

MAIN_CATEGORY	SUB_CATEGORY	PID_NAME	GENE
Autoinflammatory disorders	Blau syndrome	Caspase recruitment domain-containing protein 15 deficiency (CARD15)	CARD15
Autoinflammatory disorders	CINCA syndrome	CINCA syndrome	CIAS1 (NLRP3)
Autoinflammatory disorders	Defects affecting the inflammasome	CRIA syndrome	RIPK1
Autoinflammatory disorders	Familial cold autoinflammatory syndrome	Familial cold autoinflammatory syndrome	CIAS1 (NLRP3)
Autoinflammatory disorders	Familial mediterranean fever (FMF)	Familial mediterranean fever defect (FMF)	MEFV
Autoinflammatory disorders	Familial periodic fever	Hyper IgD syndrome (MVK)	MVK
Autoinflammatory disorders	Familial periodic fever	TNF-receptor associated periodic fever syndrome (TRAPS)	TNFRSF1A
Autoinflammatory disorders	Muckle-Wells syndrome	Muckle-Wells syndrome	CIAS1 (NLRP3)
Autoinflammatory disorders	Non-Inflammasome Related Conditions	COPA defect	COPA
Autoinflammatory disorders	Non-Inflammasome Related Conditions	Tim-3 deficiency	HAVCR2 (TIM-3)
Autoinflammatory disorders	Other autoinflammatory diseases with known genetic defect	A20 deficiency	TNFAIP3
Autoinflammatory disorders	Other autoinflammatory diseases with known genetic defect	ADA2-Deficiency (DADA2) / Category: Autoinflammatory	CECR1
Autoinflammatory disorders	Other autoinflammatory diseases with known genetic defect	CDC42 deficiency	CDC42
Autoinflammatory disorders	Other autoinflammatory diseases with known genetic defect	Other autoinflammatory diseases with known genetic defect	CAD14
Autoinflammatory disorders	Other autoinflammatory diseases with known genetic defect	Other autoinflammatory diseases with known genetic defect	CECR1
Autoinflammatory disorders	Other autoinflammatory diseases with known genetic defect	Other autoinflammatory diseases with known genetic defect	HOIL1, RBCK1
Autoinflammatory disorders	Other autoinflammatory diseases with known genetic defect	Other autoinflammatory diseases with known genetic defect	IL1RN
Autoinflammatory disorders	Other autoinflammatory diseases with known genetic defect	Other autoinflammatory diseases with known genetic defect	IL36RN
Autoinflammatory disorders	Other autoinflammatory diseases with known genetic defect	Other autoinflammatory diseases with known genetic defect	LPIN2
Autoinflammatory disorders	Other autoinflammatory diseases with known genetic defect	Other autoinflammatory diseases with known genetic defect	PLCG2
Autoinflammatory disorders	Other autoinflammatory diseases with known genetic defect	Other autoinflammatory diseases with known genetic defect	PSMB8
Autoinflammatory disorders	Other autoinflammatory diseases with known genetic defect	Other autoinflammatory diseases with known genetic defect	SH3BP2
Autoinflammatory disorders	Other autoinflammatory diseases with known genetic defect	Other autoinflammatory diseases with known genetic defect	SLC29A3
Autoinflammatory disorders	Pyogenic sterile arthritis pyoderma gangrenosum and acne	Proline/serine/threonine phosphatase-interacting protein 1 deficiency (PSTPIP1)	PSTPIP1
Autoinflammatory disorders	Type 1 interferonopathies	Aicardi-Goutieres syndrome 7 (AGS7)	IFIH1 (GOF)
Autoinflammatory disorders	Type 1 interferonopathies	Type 1 interferonopathies	ACP5
Autoinflammatory disorders	Type 1 interferonopathies	Type 1 interferonopathies	ADAR1
Autoinflammatory disorders	Type 1 interferonopathies	Type 1 interferonopathies	RNASEH2A
Autoinflammatory disorders	Type 1 interferonopathies	Type 1 interferonopathies	RNASEH2B
Autoinflammatory disorders	Type 1 interferonopathies	Type 1 interferonopathies	RNASEH2C
Autoinflammatory disorders	Type 1 interferonopathies	Type 1 interferonopathies	SAMHD1
Autoinflammatory disorders	Type 1 interferonopathies	Type 1 interferonopathies	STING (TMEM173)
Autoinflammatory disorders	Type 1 interferonopathies	Type 1 interferonopathies	TREX1
Autoinflammatory disorders	Unclassified autoinflammatory diseases	Unclassified autoinflammatory diseases	none
Bone marrow failure	Bone marrow failure	BMFS2 (Hebo deficiency)	ERCC6L2
Combined immunodeficiencies	Atypical Severe Combined Immunodeficiency (Atypical SCID)	Atypical Severe Combined Immunodeficiency (Atypical SCID)	ADA
Combined immunodeficiencies	Atypical Severe Combined Immunodeficiency (Atypical SCID)	Atypical Severe Combined Immunodeficiency (Atypical SCID)	Artemis
Combined immunodeficiencies	Atypical Severe Combined Immunodeficiency (Atypical SCID)	Atypical Severe Combined Immunodeficiency (Atypical SCID)	CARD11
Combined immunodeficiencies	Atypical Severe Combined Immunodeficiency (Atypical SCID)	Atypical Severe Combined Immunodeficiency (Atypical SCID)	CD3d
Combined immunodeficiencies	Atypical Severe Combined Immunodeficiency (Atypical SCID)	Atypical Severe Combined Immunodeficiency (Atypical SCID)	CD3e
Combined immunodeficiencies	Atypical Severe Combined Immunodeficiency (Atypical SCID)	Atypical Severe Combined Immunodeficiency (Atypical SCID)	CD3z
Combined immunodeficiencies	Atypical Severe Combined Immunodeficiency (Atypical SCID)	Atypical Severe Combined Immunodeficiency (Atypical SCID)	Coronin-1A
Combined immunodeficiencies	Atypical Severe Combined Immunodeficiency (Atypical SCID)	Atypical Severe Combined Immunodeficiency (Atypical SCID)	IL21R
Combined immunodeficiencies	Atypical Severe Combined Immunodeficiency (Atypical SCID)	Atypical Severe Combined Immunodeficiency (Atypical SCID)	IL2RG
Combined immunodeficiencies	Atypical Severe Combined Immunodeficiency (Atypical SCID)	Atypical Severe Combined Immunodeficiency (Atypical SCID)	IL7Ralpha
Combined immunodeficiencies	Atypical Severe Combined Immunodeficiency (Atypical SCID)	Atypical Severe Combined Immunodeficiency (Atypical SCID)	JAK3
Combined immunodeficiencies	Atypical Severe Combined Immunodeficiency (Atypical SCID)	Atypical Severe Combined Immunodeficiency (Atypical SCID)	PNP
