ESID Online Registry: Diseases and Genes

as of 14.12.2022 - Please send ideas and questions to esid-registry@uniklinik-freiburg.de

Main Category	Sub Category	IEI Diagnosis (PID Diagnosis)	Gene
Autoinflammatory disorders	**Blau syndrome**	Caspase recruitment domain-containing protein 15 deficiency (CARD15)	CARD15
	CINCA syndrome	CINCA syndrome	CIAS1 (NLRP3)
	Covid-19 hyperinflammation	Covid-19 related hyperinflammation syndrome	none
	Familial cold autoinflammatory syndrome	Familial cold autoinflammatory syndrome	CIAS1 (NLRP3)
	Familial mediterranean fever (FMF)	Familial mediterranean fever defect (FMF)	MEFV
	Familial periodic fever	Hyper IgD syndrome (MVK)	MVK
		TNF-receptor associated periodic fever syndrome (TRAPS)	TNFRSF1A
	Muckle-Wells syndrome	Muckle-Wells syndrome	CIAS1 (NLRP3)
	Other autoinflammatory diseases with known genetic defect	Other autoinflammatory diseases with known genetic defect	CAD14
			CECR1
			HOIL1
			IL1RN
			IL36RN
			LPIN2
			PLCG2
			PSMB8
			SH3BP2
			SLC29A3
		A20 deficiency	TNFAIP3
		ADA2 deviciency (DADA2)	CECR1
	Pyogenic sterile arthritis pyoderma gangrenosum and acne	Proline/serine/threonine phosphatase-interacting protein 1 deficiency (PSTPIP1)	PSTPIP1
	Non-Inflammasome Related Conditions	COPA defect	COPA
		Tim-3 deficiency	HAVCR2
	Type 1 Interferonopathies	Aicardi-Goutieres syndrome 7 (AGS7)	*IFIH1 (GOF)*
	Unclassified autoinflammatory diseases	Unclassified autoinflammatory diseases	none
Combined immunodeficiencies	**Atypical Severe Combined Immunodeficiency (Atypical SCID)**	Atypical Severe Combined Immunodeficiency (Atypical SCID)	ADA
			Artemis
			CARD11
			CD3d
			CD3e
			CD3z
			Coronin-1A
			IL2RG
			IL21R
			IL7Ralpha
			JAK3
			PNP
			RAG1
			RAG2
	CD4-deficiency	Selective CD4 cell deficiency	CD4
	CD8-deficiency	CD8 deficiency	CD8A
	Combined immunodeficiency (CID)	Combined immunodeficiency	ADA
			ARPC1B
			Artemis
			CARD11
			Caspase 8
			CD3G
			CD27
			CD70
			Cernunnos
			C-REL
			CTLA-4
			DNA-PKcs
			DOCK2
			DOCK8
			IKBKB
			IL21R
			IL2Ralpha
			IRF4

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Combined immunodeficiencies	**Combined immunodeficiency (CID)**	Combined immunodeficiency	ITK
			LCK
			LIG4
			LRBA
			MAGT1
			MALT1
			MSN (moesin)
			MST1 (STK4)
			NFKB1
			Orai1 (TMEM142A)
			OX40
			PGM3
			PIK3CD (PI3K-delta)
			PIK3CG
			PIK3R1
			PRKDC
			PNP
			RECQL4
			RhoH
			RLTPR (CARMIL2)
			RAG1
			RAG2
			RELB
			RIPK1
			STAT1
			STAT5
			STAT5a
			STAT5b
			STIM1
			TACI
			TBX1
			TRAC
			TTC7A
			UNC119D
			ZAP70
		Activated PI3K-delta syndrome (APDS)	PIK3CD (PI3K-delta)
			PIK3R1
		DIAPH1 deficiency	*DIAPH1*
		EXTL3 deficiency	EXTL3
		NCKAPL1 deficiency	*NCKAPL1*
		Polymerase d 2 deficiency	POLD2
		SASH3 deficiency	*SASH3*
	HLA class I deficiency	HLA class I deficiency	TAP1
			TAP2
			TAPBP
	HLA class II deficiency	HLA class II deficiency	MHC2TA
			RFX5
			RFXANK
			RFXAP
	Kabuki syndrome 1	Kabuki syndrome 1	KMT2D (MLL2)
	Kabuki syndrome 2	Kabuki syndrome 2	KDM6A
	NUDE/SCID	Winged-helix nude deficiency (FOXN1)	FOXN1
	Omenn syndrome	Omenn Syndrome	ADA
			Artemis
			CD45
			Coronin-1A
			Del 22q11.2
			IL2RG
			IL21R
			IL2Ralpha

