JMF GLOSSARY OF TERMS

Term	Definition
Abscess	A collection of pus in the body that usually causes inflammation and swelling. Abscesses can develop anywhere in the body. They can appear as tender, raised masses. Deep abscesses in the body can cause organ damage.
Adenosine Deaminase	An enzyme produced by the ADA gene, most active in lymphocytes. The purpose of this enzyme is to eliminate deoxyadenosine, which is formed when DNA breaks down and is harmful to lymphocyte function.
Agammaglobulinemia	A Primary Immunodeficiency characterized by the absence of B cells, dysfunction of antibodies, and Btk gene abnormalities. This results in a vulnerable immune system, leaving an afflicted individual more susceptible to infections.
Allele	An allele is 1 of 2 versions of a gene. Individuals inherit 1 allele from each parent. Different alleles are termed heterozygous and are responsible for differences in phenotypes.
Anaphylaxis	A severe allergic reaction to an allergen. After exposure to an allergen, the body releases antihistamine, causing airways to close. The flood of chemicals from the immune system causes symptoms like shock, an instant drop in blood pressure, swelling of the airways, and a skin rash. If not treated immediately, this reaction can be fatal.
Anemia	A condition that causes a lack of healthy red blood cells that would normally provide enough oxygen to the body's tissue. This condition ranges in severity and is not always permanent. Symptoms include feelings of fatigue, irregular heartbeat, pale skin, shortness of breath, chest pains, and headache.
Antibody	Also known as immunoglobulins, this protective protein is produced by the white blood cell known as a B cell or B lymphocyte. This immune function is produced as soon as an antigen enters the body. When the immune system lacks these proteins, the afflicted individual is very susceptible to infection.
Antigen	An antigen is a foreign substance or toxin that, upon entry to the body, causes the immune system to produce antibody. Antigens include bacteria, viruses, parasites, fungi, etcetera.
Antigen-Presenting Cell	A type of cell that displays antigen, which is recognized by T cells. Antigen-presenting cells are part of the major histocompatibility complex (MHC). Antigen-presenting cells include dendritic cells, macrophages, and certain B cells and epithelial cells.
Apoptosis	Programmed cell death that occurs normally to remove cells that are unwanted or damaged. This process is essential to maintaining the health of the body. If this process is irregular, it can be the cause of many different diseases (e.g. tumor growth).

Autoantibodies	An antibody that attacks self-antigens of the body. Autoantibodies are harmful and can cause damage to cells and tissues.
Autoimmunity	The failure of the immune system to recognize “self” components, leading to attacks of its own cells and tissues. Autoimmunity can cause diseases such as Celiac Disease, Graves’ Disease, and Diabetes Mellitus Type 1, amongst others.
Autoinflammatory Diseases	Characterized by intense, prolonged periods of inflammation, including symptoms such as fever, swelling, edema, and development of rashes. This category of diseases is caused by deficiencies in the innate immune system, which react to cause inflammation without being exposed to antigens or autoantibodies.
Autosomal Dominant	A pattern of genetic inheritance in which only 1 copy of a gene is needed in order for a certain phenotype to be expressed. Genes of the 22 pairs of autosomes (non-sex chromosomes) can be inherited in dominant patterns.
Autosomal Recessive	A pattern of genetic inheritance in which 2 copies of a gene are needed in order for a certain phenotype to be expressed. Genes of the 22 pairs of autosomes (non-sex chromosomes) can be inherited in recessive patterns.
B Cell	A type of lymphocyte, or white blood cell, that produces antibodies. The B cell is a vital part of the immune system. B cells are an important component of humoral immunity within the adaptive immune system. B cells produce antigen-specific antibodies. Through cell differentiation, the B cell can either become a plasma B cell or a memory B cell.
Basophil	A type of granulocyte that assists in the inflammatory response. Basophils contain histamine that promotes proper blood flow. Additionally, the basophil contains heparin, which helps prevent the blood from clotting too rapidly. Basophils can bind IgE and play an important role in allergies and parasitic infection.
Blood Plasma	The pale yellow liquid component of blood that contains red and white blood cells in suspension. Plasma transports blood cells, nutrients, antibodies, proteins, and chemical messengers throughout the body to maintain proper function and fluid balance. Plasma plays an essential role in protecting from infection.
Bone Marrow Transplant	A procedure to replace damaged or broken-down bone marrow located in healthy tissue. Bone marrow is the soft tissue found inside our bones. There are 3 types of bone marrow transplants: autologous, allogenic, and umbilical cord blood.
Candidiasis	A fungal infection caused by a species of Candida yeast. Candidiasis can affect different parts of the body. Some examples include the mouth and throat (thrush), vagina (yeast infection), or the bloodstream.
Carrier	An individual with 1 recessive, abnormal copy of an allele and a normal, healthy copy on the same locus of a pair of chromosomes. Carriers have the capability to pass an abnormal copy on to offspring. Depending on inheritance from the other parent, a disease phenotype may be expressed.

