



The global organisation working to improve the quality of life for people
with primary immunodeficiencies.

PRIMARY IMMUNODEFICIENCY GLOSSARY

The purpose of this glossary is to give primary immunodeficiency patients a possibility to understand the contents of a diagnosis and the instructions given in connection with treatment and care.

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Acquired immune deficiency		An immune deficiency acquired during a person's lifetime. Can be caused by e.g.: an infection, medication or radiation.
Acquired immune deficiency syndrome	AIDS	A secondary immunodeficiency caused by the HIV Virus.
Acute:		A descriptive term used to describe an illness which is usually short in duration and of recent onset.
Adenosine Deaminase	ADA	An enzyme essential for the development of the immune system.
Agammaglobulinemia		An almost total lack of immunoglobulin or antibodies.
Amniocentesis:		The withdraw of amniotic fluid surrounding a fetus in order to perform prenatal genetic testing.
Androgen:		A male sex hormone.
Anemia:		A condition in which the blood is deficient in red blood cells, in hemoglobin, or in total volume.
Antibodies:		Protein molecules that are produced and secreted by plasma cells, after differentiation of the B-lymphocytes (B-cells) in response to stimulation by an antigen; their primary function is to fight bacteria, viruses, toxins, and other substances foreign to the body.
Aspergillus:		A kind of fungi which includes many common molds.
Antigen:		Any foreign substance that provokes an immune response when introduced into the body; the immune response usually involves both T-lymphocytes and B-lymphocytes.
Ataxia		An unsteady gait caused by neurological abnormalities.
Autoantibody		An immunoglobulin, which reacts against the person's own tissue
Autoimmune disease		A disease that results when the body's immune system reacts against a person's own tissue.
Autosomal recessive inheritance		A form of inheritance where the characteristic, or disease, is inherited from both parents.
Autosomes		Any chromosome other than the sex chromosome.
Bacteria		Single cell organisms (microorganisms) that can be seen only under a microscope. While bacteria can be useful, many bacteria can cause disease in humans.
Batch release testing		Testing of end products by regulatory authorities before official release to ensure that the product specification is met.
B-lymphocytes (B-cells):		White blood cells of the immune system derived from bone marrow and involved in the production of antibodies.
Blood bank		Hospital unit, which provides all aspects of blood transfusion services, incl. delivery of blood components of high international standards 24 hours a day.
Blood serum		Is blood plasma without fibrinogen or the other clotting factors (i.e., whole blood minus both the cells <i>and</i> the clotting factors)
Blood plasma		Is the yellow liquid component of blood, in which the blood cells in whole blood would normally be suspended. It makes up about 55% of the total blood volume. It is the <i>intravascular fluid</i> part of extracellular fluid. It is mostly water (90% by volume) and contains dissolved proteins, glucose, clotting factors, mineral ions, hormones and carbon dioxide (plasma being the main medium for excretory product lasmaphetation).
Blood products		In order to provide maximum benefit from each blood donation and to extend shelf-life, blood banks fractionate some whole blood into several products. The most common of these products are packed Red Blood Cells (RBC), plasma, platelets, cryoprecipitate, and fresh frozen plasma (FFP). FFP is quick-frozen to retain the labile clotting factors V and VIII.
Bone marrow		Soft tissue located in the hollow centers of most bones that contain developing red blood cells, white cells, platelets and cells of the immune system.
Bone marrow transplantation	BMT	A treatment by the means of which bone marrow from one person is transplanted to another person
Bronchiectasis		A dilation of the tubes (bronchi) leading to the air sacs of the lung; usually the consequence of recurrent infection.
Carrier detection		The detection of a genetic characteristic in a person who carries the characteristic (or disease) in their genes but shows no clinical evidence.

