Glossary

Note – More detailed information of each item may be found online.

Acquired immune deficiency syndrome (AIDS): A secondary immunodeficiency caused by infection with the HIV (human immunodeficiency virus).ituximab and ocrelizumab, etc.) may exacerbate an underlying immunodeficiency by destroying lymphocytes which produce immunoglobulins.

Acute: An adjective describing 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 and normal brain function.

Agammaglobulinemia: An almost total lack of immunoglobulin or antibodies.

Amniocentesis: The withdrawal of amniotic fluid surrounding a fetus in order to perform prenatal genetic testing.

Anaphylaxis: A life-threatening systemic allergic reaction.

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 certain types of white blood cells (B lymphocytes, plasma cells) into the tissues and serum 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 species of fungi (mold) which is particularly a problem for individuals with Chronic Granulomatous Disease and/or some T cell defects.

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 or general incoordination caused by neurological abnormalities.

Autoantibody: An antibody produced by the immune system in reaction to any of its own cells, cell products and tissues.

Autoimmune disease: A disease that results when the body's immune system reacts against the person's own cells or tissues.

Autosomal recessive inheritance: A form of inheritance where the characteristic, or disease, is inherited from both parents.

Autosomes: Any chromosome other than the sex chromosomes (X and Y).

Bacteria: Single cell organisms (microorganisms) that can be seen only under a microscope. Although there are thousands of different kinds of bacteria in our environment and in or on our bodies, only a few actually cause disease in human beings. Patients with certain kinds of immune defect may have problems with specific kinds of bacteria that do not cause disease in individuals with a normal immune system. Certain other kinds of bacteria infect both immune deficient and normal individuals, but the immune deficient individuals have more trouble clearing this infection and therefore the infection may progress to develop organ damage or other serious consequences.

B lymphocytes (B cells): White blood cells of the immune system derived from the bone marrow and involved in the production of antibodies.

Bone marrow: Soft tissue located in the hollow centers of most bones; the marrow contains developing red blood cells, white cells, platelets and cells of the immune system.

Bradykinin: A protein that causes blood vessels to dilate (enlarge) and results in a decrease in blood pressure.

Bronchiectasis: A dilation and disruption of the tubes (bronchi) leading to the air sacs of the lung; usually the consequence of recurrent (chronic) lung infections.

Carrier detection: The detection of a genetic characteristic in a person who carries the characteristic (or disease) in their genes but does not have the disease.

CD 40 ligand: A protein found on the surface of T lymphocytes; some individuals with X-linked Hyper IgM Syndrome have a deficiency in this protein.

Cellular immunity: Immune protection provided by the direct action of some immune cells, usually referring to T cell immunity.

Chromosomes: Physical structures in the cell's nucleus that carry genes; each human cell 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): Retrieval of a sample of the developing placenta from the womb in order to perform prenatal genetic testing.

Combined immunodeficiency: Immunodeficiency when both T- and B lymphocytes are inadequate or lacking, or not functioning properly.

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 test that includes separate counts for red and white blood cells and platelets.

Congenital: Present at birth or born with the health problem.

Consanguineous (or Consanguinous): Descended from the same family or ancestors.

Cord blood: Blood obtained from the umbilical cord and 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 that affect the activity of other cells and are important in controlling immune responses and inflammation. Interleukins and interferons are cytokines.

DNA (deoxyribonucleic acid): Found in the cell nucleus, DNA carries genetic information.

Eczema: Skin inflammation with redness, itching, oozing and scaling. Also called atopic dermatitis, and is usually seen in allergic individuals but also can occur in people with PI.

Endocrine system: The glands in the body that produce hormones.

Eosinophilia: An increase in the number of granular white blood cells that stain with the dye eosin (eosinophils), which occurs with some allergies and parasitic diseases.

Febrile illness: An illness accompanied by fever.

Ficolins: Soluble molecules of the innate immune system, which recognize carbohydrate molecules on pathogens, apoptotic and necrotic cells and activate the complement system.

Fungus: Member of a class of relatively primitive microorganisms including mushrooms, yeast and molds.

Gammaglobulin (or Gamma globulin): The protein fraction of blood that contains immunoglobulins or antibodies. It usually refers to the IgG fraction of immunoglobulins as defined by their position in serum protein electrophoresis methods.