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Combined immunodeficiencies	**Omenn syndrome**	Omenn Syndrome	IL7Ralpha
			JAK3
			RAG1
			RAG2
			RMRP
			TBX1
	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	ADA
			Artemis
			CD3d
			CD3e
			CD3z
			CD45
			Coronin-1A
			Del 22q11.2
			DNA-PKcs
			IL2RG
			IL21R
			IL7Ralpha
			JAK3
			LIG4
			NHEJ1
			PGM3
			RAG1
			RAG2
			TBX1
			ZAP70
		Cernunnos/XLF deficiency	NHEJ1
		ITPKB deficiency	ITPKB
		Reticular Dysgenesis - AK2 (SCID)	AK2
		Reticular Dysgenesis - UNK (SCID)	none
	T-B- SCID	DNA ligase IV deficiency	*LIG4*
	Combined immunodeficiency with associated or syndromic features	Chromosome 11q deletion syndrome (Jacobsen syndrome)	11q23del
		BCL11B deficiency	*BCL11B*
		EPG5 deficiency	EPG5
		FNIP1- associated IEI	*FNIP1*
		Immunodeficiency, developmental delay and hypohomocysteinemia, IMDDHH	NFE2L2
		MIRAGE syndrome	SAMD9 (GOF)
		HOIP deficiency	*RNF31*
Complement deficiencies	**Complement deficiency**	Acquired angioedema	none
		C3b inactivator deficiency	C3b inactivator
		CD59 antigen P18-20 deficiency (CD59)	CD59
		Complement component 1q deficiency	C1Q-alpha
			C1Q-beta
			C1Q-gamma
			Q, subunit unknown
		Complement component 1r deficiency	C1r
		Complement component 1s deficiency	C1s
		Complement component 2 deficiency	C2
		Complement component 3 deficiency	C3
		Complement component 4 deficiency	C4
		Complement component 5 deficiency	C5
		Complement component 6 deficiency	C6
		Complement component 7 deficiency	C7
		Complement component 8 deficiency	C8
		Complement component 9 deficiency	C9
		Complement factor B deficiency	Factor B
		Complement factor H deficiency	Factor H
		Decay-accelerating factor for complement deficiency (DAF CD55)	CD55
		Factor D deficiency	Factor D
		Factor I deficiency	Factor I

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		Ficolin3 deficiency	FCN3
		Hereditary Angioedema (C1inh)	C1 Inhibitor
		Mannan-binding lectin serine protease (MASP) deficiency	COLEC11
			MASP1

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Complement deficiencies	**Complement deficiency**	Mannan-binding lectin serine protease (MASP) deficiency	MASP2
		Membrane Attack Complex Inhibitor (CD59) deficiency	CD59
		Membrane Cofactor Protein (CD46) deficiency	CD46
		Properdin P factor complement deficiency (PFC)	Properdin
		Thrombomodulin deficiency	THBD
		Factor H Related Protein Deficiency	CFHR1
			CFHR2
			CFHR3
			CFHR4
			CFHR5
	Mannose-binding lectin (MBL)	Mannose-binding lectin deficiency (MBL)	MBL
	Unclassified complement deficiencies	Unclassified complement deficiencies	none
Defects in innate immunity	**CARD 9 deficiency**	CARD9 deficiency	CARD9
	Chronic mucocutaneous candidiasis (CMC)	Chronic mucocutaneous candidiasis (CMC)	ACT1
			AIRE
			IL-17 F
			IL-17 receptor alpha
			STAT1
			STAT1 (GOF)
	DBR1 deficiency	DBR1 deficiency	DBR1
	Defects of TLR/NFkappa-B signalling	Defects of TLR/NFkappa-B signalling	IKK-gamma (NEMO, IKBKG)
			IRAK4
			MyD88
			NFKBIA
			TIRAP
		IRAK1 deficiency	MECP2
	Epidermodysplasia verruciformis	Epidermodysplasia verruciformis	TMC6 (EVER1)
			TMC8 (EVER2)
			CIB1 (EVER3)
	Herpetic encephalitis	Herpetic encephalitis (HSE)	DBR1
			IRF3
			TBK1
			TLR3
			TRAF3
			TRIF
			UNC93B1
			none
	HOIL1 deficiency	HOIL1 deficiency	HOIL1 , RBCK1
	IRF9 deficiency	IRF9 deficiency	IRF9
	NBAS deficiency	Acute liver failure due to NBAS deficiency	NBAS
	Predisposition to severe viral infection	Predisposition to severe viral infection	MCM4
			STAT2
		IFNAR1 deficiency	IFNAR1
		IRF7 deficiency	IRF7
	Trypanosomias	Trypanosomias	APOL-I
	Unclassified defects in innate immunity	Unclassified defects in innate immunity	none
	Warts hypogammaglobulinemia infections and myelokathexis (WHIM)	Warts hypogammaglobulinemia infections and myelokathexis (WHIM)	CXCR4