Combined immunodeficiencies	Atypical Severe Combined Immunodeficiency (Atypical SCID)	Atypical Severe Combined Immunodeficiency (Atypical SCID)	RAG1
Combined immunodeficiencies	Atypical Severe Combined Immunodeficiency (Atypical SCID)	Atypical Severe Combined Immunodeficiency (Atypical SCID)	RAG2
Combined immunodeficiencies	CD4-deficiency	Selective CD4 cell deficiency	CD4
Combined immunodeficiencies	CD8-deficiency	CD8 deficiency	CD8A
Combined immunodeficiencies	CID, Generally Less Profound than SCID	IKZF2 (Helios) deficiency	IKZF2
Combined immunodeficiencies	CID, Generally Less Profound than SCID	Polymerase delta 1 deficiency	POLD1
Combined immunodeficiencies	CID, Generally Less Profound than SCID	RelA haplosufficiency	RELA
Combined immunodeficiencies	Combined immunodeficiency (CID)	Activated PI3K-delta syndrome (APDS)	PIK3CD (PI3K-delta)
Combined immunodeficiencies	Combined immunodeficiency (CID)	Activated PI3K-delta syndrome (APDS)	PIK3R1
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	ADA
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	ARPC1B
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	Artemis
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	CARD11
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	Caspase 8
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	CD27
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	CD3G
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	CD70
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	Cernunnos
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	C-REL
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	CTLA-4
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	DNA-PKcs
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	DOCK11
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	DOCK2
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	DOCK8
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	IKKB
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	IL21R
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	IL2Ralpha
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	IRF4
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	ITK
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	JAK3
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	LCK
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	LIG4
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	LRBA
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	MAGT1
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	MALT1
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	MSN (moesin)
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	MST1 (STK4)
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	NFKB1
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	Orai1 (TMEM142A)
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	OX40
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	PGM3
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	PIK3CD (PI3K-delta)
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	PIK3CG
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	PIK3R1
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	PNP
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	PRKDC

Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	RAG1
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	RAG2
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	RECQL4
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	RELB
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	RhoH
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	RIPK1
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	RLTPR (CARMIL2)
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	STAT1
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	STAT5
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	STAT5a
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	STAT5b
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	STIM1
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	TACI
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	TBX1
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	TRAC
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	TTCTA
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	UNC119D
Combined immunodeficiencies	Combined immunodeficiency (CID)	Combined immunodeficiency (CID)	ZAP70
Combined immunodeficiencies	Combined immunodeficiency (CID)	DIAPH1 deficiency	DIAPH1
Combined immunodeficiencies	Combined immunodeficiency (CID)	EXTL3 deficiency	EXTL3
Combined immunodeficiencies	Combined immunodeficiency (CID)	ICOS deficiency	ICOS
Combined immunodeficiencies	Combined immunodeficiency (CID)	JAK3 deficiency	JAK3
Combined immunodeficiencies	Combined immunodeficiency (CID)	NCKAP1 deficiency	NCKAP1
Combined immunodeficiencies	Combined immunodeficiency (CID)	PI4KA deficiency	PI4KA
Combined immunodeficiencies	Combined immunodeficiency (CID)	Polymerase d 2 deficiency	POLD2
Combined immunodeficiencies	Combined immunodeficiency (CID)	SASH3 deficiency	SASH3
Combined immunodeficiencies	Combined immunodeficiency (CID)	STAT6 GOF	STAT6 (GOF)
Combined immunodeficiencies	Combined immunodeficiency with associated or syndromic features	AIOLOS deficiency	IKZF3
Combined immunodeficiencies	Combined immunodeficiency with associated or syndromic features	BCL11B deficiency	BCL11B
Combined immunodeficiencies	Combined immunodeficiency with associated or syndromic features	Chromosome 11q deletion syndrome (Jacobsen syndrome)	11q23
Combined immunodeficiencies	Combined immunodeficiency with associated or syndromic features	Chromosome 11q deletion syndrome (Jacobsen syndrome)	11q24
Combined immunodeficiencies	Combined immunodeficiency with associated or syndromic features	EPG5 deficiency (Vici syndrome)	EPG5
Combined immunodeficiencies	Combined immunodeficiency with associated or syndromic features	HOIP deficiency	RNF31
Combined immunodeficiencies	Combined immunodeficiency with associated or syndromic features	IMDDHH	NFE2L2
