Diseases in the ESID Online Database

as of Nov 16, 2011 - please send ideas and questions to registry@esid.org

Main Category	Sub-category	Sub-registry	Mutation Entries
Predominantly antibody disorders	**Agammaglobulinemias**	Agammaglobulinemia	Btk. (x-linked), BLNK/SLP65, CD79A, IGLL1, IGHM, LRRC8, none/unknown
	Class switch recombination defects (CSR) / HIGM syndromes	Activation-induced cytidine deaminase deficiency (AID)	AID
		CD40 deficiency (TNFRSF5)	CD40 (TNFRSF5)
		CD40 ligand deficiency (CD154)	CD40L (CD154)
		CSR defects and HIGM syndromes with unknown genetic cause	none/unknown
		PMS2 deficiency (HIGM phenotype)	PMS2
		Uracil-DNA glycosylase deficiency (UNG)	UNG
	Hypogammaglobulinemias	Common variable immunodeficiency (CVID)	BAFFR, CD19, CD21, CD81, ICOS, TACI, none/unknown
		Deficiency of specific IgG	none/unknown
		Dystrophia myotonica type2 (PROMM/ZNF9)	ZNF9
		IgA with IgG subclass deficiency	TACI, none/unknown
		Immunoglobulin chain deficiencies	Heavy chain, Kappa light chain, Lambda light chain
		Isolated IgG subclass deficiency	BAFFR, CD19, CD21, CD81, ICOS, TACI, none/unknown
		Other Hypogammaglobulinemias	none/unknown, 11q23
		Other immunoglobulin gene deletions	none/unknown
		Secondary hypogammaglobulinemia	none/unknown
		Secondary selective IgA deficiency	none/unknown
		Selective IgA deficiency	TACI, none/unknown
		Selective IgM deficiency	none/unknown
		Thymoma with immunodeficiency	none/unknown
		Transcobalamin II deficiency	Transcobalamine II
		Transient hypogammaglobulinemia of infancy	none/unknown

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Predominantly T-cell deficiencies	**CD4-deficiency**	Selective CD4 cell deficiency	CD4
	CD8-deficiency	CD 8 deficiency	CD8A
	Combined immunodeficiency (CID)	Atypical Severe Combined Immunodeficiency (Atypical SCID)	none/unknown
		Calcium channel dysfunction	Orai1 (TMEM142A), STIM1
		Caspase 8 deficiency (T-cell deficiency)	Caspase 8
		CD3 gamma deficiency	CD3G, none/unknown
		Cernunnos/XLF deficiency	Cernunnos
		DNA-ligase 4 ATP-dependent deficiency (LIG4)	LIG4
		DOCK8 deficiency	DOCK8
		Interleukin 2 receptor alpha deficiency (CD25) (T-cell deficiency)	IL2Ralpha
		ITK deficiency	ITK
		Nucleoside phosphorylase deficiency (PNP)	PNP
		Signal transducer and activator of transcription 5 deficiency (STAT5)	STAT5
	HLA class I deficiency	HLA class I deficiency	TAP1, TAP2, TAPBP, none/unknown
	HLA class II deficiency	HLA class II deficiency	MHC2TA, RFX5, RFXANK, RFXAP, none/unknown
	NUDE/SCID	Winged-helix nude deficiency (FOXN1)	FOXN1
	Omenn syndrome	Omenn syndrome	ADA, Artemis, CD45, Del 22q11.2, Gamma C (x-linked), IL2Ralpha, IL7Ralpha, JAK3, RAG1, RAG2, RMRP, Reticular dysgenesia, none/unknown
	Other unclassified T-cell disorders	Other unclassified T-cell disorders	RECQL4 (Poikilodermia congenita), none/unknown
	Other unclassified T-cell disorders	Secondary T-cell deficiency	none/unknown
	T-B- Severe combined immunodeficiency (SCID)	T-B- Severe combined immunodeficiency (SCID)	ADA, Artemis, LIG4, RAG1, RAG2, Reticular dysgenesia (AP2), none/unknown
	T-B+ Severe combined immunodeficiency (SCID)	T-B+ Severe combined immunodeficiency (SCID)	IL7Ralpha, JAK3, CD45, Gamma C (x-linked), CD3D, CD3E, CD3Z, none/unknown
	T+B+ Combined immunodeficiency (CID)	Atypical Combined Immunodeficiency	ADA, Artemis, Gamma C (x-linked), IL7Ralpha, JAK3, PNP, RAG1, RAG2, Reticular dysgenesia
	ZAP deficiency	Zeta-chain-associated protein kinase deficiency (ZAP70)	ZAP70

