ESID Online Registry: Diseases and Genes

as of 30.07.2015 - Please send ideas and questions to esid.registry@kenes.com

Main category	Sub category	PID Diagnosis	Gene
Autoinflammatory disorders	**Blau syndrome**	Caspase recruitment domain-containing protein 15 deficiency (CARD15)	CARD15
	CINCA syndrome	CINCA syndrome	CIAS1 (NLRP3)
	Familial cold autoinflammatory syndrome	Familial cold autoinflammatory syndrome	CIAS1 (NLRP3)
	Familial mediterranean fever	Familial mediterranean fever defect	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
			HOIL1
			IL1RN
			IL36RN
			LPIN2
			PLCG2
			PSMB8
			SH3BP2
			SLC29A3
	Periodic fever, unclassified	Periodic fever, unclassified	none
	PFAPA	Periodic fever aphtous stomatitis, pharyngitis and adenopathy (PFAPA)	none
	Pyogenic sterile arthritis pyoderma gangrenosum and acne	Proline/serine/threonine phosphatase-interacting protein 1 deficiency (PSTPIP1)	PSTPIP1
	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
			Gamma C (x-linked)
			IL21R
			IL7Ralpha
			JAK3
			PNP
			RAG1
			RAG2
			Reticular dysgenesia
	CD4-deficiency	Selective CD4 cell deficiency	CD4
	CD8-deficiency	CD8 deficiency	CD8A

Combined immunodeficiency (CID)	Combined immunodeficiency	Artemis
		CARD11
		Caspase 8
		CD3G
		Cernunnos
		CTLA-4
		DNA-PKcs
		DOCK8
		IKBKB
		IL21R
		IL2Ralpha
		ITK
		LCK
		LIG4
		MAGT1
		MALT1
		MSN (moesin)
		MST1 (STK4)
		Orai1 (TMEM142A)
		OX40
		PIK3CD (PI3K-delta)
		PIK3R1
		PRKDC
		PNP
		RECQL4 (Poikilodermia congenita)
		RhoH
		STAT5
		STAT5a
		STAT5b
		STIM1
		TRAC
		TTC7A
		UNC119D
		ZAP70
HLA class I deficiency	HLA class I deficiency	TAP1
		TAP2
		TAPBP
HLA class II deficiency	HLA class II deficiency	MHC2TA
		RFX5
		RFXANK
		RFXAP
NUDE/SCID	Winged-helix nude deficiency (FOXN1)	FOXN1

	Omenn syndrome	Omenn syndrome	ADA
			Artemis
			CD45
			Coronin-1A
			Del 22q11.2
			Gamma C (x-linked)
			IL21R
			IL2Ralpha
			IL7Ralpha
			JAK3
			RAG1
			RAG2
			Reticular dysgenesia
			RMRP
	Severe combined immunodeficiency (SCID)	Severe combined immunodeficiency (SCID)	ADA
			Artemis
			CD3d
			CD3e
			CD3z
			CD45
			Coronin-1A
			Del 22q11.2
			DNA-PKcs
			Gamma C (x-linked)
			IL21R
			IL7Ralpha
			JAK3
			LIG4
			RAG1
			RAG2
			Reticular dysgenesia
Complement deficiencies	**Complement deficiency**	Acquired angioedema	none
		C3b inactivator deficiency	C3b inactivator
		CD59 antigen P18-20 deficiency (CD59)	CD59
		Complement component 1 deficiency	C1Q-alpha
			C1Q-beta
			C1Q-gamma
			C1r
			C1s
			Q, subunit unknown
		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
		Ficolin3 deficiency	FCN3
		Hereditary Angioedema (C1inh)	C1 Inhibitor
		Mannan-binding lectin serine protease (MASP) deficiency	COLEC11
			MASP1
			MASP2
		Membrane Attack Complex Inhibitor (CD59) deficiency	CD59
		Membrane Cofactor Protein (CD46) deficiency	CD46
		Properdin P factor complement deficiency (PFC)	Properdin
		Thrombomodulin deficiency	THBD
	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
	Defects of TLR/NFkappa-B signalling	Defects of TLR/NFkappa-B signalling	IKK-gamma (NEMO, IKBKG)
			IRAK4
			MyD88
			NFKBIA
			TIRAP
	Epidermodysplasia verruciformis	Epidermodysplasia verruciformis	TMC6 (gene 1)
			TMC8 (gene 2)
	Herpetic encephalitis	Herpetic encephalitis (HSE)	TBK1
			TLR3
			TRAF3
			TRIF
			UNC93
	HOIL1 deficiency	HOIL1 deficiency	none
	Predisposition to several viral infection	Predisposition to several viral infection	MCM4
			STAT2
	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