Combined immunodeficiencies	Combined immunodeficiency with associated or syndromic features	MIRAGE syndrome	SAMD9 (GOF)
Combined immunodeficiencies	DNA-repair defects	X-linked reticulate pigmentary disorder-POLA1	POLA1
Combined immunodeficiencies	HLA class I deficiency	HLA class I deficiency	TAP1
Combined immunodeficiencies	HLA class I deficiency	HLA class I deficiency	TAP2
Combined immunodeficiencies	HLA class I deficiency	HLA class I deficiency	TAPBP
Combined immunodeficiencies	HLA class II deficiency	HLA class II deficiency	MHC2TA
Combined immunodeficiencies	HLA class II deficiency	HLA class II deficiency	RFX5
Combined immunodeficiencies	HLA class II deficiency	HLA class II deficiency	RFXANK
Combined immunodeficiencies	HLA class II deficiency	HLA class II deficiency	RFXAP
Combined immunodeficiencies	Kabuki syndrome 1	Kabuki syndrome 1	KMT2D (MLL2)
Combined immunodeficiencies	Kabuki syndrome 2	Kabuki syndrome 2	KDM6A
Combined immunodeficiencies	NUDE/SCID	Winged-helix nude deficiency (FOXN1)	FOXN1
Combined immunodeficiencies	Omenn syndrome	Omenn syndrome	ADA
Combined immunodeficiencies	Omenn syndrome	Omenn syndrome	Artemis
Combined immunodeficiencies	Omenn syndrome	Omenn syndrome	CD45
Combined immunodeficiencies	Omenn syndrome	Omenn syndrome	Coronin-1A
Combined immunodeficiencies	Omenn syndrome	Omenn syndrome	Del 22q11.2
Combined immunodeficiencies	Omenn syndrome	Omenn syndrome	IL21R
Combined immunodeficiencies	Omenn syndrome	Omenn syndrome	IL2Ralpha
Combined immunodeficiencies	Omenn syndrome	Omenn syndrome	IL2RG
Combined immunodeficiencies	Omenn syndrome	Omenn syndrome	IL7Ralpha
Combined immunodeficiencies	Omenn syndrome	Omenn syndrome	JAK3
Combined immunodeficiencies	Omenn syndrome	Omenn syndrome	RAG1
Combined immunodeficiencies	Omenn syndrome	Omenn syndrome	RAG2
Combined immunodeficiencies	Omenn syndrome	Omenn syndrome	RMRP
Combined immunodeficiencies	Omenn syndrome	Omenn syndrome	TBX1
Combined immunodeficiencies	Otofaciocervical syndrome type 2	PAX1 deficiency	PAX1
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Cernunnos/XLF deficiency	NHEJ1
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	ITPKB deficiency	ITPKB
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Reticular Dysgenesis - AK2 (SCID)	AK2
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Reticular Dysgenesis - AK2 (SCID)	none
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	ADA
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	Artemis
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	CD3d
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	CD3e
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	CD3z
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	CD45
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	Coronin-1A
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	Del 22q11.2
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	DNA-PKcs
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	IL21R
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	IL2RG
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	IL7Ralpha
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	JAK3
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	LIG4
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	NHEJ1
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	PGM3
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	RAG1
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	RAG2
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	TBX1
Combined immunodeficiencies	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	ZAP70
Combined immunodeficiencies	T-B- SCID	DNA ligase IV deficiency	LIG4
Complement deficiencies	Complement deficiency	Acquired angioedema	none
Complement deficiencies	Complement deficiency	C3b inactivator deficiency	C3b inactivator
Complement deficiencies	Complement deficiency	CD59 antigen P18-20 deficiency (CD59)	CD59
Complement deficiencies	Complement deficiency	Complement component 1 deficiency	C1Q-alpha
Complement deficiencies	Complement deficiency	Complement component 1 deficiency	C1Q-beta
Complement deficiencies	Complement deficiency	Complement component 1 deficiency	C1Q-gamma
Complement deficiencies	Complement deficiency	Complement component 1 deficiency	C1r
Complement deficiencies	Complement deficiency	Complement component 1 deficiency	C1s

Complement deficiencies	Complement deficiency	Complement component 1 deficiency	Q, subunit unknown
Complement deficiencies	Complement deficiency	Complement component 1q deficiency (C1q deficiency)	C1Q-alpha
Complement deficiencies	Complement deficiency	Complement component 1q deficiency (C1q deficiency)	C1Q-beta
Complement deficiencies	Complement deficiency	Complement component 1q deficiency (C1q deficiency)	C1Q-gamma
Complement deficiencies	Complement deficiency	Complement component 1q deficiency (C1q deficiency)	Q, subunit unknown
Complement deficiencies	Complement deficiency	Complement component 1r deficiency (C1r deficiency)	C1r
Complement deficiencies	Complement deficiency	Complement component 1s deficiency (C1s deficiency)	C1s
Complement deficiencies	Complement deficiency	Complement component 2 deficiency	C2
Complement deficiencies	Complement deficiency	Complement component 3 deficiency (C3)	C3
Complement deficiencies	Complement deficiency	Complement component 4 deficiency	C4
Complement deficiencies	Complement deficiency	Complement component 5 deficiency	C5
Complement deficiencies	Complement deficiency	Complement component 6 deficiency	C6
Complement deficiencies	Complement deficiency	Complement component 7 deficiency	C7
Complement deficiencies	Complement deficiency	Complement component 8 deficiency	C8
Complement deficiencies	Complement deficiency	Complement component 9 deficiency	C9
Complement deficiencies	Complement deficiency	Complement factor B deficiency	Factor B
