Glossary

Sources: National Library of Medicine (https://www.nlm.nih.gov), National Cancer Institute (https://www.cancer.gov/), Merriam Webster Medical Dictionary (https://www.merriam-webster.com/), U.S. Food and Drug Administration (https://www.fda.gov/). Accessed March 2022.
<Sharp brackets> mean “also known as.”

Absolute Neutrophil Count (ANC): A measure of the number of neutrophils in the blood. Neutrophils are a type of white blood cell that help the body fight infection.

Acute Myeloid Leukemia (AML): A quickly progressive cancer of the myeloid line of blood cells, characterized by the rapid growth of immature white blood cells (also called myeloblasts) that accumulate in the bone marrow and interfere with the production of normal blood cells.

Adenoma: A benign tumor of a glandular structure or of glandular origin.

Adermatoglyphia: The absence of ridges on the skin on the pads of the fingers and toes, as well as on the palms of the hands and soles of the feet.

Adnexa: The appendages, or associated anatomical parts, of an organ.

Alanine Aminotransferase (ALT): An enzyme found in highest amounts in the liver. Injury to the liver results in release of ALT into the blood.

Alkylating Agent: A substance with mutagenic activity that inhibits cell division and growth and is used to treat some cancers.

Allele: A copy or alternate format of a gene.

Allogeneic: Involving, derived from, or being individuals of the same species that are sufficiently unlike genetically to interact antigenically.

Alopecia: Loss of hair.

Androgens: A synthesized, male sex hormone (testosterone).

Anemia: A condition in which the blood is deficient in red blood cells, in hemoglobin, or in total volume.

Anonychia: Congenital absence of the nails.

Antibody: Any of a large number of proteins produced normally by specialized B cells after stimulation by an antigen and act specifically against the antigen in an immune response. Some are produced abnormally by some cancer cells, and typically consist of four subunits including two heavy chains and two light chains. <immunoglobulin>

Antigen: Any substance foreign to the body that evokes an immune response either alone or after forming a complex with a larger molecule (as a protein) and that is capable of binding with a product (as an antibody or T cell) of the immune response.

Aplastic Anemia: A condition in which the bone marrow does not make enough blood cells.

Apoptosis: A genetically determined process of cell self-destruction that is marked by the fragmentation of nuclear DNA. It is activated either by the presence of a stimulus or by the removal of a stimulus or suppressing agent and is a normal physiological process eliminating DNA-damaged, superfluous, or unwanted cells. When halted (as by genetic mutation), it may result in uncontrolled cell growth and tumor formation. <programmed cell death>

Arteriovenous Malformations: Vascular anomaly characterized by abnormal connections between the arteries and the veins.

Ascites: Abnormal accumulation of serous fluid in the spaces between tissues and organs in the cavity of the abdomen. <hydroperitoneum>

Assisted reproductive technologies (ART): A broad term used to describe the range of technology used to assist human reproduction in the treatment of infertility.

Aspartate Aminotransferase (AST): An enzyme found in highest amounts in the liver. Injury to the liver results in release of AST into the blood. <aspartate transaminase>, <glutamic-oxaloacetic transaminase>

Ataxia: The inability to coordinate voluntary muscular movements.

Autosomal Dominant (AD): A trait or disorder that requires only one copy of the genetic mutation at a particular locus in order to express observable phenotype.

Autosomal Recessive (AR): Describes a trait or disorder that occurs when one variant is present on both alleles (copies) of a given gene in order to express observable phenotype.

Avascular Necrosis: Death of bone tissue due to impaired or disrupted blood supply (as that caused by traumatic injury or disease) and marked by severe pain in the affected region and by weakened bone that may flatten and collapse. <osteonecrosis>

B cells: A type of white blood cell that makes antibodies; are part of the immune system and develop from stem cells in the bone marrow. <B lymphocyte>

Biallelic Mutations: Mutations that occur on both copies of a single gene.

Blepharitis: Inflammation of the eyelids and especially of their margins.

Bone Marrow: The soft tissue filling the cavities of bones. Bone marrow exists in two types, yellow and red. Yellow marrow is found in the large cavities of large bones and consists mostly of fat cells and a few primitive blood cells. Red marrow is a hematopoietic tissue and is the site of production of erythrocytes and granular leukocytes. Bone marrow is made up of a framework of connective tissue containing branching fibers with the frame being filled with marrow cells.

Buccal Mucosa / Oral Mucosa: The inner lining of the cheeks.