CD 40 ligand		A protein found on the surface of T-lymphocytes; individuals with X-linked hyper IgM syndrome have a deficiency in this protein.
Characterization		Analytical measurements which allow detailed understanding of the composition and other attributes of a product.
Cellular immunity		Immune protection provided by the direct action mainly of the T- and NK-cells.
Chemokine		Polypeptides(chains of aminoacids) controlling the activities of the leucocytes
Chromosomes		Physical structures in the cell's nucleus that carry genes; each human cells has 23 pairs of chromosomes.
Chronic		Descriptive term used to describe an illness or infection that may be recurrent or last a long time.
Chorionic villus sampling	CVS	Involves the retrieval of a sample of the developing placenta from the womb in order to perform prenatal genetic testing.
Clinical immunology		Comprises: <u>Blood bank activities</u> to include all aspects of blood transfusion services. <u>Tissue typing activities</u> to include HLA class I and class II typing, pre- and post transplant for BMT, HPA platelet gene typing and anti-platelet antibody screening and specificity detection, immunodeficiency screening and diagnosis, monitoring of treatment, leukemia diagnosis and monitoring of treatment.
Combined immunodeficiency		Immunodeficiency when both T- and B-lymphocytes cells are inadequate or lacking.
Complement		A complex series of blood proteins that act in a definite sequence to affect the destruction of bacteria, viruses and fungi.
Complete blood count		A blood count that includes separate counts for red and white blood cells.
Compensated/ remunerated donor		A donor is considered compensated/remunerated if he/she receives any kind of compensation for travel, work time, inconvenience or gift as payment for his/her blood donation, even if the donation is done voluntarily
Congenital		Present at birth
Consanguineous		Descended from the same family or ancestors.
Cord blood		Blood obtained from the placenta at birth.
Cryptosporidium		An organism that can cause gastrointestinal symptoms and liver disease; may be present in drinking water.
Cytokines		Proteins secreted by cells of the lymph system that affects the activity of other cells and is important in controlling inflammatory responses. Interleukins and interferons are cytokines.
Database		A body of information stored in a computer, and from which particular pieces of information can be retrieved when required
Deoxyribonucleic acid	DNA	The carrier of genetic information found in the cell nucleus.
Donor screening		Individual donations of blood are screened to ensure that blood-borne viruses do not enter the plasma pool. Screening is currently available for HBV, HCV and HIV
Donor selection		Procedures designed to identify and exclude donors at risk of being infected with viruses that can be transmitted by blood transfusion
Eczema		Skin inflammation with redness, itching, encrustations, and scaling.
Endocrine system		A series of glands in the body that produce hormones.
Enveloped/lipid enveloped viruses		The common transfusion transmitted viruses HIV, HCV and HBV, which are all characterized by a lipid viral enveloped and are highly infectious
Enzyme		A protein facilitating chemical reactions
Eosinophilia		An increase in the number of granular white blood cells that stain with the dye eosin, occurring in some allergies and parasitic diseases.
Finished product testing		Testing done on final product to allow manufacturers to characterize their products and to demonstrate compliance of every batch with the licensed specification
Fractionation		The process of separating and processing human blood plasma into a range of products for therapeutic use
Fungus		Member of a class of relatively primitive microorganisms including mushrooms, yeast, and molds.
Gamma globulins		The protein fraction of blood that contains immunoglobulins or