Complement deficiencies	Complement deficiency	Complement factor H deficiency	Factor H
Complement deficiencies	Complement deficiency	Decay-accelerating factor for complement deficiency (DAF CD55)	CD55
Complement deficiencies	Complement deficiency	Factor D deficiency	Factor D
Complement deficiencies	Complement deficiency	Factor H Related Protein Deficiency	CFHR1
Complement deficiencies	Complement deficiency	Factor H Related Protein Deficiency	CFHR2
Complement deficiencies	Complement deficiency	Factor H Related Protein Deficiency	CFHR3
Complement deficiencies	Complement deficiency	Factor H Related Protein Deficiency	CFHR4
Complement deficiencies	Complement deficiency	Factor H Related Protein Deficiency	CFHR5
Complement deficiencies	Complement deficiency	Factor I deficiency	Factor I
Complement deficiencies	Complement deficiency	Ficolin3 deficiency	FCN3
Complement deficiencies	Complement deficiency	Hereditary Angioedema (HAE, C1inh)	C1 Inhibitor
Complement deficiencies	Complement deficiency	Mannan-binding lectin serine protease (MASP) deficiency	COLEC11
Complement deficiencies	Complement deficiency	Mannan-binding lectin serine protease (MASP) deficiency	MASP1
Complement deficiencies	Complement deficiency	Mannan-binding lectin serine protease (MASP) deficiency	MASP2
Complement deficiencies	Complement deficiency	Membrane Attack Complex Inhibitor (CD59) deficiency	CD59
Complement deficiencies	Complement deficiency	Membrane Cofactor Protein (CD46) deficiency	CD46
Complement deficiencies	Complement deficiency	Properdin P factor complement deficiency (PFC)	Properdin
Complement deficiencies	Complement deficiency	Thrombomodulin deficiency	THBD
Complement deficiencies	Mannose-binding lectin (MBL)	Mannose-binding lectin deficiency (MBL)	MBL
Complement deficiencies	Unclassified complement deficiencies	Unclassified complement deficiencies	none
Defects in innate immunity	CARD 9 deficiency	CARD9 deficiency	CARD9
Defects in innate immunity	Chronic mucocutaneous candidiasis (CMC)	Chronic mucocutaneous candidiasis (CMC)	ACT1
Defects in innate immunity	Chronic mucocutaneous candidiasis (CMC)	Chronic mucocutaneous candidiasis (CMC)	AIRE
Defects in innate immunity	Chronic mucocutaneous candidiasis (CMC)	Chronic mucocutaneous candidiasis (CMC)	IL-17 F
Defects in innate immunity	Chronic mucocutaneous candidiasis (CMC)	Chronic mucocutaneous candidiasis (CMC)	IL-17 receptor alpha
Defects in innate immunity	Chronic mucocutaneous candidiasis (CMC)	Chronic mucocutaneous candidiasis (CMC)	STAT1
Defects in innate immunity	Chronic mucocutaneous candidiasis (CMC)	Chronic mucocutaneous candidiasis (CMC)	STAT1 (GOF)
Defects in innate immunity	DBR1 deficiency	DBR1 deficiency	DBR1
Defects in innate immunity	Defects of TLR/NFkappa-B signalling	Defects of TLR/NFkappa-B signalling	IKK-gamma (NEMO, IKBKG)
Defects in innate immunity	Defects of TLR/NFkappa-B signalling	Defects of TLR/NFkappa-B signalling	IRAK4
Defects in innate immunity	Defects of TLR/NFkappa-B signalling	Defects of TLR/NFkappa-B signalling	MyD88
Defects in innate immunity	Defects of TLR/NFkappa-B signalling	Defects of TLR/NFkappa-B signalling	NFKBIA
Defects in innate immunity	Defects of TLR/NFkappa-B signalling	Defects of TLR/NFkappa-B signalling	TIRAP
Defects in innate immunity	Defects of TLR/NFkappa-B signalling	Defects of TLR/NFkappa-B signalling	TLR8 (GOF)
Defects in innate immunity	Defects of TLR/NFkappa-B signalling	Defects of TLR7	TLR7
Defects in innate immunity	Defects of TLR/NFkappa-B signalling	IRAK1 deficiency	MECP2
Defects in innate immunity	Defects of TLR/NFkappa-B signalling	TLR8 GOF	TLR8 (GOF)
Defects in innate immunity	Epidermodysplasia verruciformis	Epidermodysplasia verruciformis	C1B1 (EVER3)
Defects in innate immunity	Epidermodysplasia verruciformis	Epidermodysplasia verruciformis	TMC6 (EVER1)
Defects in innate immunity	Epidermodysplasia verruciformis	Epidermodysplasia verruciformis	TMC8 (EVER2)
Defects in innate immunity	Herpetic encephalitis	Herpetic encephalitis (HSE)	DBR1
Defects in innate immunity	Herpetic encephalitis	Herpetic encephalitis (HSE)	IRF3
Defects in innate immunity	Herpetic encephalitis	Herpetic encephalitis (HSE)	TBK1
Defects in innate immunity	Herpetic encephalitis	Herpetic encephalitis (HSE)	TLR3
Defects in innate immunity	Herpetic encephalitis	Herpetic encephalitis (HSE)	TRAF3
Defects in innate immunity	Herpetic encephalitis	Herpetic encephalitis (HSE)	TRIF
Defects in innate immunity	Herpetic encephalitis	Herpetic encephalitis (HSE)	UNC93B1
Defects in innate immunity	Herpetic encephalitis	TRIF deficiency	TICAM1
Defects in innate immunity	HOIL1 deficiency	HOIL1 deficiency	HOIL1, RBCK1
Defects in innate immunity	NBAS deficiency	Acute liver failure due to NBAS deficiency	NBAS
Defects in innate immunity	Predisposition to severe viral infection	IFNAR1 deficiency	IFNAR1
Defects in innate immunity	Predisposition to severe viral infection	IRF7 deficiency	IRF7
Defects in innate immunity	Predisposition to severe viral infection	Predisposition to several viral infection	MCM4
Defects in innate immunity	Predisposition to severe viral infection	Predisposition to several viral infection	STAT2
Defects in innate immunity	Predisposition to severe viral infection	ZNF31 deficiency	ZNF31
Defects in innate immunity	Predisposition to Severe Viral Infection	IRF9 deficiency	IRF9
Defects in innate immunity	Trypanosomias	Trypanosomias	APOL-I
Defects in innate immunity	Unclassified defects in innate immunity	Unclassified defects in innate immunity	none
Defects in innate immunity	Warts hypogammaglobulinemia infections and myelokathexis (WHIM)	Warts hypogammaglobulinemia infections and myelokathexis (WHIM)	CXCR4
Diseases of immune dysregulation	Autoimmune lymphoproliferative syndrome (ALPS)	ALPS-like disease	FADD
Diseases of immune dysregulation	Autoimmune lymphoproliferative syndrome (ALPS)	ALPS-like disease	LRBA