Gamma interferon: A cytokine primarily produced by T lymphocytes that improves bacterial killing by phagocytes; used in the 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: Correction of genetic diseases by providing a correct or normal form of the abnormal gene which is causing the disease.

Genotype: One gene or a set of genes that determine certain traits or disease characteristics (phenotype). (Also see entry "phenotype.")

Graft-versus-host disease (GVHD): A medical reaction that occurs following transplanted tissues from a genetically different person. Transplanted immune lymphocytes in the donated tissue or bone marrow recognize the recipient as foreign and attacks the tissues of the recipient.

Graft rejection: The immune response of the recipient to the transplanted organ or tissue resulting in its rejection.

Granulocyte: A white blood cell of the immune system characterized by the presence of granules in the cytoplasm. Neutrophils, eosinophils and basophils are examples of granulocytes. Some granulocytes (especially neutrophils) ingest (phagocytose) foreign material such as germs (bacteria). (Also see entry on "phagocyte.")

Granuloma (pl. Granulomata): A mass of granulation tissue typically produced in response to infection, inflammation or the presence of a foreign substance.

HLA Haplotype: A particular combination of genes clustered on the sixth human chromosome that determines histocompatibility. (Also see entries for "histocompatibility antigens" and "MHC.")

Human leukocyte antigen (HLA): The name for a large group of genes in humans on chromosome 6 that code for cell surface proteins that determine tissue type (histocompatibility antigens). This complex of genes is also called the major histocompatibility complex (MHC). (See entries on "histocompatibility antigens" and "Major histocompatibility complex.")

Helper T lymphocytes (Helper T cells): A subset of T lymphocytes that help B lymphocytes and T lymphocytes to become activated to exert their protective functions.

Heterozygous mutation: Each diploid cell (most cells of the body except sperm and eggs) has two copies of every gene, one inherited from the mother and one from the father. Any given gene may contain a mutation. If only one of the two copies of the gene contains the mutation it is called a heterozygous mutant.

Histocompatibility antigens: Proteins 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 in providing a signal to the immune system whether a cell is part of the self or foreign like an invading organism. (Also see entries for "human leukocyte antigen" and "MHC.")

Homozygous mutation: Each diploid cell (most cells of the body except sperm and eggs) has two copies of every gene. Any given gene may contain a mutation. If both copies of the gene contain the same mutation it is called a homozygous mutation.

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Humoral immunity: Immune protection provided by soluble factors, such as antibodies which circulate in the body's fluids.

Hypocalcemia: An abnormally low concentration of calcium in the blood.

Hypogammaglobulinemia: Lower than normal levels of immunoglobulins (or antibodies) in the blood.

Hypoparathyroidism: A disorder in which the parathyroid glands in the neck do not produce enough parathyroid hormone (PTH).

Hypoplasia: The failure of an organ or body part to grow or develop fully.

IgA: An immunoglobulin found in blood but mostly and secreted into tears, saliva and on the mucous membranes of respiratory and intestinal tracks.

IgD: An immunoglobulin that makes up only 0.25% of the immunoglobulins in the plasma. IgD on the surface of B-lymphocytes are important in B-cell differentiation. The function of IgD is poorly understood but may have a role in the allergic response.

IgE: An immunoglobulin found in trace amounts in the blood and responsible for allergic reactions.

IgG: The most abundant and common of the immunoglobulins. It is the only antibody that can cross the placenta from the mother to the developing fetus. It is the only immunoglobulin class that is commercially available for treating immunodeficiency.

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 microbial organisms and foreign substances that threatens the body.

Immunocompetent: Capable of developing an immune response.

Immunocompromised: A state in which a person's immune system is weakened or absent. Individuals who are immunocompromised are less capable of battling infections because of an immune response that is not properly functioning.

Immunodeficiency: A state of either a congenital (present at birth) or an acquired abnormality of the immune system that prevents adequate immune responsiveness. (Also see entries for "primary immunodeficiency" and "secondary immunodeficiency.")

Immunoglobulin replacement therapy: The intravenous, intramuscular or subcutaneous injection of immunoglobulin on a regular basis to treat antibody deficiency.