		antibodies.
Gamma interferon		A cytokine primarily produced by T-lymphocytes that improves bacterial killing by phagocytes; used as treatment for chronic granulomatous disease.
Gene		A unit of genetic material (DNA).
Gene (or genetic) testing		Testing performed to determine if an individual possesses a specific gene or genetic trait.
Gene therapy		Treatment of genetic diseases by providing the correct or normal form of the abnormal gene causing the disease.
Graft-versus-host disease	GVHD	A reaction in which transplanted immunocompetent cells attack the tissue of the recipient.
Graft rejection		The immunologic response of the recipient to the transplanted organ or tissue resulting in rejection of the transplanted organ or tissue.
Granulocyte		A white cell of the immune system characterized by the ability to ingest (phagocytize) foreign material; neutrophils, eosinophils, and basophils are examples of granulocytes.
Granulocyte colony-stimulating factor	G-CSF	A cytokine which stimulates proliferation, development and function of granulocytes
Granulocyte-macrophage colony-stimulating factor	GM-CSF	A cytokine stimulating proliferation, development and function of granulocytes and macrophages.
Good manufacturing practices	GMPs	All the elements in established practice that will collectively lead to final products that consistently meet expected requirements as reflected in product specification. These include traceability, segregation of manufacturing steps to avoid cross-contamination, training, documentation, change control, deviation reporting
Haplotype		A series of gene clusters on the sixth human chromosome that determines histocompatibility antigens (tissue type), inherited from one parent.
Helper lymphocytes (Helper T-cells):		A subset of T-lymphocytes that help B-lymphocytes and T-lymphocytes to function more optimally.
Histocompatibility antigens		Chemicals on the surface of many cells of the body, including the cells of the immune system, which are relatively unique to each individual and are responsible for our tissue type.
HLA-system		The human tissue type system. Divided into two classes: Class I (HLA-A, -B, -C) and class II (HLA-DQ, -DP, -DR)
Humoral immunity		Immune protection provided by soluble factors, such as antibodies, which circulate in the body's fluids.
Hypogammaglobulinemia		Lower than normal levels of gamma globulins or immunoglobulins (or antibodies) in the blood.
Hypo-		Under normal
Hypoplasia		The failure of an organ or body part to grow or develop fully.
Inventory hold		The retention in storage of plasma for fractionation while processes designed to assure donor safety are undertaken
Immunglobulin A	IgA	An immunoglobulin found in blood and secreted into tears, saliva, and on the mucous membranes of respiratory and intestinal tracks.
Immunglobulin D	IgD	An immunoglobulin whose function is poorly understood at this time.
Immunglobulin E	IgE	An immunoglobulin found in trace amounts in the blood and lasmaphble, when in excess, for allergic reactions
Immunglobulin G	IgG	The most abundant and common of the immunoglobulins. IgG functions mainly against bacteria and some viruses. It is the only antibody that can cross the placenta from the mother to the developing fetus.
Immunglobulin M	IgM	An immunoglobulin found in the blood. IgM functions in much the same way as IgG but is formed earlier in the immune response. It is also very efficient in activating complement.
Immune response		The response of the immune system against foreign substances.
Immunocompetent:		Capable of developing an immune response.
Immunodeficiency	ID	A state of either a congenital (present at birth) or an acquired abnormality of the immune system that prevents adequate immune responsiveness.
Immunoglobulin		The intravenous or subcutaneous injection of immunoglobulin.

replacement therapy		
Immunoglobulins	Ig	Another name for antibody; there are five classes: IgA, IgD, IgG, IgM, and IgE.
Incidence		The frequency with which something, such as a disease, appears in a particular population or area. In disease epidemiology, the incidence is the number of newly diagnosed cases during a specific time period. The incidence is distinct from the prevalence which refers to the number of cases alive on a certain date
Incubation period		The period between the infection of an individual by a pathogen and the manifestation of the disease it causes.
Inducible T-cell co-stimulator	ICOS	A protein that in humans is encoded by the <i>ICOS</i> gene. CD278 or ICOS (Inducible T-cell CO Stimulator) is a CD28-superfamily costimulatory molecule that is expressed on activated T cells. It is thought to be important for Th2 cells in particular. The protein encoded by this gene belongs to the CD28 and CTLA-4 cell-surface receptor family. It forms homodimers and plays an important role in cell-cell signaling, immune responses, and regulation of cell proliferation.
In vitro		Outside of a living environment; refers to a process or study taking place in test tubes, etc.
In vivo		Inside a living environment; refers to a process or study taking place in the body.
Infection		Disease caused by a pathogen
Inflammation		Heat of a part of the body, with pain, redness and swelling
Interleukin	IL	Subset of cytokine, acts as a signaling protein. Mainly produced by the T cells and macrophages
Interferon	INF	Subset of cytokine
Intravenous immunoglobulin infusion	IVIG	Immunoglobulin (gamma globulin) therapy injected directly into the vein.
Killer lymphocytes		T-lymphocytes that directly kill microorganisms or cells that are infected with microorganisms.
Labile blood components		Products that are extracted from donated blood, either directly or in one or few manufacturing steps, and which quickly lose their therapeutic potential. (E.g. cell preparations and plasma).
Leukemia		Type of cancer affecting the cells of the immune system.
Leukocyte (white blood cell)		Group of small colorless blood cells that play a major role in the body's immune system. There are five basic leukocytes: monocytes, lymphocytes, neutrophils, eosinophils, and basophils.
Limit testing		Testing of the plasma pool using NAT in which a maximum level of viral contamination, rather than an absolute elimination, is the aim
Live vaccines		Live viruses are used in the vaccine; live vaccines (particularly oral polio) can transmit the disease they were designed to prevent in immunocompromised individuals.
Lymph		Fluid made up of various components of the immune system that flows throughout tissues of the body via the lymph nodes and lymphatic vessels.
Lymph nodes		Small bean-sized organs of the immune system, distributed widely throughout the body. Each lymph node contains a variety of specialized compartments that house B-lymphocytes, T-lymphocytes, and macrophages. Lymph nodes unite in one location the several factors needed to produce an immune response.
Lymphocytes		Small white cells, normally present in the blood and in lymphoid tissue, that bear the major responsibility for carrying out the functions of the immune system. There are two major forms of lymphocytes, B-lymphocytes, and T-lymphocytes, which have distinct but related functions in generating an immune response.
Lymphoma		Type of cancer of the lymphocytes of the immune system.
Lyophilization		The process of isolating a solid substance from solution by freezing the solution and evaporating the ice under vacuum. Freeze-drying
Macrophages		A phagocytic tissue cell of the immune system that functions in the destruction of foreign antigens (as bacteria and viruses), and serves as an antigen-presenting cell.