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Diseases of immune dysregulation	**Autoimmune lymphoproliferative syndrome (ALPS)**	Autoimmune lymphoproliferative syndrome (ALPS)	CARD11
			Caspase 8 (ALPS IIB)
			Caspase 10 (ALPS IIA)
			CD95 (germline - ALPS IA)
			CD95 (somatic - ALPS Im)
			FAS (TNFRSF6)
			FASL (CD178) (ALPS IB)
			PRKCD
		ALPS-like disease	LRBA
			FADD
		Ras associated lymphoproliferative disease (RALD)	K-Ras
			N-Ras
	Autoimmunity with or without lymphoproliferation	JAK1 GOF	JAK1 GOF
		Tripeptidyl-Peptidase II deficiency	TPP2
	Autoimmune polyendocrinopathy candidiasis ectodermal dystrophy (APECED)	Autoimmune polyendocrinopathy candidiasis ectodermal dystrophy (APECED)	AIRE
			ITCH
	Early-onset inflammatory bowel disease	Early-onset inflammatory bowel disease	ILR10
			ILR10 receptor alpha chain
			ILR10 receptor beta chain
		IBD-like	LRBA
	Hemophagocytic Lymphohistiocytosis (HLH)	CD27 deficiency	CD27
		CD70-deficiency	CD70
		Chediak Higashi syndrome	LYST
		Familial hemophagocytic lymphohistiocytosis syndromes (FHLH)	PRF1
			STX11
			STXBP2 (Munc 18-2)
			UNC13D
		Griscelli syndrome type 2	RAB27A
		Hermansky-Pudlak syndrome	AP3B1
		ITK deficiency (HLH phenotype)	ITK
		X-linked lymphoproliferative syndrome (XLP)	BIRC4/XIAP (XLP2)
			SH2D1A (XLP1)
	IPEX	FOXP3 deficiency (IPEX)	FOXP3
		Interleukin 2 receptor alpha deficiency (CD25) (IPEX phenotype)	IL2Ralpha
		IPEX-like disease	LRBA
	Type 1 interferonopathies	Type 1 interferonopathies	ACP5
			ADAR1
			RNASEH2A
			RNASEH2B
			RNASEH2C
			SAMHD1
			TREX1
			STING (TMEM173)
	Early-onset multi-organ autoimmune disease	Early-onset multi-organ autoimmune disease	CTLA-4
			STAT3 GOF
		Infancy onset STING-associated vasculopathy	STING (TMEM173)
	Regulatory T-cell defects	LRBA-deficiency	LRBA
		DEF6 deficiency	DEF6
		BACH2 deficiency	BACH2
	Susceptibility to EBV and lymphoproliferative conditions	CTPS1 deficiency	CTPS1
		CD137 deficiency (41BB)	TNFRSF9
	Unclassified disorders of immune dysregulation	Unclassified disorders of immune dysregulation	none

