Glossary

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 withdrawing of amniotic fluid surrounding a fetus in order to perform prenatal genetic testing.

Anaphylaxis: A life-threatening type of 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 cells (B-lymphocytes, plasma 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 fungus which is particularly a problem for individuals with CGD 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 caused by neurological abnormalities.

Autoantibody: An antibody produced by the body in reaction to any of its own cells or cell products.

Autoimmune disease: A disease that results when the body’s immune system reacts against the 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. 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 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 peptide 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): Involves the 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 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 (CBC): A blood test that includes separate counts for red and white blood cells.

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 that affect the activity of other cells and are important in controlling inflammatory responses. Interleukins and interferons are cytokines.

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

Eczema: Skin inflammation with redness, itching, encrustations and scaling.

Endocrine system: A series of glands in the body that produce hormones.

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

Febrile illness: An illness accompanied by fever.

Ficolins: Ficolins are humoral molecules of the innate immune system, which recognize carbohydrate molecules on pathogens, apoptotic and necrotic cells.

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

Gamma globulin: 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 in the treatment for Chronic Granulomatous Disease (CGD).

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 which is causing the disease.

Graft-versus-host disease: 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.

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

Haplotype: A set of genetic determinants located on a single chromosome, often used to describe the series of genes clustered on the sixth human chromosome that determines the major histocompatibility complex (MHC), the tissue antigens involved in the “tissue types” important in transplantation of organs and bone marrow.

Helper lymphocytes (Helper T-cells): A subset of T-lymphocytes that help B-lymphocytes and T-lymphocytes to function more optimally.
Glossary

**Heterozygous Mutation:** Each diploid cell has two copies of every gene. 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:** 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.

**Homozygous Mutation:** Each diploid cell has two copies of every gene. Any given gene may contain a mutation. If both copies of the gene contain the mutation it is called a homozygous mutation.

**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 and secreted into tears, saliva and on the mucous membranes of respiratory and intestinal tracks.

**IgD:** An immunoglobulin whose function is poorly understood at this time.

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

**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.

**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.

**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.

**Immunoglobulin replacement therapy:** The intravenous, intramuscular or subcutaneous injection of immunoglobulin to provide antibodies that the immunodeficient person cannot make themself.

**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.

**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 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):** 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 or bacteria are used in some vaccines; live vaccines (particularly oral polio) can transmit the disease they were designed to prevent when given to seriously 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.

**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.

**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:** Type of cancer of the lymphocytes.

**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:** 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.

**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.

**Microorganisms:** Minute living organisms, usually one-cell organisms, which include bacteria, protozoa, and fungi.

**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.

**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.
Glossary

**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 includes the bacterium that causes 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.

**Nystagmus:** Involuntary, usually rapid movement of the eyeballs.

**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 control the normal metabolism and blood levels of calcium.

**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.

**Phagosomes:** A cellular compartment in which pathogenic microorganisms can be killed and digested.

**Phenotypic Variability:** The range of differences seen from individual to individual in the effect that any particular single gene may produce. Often used to describe differences in disease severity amongst family members who all have inherited the same mutant gene.

**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-producing cells descended from B-lymphocytes.

**Plasmapheresis:** A process in which blood taken from a patient is treated to extract the cells and corpuscles, which are then added to another fluid and then returned to the patient’s body.

**Platelets:** Smallest and most fragile of the blood cells; primary function is associated with the process of blood clotting.

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

**Polysaccharides:** Complex sugars.

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

**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 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: Infections, such as otitis, sinusitis, pneumonia, deep-seated abscess, osteomyelitis, bacteremia or meningitis that occur repeatedly.

Secondary immunodeficiency: 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 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.

Staphylococcal: Staph is short for *Staphylococcus*, a type of bacteria. There are over 30 types, but *Staphylococcus aureus* causes most staph infections.

Stem cells: Cells from which all blood cells and immune cells are derived, bone marrow is rich in stem cells.

Subcutaneous immunoglobulin (SCIG) infusion: Administration of immunoglobulin directly under the skin.

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.

Titer: A measurement of the amount or concentration of a substance in a solution. It usually refers to the amount of some antibodies found in a patient’s blood.

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.

Vectors: Modified viruses containing normal genes; 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).