Cell Differentiation	The process in which a cell develops into a specific role. During growth and development, each cell is programmed or given a specific role in the body. Cell differentiation is important to the development of cell function and function of the organism.
Cell-Mediated Immune Response	An immune response that occurs when T cells attack foreign antigens through cytotoxicity, activation of macrophages, or production of lymphokines.
Chromosome	A strand of tightly coiled DNA and proteins that contain hereditary information (genes), which provides instructions for gene expression. Each chromosome contains a centromere (constriction point), 2 short arms (p arm), and 2 long arms (q arm).
Complement System	A part of the immune system that is composed of many plasma proteins that cause an inflammatory response to fight infection. This system "complements" antibodies and phagocytes by assisting in clearing pathogens.
Consanguinity	Relationship by blood or kinship. This term specifically refers to mating of 2 individuals with 1 or more recent ancestors in common.
Cord Blood	Blood contained in a newborn's umbilical cord, which contains stem cells that can renew themselves. Cord blood can be used to strengthen the immune system and replace damaged cells.
Cytokine	A group of proteins, glycoproteins, and peptides that provide regulatory and signaling functions. They help to regulate host defense mechanisms in response to infection, inflammation, or trauma to the body. Interferon and interleukins are examples of cytokines.
Cytotoxicity	The level to which an agent has the potential to cause harmful, destructive action on a cell. Examples of cytotoxic agents include autoantibodies and venom.
Dendritic Cells	Antigen-presenting leukocytes typically found in tissue that has direct interaction with the external environment like the skin, inner lining of the nose, stomach, and lungs. The primary function of this immune cell is to assess entering antigens and let the other immune cells, B cells, and T cells know about the danger and how to formulate an attack. It is the primary messenger between the innate and adaptive immune systems.
DNA	The study of genetics starts with understanding DNA, which is short for deoxyribonucleic acid. DNA is the code in our cells that determines how our bodies develop and function. DNA is a complicated structure. It is helpful to think about DNA as really long ladder-like chains that coil around on themselves. Each step in the ladder consists of pieces we call bases. There are only four bases in all of our DNA. These are represented by the letters A, C, G, & T. These four bases are repeated in different sequences throughout the DNA ladder. Specific sections of bases of DNA are GENES.

Eczema	An inflammatory skin reaction that presents itself as bumps, redness, swelling, and scaling. It is a rather common problem in many individuals; however, it can be more severe in an immunodeficient patient.
Edema	Swelling of the skin caused by excess fluid found in your tissue. This can affect any part of the body; however, it usually presents itself on the hands, arms, feet, ankles, and legs. Causes range from medication, pregnancy, and a possible underlying disease like heart failure or kidney disease.
Enzyme Replacement Therapy	A medical therapy in which old, defective, or absent enzymes are replaced through regular infusions.
Eosinophil	A type of granulocyte formed in the bone marrow. It contains cytoplasm filled with granules that contain proteins and enzymes. Eosinophils are present in the inflammatory response, trapping and killing foreign substances. They may also play a role in gland development. Overactivity of eosinophils can be harmful to the body.
Epidemiology	The study of the patterns, distribution, and causes of diseases as they appear in different populations, and with the goal to assist affected populations and identify future prevention methods.
Erythrocyte	A red blood cell, the most common type of cell, that transports oxygen and carbon dioxide throughout the body, to and from tissues. Red blood cells also contain hemoglobin, which presents red pigmentation.
Family History	A record of health information about an individual and his or her family members. Based on what close kin have been diagnosed with in the past, physicians analyze patterns, giving them the ability to not only assess the individual's risk factor for certain diseases, but to also predict the possibility of a disease for the individual's future generations.
Gamete	A mature sex cell, such as a sperm or egg, which contains a haploid number of chromosomes and is capable of fusing with another sex cell.
Gene Expression	The manifestation of a phenotype attributed to a specific gene through transcription and genetic translation.
Gene Mutation	An alteration in the DNA sequence that composes a gene, sometimes causing disease. Mutations can be inherited or acquired throughout the lifespan. Genetic mutations can be categorized as missense, nonsense, insertion, deletion, duplication, frameshift, or repeat expansion.
Gene Regulation	The process in which genes are turned on and off. This is important in an organism's development stages as the proper genes are expressed at the appropriate time.
Gene Therapy	A type of medical treatment that uses genes as a method to treat disease. Researchers are approaching this type of therapy in a variety of different ways, such as replacing a mutated gene with a new gene, inactivating a mutated gene that is not functioning properly, and introducing a new gene in order to fight disease.