Bullae: Large blisters on the skin that are filled with clear fluid.

Carcinogen: Any substance that causes cancer.

Carcinoma: A malignant tumor of epithelial origin

Carcinoma *in situ* (CIS): Carcinoma in the stage of development when the cancer cells are still within their site of origin (as the mouth or uterine cervix).

Carrier: In classical genetics, a carrier refers to an individual who carries one deleterious allele for an autosomal recessive disorder. In clinical discussions, a carrier may refer to an individual who carries a deleterious allele that predisposes to disease.

Cerebellar Hypoplasia: A neurological condition in which the cerebellum is smaller than usual or not completely developed.

Chemotherapy: The use of chemical agents in the treatment or control of disease. Especially the administration of treatment that uses drugs to stop the growth of cancer cells, either by killing the cells or by stopping them from dividing.

Cholestasis: Any condition in which the flow of bile from the liver is slow or blocked.

Clinical Trial: A type of research study that tests how well new medical approaches work in people. These studies test new methods of screening, prevention, diagnosis, or treatment of a disease. <clinical study>

ClinicalTrials.Gov: An online registry of clinical trials that are being conducted around the world. ClinicalTrials.gov is operated by the National Library of Medicine at the National Institutes of Health and can be accessed by anyone who has access to the internet.

Clubbing: Changes in the areas under and around the toenails and fingernails that may include the softening of the nail bed, forming of a sharper angle with the cuticle, bulging of the last part of the finger, and curving downward of the nail. <Digital Clubbing>

Cirrhosis: Widespread disruption of normal liver structure by fibrosis and the formation of regenerative nodules that is caused by any of various chronic progressive conditions affecting the liver.

Coats Retinopathy: The disorder causes blood vessels in the retina to be abnormally enlarged (dilated) and twisted. The abnormal vessels leak fluid, which can eventually cause the layers of the retina to separate (retinal detachment). These eye abnormalities often result in vision loss.

Common Variable Immunodeficiency (CVID): A disorder characterized by low levels of immunoglobulin (antibodies) and an increased risk of infection.

Comorbidity: A medical condition that exists simultaneously with and usually independently of another medical condition.

Complete Blood Count (CBC): A measure of the number of red blood cells, white blood cells, and platelets in the blood.

Compound Heterozygous: Usually refers to autosomal recessive disorders where an individual has two different abnormal alleles at a particular locus, one of each chromosome of a pair.

Conditioning Regimen: The treatments used to prepare a patient for stem cell transplantation that help make room in the patient's bone marrow for new blood stem cells to grow, prevent the patient's body from rejecting the transplanted cells, and help kill any cancer cells that are in the body.

Conjunctival Fornix: The loose arching folds connecting the conjunctival membrane lining the inside of the eyelid with the conjunctival membrane covering the eyeball.

Contraception: The deliberate prevention of conception.

Corpus Callosum: The band of commissural fibers uniting the cerebral hemispheres.

Corticosteroids: Any steroid hormone made in the adrenal cortex or in a laboratory setting; used medically as hormone replacement, to suppress the immune system, and to treat some side effects of cancer and its treatment.

Cutaneous: Relating to the skin.

Cytopenias: Deficiency of the cellular elements of the blood.

Cytotoxic: Toxic to cells.

Danazol: A synthetic sex hormone used in some studies of attenuation of accelerated telomere attrition.

***De novo* Variant:** An alteration in a gene that is present for the first time in one family member as a result of a mutation in a germ cell (egg or sperm) of one of the parents or in the fertilized egg itself. <*de novo* mutation>

Diffusing Capacity of the Lungs for Carbon Monoxide (DL_{CO}): A measurement to assess the lungs' ability to transfer gas from inspired air to the bloodstream.

Dilation: The act or action of stretching, widening, or enlarging a part of the body.

Deoxyribonucleic acid (DNA): The molecule inside cells that contains the genetic information responsible for the development and function of an organism. DNA molecules allow this information to be passed from one generation to the next. DNA is made up of a double-stranded helix held together by weak hydrogen bonds between purine-pyrimidine nucleotide base pairs: adenine (A) paired with thymine (T), and guanine (G) paired with cytosine (C).

Dual energy X-ray absorptiometry (DEXA or DXA): A procedure that measures the amount of calcium and other minerals in a bone by passing x-rays with two different energy levels through the bone; shows the strength and thickness of a bone.