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Other well defined PIDs	**Asplenia syndrome**	Asplenia syndrome (Ivemark syndrome)	none
		Isolated congenital asplenia	RPSA
	Cartilage hair hypoplasia	Cartilage hair hypoplasia	RMRP
	CHARGE syndrome	CHARGE syndrome	CHARGE-CHD7
			SEMA3E
	Congenital sideroblastic anemia with immunodeficiency, fevers, and developmental delay (SIFD)	Congenital sideroblastic anemia with immunodeficiency, fevers, and developmental delay (SIFD)	TRNT1
	Defects of Vitamin B12 and Folate metabolism	Defects of Vitamin B12 and Folate metabolism	MTHFD1
			SLC46A1
			TCN2
	DiGeorge syndrome	DiGeorge syndrome	Del 10p
			Del 22q11.2
			TBX1
	DNA-breakage disorder	AT-like disorder	MRE11
		Ataxia telangiectasia (ATM)	ATM
		Bloom syndrome	BLM Helicase
			none
		DNA-ligase 1 ATP-dependent deficiency (LIG1)	LIG1
		Immunodeficiency centromeric instability facial anomalies syndrome (ICF)	CDCA7
			DNMT3B
			HELLS
			ZBTB24
		MCM4 deficiency	MCM4
		Nijmegen breakage syndrome (NBS1)	NBS1
		Other DNA-breakage disorder	none
		Post-Meiotic Segregation 2 (PMS2) deficiency	PMS2
		RNF168 deficiency	RNF168
		Seckel syndrome	none
	Dyskeratosis congenita	Dyskeratosis congenita	DKC1
			GAR1
			NAF1
			NHP2
			NOP10
			PARN
			RTEL1
			TERC
			TERT
			TINF2
		Hoyeraal-Hreidarsson syndrome	APOLLO (snm1b def)
			DKC1
			NHP2
			NOP10
			PARN
			RTEL1
			TERC
			TERT
			TINF2
	Fc receptor deficiencies	Fc receptor deficiencies	FCGR1A
			FCGR2A
			FCGR2B
			FCGR3A
			FCGR3B
			FCGRT
	FILS syndrome	Facial dysmorphism, immunodeficiency, livedo, and short stature (FILS syndrome)	POLE1

Main Category	Sub Category	IEI Diagnosis (PID Diagnosis)	Gene
Other well defined PIDs	**GINS1 deficiency**	GINS1 deficiency	GINS1
	Hyper IgE syndromes	Hyper IgE syndrome (HIES)	DOCK8
			IL6ST
			STAT3 DN
			Tyk2
			ZNF341
	IKAROS deficiency	IKAROS deficiency	IKAROS
	Immunodeficiencies with multiple intestinal atresias	Immunodeficiencies with multiple intestinal atresias	TTC7A
	MonoMAC (WILD)	Monocytopenia and mycobacterial infection (MonoMAC (WILD))	GATA2
	MYSM1 deficiency	MYSM1 deficiency	MYSM1
	Netherton syndrome	Netherton syndrome	SPINK5
	Roifman syndrome	Roifman syndrome	RNU4ATAC
	Schimke disease	Schimke disease	SMARCAL1
	Trichohepatoenteric syndrome (Giraud syndrome)	Trichohepatoenteric syndrome	SKIV2L
			TTC37
	Unclassified syndromic immunodeficiencies	Unclassified syndromic immunodeficiencies	none
	SIFD	Congenital sideroblastic anemia with immunodeficiency, fevers, and developmental delay (SIFD)	TNRT1
	VODI	Hepatic venoocclusive disease with immunodeficiency (VODI)	SP110
	Wiskott-Aldrich syndrome (WAS)	WIP deficiency	WIPF1
		Wiskott-Aldrich syndrome (WAS)	WAS
		X-linked thrombocytopenia with mutations in WAS	WAS

