion of chromosome 22q11.2 including genes responsible for physical malformations	Seizures at the birth because of hypocalcemia (parathyroid hypoplasia), thymic hypoplasia, derivatives of the pharyngeal arches, congenital heart defects, etc.
DNA reparation defect	*Ataxia- telangiectasia (Louis-Bar syndrome)*	ATM (Ataxia-telangiectasia mutated gene) on 11q22.3 that responsible for cell cycle disorder	Clinical triad: (1) progressive cerebellar ataxia, (2) oculocutaneous telangiectases, (3) chronic sinopulmonary infections as well as increased incidence of tumors
DNA reparation defect	*Nijmegen Breakage Syndrome*	NBN (nibrin gene) on 8q21.3 responsible for cell cycle disorder	Microcephaly, growth retardation, early infections, and predisposition to cancer
Many cells	*Immunodeficiencies 27A/27B*	IFNGR1 (CD119) on 6q23.3 that leads to defects of phagocytosis and development of inflammation in response to Mycobacteria, and diferrent pathogens	Disseminated BCG infection, SCID-like and CGD-like symptoms
NK cells and other cells	*Immunodeficiency 20*	CD16 (FCGR3A) gene on 1q23.3 that leads to decreased NK cell mediated cytotoxicity	Recurrent viral infections *Epstein Barr Virus (EBV), Herpes Zoster Virus (HZV)* and other herpesviruses, and *Human Papilloma*

			Viruses (HPV)
Neutrophils	X-Linked *Chronic Granulomatous Disease (CGD)*	CYBB on Xp21.1-p11.4, which encodes p22phox for phagocyte oxidase that leads to inability to generate ROS	Abscesses of the skin, lymph nodes and liver in male babies
Leukocytes	Type 1 *Leukocyte Adhesion Deficiency* (LAD1)	ITGB2 (CD18) on 21q22.3 that leads to inability to synthesize CD18 (a β unit of integrin)	Repeated bacterial and fungal infections, intestinal or perianal fistulae, delayed wound healing, and high leukocytosis
Leukocytes	Type 2 *Leukocyte Adhesion Deficiency* (LAD2)	SLC35C1 on 11p11.2 that results in the inability to synthesize sialylated Lewis^{x}	Repeated bacterial and fungal infections, intestinal or perianal fistulae, delayed wound healing, and high leukocytosis
Leukocytes	Type 3 *Leukocyte Adhesion Deficiency* (LAD3)	FERMT3 on 11q13.1 that leads to inability to the provision of "inside-out" signaling through β unit of integrins	Repeated bacterial and fungal infections, intestinal or perianal fistulae, delayed wound healing, bleeding, and high leukocytosis
Comple-ment	*C3 deficiency*	C3 on 19p13.3	Recurrent pyogenic infections, susceptibility to *Neisseria meningitidis* and other gram-negative bacteria
Comple-ment	*Hereditary angioedema* (C1 inhibitor deficiency)	C1NH on 11q12.1	Recurrent episodes of angioedema at any age