Diseases of immune dysregulation	**Autoimmune lymphoproliferative syndrome (ALPS)**	Autoimmune lymphoproliferative syndrome (ALPS)	CARD11
			Caspase 10 (ALPS IIA)
			Caspase 8 (ALPS IIB)
			CD95 (germline - ALPS IA)
			CD95 (somatic - ALPS Im)
			FADD (TNFRSF6)
			FASL (CD178) (ALPS IB)
			PRKCD
		Ras associated lymphoproliferative disease (RALD)	K-Ras
			N-Ras
	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
	Hemophagocytic Lymphohistiocytosis (HLH)	CD27 deficiency	CD27
		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	none
	Type 1 interferonopathies	Type 1 interferonopathies	ACP5
			ADAR1
			RNASEH2A
			RNASEH2B
			RNASEH2C
			SAMHD1
			TREX1
	Early-onset multi-organ autoimmune disease	Early-onset multi-organ autoimmune disease	STAT3
	Unclassified disorders of immune dysregulation	Unclassified disorders of immune dysregulation	none

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)	TNRT1
	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
	DNA-breakage disorder	AT-like disorder	MRE11
		Ataxia telangiectasia (ATM)	ATM
		Bloom syndrome (RECQ2)	BLM Helicase
		DNA-ligase 1 ATP-dependent deficiency (LIG1)	LIG1
		Immunodeficiency centromeric instability facial anomalies syndrome (ICF)	DNMT3B
			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
			NHP2
			NOP10
			RTEL1
			TERC
			TERT
			TINF2
		Hoyeraal-Hreidarsson syndrome	APOLLO (snm1b def)
			DKC1
			NHP2
			NOP10
			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
	Hyper IgE syndromes	Hyper IgE syndrome (HIES)	DOCK8
			STAT3
			Tyk2
	IKAROS deficiency	IKAROS deficiency	IKAROS
	Immunodeficiencies with multiple intestinal atresias	Immunodeficiencies with multiple intestinal atresias	TTC7A
	MonoMAC	Monocytopenia and mycobacterial infection (MonoMAC)	GATA2
	MYSM1 deficiency	MYSM1 deficiency	MYSM1
	Netherton syndrome	Netherton syndrome	SPINK5
	Schimke disease	Schimke disease	SMARCAL1
	Trichohepatoenteric syndrome (Giraud syndrome)	Trichohepatoenteric syndrome	SKIV2L
			TTC37
	Unclassified syndromic immunodeficiencies	Unclassified syndromic immunodeficiencies	none
	VODI	Hepatic venoocclusive disease with immunodeficiency (VODI)	SP110
	WILD syndrome	Warts, immunodeficiency, lymphedema, dysplasia (WILD) syndrome	none
	Wiskott-Aldrich syndrome (WAS)	WIP deficiency	WIPF1
		Wiskott-Aldrich syndrome (WAS)	WASP (x-linked)
		X-linked thrombocytopenia with mutations in WASP	WASP (x-linked)

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)
			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
	Congenital neutropenia	Congenital neutropenia	CSF3R
			ELA2
			G6PC3
			GATA2
			GFI1
			HAX1
			JAGN1
			P14
			WASP (x-linked)
	Cyclic neutropenia	Cyclic neutropenia	ELA2
	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
			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	SBDS
	Specific granule defect	CCAAT/enhancer binding protein epsilon deficiency (CEBPE)	CEBPE
	Unclassified phagocytic disorders	Unclassified phagocytic disorders	none

Predominantly antibody disorders	**Agammaglobulinemias**	Agammaglobulinemia	BLNK/SLP65
			Btk. (x-linked)
			CD79A
			CD79B
			IGHM
			IGLL1
			LRRC8
			PIK3R1
			TCF3
	Class switch recombination defects (CSR) / HIGM syndromes	CSR defects and Hyper IgM (HIGM) syndromes	AID
			CD40 (TNFRSF5)
			CD40L (CD154)
			PMS2
			UNG
	Hypogammaglobulinemias	Common variable immunodeficiency disorders (CVID)	BAFFR
			CD19
			CD20
			CD21
			CD81
			ICOS
			LRBA
			NFKB2
			TACI
			TWEAK
		Deficiency of specific IgG (Specific antibody deficiency - SPAD)	none
		Dystrophia myotonica	ZNF9
		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
	Unclassified antibody deficiency	Unclassified antibody deficiency	none
			11q23
			LRBA
			PIK3CD (PI3K-delta)
			PRKCD
Unclassified Immunodeficiencies	**Unclassified immunodeficiencies**	Unclassified immunodeficiencies	none