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Phagocytic disorders	**Actin beta deficiency**	Actin beta deficiency (ACTB)	Actin beta
	Barth syndrome	Barth syndrome	TAZ
	Chronic granulomatous disease (CGD)	Chronic granulomatous disease (CGD)	GP91-phox (CYBB)
			P22-phox (CYBA)
			CYBC1 EROS
			P40-phox (NCF4)
			P47-phox (NCF1)
			P67-phox (NCF2)
	Clericuzio-type poikiloderma with neutropenia syndrome	Clericuzio-type poikiloderma with neutropenia syndrome	C16orf57
	COHEN syndrome	COHEN syndrome	COH1
			VPS13B
	Congenital neutropenia	Congenital neutropenia	CSF3R
			ELA2
			G6PC3
			GATA2
			GFI1
			HAX1
			JAGN1
			P14
			SRP54
			WAS
		VPS45 deficiency (SCN5)	VPS45
	Cyclic neutropenia	Cyclic neutropenia	ELA2
	Defects of Motility	MKL1 deficiency	MKL1
	Defects with susceptibility to mycobacterial infection (MSMD)	Defects with susceptibility to mycobacterial infection (MSMD)	GP91-phox (CYBB)
			IFNGR1
			IFNGR2
			IL12B
			IL12R beta-1
			IL18
			IL23-alpha
			IRAK4
			IRF8
			ISG15
			SPPL2A
			STAT1
			STAT5
	Glycogen storage disease type 1b (GS1b)	Glycogen storage disease type 1b (GS1b)	G6PT1
	Leukocyte adhesion deficiency (LAD)	Leukocyte adhesion deficiency (LAD)	LAD1 / ITGB2
			LAD2 / FUCT1
			LAD3
	Localized juvenile peridontitis	Localized juvenile peridontitis	Formyl peptide receptor
	Myeloperoxidase deficiency (MPO)	Myeloperoxidase deficiency (MPO)	MPO
	Neutrophil glucose-6-phosphate dehydrogenase	Glucose-6-phosphate dehydrogenase deficiency (G6PD)	G6PD
	Papillon-Lefevre syndrome	Papillon-Lefevre syndrome	CTSC
	PID with partial albinism	Partial albinism and immunodeficiency syndrome	none
	Pulmonary alveolar proteinosis	Pulmonary alveolar proteinosis	CSF2RA
	RAC2-GTPase defect	RAS-related C3 Bolutinum toxin substrate 2 deficiency (RAC2)	RAC2
	Shwachman-Diamond-syndrome	Shwachman-Diamond-syndrome	DNAJC21
			EFL1
			SBDS
			SRP54
	Specific granule defect	CCAAT/enhancer binding protein epsilon deficiency (CEBPE)	CEBPE
	Unclassified phagocytic disorders	Unclassified phagocytic disorders	none

Main Category	Sub Category	IEI Diagnosis (PID Diagnosis)	Gene
Predominantly antibody disorders	**Agammaglobulinemias**	Agammaglobulinemia	BLNK/SLP65
			BTK
			CD79A
			CD79B
			IGHM
			IGLL1
			PIK3R1
			TCF3
	Class switch recombination defects (CSR) / HIGM syndromes	CSR defects and Hyper IgM (HIGM) syndromes	AID
			CD40 (TNFRSF5)
			CD40L (CD154)
			PMS2
			UNG
	Hypogammaglobulinemias	ADA2 Deficiency (DADA2)	CECR1
		ARHGEF1-deficiency	ARHGEF1
		Common variable immunodeficiency disorders (CVID)	ATP6AP1
			BACH2
			BAFFR
			CD19
			CD20
			CD21
			CD81
			CTLA-4
			ICOS
			IKZF1
			IRF2BP2
			LRBA
			MOGS
			NFKB1
			NFKB2
			PTEN
			TACI
			TRNT1
			TTC37
			TWEAK
		Deficiency of specific IgG (Specific antibody deficiency - SPAD)	none
			NFKBIA
		IgA with IgG subclass deficiency	TACI
		Immunoglobulin chain deficiencies	Heavy chain
			Kappa light chain
			Lambda light chain
		Isolated IgG subclass deficiency	BAFFR
			CD19
			CD21
			CD81
			ICOS
			TACI
		Other immunoglobulin gene deletions	none
		Selective IgA deficiency	TACI
		Selective IgM deficiency	none
		Thymoma with immunodeficiency	none
		Transcobalamin II deficiency	Transcobalamine II
		Transient hypogammaglobulinemia of infancy	none
	CVID Phenotype	NFKB1 deficiency	NFKB1
	Unclassified antibody deficiency	Unclassified antibody deficiency	none
Bone marrow failure	**Bone marrow failure**	BMFS2 (Hebo deficiency)	ERCC6L2
Unclassified Immunodeficiencies	**Unclassified immunodeficiencies**	Unclassified immunodeficiencies	none