Immunoglobulins (Ig): Another name for antibody; there are five classes: IgA, IgD, IgG, IgM and IgE.

Incubation period: The period between the infection of an individual by a pathogen and the manifestation of the disease it causes.

Insertional mutagenesis: Mutation caused by the insertion of new genetic material into a normal gene.

Intention tremor: A slow tremor of the extremities that increases on attempted voluntary movement and is observed in certain diseases of the nervous system (e.g., Parkinson's disease).

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.

Intravenous immunoglobulin (IVIG) infusion: Immunoglobulin (gamma globulin) therapy injected directly into the vein.

Killer lymphocytes: T lymphocytes or natural killer cells that directly kill microorganisms or cells that are infected with microorganisms.

Kinin: Any of various polypeptides that are formed locally in the tissues and cause dilation of blood vessels and contraction of smooth muscle.

Leukemia: Type of cancer affecting the white blood cells.

Leukocyte (white blood cell): A group of small colorless blood cells that play a major role in the body's immune response. There are five basic types of leukocytes: monocytes, lymphocytes, neutrophils, eosinophils and basophils.

Live vaccines: Live viruses and bacteria are contained in the vaccine. In seriously immunocompromised individuals, live vaccines can sometimes transmit the disease they were designed to prevent. Examples of common live vaccines include oral polio, rotavirus, measles, mumps, rubella, BCG and chicken pox.

Lymph: Blood plasma-like fluid that contains lymphocytes and granulocytes, but no red blood cells, that circulates through the tissues of the body in small vessels called lymphatics. Lymph is collected from the tissues and lymph nodes via the lymphatic vessels and is rejoined with the circulating blood in a venous connection found near the neck called the thoracic duct.

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, dendritic cells and macrophages.

Lymphocytes: Small white blood cells, normally present in the blood and in lymphoid tissues 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.

Lymphokines: A class of cytokines specifically secreted by lymphoid cells that are important in regulating inflammation and immune responses and for recruiting other cells to participate in immune and inflammatory responses.

Lymphoma: A type of cancer of the lymphocytes.

Macrophages: A phagocytic tissue cell of the immune system that functions as a scavenger in the destruction of foreign antigens (as bacteria and viruses) and serves as an antigen-presenting cell. (See entry on "antigen presenting cell.")

Major histocompatibility complex: (MHC) A series of genes on chromosome 6 that direct the synthesis of the proteins 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.

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 molecules.

Microorganisms: Very, small living organisms that can only be seen with a microscope, usually one-cell organisms, which include bacteria, protozoa, and fungi.

Molecules: The smallest unit of a chemical 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 mucous membranes of the mouth, nose, gastrointestinal tract, eyes, etc; IgA antibodies protect these surfaces, or mucus membranes, from infection.

Mucocutaneous Candidiasis: A group of syndromes with common features including chronic noninvasive Candida infections of the skin, nails, and mucous membranes and associated autoimmune manifestations. It is caused by genetic faults in the immune system

Multifactorial immune disorders: Conditions or diseases arising from a combination of genetic and non-genetic causes, including environmental factors.

Neurology: A branch of medicine concerned with the structure, functions, and diseases of the nervous system.

Neisseria: A group of bacteria that colonize the mucosal surfaces and includes different bacteria that cause meningitis, gonorrhea and other illnesses.

Neonate: A newborn baby, specifically a baby in the first 4 weeks after birth.

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. The major cellular component of pus. (See entry on "granulocyte.")

Nystagmus: Involuntary, usually rapid movement of the eyes.

Opportunistic infection: An infection that occurs only under certain conditions, such as in immunodeficient individuals. Not normally a pathogen for individuals with intact immune systems.

Organism: An individual living thing.

Osteomyelitis: Infection in 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 secrete parathormone and control the normal metabolism and blood levels of calcium. (Also see entry on "hypoparathyroidism.")

Petechiae: Pinhead-sized red spots resulting from bleeding into the skin.

Phagocyte (also phagocytic cell): A general class of white blood cells that ingest microbes and other cells and foreign particles; monocytes, macrophages and neutrophils are types of phagocytes. (Also see entry on "granulocyte.")