Dyskeratosis Congenita (DC): A rare condition classified under a broad spectrum of genetic disorders known as telomere diseases. These diseases can often cause bone marrow failure and lung disease. People with DC frequently develop unusual skin pigmentation patterns, nail discoloration, white patches in the mouth (oral leukoplakia) and are especially susceptible to conditions that impair bone marrow function. DC may also cause pulmonary fibrosis, a condition that leads to the accumulation of scar tissue in the lungs, decreasing the flow of oxygen into the bloodstream. Although congenital (present at birth), the signs and symptoms of DC often may not appear until late childhood or early adolescence, and in some cases, not until adulthood.

Dyskerin: A protein involved in maintaining telomeres.

Dyslipidemia: A condition marked by abnormal concentrations of lipids or lipoproteins in the blood.

Dysphagia: Difficulty in swallowing.

Dysplasia: Cells that look abnormal under a microscope but are not cancer.

Dysplastic Nails: Ridging, flaking or poor growth of the nails.

Dyspnea: Difficult, painful breathing or shortness of breath.

Ectropion: The turning out of the eyelid so that the inner surface is exposed.

Edema: An abnormal excess accumulation of serous fluid in connective tissue or in a serous cavity.

Effusion: Escape of fluid from an anatomical vessel due to rupture.

Elastography: A type of imaging test that checks the liver for fibrosis. <liver elastography>

Endocrinologist: Physician treating problems associated with the endocrine system.

Engraftment: The process by which donor stem cells establish themselves successfully within the recipient.

Enterocolitis: Inflammation affecting both the small and large intestine.

Enteropathy: Disease of the intestinal tract.

Epigenetics: The study of heritable changes that do not affect the DNA sequence but influence gene expression.

Esophageal Stenosis: A narrowing of the esophagus that may interfere with swallowing.

Esophageal Web: Membranous structure in which a thin fold of tissue creates at least a partial obstruction of the esophageal lumen.

Esophagram: X-ray based test that takes pictures of the esophagus.

Exception points: A system to award increased waitlist priority to those patients whose severity of illness or risk of complications are not captured by the MELD score.

Excision: In a biopsy, the removal of the entire lump or suspicious area.

Exudative Retinopathy: A condition where blood vessels in the retina become abnormally enlarged and twisted. The abnormal vessels leak fluid, which can eventually cause the layers of the retina to separate (retinal detachment). This eye abnormality often results in vision loss.

Familial: Tending to occur in more members of a family than expected by chance alone.

Fibrosis: An increase of interstitial fibrous tissue.

FISH: A technique used to identify the presence of specific chromosomes or chromosomal regions through hybridization (attachment) of fluorescently-labeled DNA probes to denatured chromosomal DNA. Examination through a microscope under fluorescent lighting detects the presence of the colored hybridized signal (and hence presence of the chromosome material) or absence of the hybridized signal (and hence absence of the chromosome material). Also called fluorescence in situ hybridization.

Forced Vital Capacity (FVC): The amount of air that can be forcibly exhaled from the lungs after taking the deepest breath possible. This value is measured by spirometry, which is a common breathing test to check lung function.

Gene: The basic unit of heredity that occupies a specific location on a chromosome. Most genes code for a specific protein or segment of protein leading to a particular characteristic or function.

Genetic Anticipation: The phenomenon in genetic diseases where symptoms occur earlier and with increased severity in succeeding generations.

Germline: The cellular lineage of a sexually reproducing organism from which eggs and sperm are derived. The genetic material contained in this cellular lineage can be passed to the next generation.

Genotype: At its broadest level, genotype includes the entire genetic constitution of an individual. It is often applied more narrowly to the set of alleles present at one or more specific loci.

Graft Versus Host Disease (GVHD): A complication that can occur when T cells from a tissue or organ transplant, especially a bone marrow transplant, react immunologically against the recipient's antigens attacking cells and tissues.

Hematologic: Of or relating to blood or to hematology.

Hematopoiesis: The formation of blood or of blood cells in the living body.

Hematopoietic Growth Factor: Any of a group of glycoproteins that promote the proliferation and maturation of blood cells.

Hematopoietic Stem Cell Transplant (HCT or HSCT): The intravenous infusion of autologous or allogeneic stem cells collected from bone marrow, peripheral blood, or umbilical cord blood to reestablish hematopoietic function in patients whose bone marrow or immune system is damaged or defective.

Hemorrhage: A copious or heavy discharge of blood from the blood vessels.