Diseases of immune dysregulation	Autoimmune lymphoproliferative syndrome (ALPS)	Autoimmune lymphoproliferative syndrome (ALPS)	Caspase 10 (ALPS IIA)
Diseases of immune dysregulation	Autoimmune lymphoproliferative syndrome (ALPS)	Autoimmune lymphoproliferative syndrome (ALPS)	FADD (combined germline and somatic)
Diseases of immune dysregulation	Autoimmune lymphoproliferative syndrome (ALPS)	Autoimmune lymphoproliferative syndrome (ALPS)	FAS (TNFRSF6) germline
Diseases of immune dysregulation	Autoimmune lymphoproliferative syndrome (ALPS)	Autoimmune lymphoproliferative syndrome (ALPS)	FAS (TNFRSF6) somatic
Diseases of immune dysregulation	Autoimmune lymphoproliferative syndrome (ALPS)	Autoimmune lymphoproliferative syndrome (ALPS)	FASL biallelic
Diseases of immune dysregulation	Autoimmune lymphoproliferative syndrome (ALPS)	Ras associated lymphoproliferative disease (RALD)	K-Ras
Diseases of immune dysregulation	Autoimmune lymphoproliferative syndrome (ALPS)	Ras associated lymphoproliferative disease (RALD)	N-Ras
Diseases of immune dysregulation	Autoimmune polyendocrinopathy candidiasis ectodermal dystrophy (APECED)	APECED / APS1 with CMC - Autoimmune polyendocrinopathy candidiasis ectodermal dystrophy (APECED)	AIRE
Diseases of immune dysregulation	Autoimmune polyendocrinopathy candidiasis ectodermal dystrophy (APECED)	APECED / APS1 with CMC - Autoimmune polyendocrinopathy candidiasis ectodermal dystrophy (APECED)	ITCH
Diseases of immune dysregulation	Autoimmunity with or without lymphoproliferation	JAK1 GOF	JAK1 (GOF)
Diseases of immune dysregulation	Autoimmunity with or without lymphoproliferation	Tripeptidyl-Peptidase II deficiency	TPP2
Diseases of immune dysregulation	Early-onset inflammatory bowel disease	Early-onset inflammatory bowel disease	ILR10
Diseases of immune dysregulation	Early-onset inflammatory bowel disease	Early-onset inflammatory bowel disease	ILR10 receptor alpha chain
Diseases of immune dysregulation	Early-onset inflammatory bowel disease	Early-onset inflammatory bowel disease	ILR10 receptor beta chain
Diseases of immune dysregulation	Early-onset inflammatory bowel disease	Inflammatory Bowel Disease-like	LRBA
Diseases of immune dysregulation	Early-onset multi-organ autoimmune disease	Early-onset multi-organ autoimmune disease	CTLA-4
Diseases of immune dysregulation	Early-onset multi-organ autoimmune disease	Early-onset multi-organ autoimmune disease	SOCS1

Diseases of immune dysregulation	Early-onset multi-organ autoimmune disease	Early-onset multi-organ autoimmune disease	STAT3
Diseases of immune dysregulation	Early-onset multi-organ autoimmune disease	Early-onset multi-organ autoimmune disease	STAT3-GOF
Diseases of immune dysregulation	Early-onset multi-organ autoimmune disease	Infancy onset STING-associated vasculopathy	STING (TMEM173)
Diseases of immune dysregulation	Hemophagocytic Lymphohistiocytosis (HLH)	CD27 deficiency	CD27
Diseases of immune dysregulation	Hemophagocytic Lymphohistiocytosis (HLH)	CD70-deficiency	CD70
Diseases of immune dysregulation	Hemophagocytic Lymphohistiocytosis (HLH)	Chediak Higashi syndrome	LYST
Diseases of immune dysregulation	Hemophagocytic Lymphohistiocytosis (HLH)	Familial hemophagocytic lymphohistiocytosis syndromes (FHLH)	PRF1
Diseases of immune dysregulation	Hemophagocytic Lymphohistiocytosis (HLH)	Familial hemophagocytic lymphohistiocytosis syndromes (FHLH)	STX11
Diseases of immune dysregulation	Hemophagocytic Lymphohistiocytosis (HLH)	Familial hemophagocytic lymphohistiocytosis syndromes (FHLH)	STXBP2 (Munc 18-2)
Diseases of immune dysregulation	Hemophagocytic Lymphohistiocytosis (HLH)	Familial hemophagocytic lymphohistiocytosis syndromes (FHLH)	UNC13D
Diseases of immune dysregulation	Hemophagocytic Lymphohistiocytosis (HLH)	Griscelli syndrome, type 2	RAB27A
Diseases of immune dysregulation	Hemophagocytic Lymphohistiocytosis (HLH)	Hermansky-Pudlak syndrome	AP3B1
Diseases of immune dysregulation	Hemophagocytic Lymphohistiocytosis (HLH)	ITK deficiency (HLH phenotype)	ITK
Diseases of immune dysregulation	Hemophagocytic Lymphohistiocytosis (HLH)	X-linked lymphoproliferative syndrome (XLP)	BIRC4/XIAP (XLP2)
Diseases of immune dysregulation	Hemophagocytic Lymphohistiocytosis (HLH)	X-linked lymphoproliferative syndrome (XLP)	SH2D1A (XLP1)
Diseases of immune dysregulation	IPEX	FOXP3 deficiency (IPEX)	FOXP3
Diseases of immune dysregulation	IPEX	Interleukin 2 receptor alpha deficiency (CD25) (IPEX phenotype)	IL2Ralpha
Diseases of immune dysregulation	IPEX	IPEX-like disease	LRBA
Diseases of immune dysregulation	Regulatory T-cell defects	BACH2 deficiency	BACH2
Diseases of immune dysregulation	Regulatory T-cell defects	CD122 deficiency	IL2Rbeta
Diseases of immune dysregulation	Regulatory T-cell defects	DEF6 deficiency	DEF6
Diseases of immune dysregulation	Regulatory T-cell defects	LRBA-deficiency	LRBA
Diseases of immune dysregulation	Susceptibility to EBV and lymphoproliferative conditions	CD137 deficiency (41BB)	TNFRSF9
Diseases of immune dysregulation	Susceptibility to EBV and lymphoproliferative conditions	CTPS1 deficiency	CTPS1
Diseases of immune dysregulation	Susceptibility to EBV and lymphoproliferative conditions	PRKCD deficiency	PRKCD
Diseases of immune dysregulation	Unclassified disorders of immune dysregulation	Unclassified disorders of immune dysregulation	none
Other well defined PIDs	Asplenia syndrome	Asplenia syndrome (Ivemark syndrome)	none
Other well defined PIDs	Asplenia syndrome	Isolated congenital asplenia	RPSA
Other well defined PIDs	Cartilage hair hypoplasia	Cartilage hair hypoplasia	RMRP
Other well defined PIDs	CHARGE syndrome	CHARGE syndrome	CHARGE-CHD7
Other well defined PIDs	CHARGE syndrome	CHARGE syndrome	SEMA3E
Other well defined PIDs	Defects of Vitamin B12 and Folate metabolism	Defects of Vitamin B12 and Folate metabolism	MTHFD1
Other well defined PIDs	Defects of Vitamin B12 and Folate metabolism	Defects of Vitamin B12 and Folate metabolism	SLC46A1