Major histocompatibility complex	MHC	A series of genes on chromosome 6 that direct the synthesis of the chemicals on the surface of many cells of the body, including the cells of the immune system, which are relatively unique to each individual and provide our tissue type.
Malignancy		Cancer.
Marketing authorization		The formal permit from a regulatory authority allowing a manufacturer to market a product following that authority's scrutiny
Metabolism		A general term which summarizes the chemical changes within a single cell, and the body as a whole, which results in either the building up or breaking down of living material.
Microbes		Very small living organisms. Normally single cells. Includes bacteria, protozoa and fungi
Microorganisms		Minute living organisms, usually one-cell organisms, which include bacteria, protozoa, and fungi.
Minipools		Plasma samples pooled from several donations, and then tested for viral markers
Molecules		The smallest unit of matter of an element or compound.
Monocyte		Phagocytic cell found in the blood that acts as a scavenger, capable of destroying invading bacteria or other foreign material; these cells develop into macrophages in tissues.
Monokines		Chemical messengers produced and secreted by monocytes and macrophages.
Mucosal surfaces		Surfaces that come in close contact with the environment, such as the mucus membranes of the mouth, nose, gastrointestinal tract, eyes, etc; IgA antibodies protect these surfaces, or mucus membranes, from infection.
Nanofiltration		A process whereby protein solutions are passed over small pore filters which can remove viruses while allowing therapeutic proteins to pass through
Neurology		A branch of medicine concerned especially with the structure, functions, and diseases of the nervous system.
Neutropenia		A lower than normal amount of neutrophils in the blood.
Neutrophils		A type of granulocyte, found in the blood and tissues that can ingest microorganisms.
NK cell		Natural killer cell
Non-compensated/ Non-remunerated voluntary donor		A donor is considered voluntary and non-remunerated if the person gives blood, blood plasma or cellular components of his/her own free will and receives no payment for it, either in the form of cash, or in some kind which would be considered a money substitute
Non-enveloped/non-lipid enveloped viruses		Pathogenic viruses (for example, HAV or parvovirus B19) which lack a lipid envelope and therefore are not susceptible to viral inactivation techniques such as solvent-detergent treatment
Nucleic acid testing	NAT	Testing for viral nucleic acid, thus allowing detection of a virus which may cause disease before the development of immunological markers of infection
Nystagmus		Involuntary usually rapid movement of the eyeballs.
Opportunistic infection		An infection that occurs only under certain conditions, such as in immunodeficient individuals.
Opsonin		Antibodies and complement factors, which bind microbes to phagocytes
Organism		An individual living thing.
Osteomyelitis		Infection of the bone.
Parasite		A plant or animal that lives, grows, and feeds on or within another living organism.
Parathyroid gland		Small glands found in the neck near the thyroid that control the normal metabolism and blood levels of calcium.
Patientlist		A list of information on patients either in a written form or in an electronic form
Petechiae		Pinhead-sized red spots resulting from bleeding into the skin.
Phagocyte		A general class of white blood cells that ingest microbes and other cells and foreign particles; monocytes, macrophages, and neutrophils are types of phagocytes.