Hepatic: Of, relating to, affecting, or associated with the liver. <hepatic injury> <hepatic insufficiency>

Hepatocellular: Of or involving hepatocytes. <hepatocellular carcinomas> <hepatocellular necrosis>

Hepatopulmonary syndrome: A condition caused by blood vessels in the lungs expanding (dilating) and increasing in number, making it difficult for red blood cells to properly absorb oxygen.

Heterozygous: The presence of two different alleles at a particular gene locus.

Hirsutism: Excessive growth of hair of normal or abnormal distribution.

Histology: The study of tissues and cells under a microscope.

Homozygous: The presence of two identical alleles at a particular gene locus.

Hoyeraal-Hreidarsson Syndrome (HH): A clinically severe Telomere Biology Disorder variant characterized by intellectual disability, intrauterine growth restriction, microcephaly, cerebellar hypoplasia, progressive combined immune deficiency, and/or bone marrow failure.

Human Papillomavirus (HPV): A sexually transmitted virus that is passed on through genital contact (such as vaginal and anal sex) as well as skin-to-skin contact.

Hyperhidrosis: Generalized or localized excessive sweating.

Hyperkeratosis: Excessive thickening of the outer layer of the skin.

Hypogammaglobulinemia: A deficiency of gamma globulins and especially antibodies in the blood.

Hypogonadism: Functional incompetence of the gonads especially in the male with subnormal or impaired production of hormones and germ cells.

Hypothyroidism: The clinical syndrome that results from deficient activity of the thyroid gland. It leads to lowered metabolic rate and general loss of vigor

Iatrogenic: Induced inadvertently by a physician or surgeon or by medical treatment or diagnostic procedures.

Idiopathic: Arising spontaneously or from an obscure or unknown cause.

Immunodeficiency: Inability to produce a normal complement of antibodies or immunologically sensitized T cells especially in response to specific antigens.

Immunoglobulin: A protein that is made by B cells and plasma cells (types of white blood cells) and helps the body fight infection. They are classified by structure and activity into five classes (immunoglobulin A; immunoglobulin D; immunoglobulin E; immunoglobulin G; immunoglobulin M).

Immunosuppression: Suppression (as by drugs) of natural immune responses.

***In vitro*:** Outside the living body and in an artificial environment.

In vivo: In the living body of a plant or animal.

Incomplete Penetrance: Occurs when individuals who carry the pathogenic variant express the associated trait while others do not.

Inspiratory Rales: A fine, high-pitched crackling or rattling sound that can occur when someone inhales.

Intrauterine growth restriction: The failure of a fetus to attain its expected fetal growth at any gestational age.

Iron Chelation: Removal of iron from the blood through medication or phlebotomy.

Low-density Lipoprotein (LDL): A lipoprotein of blood plasma that is composed of a moderate proportion of protein with little triglyceride and a high proportion of cholesterol and that is associated with increased probability of developing atherosclerosis <bad cholesterol>, <beta-lipoprotein>, <low- density lipoprotein>

Leukemia: An acute or chronic blood cancer characterized by the type of white blood cell most prominently involved.

Leukocyte: A type of blood cell that is made in the bone marrow and found in the blood and lymph tissue. Leukocytes are part of the body's immune system. They help the body fight infection and other diseases. Types of leukocytes are granulocytes (neutrophils, eosinophils, and basophils), monocytes, and lymphocytes (T cells and B cells).

Leukoencephalopathy: Any of various diseases affecting the brain's white matter.

Leukoplakia: A white patch lesion found on a mucous membrane that cannot be scraped off. Leukoplakia is generally considered a precancerous condition, however its appearance may also result from a variety of hereditary diseases.

Luteinizing hormone (LH): A female hormone that, in combination with follicle stimulating hormone, stimulates the secretion of estrogen from ovarian follicles. In men, it is important in the development of interstitial tissue in the testis and for the secretion of testosterone <interstitial-cell stimulating hormone> <lutropin>

Lymphocyte: Any of the colorless weakly motile cells that originate from stem cells and differentiate in lymphoid tissue (as of the thymus or bone marrow), that are the typical cellular elements of lymph, that include the cellular mediators of immunity, and that constitute 20 to 30 percent of the white blood cells of normal human blood.

Lymphopenia: Reduction in the number of lymphocytes circulating in the blood of humans or animals.

Macrocytosis: The occurrence of larger-than-normal red blood cells.

Mammogram: An x-ray of the breast.

Menarche: The beginning of the menstrual function marked by the first menstrual period of an individual.