Other well defined PIDs	Defects of Vitamin B12 and Folate metabolism	Defects of Vitamin B12 and Folate metabolism	TCN2
Other well defined PIDs	DiGeorge syndrome	DiGeorge syndrome (DGS)	Del 10p
Other well defined PIDs	DiGeorge syndrome	DiGeorge syndrome (DGS)	Del 22q11.2
Other well defined PIDs	DiGeorge syndrome	DiGeorge syndrome (DGS)	TBX1
Other well defined PIDs	DNA-breakage disorder	Ataxia telangiectasia (ATM)	ATM
Other well defined PIDs	DNA-breakage disorder	AT-like disorder	MRE11
Other well defined PIDs	DNA-breakage disorder	Bloom syndrome	BLM Helicase
Other well defined PIDs	DNA-breakage disorder	Bloom syndrome	none
Other well defined PIDs	DNA-breakage disorder	DNA-ligase 1 ATP-dependent deficiency (LIG1)	LIG1
Other well defined PIDs	DNA-breakage disorder	Immunodeficiency centromeric instability facial anomalies syndrome (ICF)	CDC47
Other well defined PIDs	DNA-breakage disorder	Immunodeficiency centromeric instability facial anomalies syndrome (ICF)	DNMT3B
Other well defined PIDs	DNA-breakage disorder	Immunodeficiency centromeric instability facial anomalies syndrome (ICF)	HELLS
Other well defined PIDs	DNA-breakage disorder	Immunodeficiency centromeric instability facial anomalies syndrome (ICF)	ZBTB24
Other well defined PIDs	DNA-breakage disorder	MCM4 deficiency	MCM4
Other well defined PIDs	DNA-breakage disorder	Nijmegen breakage syndrome (NBS1)	NBS1
Other well defined PIDs	DNA-breakage disorder	Other DNA-breakage disorder	none
Other well defined PIDs	DNA-breakage disorder	Post-Meiotic Segregation 2 (PMS2) deficiency	PMS2
Other well defined PIDs	DNA-breakage disorder	RNF168 deficiency	RNF168
Other well defined PIDs	DNA-breakage disorder	Seckel syndrome	none
Other well defined PIDs	Dyskeratosis congenita	Dyskeratosis congenita	DKC1
Other well defined PIDs	Dyskeratosis congenita	Dyskeratosis congenita	NAF1
Other well defined PIDs	Dyskeratosis congenita	Dyskeratosis congenita	NHP2
Other well defined PIDs	Dyskeratosis congenita	Dyskeratosis congenita	NOP10
Other well defined PIDs	Dyskeratosis congenita	Dyskeratosis congenita	PARN
Other well defined PIDs	Dyskeratosis congenita	Dyskeratosis congenita	RTEL1
Other well defined PIDs	Dyskeratosis congenita	Dyskeratosis congenita	TERC
Other well defined PIDs	Dyskeratosis congenita	Dyskeratosis congenita	TERT
Other well defined PIDs	Dyskeratosis congenita	Dyskeratosis congenita	TINF2
Other well defined PIDs	Dyskeratosis congenita	Hoyeraal-Hreidarsson syndrome	APOLLO (snm1b def)
Other well defined PIDs	Dyskeratosis congenita	Hoyeraal-Hreidarsson syndrome	DKC1
Other well defined PIDs	Dyskeratosis congenita	Hoyeraal-Hreidarsson syndrome	NHP2
Other well defined PIDs	Dyskeratosis congenita	Hoyeraal-Hreidarsson syndrome	NOP10
Other well defined PIDs	Dyskeratosis congenita	Hoyeraal-Hreidarsson syndrome	PARN
Other well defined PIDs	Dyskeratosis congenita	Hoyeraal-Hreidarsson syndrome	RTEL1
Other well defined PIDs	Dyskeratosis congenita	Hoyeraal-Hreidarsson syndrome	TERC
Other well defined PIDs	Dyskeratosis congenita	Hoyeraal-Hreidarsson syndrome	TERT
Other well defined PIDs	Dyskeratosis congenita	Hoyeraal-Hreidarsson syndrome	TINF2
Other well defined PIDs	Fc receptor deficiencies	Fc receptor deficiencies	FCGR1A
Other well defined PIDs	Fc receptor deficiencies	Fc receptor deficiencies	FCGR2A
Other well defined PIDs	Fc receptor deficiencies	Fc receptor deficiencies	FCGR2B
Other well defined PIDs	Fc receptor deficiencies	Fc receptor deficiencies	FCGR3A
Other well defined PIDs	Fc receptor deficiencies	Fc receptor deficiencies	FCGR3B
Other well defined PIDs	Fc receptor deficiencies	Fc receptor deficiencies	FCGRT
Other well defined PIDs	FILS syndrome	Facial dysmorphism, immunodeficiency, livedo, and short stature (FILS syndrome)	POLE1
Other well defined PIDs	FOXN1 Haploinsufficiency	FOXN1 Haploinsufficiency	FOXN1
Other well defined PIDs	GINS 1 deficiency	GINS 1 deficiency	GINS1
Other well defined PIDs	Hyper IgE syndromes	Hyper IgE syndrome (HIES)	DOCK8
Other well defined PIDs	Hyper IgE syndromes	Hyper IgE syndrome (HIES)	IL6ST
Other well defined PIDs	Hyper IgE syndromes	Hyper IgE syndrome (HIES)	STAT3
Other well defined PIDs	Hyper IgE syndromes	Hyper IgE syndrome (HIES)	STAT3-DN
Other well defined PIDs	Hyper IgE syndromes	Hyper IgE syndrome (HIES)	Tyk2
Other well defined PIDs	Hyper IgE syndromes	Hyper IgE syndrome (HIES)	ZNF341
Other well defined PIDs	IKAROS deficiency	IKAROS deficiency	IKAROS
Other well defined PIDs	Immunodeficiencies with multiple intestinal atresias	Immunodeficiencies with multiple intestinal atresias	TTC7A
Other well defined PIDs	MonoMAC (WILD)	Monocytopenia and mycobacterial infection (MonoMAC (WILD))	GATA2
Other well defined PIDs	MYSM1 deficiency	MYSM1 deficiency	MYSM1
Other well defined PIDs	Netherton syndrome	Netherton syndrome	SPINK5
Other well defined PIDs	Roifman syndrome	Roifman syndrome	RNU4ATAC
Other well defined PIDs	Schimke disease	Schimke disease	SMARCAL1
Other well defined PIDs	SIFD	Congenital sideroblastic anemia with immunodeficiency, fevers, and developmental delay (SIFD)	TRNT1
Other well defined PIDs	Trichohepatoenteric syndrome (Giraud syndrome)	Trichohepatoenteric syndrome	SKIV2L

Other well defined PIDs	Trichohepatoenteric syndrome (Giraud syndrome)	Trichohepatoenteric syndrome	TTC37
Other well defined PIDs	Unclassified syndromic immunodeficiencies	Unclassified syndromic immunodeficiencies	none
Other well defined PIDs	VODI	Hepatic venoocclusive disease with immunodeficiency (VODI)	SP110
Other well defined PIDs	Wiskott-Aldrich syndrome (WAS)	WIP deficiency	WIPF1
Other well defined PIDs	Wiskott-Aldrich syndrome (WAS)	Wiskott-Aldrich syndrome (WAS)	WAS
Other well defined PIDs	Wiskott-Aldrich syndrome (WAS)	X-linked thrombocytopenia with mutations in WAS	WAS
Phagocytic disorders	Actin beta deficiency	Actin beta deficiency (ACTB)	Actin beta