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Phagocytic disorders	**Actin beta deficiency**	Actin beta deficiency (ACTB)	Actin beta
	Barth syndrome	Barth syndrome	TAZ, none/unknown
	Chronic granulomatous disease (CGD)	Chronic granulomatous disease (CGD)	GP91-phox(CYBB), P22-phox (CYBA), P47-phox (NCF1), P67-phox (NCF2), P40-phox (NCF4), none/unknown
	Clericuzio-type poikiloderma with neutropenia syndrome	Clericuzio-type poikiloderma with neutropenia syndrome	C16orf57, none/unknown
	COHEN syndrome	COHEN syndrome	COH1, none/unknown
	Cyclic neutropenia	Cyclic neutropenia	ELA2, none/unknown
	Defects with susceptibility to mycobacterial infection (MSMD)	Defects with susceptibility to mycobacterial infection (MSMD)	IFNGR1, IFNGR2, IL12R beta-1, IL12B, IL18, IL23-alpha, IRAK4, STAT1, none/unknown
	Glycogen storage disease type 1b (GS1b)	Glycogen storage disease type 1b (GS1b)	G6PT1, none/unknown
	Leukocyte adhesion deficiency (LAD)	Leukocyte adhesion deficiency (LAD)	LAD2 / FUCT1, LAD1 / ITGB2, LAD3
	Localized juvenile peridontitis	Localized juvenile peridontitis	Formyl peptide receptor, none/unknown
	Myeloperoxidase deficiency (MPO)	Myeloperoxidase deficiency (MPO)	MPO
	Neutrophil glucose-6-phosphate dehydrogenase	Glucose-6-phosphate dehydrogenase deficiency (G6PD)	G6PD
	Other phagocytic disorders	Other phagocytic disorders	none/unknown
	Papillon-Lefevre syndrome	Papillon-Lefevre syndrome	CTSC, none/unknown
	PID with partial albinism	Partial albinism and immunodeficiency syndrome	none/unknown
	RAC2-GTPase defect	RAS-related C3 Bolutinum toxin substrate 2 deficiency (RAC2)	RAC2
	Severe congenital neutropenia	Severe congenital neutropenia	CSF3R, GFI1, ELA2, WASP (x-linked), HAX1, G6PC3, P14, none/unknown
	Shwachman-Diamond-syndrome	Shwachman-Diamond-syndrome	SBDS, none/unknown
	Specific granule defect	CCAAT/enhancer binding protein epsilon deficiency (CEBPE)	CEBPE

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Complement deficiencies	**Complement deficiency**	C3b inactivator deficiency	C3b inactivator
		CD59 antigen P18-20 deficiency (CD59)	CD59
		Complement component 1 deficiency	C1Q-alpha, C1Q-beta, C1Q-gamma, C1Q, subunit unknown, C1r, C1s
		Complement component 2 deficiency	C2
		Complement component 3 deficiency (C3)	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
		Hereditary Angioedema (C1inh)	C1 Inhibitor
		I Factor deficiency (IF)	Factor I
		Mannan-binding lectin serine protease 2 deficiency (MASP2)	MASP2
		Properdin P factor complement deficiency (PFC)	Properdin
	Mannose-binding lectin (MBL)	Mannose-binding lectin deficiency (MBL)	MBL

Autoinflammatory syndromes	**Blau syndrome**	Caspase recruitment domain-containing protein 15 deficiency (CARD15)	CARD15
	CINCA syndrome	CINCA syndrome	CIAS1
	Familial cold autoinflammatory syndrome	Familial cold autoinflammatory syndrome	CIAS1
	Familial mediterranean fever	Familial mediterranean fever defect	MEFV, none/unknown
	Familial periodic fever	Hyper IgD syndrome (MVK)	MVK
		TNF-receptor associated periodic fever syndrome (TRAPS)	TNFRSF1A
	Muckle-Wells syndrome	Muckle-Wells syndrome	CIAS1
	PFAPA	Periodic fever aphtous stomatitis, pharyngitis and adenopathy (PFAPA)	none/unknown
	Pyogenic sterile arthritis pyoderma gangrenosum and acne	Proline/serine/threonine phosphatase-interacting protein 1 deficiency (PSTPIP1)	PSTPIP1