Genes	A single gene may be anywhere from several hundred DNA bases long to more than 2 million bases long! Humans have somewhere around 20,000 to 25,000 genes. Each gene contains a code that gives instructions on how to make a protein or enzyme that our bodies need to develop and work correctly. These proteins and enzymes in turn are the building blocks of our bodies. They make muscle, tissue, and hair and carry messages within our body. They are the catalyst for millions of reactions that occur every single minute in a living human.
Genetic Variants	The phenotypic and genotypic differences among individuals and populations.
Genotype-Phenotype Correlation	An association that refers to the resulting phenotype as a result of the presence of a mutation in the genotype.
Graft-versus-Host Disease	This is a reaction that can occur because of newly injected stem cells and, sometimes, transplanted organs that attack the patient's tissue and antigen-attacking cells.
Granulocyte	White blood cell made up of small granules that contain proteins. The 3 types of granulocytes are called neutrophils, eosinophils, and basophils. The function of a granulocyte is to fight off antigens. Individuals who do not produce enough of this white blood cell are more prone to infection.
Granuloma	An area of inflammation in tissues caused by an accumulation of macrophages, usually due to infection.
Haplo	A term meaning simple or single. Haplo refers to haplogroups studied through maternal and paternal lines of inheritance in the mitochondria and Y-chromosome, as there is very little recombination that occurs in these lines.
Hematopoietic Stem Cell Transplant	A medical therapy that includes the infusion of stem cells derived from bone marrow, cord blood, or peripheral blood to replace and restore function in patients who have immune deficiencies.
Hemolysis	The breakdown of red blood cells resulting in the release of hemoglobin and cytoplasm into the blood plasma.
Heritability	The proportion of variability of a particular phenotype inherited through genes.
Heterogeneous	Consisting of different genes. A Heterogenous Condition is caused by a variety of different genes or alleles.
Homogeneous	Consisting of the same genetic components. A Homogenous Condition is caused by inheritance of the same molecular defect, or the inheritance of several mutations affecting the same gene.
Host Defense Mechanisms	Defenses that protect against infection, including natural barriers (skin, mucous membranes, the respiratory tract, gastrointestinal tract, and genitourinary tract), non-specific immune responses (macrophage, lymphocyte, cytokine activation, the inflammatory response), and specific immune responses (activation of the complement system).