Phagocytic disorders	Barth syndrome	Barth syndrome	TAZ
Phagocytic disorders	Chronic granulomatous disease (CGD)	Chronic granulomatous disease (CGD)	CYB1 EROS
Phagocytic disorders	Chronic granulomatous disease (CGD)	Chronic granulomatous disease (CGD)	GP91-phox(CYBB)
Phagocytic disorders	Chronic granulomatous disease (CGD)	Chronic granulomatous disease (CGD)	P22-phox (CYBA)
Phagocytic disorders	Chronic granulomatous disease (CGD)	Chronic granulomatous disease (CGD)	P40-phox (NCF4)
Phagocytic disorders	Chronic granulomatous disease (CGD)	Chronic granulomatous disease (CGD)	P47-phox (NCF1)
Phagocytic disorders	Chronic granulomatous disease (CGD)	Chronic granulomatous disease (CGD)	P67-phox (NCF2)
Phagocytic disorders	Clericuzio-type poikiloderma with neutropenia syndrome	Clericuzio-type poikiloderma with neutropenia syndrome	C16orf57
Phagocytic disorders	Clericuzio-type poikiloderma with neutropenia syndrome	Clericuzio-type poikiloderma with neutropenia syndrome	USB1
Phagocytic disorders	COHEN syndrome	COHEN syndrome	COH1
Phagocytic disorders	COHEN syndrome	COHEN syndrome	VPS13B
Phagocytic disorders	Congenital neutropenia	Congenital neutropenia	CLPB
Phagocytic disorders	Congenital neutropenia	Congenital neutropenia	CSF3R
Phagocytic disorders	Congenital neutropenia	Congenital neutropenia	ELA2
Phagocytic disorders	Congenital neutropenia	Congenital neutropenia	G6PC3
Phagocytic disorders	Congenital neutropenia	Congenital neutropenia	GATA2
Phagocytic disorders	Congenital neutropenia	Congenital neutropenia	GF11
Phagocytic disorders	Congenital neutropenia	Congenital neutropenia	HAX1
Phagocytic disorders	Congenital neutropenia	Congenital neutropenia	JAGN1
Phagocytic disorders	Congenital neutropenia	Congenital neutropenia	P14
Phagocytic disorders	Congenital neutropenia	Congenital neutropenia	SRP54
Phagocytic disorders	Congenital neutropenia	Congenital neutropenia	WAS
Phagocytic disorders	Congenital neutropenia	VPS45 deficiency (SCN5)	VPS45
Phagocytic disorders	Cyclic neutropenia	Cyclic neutropenia	ELA2
Phagocytic disorders	Defects of Motility	MKL1 deficiency	MKL1
Phagocytic disorders	Defects with susceptibility to mycobacterial infection (MSMD)	Defects with susceptibility to mycobacterial infection (MSMD)	GP91-phox(CYBB)
Phagocytic disorders	Defects with susceptibility to mycobacterial infection (MSMD)	Defects with susceptibility to mycobacterial infection (MSMD)	IFNGR1
Phagocytic disorders	Defects with susceptibility to mycobacterial infection (MSMD)	Defects with susceptibility to mycobacterial infection (MSMD)	IFNGR2
Phagocytic disorders	Defects with susceptibility to mycobacterial infection (MSMD)	Defects with susceptibility to mycobacterial infection (MSMD)	IL12B
Phagocytic disorders	Defects with susceptibility to mycobacterial infection (MSMD)	Defects with susceptibility to mycobacterial infection (MSMD)	IL12R beta-1
Phagocytic disorders	Defects with susceptibility to mycobacterial infection (MSMD)	Defects with susceptibility to mycobacterial infection (MSMD)	IL18
Phagocytic disorders	Defects with susceptibility to mycobacterial infection (MSMD)	Defects with susceptibility to mycobacterial infection (MSMD)	IL23-alpha
Phagocytic disorders	Defects with susceptibility to mycobacterial infection (MSMD)	Defects with susceptibility to mycobacterial infection (MSMD)	IRAK4
Phagocytic disorders	Defects with susceptibility to mycobacterial infection (MSMD)	Defects with susceptibility to mycobacterial infection (MSMD)	IRF8
Phagocytic disorders	Defects with susceptibility to mycobacterial infection (MSMD)	Defects with susceptibility to mycobacterial infection (MSMD)	ISG15
Phagocytic disorders	Defects with susceptibility to mycobacterial infection (MSMD)	Defects with susceptibility to mycobacterial infection (MSMD)	SPPL2A
Phagocytic disorders	Defects with susceptibility to mycobacterial infection (MSMD)	Defects with susceptibility to mycobacterial infection (MSMD)	STAT1
Phagocytic disorders	Defects with susceptibility to mycobacterial infection (MSMD)	Defects with susceptibility to mycobacterial infection (MSMD)	STAT5
Phagocytic disorders	Glycogen storage disease type 1b (GS1b)	Glycogen storage disease type 1b (GS1b)	G6PT1
Phagocytic disorders	Leukocyte adhesion deficiency (LAD)	Leukocyte adhesion deficiency (LAD)	LAD1 / ITGB2
Phagocytic disorders	Leukocyte adhesion deficiency (LAD)	Leukocyte adhesion deficiency (LAD)	LAD2 / FUCT1
Phagocytic disorders	Leukocyte adhesion deficiency (LAD)	Leukocyte adhesion deficiency (LAD)	LAD3
Phagocytic disorders	Localized juvenile peridontitis	Localized juvenile peridontitis	Formyl peptide receptor
Phagocytic disorders	Myeloperoxidase deficiency (MPO)	Myeloperoxidase deficiency (MPO)	MPO
Phagocytic disorders	Neutrophil glucose-6-phosphate dehydrogenase	Glucose-6-phosphate dehydrogenase deficiency (G6PD)	G6PD
Phagocytic disorders	Papillon-Lefevre syndrome	Papillon-Lefevre syndrome	CTSC
Phagocytic disorders	PID with partial albinism	Partial albinism and immunodeficiency syndrome	none
Phagocytic disorders	Pulmonary alveolar proteinosis	Pulmonary alveolar proteinosis	CSF2RA
Phagocytic disorders	RAC2-GTPase defect	RAS-related C3 Botulinum toxin substrate 2 deficiency (RAC2)	RAC2
Phagocytic disorders	Shwachman-Diamond-syndrome	Shwachman-Diamond-syndrome	DNAJC21
Phagocytic disorders	Shwachman-Diamond-syndrome	Shwachman-Diamond-syndrome	EFL1
Phagocytic disorders	Shwachman-Diamond-syndrome	Shwachman-Diamond-syndrome	SBDS
Phagocytic disorders	Shwachman-Diamond-syndrome	Shwachman-Diamond-syndrome	SRP54
Phagocytic disorders	Specific granule defect	CCAAT/enhancer binding protein epsilon deficiency (CEBPE)	CEBPE
Phagocytic disorders	Unclassified phagocytic disorders	Unclassified phagocytic disorders	none
Predominantly antibody disorders	Agammaglobulinemias	Agammaglobulinemia	BLNK/SLP65
Predominantly antibody disorders	Agammaglobulinemias	Agammaglobulinemia	BTK