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Other well defined PIDs	**Asplenia syndrome**	Asplenia syndrome (Ivemark syndrome)	none/unknown
		Isolated congenital asplenia	none/unknown
	Cartilage hair hypoplasia	Cartilage hair hypoplasia	RMRP, none/unknown
	CHARGE syndrome	CHARGE syndrome	CHARGE-CHD7
	DiGeorge syndrome	DiGeorge syndrome	Del 22q11.2, Del 10p, none/unknown
	DNA-breakage disorder	AT-like disorder	MRE11
		Ataxia telangiectasia (ATM)	ATM, none/unknown
		Bloom syndrome (RECQ2)	BLM Helicase, not confirmed yet
		DNA-ligase 1 ATP-dependent deficiency (LIG1)	LIG1
		Fanconi anemia	none/unknown
		Immunodeficiency centromeric instability facial anomalies syndrome (ICF)	DNMT3B, ZBTB24, none/unknown
		Nijmegen breakage syndrome (NBS1)	NBS1
		Other DNA-breakage disorder	none/unknown
		Post-Meiotic Segregation 2 (PMS2) deficiency	PMS2
		Seckel syndrome	none/unknown
	Dyskaterosis congenita	Dyskaterosis congenita	DKC1, NOP10, TERC, TERT, TINF2, unknown
		Hoyeraal-Hreidarsson syndrome	DKC1, NOP10, TERC, TERT, TINF2, APOLLO (snm1b def), unknown
	Fc receptor deficiencies	Fc receptor deficiencies	FCGR1A, FCGR3A, FCGR3B, FCGR2A, FCGR2B, FCGRT
	Hyper IgE syndromes	Hyper IgE syndrome (HIES)	STAT3, DOCK8, Tyk2, none/unknown
	MonoMAC	Monocytopenia and mycobacterial infection (MonoMAC)	GATA2, none/unknown
	Netherton syndrome	Netherton syndrome	SPINK5, none/unknown
	Osteopetrosis	Osteopetrosis	TCIRG1, CLCN7, CA2, OSTM1, none/unknown
	Other syndromic PID	Other syndromic PID	unknown
	Schimke disease	Schimke disease	SMARCAL1, none/unknown
	Trichohepatoenteric syndrome (Giraud syndrome)	Trichohepatoenteric syndrome	TTC37
	VODI	Hepatic venoocclusive disease with immunodeficiency (VODI)	SP110
	WILD syndrome	Warts, immunodeficiency, lymphedema, dysplasia (WILD) syndrome	none/unknown
	Wiskott-Aldrich syndrome (WAS)	Wiskott-Aldrich syndrome (WAS)	WASP (x-linked), none/unknown
		X-linked thrombocytopenia with mutations in WASP	WASP (x-linked)

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Autoimmune and immunedysregulation syndromes	**Autoimmune lymphoproliferative syndrome (ALPS)**	Autoimmune lymphoproliferative syndrome (ALPS)	Caspase 10 (ALPS IIA), Caspase 8 (ALPS IIB), CD95 (somatic – ALPS Im), FASL (CD178) (ALPS IB), CD95 (germline – ALPS IA), FADD (TNFRSF6), none/unknown
		Ras associated lymphoproliferative disease (RALD)	N-Ras, K-Ras
	Autoimmune polyendocrinopathy candidiasis ectodermal dystrophy (APECED)	Autoimmune polyendocrinopathy candidiasis ectodermal dystrophy (APECED)	AIRE, none/unknown
	Early-onset inflammatory bowel disease	Early-onset inflammatory bowel disease	ILR10, ILR10 receptor alpha chain, ILR10 receptor beta chain, none/unknown
	Hemophagocytic Lymphohistiocytosis (HLH)	Chediak Higashi syndrome	LYST, none/unknown
		Familial hemophagocytic lymphohistiocytosis syndromes (FHLH)	UNC13D, STXBP2, PRF1, STX11, none/unknown
		Griscelli syndrome	MYO5A, RAB27A, MLPH, none/unknown
		Hermansky-Pudlak syndrome	AP3B1, none/unknown
		ITK deficiency (HLH phenotype)	ITK
		X-linked lymphoproliferative syndrome (XLP)	BIRC4/XIAP, SH2D1A,
	IPEX	FOXP3 deficiency (IPEX)	FOXP3, none/unknown
		Interleukin 2 receptor alpha deficiency (CD25) (IPEX phenotype)	IL2Ralpha
		IPEX-like disease	none/unknown
	Other autoimmune and immundysregulation syndromes	Other autoimmune and immundysregulation syndromes	unknown
Defects in innate immunity	**Chronic mucocutaneous candidiasis (CMC)**	Chronic mucocutaneous candidiasis (CMC)	AIRE, none/unknown, IL-17 receptor alpha, IL-17 F, STAT1
	Defects of TLR/NFkappa-B signalling	Defects of TLR/NFkappa-B signalling	IRAK4, NFKBIA, IKK-gamma, MyD88, none/unknown
	Epidermodysplasia verruciformis	Epidermodysplasia verruciformis	TMC6, TMC8, none/unknown
	Herpetic encephalitis	Herpetic encephalitis	TLR3, TRAF3, TRIF, UNC93, none/unknown
	Warts hypogammaglobulinemia infections and myelokathexis (WHIM)	Warts hypogammaglobulinemia infections and myelokathexis (WHIM)	CXCR4, none/unknown