Humoral Immune System	Also known as the antibody-mediated system because of the active function of antibodies. In this system, macrophages first ingest pathogens and then present components of engulfed material, which trigger helper T cells. These cells help to trigger B cells, which begin to produce antibodies that bind to foreign antigens. Antibodies produce "memory" of antigens to help quickly respond to the same antigen encountered again.
Hypoplasia	Underdevelopment or incomplete development of an organ or tissues that is often the result of inadequate cell production.
Immunoglobulins	A term for antibodies, these Y-shaped glycoprotein molecules are produced by B cells to recognize and bind to specific antigens. Immunoglobulins are produced in 5 different varieties: IgA, IgD, IgG, IgE, and IgM.
Inflammatory Bowel Disease	Characterized by chronic inflammation of the gastrointestinal tract, causing abdominal discomfort and pain. Specific conditions include Crohn's Disease and Ulcerative Colitis.
Innate Immune System	A non-specific portion of the immune system that provides the first line of defense against harmful pathogens. This system relies on anatomical barriers (skin, epithelial surfaces, sweat, cilia, surfactant), humoral barriers (edema, phagocytosis, lysis, opsonization), and cellular barriers (macrophages, Natural Killer cells, neutrophils, and eosinophils).
Interleukin	Cytokines (proteins) that stimulate and mediate the activity of lymphocytes such as T cells and B cells.
Intravenous Immunoglobulin	A product containing pooled immunoglobulin from approximately 1,000 blood donors administered intravenously to patients with deficiencies in immunity.
Isohemagglutinin	An antibody that causes the agglutination of red blood cells, which develops the main blood groups. Examples include anti-A and anti-B isohemagglutinins.
Isotype Switching	A process that is a result of deletion of DNA sequences, allowing antibodies to switch from IgG and IgM to either IgE, IgA, or IgG. This process occurs during B-cell differentiation into plasma cells and is mediated by V(D)J recombinase.
Leukocyte	Another term for a white blood cell that circulates in the blood and lymph, and is comprised of B cells, T cells, granulocytes, monocytes, and macrophages.
Ligand	A substance that binds to a biomolecule, usually a protein. Ligands are often signal-triggering molecules that cause biochemical changes. Ligands may be activators, inhibitors, neurotransmitters, or substrates.
Lymphoproliferation	The production of excessive quantities of lymphocytes, often associated with autoimmune syndromes and caused by mutations in the FAS gene.
Macrophage	A white blood cell that interacts in the innate and adaptive immune systems, engulfing and digesting pathogens that enter the body.

Major Histocompatibility Complex	Cell surface molecules that mediate the interaction of white blood cells with other cells in the immune system. There are 3 different classes of MHC. Class I is associated with CD8, allowing destruction of host cells that display foreign antigen. MHC Class II is associated with specific immunity, moderating CD4-dependent responses to antigen.
Matched Related Donor	A type of bone marrow transplant in which the donor is human leukocyte antigen (HLA) identical to the recipient. The donor and recipient are usually related through common kinship (sibling, parents, or other relatives). This type of transplant is least likely to cause Graft-versus-Host Disease.
Matched Unrelated Donor	A type of bone marrow transplant in which the donor and recipient have identically matched human leukocyte antigens. Unlike a matched related donor transplant, the donor and recipient do not share a common recent relative or kinship.
Mismatched Unrelated Donor	A type of bone marrow transplant in which the donor and recipient have 5 out of 6 matched human leukocyte antigens. This type of transplant may be used if a matched related or unrelated donor is not available. The donor and recipient do not share a common recent relative or kinship.
Mitochondria	Cells that produce energy for the body to carry out cellular functions. Specifically, mitochondria control ATP production. Mitochondria are inherited maternally.
Molecular Sequencing	The process of determining the order of DNA molecules (adenine, cytosine, guanine, thymine) of individual genes, gene clusters, chromosomes, or the entire genome. Sequencing can provide information on mutations in genes and underlying mechanisms of genetic disorders.
Natural Killer Cell	A type of lymphocyte of the innate immune system that serves important roles in host defense mechanisms. These cells provide surveillance, and help to reject virally infected cells and tumor cells. Natural Killer (NK) cells contribute to co-stimulation of immune cells, production of cytokines, and signaling of cytotoxic cell activity.
Neutropenia	An abnormally low count of neutrophils leading to higher susceptibility to infection. This condition is defined as fewer than 1,700 neutrophils per microliter.
Neutrophil	A type of granulocyte that has high phagocytic activity, engulfing harmful bacteria and microorganisms that enter the body. This white blood cell helps to mediate immune responses and migrates to areas of infection through chemotaxis.
Newborn Screening	A process that identifies severe medical conditions that can affect long-term health and survival. At birth, drops of blood are collected from pricking a newborn's heel for the purpose of testing for genetic, metabolic, and/or endocrine disorders that can be detected through developed assays. Conditions identified early can be properly diagnosed and treated to prevent morbidities or even death that may occur otherwise.