Predominantly antibody disorders	Agammaglobulinemias	Agammaglobulinemia	CD79A
Predominantly antibody disorders	Agammaglobulinemias	Agammaglobulinemia	CD79B
Predominantly antibody disorders	Agammaglobulinemias	Agammaglobulinemia	IGHM
Predominantly antibody disorders	Agammaglobulinemias	Agammaglobulinemia	IGLL1
Predominantly antibody disorders	Agammaglobulinemias	Agammaglobulinemia	PIK3R1
Predominantly antibody disorders	Agammaglobulinemias	Agammaglobulinemia	TCF3
Predominantly antibody disorders	Agammaglobulinemias	FNIP1- associated IEI	FNIP1
Predominantly antibody disorders	Agammaglobulinemias	TOP2B deficiency	TOP2B
Predominantly antibody disorders	Class switch recombination defects (CSR) / HIGM syndromes	CSR defects and Hyper IgM (HIGM) syndromes	AID
Predominantly antibody disorders	Class switch recombination defects (CSR) / HIGM syndromes	CSR defects and Hyper IgM (HIGM) syndromes	CD40 (TNFRSF5)
Predominantly antibody disorders	Class switch recombination defects (CSR) / HIGM syndromes	CSR defects and Hyper IgM (HIGM) syndromes	CD40L (CD154)
Predominantly antibody disorders	Class switch recombination defects (CSR) / HIGM syndromes	CSR defects and Hyper IgM (HIGM) syndromes	PMS2
Predominantly antibody disorders	Class switch recombination defects (CSR) / HIGM syndromes	CSR defects and Hyper IgM (HIGM) syndromes	UNG
Predominantly antibody disorders	CVID Phenotype	NFKB1 deficiency	NFKB1
Predominantly antibody disorders	CVID Phenotype	PTEN Deficiency (LOF)	PTEN
Predominantly antibody disorders	CVID Phenotype	SH3KBP1 (CIN85) deficiency	SH3KBP1
Predominantly antibody disorders	CVID Phenotype	TAC1 deficiency	TNFRSF13B
Predominantly antibody disorders	Hypogammaglobulinemias	ADA2-Deficiency (DADA2) / Category: Antibody Deficiencies	CECR1
Predominantly antibody disorders	Hypogammaglobulinemias	ARHGEF1-deficiency	ARHGEF1
Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	ATP6AP1
Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	BACH2
Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	BAFFR
Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	CD19
Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	CD20
Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	CD21
Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	CD81
Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	CTLA-4
Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	ICOS
Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	IKZF1
Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	IRF2BP2

Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	LRBA
Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	MOGS
Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	NFKB1
Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	NFKB2
Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	none
Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	PTEN
Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	TACI
Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	TRNT1
Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	TTC37
Predominantly antibody disorders	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	TWEAK
Predominantly antibody disorders	Hypogammaglobulinemias	Deficiency of specific IgG (Specific antibody deficiency - SPAD)	NFKBIA
Predominantly antibody disorders	Hypogammaglobulinemias	IgA with IgG subclass deficiency	TACI
Predominantly antibody disorders	Hypogammaglobulinemias	Immunoglobulin chain deficiencies	Heavy chain
Predominantly antibody disorders	Hypogammaglobulinemias	Immunoglobulin chain deficiencies	Kappa light chain
Predominantly antibody disorders	Hypogammaglobulinemias	Immunoglobulin chain deficiencies	Lambda light chain
Predominantly antibody disorders	Hypogammaglobulinemias	Isolated IgG subclass deficiency	BAFFR
Predominantly antibody disorders	Hypogammaglobulinemias	Isolated IgG subclass deficiency	CD19
Predominantly antibody disorders	Hypogammaglobulinemias	Isolated IgG subclass deficiency	CD21
Predominantly antibody disorders	Hypogammaglobulinemias	Isolated IgG subclass deficiency	CD81
Predominantly antibody disorders	Hypogammaglobulinemias	Isolated IgG subclass deficiency	ICOS
Predominantly antibody disorders	Hypogammaglobulinemias	Isolated IgG subclass deficiency	TACI
Predominantly antibody disorders	Hypogammaglobulinemias	Other immunoglobulin gene deletions	none
Predominantly antibody disorders	Hypogammaglobulinemias	Selective IgA deficiency	TACI
Predominantly antibody disorders	Hypogammaglobulinemias	Selective IgM deficiency	none
Predominantly antibody disorders	Hypogammaglobulinemias	Thymoma with immunodeficiency	none
Predominantly antibody disorders	Hypogammaglobulinemias	Transcobalamin II deficiency	Transcobalamin II
Predominantly antibody disorders	Hypogammaglobulinemias	Transient hypogammaglobulinemia of infancy	none
Predominantly antibody disorders	Isotype, Light Chain, or Functional Deficiencies with Generally Normal Numbers of B Cells	CARD11 GOF	CARD11
Predominantly antibody disorders	Unclassified antibody deficiency	Steinert-Myotonia Dystrophia	ZNF9
Predominantly antibody disorders	Unclassified antibody deficiency	Unclassified antibody deficiency	11q23
Predominantly antibody disorders	Unclassified antibody deficiency	Unclassified antibody deficiency	LRBA
Predominantly antibody disorders	Unclassified antibody deficiency	Unclassified antibody deficiency	NFKB1
Predominantly antibody disorders	Unclassified antibody deficiency	Unclassified antibody deficiency	NFKB2
Predominantly antibody disorders	Unclassified antibody deficiency	Unclassified antibody deficiency	none
Predominantly antibody disorders	Unclassified antibody deficiency	Unclassified antibody deficiency	PIK3CD (PI3K-delta)
Unclassified Immunodeficiencies	Unclassified immunodeficiencies	Unclassified immunodeficiencies	none
Unclassified Immunodeficiencies	Unclassified immunodeficiencies	Unclassified immunodeficiencies	TACI