Phagosomes: A compartment inside a phagocyte in which pathogenic microorganisms can be killed and digested.

Phenotype: The constellation of traits that result from one or more specific genes (genotype). (Also see entry on "genotype.")

Phenotypic variability: The differences in certain traits or disease characteristics that can be associated with a single genotype. (Also see entry on "genotype.")

Phenylketonuria (PKU): A genetic disorder in which the body cannot normally process the amino acid phenylalanine (Phe), part of many proteins that are found in certain foods.

Plasma cells: Antibody (immunoglobulin)-producing cells that develop from B lymphocytes.

Plasmapheresis: A process in which whole blood is taken from an individual's circulation and then treated, usually by centrifugation, to separate the plasma (that is saved) from the cells and corpuscles, which are then then returned to the individual's body.

Platelets: Smallest of the blood cells; their primary function is associated with the process of blood clotting.

Pneumatocele: An air or gas filled cyst that most often develops within lung tissue.

Polymorphism: The quality or state of existing in or assuming different forms.

Polysaccharides: Complex sugars.

Primary immunodeficiency: Immunodeficiency that is intrinsic to the cells and tissues of the immune system, not due to another illness, medication or outside agent damaging the immune system.

Prophylaxis: 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 one example of proteins.

Pyogenic infection: Any infection that results in pus production.

Purpura: Bluish spots (bruises) on the skin occurring in individuals with low blood platelets (thrombocytopenic purpura) or severe blood stream infections (septic purpura).

Recurrent infections: Any infections that occur repeatedly, usually (but not always) affecting the same set of organs in one individual.

Secondary immunodeficiency (SID): Immunodeficiency due to another illness or agent, such as human immunodeficiency virus (HIV), cancer, or chemotherapy.

Sepsis: An infection of the blood.

Sinopulmonary: Of or relating to the paranasal sinuses, the middle ear and the pulmonary airway from the nose down to the terminal bronchi and air sacs in the lungs.

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.

Staph or Staphylococcal: Staphylococcus, a type of bacteria, is commonly referred to as "staph." There are more than 30 types, but Staphylococcus aureus causes most staph infections.

Stem cells: Cells from which all other cell types are derived. There are also stem cells that only give rise to a limited number of other cells such as the hematopoietic stem cell (HSC) from which blood cells and immune cells are derived. The bone marrow is rich in hematopoietic stem cells.

Subcutaneous immunoglobulin (SCIG) infusion: Administration of gamma globulin directly under the skin into the subcutaneous tissues.

T cell anergy: A tolerance mechanism in which the lymphocyte is intrinsically functionally inactivated following an antigen encounter, but remains alive for an extended period of time in a hyporesponsive state.

Telangiectasia: Dilation of the small blood vessels, usually in the skin and eyes.

Thrombocytopenia: Low platelet count.

Thrush: A fungal disease on mucous membranes of the mouth caused by Candida.

Thymus gland: A lymphoid organ located behind the upper portion of the sternum (breastbone). The thymus is the chief source and educator of T lymphocytes. This organ increases in size from infancy to adolescence and then begins to shrink.

Titer: A measurement of the amount or concentration of a substance in a solution. It usually refers to the level in the blood of antibodies binding a specific antigen.

T lymphocytes (or T cells): Surfaces that come in close contact with the environment, such as the mucous membranes of the mouth, nose, gastrointestinal tract, eyes, etc; IgA antibodies protect these surfaces, or mucus membranes, from infection.

Unusual infectious agents: These are normally non-pathogenic agents or those not generally found in humans, which can cause serious disease in immunocompromised patients. (Also see entry for "opportunistic infection.")

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 containing fluid in the cytoplasm of a cell.

Vectors: Modified viruses or other carriers used to deliver genetic material into a cell; used in gene therapy to insert normal genes in cells.

Venipuncture: The collection of blood from a vein, usually for laboratory testing.

Virus: A submicroscopic microbe causing infectious disease; can reproduce only in living cells.

White blood cells: See "leukocyte."

X-linked recessive inheritance: A form of inheritance where the characteristic, or disease, is inherited on the X-chromosome. As such, it almost always is only seen in boys (male offspring).