Passive Immunity	Immunity acquired from another individual through the transfer of antibodies. This can refer to antibodies passed naturally from mother to offspring, or artificially through injection of antibodies.
Pathogen	Any agent that can cause disease. Most commonly a pathogen refers to bacteria, virus, fungi, or parasite.
Phagocyte	White blood cells that engulf harmful pathogens that enter the body, or any dead or decaying cells.
Phagocytosis	The process in which a phagocyte engulfs and digests a harmful pathogen, or dead or decaying cellular material.
Pharyngeal	Relating to or located in the pharynx (mouth, throat), which is posterior to the nasal cavity and superior to the esophagus.
Recombination	The exchange of DNA during meiosis between 2 homologous chromosomes to provide genetic variety in offspring.
Recurrent Infections	A breakdown in the body's immune defense, causing abnormally frequent illness. Recurrent infections may be due to an overactive (allergies) or underactive immune system (Primary Immunodeficiency).
Septicemia	A serious, life-threatening infection that can affect all body systems. Symptoms include body chills, high fever, rapid heartbeat, and can progress quickly.
Sex-Linked	The phenotypic expression of genetic material that is located on or linked to a sex chromosome (X or Y).
Signaling	Communication between cells to activate and coordinate function. Signaling plays an important role in cell development, immunity, tissue repair, and normal function. Errors in cell signaling can result in abnormal function, causing disease.
Sinopulmonary	Affecting, relating to, or involving the nasal sinuses and the airway of the lungs.
Sinusitis	Swelling or inflammation of the tissue in the sinuses, which causes susceptibility to bacterial, fungal, or viral growth. Common conditions include Allergic Rhinitis, the Common Cold, and Nasal Polyps.
Somatic Cell	A type of cell that forms a multicellular organism and contains chromosomes. Somatic cells are not gametes, germ cells, or stem cells. Somatic cells compose blood, bones, connective tissue, internal organs, and skin.
Stem Cell	A biological cell that can divide to create additional stem cells and differentiate into a specialized cell. There are 2 types of stem cells: embryonic and adult. Stem cells have restorative and regenerative function.
Subcutaneous Immunoglobulin	A product containing pooled immunoglobulin from approximately 1,000 blood donors administered under the skin in fatty tissues of the abdomen, lower back, thighs, and upper arms to patients with deficiencies in immunity. Subcutaneous

	Immunoglobulin infusions are typically administered at home and more frequently; biweekly, weekly, and even daily based on a patient's need.
T Cell	A lymphocyte, or white blood cell, that matures in the thymus, contains a distinct T-cell receptor, and plays an essential role in cell-mediated immune function. There are several types of T cells including cytotoxic, helper, memory, natural killer, and regulatory T cells.
T-Cell Receptor Excision Circles (TREC)	A byproduct of DNA produced during the rearrangement of T-cell receptor (TCR) genes. An analysis of TREC can determine thymic function and competency of the T-cell effector immune response.
Thrombocytopenia	A condition characterized by an abnormally low number of platelets, which help the blood to clot. This condition can be present in bone marrow, the bloodstream, or the liver or spleen.
Thrush	A condition that occurs when a fungus, known as Candida, grows abnormally in the mouth and throat. Symptoms appear as large white sores that may bleed in the mouth or on the tongue. Long-lasting thrush can signify weakness or dysfunction of the immune system.
Thymus	An organ of the immune system where T cells are produced and matured. The thymus is located behind the sternum, in front of the heart. It is distinguished by 2 identical lobes.
Thymus Transplantation	An organ transplant, in which the thymus is removed and replaced with a donated matched thymus. Individuals with DiGeorge Syndrome, characterized by absent thymus or hypoplasia of the thymus, require transplantation.
V(D)J Recombination	Genetic recombination of immunoglobulin and T-cell receptors to provide diversity of the immune system and to encode proteins to provide defense against bacteria, viruses, parasites, and other harmful antigens. Variable, Diverse, and Joining segments refers to the random process of selection to create this gene diversity.
Whole Exome Sequencing	Whole Exome Sequencing is a genetic testing technique that zeroes in on the protein-coding portion or exons of the genome.
Whole Genome Sequencing	A type of DNA sequencing that provides complete and comprehensive information about the genome. Also known as entire genome sequencing, this process allows sequencing of all of an organism’s chromosomes, providing specific information on the 6 billion nucleotides that comprise human DNA. Whole genome sequencing provides distinct and vast amounts of information that can be used to analyze underlying disease mechanisms.
X-Linked Recessive	A type of pattern inheritance in which a gene is passed on through the X-chromosome. A mutation on the X chromosome will cause the phenotype to be expressed in male individuals, since they only carry one X chromosome. Females are more common carriers; however, if they inherit the same mutation on the X chromosome from both parents, they will express the phenotype.