Pharmacokinetics		The action of drugs in the body over a period of time, including the processes of absorption, localization in tissues, biotransformation and excretion
Plasma cells		Antibody-producing cells descended from B-lymphocytes.
Plasmapheresis		Is a medical technology in which the blood of a donor or patient is passed through an apparatus that separates out the plasma and returns the remainder to the circulation. It is thus an extracorporeal therapy
Plasma master file	PMF	A dossier of information compiled according to European guidelines, which allows the manufacturer of plasma derivatives to fully describe the source material
Plasma pool		Plasma from a number of donors to be used to make one lot of products
Plasma Proteins		Plasma contains a large variety of proteins including albumin, immunoglobulins, and clotting proteins such as fibrinogen. Albumin constitutes about 60% of the total protein in plasma and is present at concentrations between 35 and 55 mg/mL. It is the main contributor to osmotic pressure of the blood and it functions as a carrier molecule for molecules with low water solubility such as lipid soluble hormones, enzymes, fatty acids, metal ions, and pharmaceutical compounds. Albumin is structurally stable due to its seventeen disulfide bonds and unique in that it has the highest water solubility and the lowest isoelectric point (pI) of the plasma proteins. Due to the structural integrity of albumin it remains stable under conditions where most other proteins denature.
Plasma Components/ Plasma derived products		Factor VIII Factor IX complex immunoglobulin antithrombin III alpha-1-antitrypsin
Platelets		Smallest and most fragile of the blood cells; primary function is associated with the process of blood clotting.
PLUS		Plasma Users: A cooperation between EHC, WFH, IPOPI, Alpha 1 Europe, GBS/CIDP, HAEI, ITP the members of which are depending on treatment with plasma derived products
Polymorphism		The quality or state of existing in or assuming different forms.
Polysaccharides		Complex sugars.
Polyethylene glycol- adenosine deaminase	PEG- ADA	A replacement enzyme, which is able to replace certain immune functions
Polymerase Chain Reaction	PCR	A technique for amplifying DNA sequences in vitro by separating the DNA into two strands and incubating it with oligonucleotide primers and DNA polymerase. It can amplify a specific sequence of DNA by as many as one billion times and is important in biotechnology, forensics, medicine, and genetic research.
Polypeptides		Chains of amino acids
Potency		The biological activity which may be measured in the laboratory which is best related to a product's actual therapeutic effect
Prevalence		The proportion of individuals in a population having a disease. Prevalence is a statistical concept referring to the number of cases of a disease that are present in a particular population at a given time.
Primary immunodeficiency	PI	Immunodeficiency that is congenital, not due to another illness, medication or outside agent damaging the immune system.
Products specification		The properties of a product. They can be measured in the laboratory, allowing a manufacturer to assess and demonstrate fitness of purpose
Prophylactic		Medical therapy initiated to prevent or guard against disease or infection.
Protein		A class of chemicals found in the body made up of chains of amino acids (building blocks); immunoglobulins (antibodies) are proteins.
Protozoa		A small parasite
Purity		The proportion of the desired ingredient (e.g. factor VIII) in concentrates, relative to other ingredients present
Quality assurance system		A mechanism for achieving, sustaining, and improving product quality
Recovered plasma		Plasma taken from fractionated whole blood

Recurrent infections		Infections, such as otitis, sinusitis, pneumonia, deep-seated abscess, osteomyelitis, bacteremia or meningitis that occur repeatedly.
Registry		Registration; an office or place where a register is kept
Register		Written record or official list regularly kept
Secondary immunodeficiency		Immunodeficiency due to another illness or agent, such as human immunodeficiency virus (HIV), cancer, or chemotherapy.
Sepsis		An infection of the blood.
Sex chromosomes		Two chromosomes: X and Y. XX for female and XY for male
Shelf life		The period of time during which a products may be stored under specified conditions and retain its characteristics
Source plasma		The fluid portion of human blood collected by plasmapheresis and intended as source material for further manufacturing use.
Spleen		An organ in the abdominal cavity; it is directly connected to the blood stream and like lymph nodes contains B-lymphocytes, T-lymphocytes, and macrophages.
Splenectomy		The surgical removal of the spleen, which is an organ that is part of the lymphatic system. The spleen is a dark-purple, bean-shaped organ located in the upper left side of the abdomen, just behind the bottom of the rib cage. In adults, the spleen is about 4.8 2.8 1.6 in (12 7 4 cm) in size, and weighs about 4–5 oz (113–142 g). Its functions include a role in the immune system; filtering foreign substances from the blood; removing worn-out blood cells from the blood; regulating blood flow to the liver; and sometimes storing blood cells. The storage of blood cells is called sequestration. In healthy adults, about 30% of blood platelets are sequestered in the spleen.
Stable blood products		Semi-stable or stable blood products have expiry term of more than a year and can tolerate less strict storage conditions without any impact on quality or safety.
Stem cells		Cells from which all blood cells and immune cells are derived, bone marrow is rich in stem cells.
Stem Cell Transplantation	SCT	A stem cell transplant is the infusion of healthy stem cells into your body. A stem cell transplant can help the body make healthy white blood cells, red blood cells or platelets, and reduce your risk of life-threatening infections, anemia and bleeding.
Subcutaneous infusion	SCIG	Administration of gamma globulin in which it is infused slowly directly under the skin with a small pump.
Telangiectasia		Dilation of the blood vessels.
Thrombocytopenia		Low platelet count.
Thrush		A fungal disease on mucous membranes of the mouth caused by Candida infections.
Thymus gland		A lymphoid organ located behind the upper portion of the sternum (breastbone). The thymus is the chief educator of T-lymphocytes. This organ increases in size from infancy to adolescence and then begins to shrink.
Tissue Typing Laboratory		Clinical Immunology Laboratory to incl.: - HLA class I and II typing and matching, - Pre- and post transplant monitoring for bone marrow transplantation, - HPA platelet gene typing and anti-platelet antibody screening and specificity detection, - Immunodeficiency: Screening and diagnosis, monitoring of treatment, - Leukemia diagnosis and monitoring of treatment.
T-lymphocytes (or T-cells)		Lymphocytes that are processed in the thymus; they are responsible in part for carrying out the immune response.
Unusual infectious agents		These are normally non-pathogenic agents or those not generally found in humans which can cause serious disease in immunocompromised patients.
Vaccine		A substance that contains components from an infectious organism which stimulates an immune response in order to protect against subsequent infection by that organism.
Vacuole		A cavity or vesicle in the cytoplasm of a cell containing fluid.
Validation		The action of proving that any material, process, procedure, activity, system, or equipment used in manufacture or control can and will

		reliably achieve the desired and intended results
Vectors		The majority are modified viruses containing normal genes; used in gene therapy to insert normal genes in cells.
Window period		The period between when a donor is infected with a virus or disease-causing agent and when infection can be detected by an immunological marker. During this period the donor is infectious but infection is undetectable. With nucleic acid testing (NAT), the window period is shortened
Virus		A submicroscopic microbe causing infectious disease; can reproduce only in living cells.
White blood cells		See leukocyte.
whole blood		whole blood is the term used in transfusion medicine for human blood from a standard blood donation. The blood is typically combined with an anticoagulant during the collection process, but is generally otherwise unprocessed. In the US, the capitalized " Whole Blood " means a specific standardized product for transfusion or further processing.
X-linked recessive inheritance		A form of inheritance where the characteristic, or disease, is inherited on the X-chromosome.