Menopause: The natural cessation of menstruation that usually occurs between the ages of 45 and 55. <climacteric>

Microcephaly: A condition of abnormal smallness of the head usually associated with intellectual delays.

Model for End-Stage Liver Disease (MELD) / Pediatric End Stage Liver Disease (PELD) Score: A scoring system to assess the severity of chronic liver disease.

Mosaicism: The occurrence of 2 or more cell lines with different genetic or chromosomal make-up, within a single individual or tissue.

Mucocutaneous Triad: Reticulated skin pigmentation, nail dystrophy, and oral leukoplakia.

Multifactorial: Caused or marked by a mode of inheritance dependent on a number of genes at different loci.

Myeloablative: The depletion of bone marrow cells, such as through the administration of chemotherapy and radiation therapy prior to a stem cell transplant.

Myelodysplastic Syndrome (MDS): A type of cancer in which the bone marrow does not make enough healthy blood cells (white blood cells, red blood cells, and platelets) and there are abnormal cells in the blood and/or bone marrow. Sometimes, myelodysplastic syndrome can transform into acute myeloid leukemia (AML).

Nail Dystrophy: This general term is used to describe changes in the shape, color, texture, and growth of the fingernails or toenails. The nails are often ridged, pitted, or abnormally curved.

Nasolacrimal Duct: The duct transmits tears from the lacrimal sac to the inferior meatus of the nose. It is also called the nasal duct.

Neoplasia: A process of tumor formation.

Neutropenia: Neutropenia is an abnormal decrease in the number of neutrophils, a type of white blood cells.

Neutrophil: A granulocyte that is the chief phagocytic white blood cell is called a neutrophil.

Next-Generation Sequencing (NGS) Technologies: This technology has been developed to speed up the process to sequence a human genome, DNA.

Nodular regenerative hyperplasia: A condition in which normal liver tissue transforms into multiple, small clusters (nodules) of replicating liver cells.

Ophthalmic: Structures that are in the region of the eye or that supply or drain the eye.

Opportunistic Infection: An infection caused by bacterial, viral or fungal pathogens that occurs more frequently and are more severe in individuals with weakened immune systems is said to be opportunistic.

Osteonecrosis: This disease is caused by reduced blood flow to the bones and joints. The lack of blood causes the bone to break down faster than the body can make enough new bone. The bone starts to die and may break down.

Osteopenia: This can cause bones to be weak and brittle, and increases the risk for broken bones. It is defined by a decrease in the amount of calcium and phosphorus in the bone. <bone loss>

Palliative Treatment: Care given to improve the quality of life and reduce pain. The goal of palliative care is to prevent or treat, as early as possible, disease symptoms and treatment side effects. It also attends to the psychological, social, and spiritual problems caused by the disease or its treatment and is often given with other treatments.

Pancytopenia: It is the reduction in the number of red and white blood cells as well as platelets. It results in fatigue due to the low numbers of red blood cells, frequent infections due to the low number of white blood cells, clotting problems due to the low number of platelets.

Pathogenic Variant: A genetic alteration that increases an individual's susceptibility or predisposition to a certain disease or disorder. When such a variant (or mutation) is inherited, development of symptoms is more likely, but not certain. <deleterious mutation> <disease-causing mutation> <predisposing mutation> <susceptibility gene mutation>

Patient registry: An organized system that uses observational study methods to collect uniform data (clinical and other) to evaluate specified outcomes for a population defined by a particular disease, condition, or exposure, and that serves a predetermined scientific, clinical, or policy purpose(s).

Parathyroid Hormone (PTH): The hormone of the parathyroid gland regulates the metabolism of calcium and phosphorus in the body. <parathormone.>

Phenotype: A phenotype is an individual's observable traits, such as height, eye color, and blood type. The genetic contribution to the phenotype is called the genotype.

Phimosis: Tightness or constriction of the orifice of the foreskin that prevents retraction of the foreskin over the glans.

Platelet: Are pieces of very large cells in the bone marrow called megakaryocytes. They help form blood clots to slow or stop bleeding and to help wounds heal. <thrombocyte>

Poikiloderma: Several disorders that are characterized by patchy discoloration of the skin.

Polypharmacy: The concurrent use of multiple medications by a patient to treat usually coexisting conditions and which may result in adverse drug interactions

Portal Hypertension: This type of hypertension in the hepatic portal system is caused by venous obstruction or occlusion that produces splenomegaly and ascites in its later stages.

Pulmonary Fibrosis: Pulmonary fibrosis is a condition in which the tissue deep in your lungs becomes damaged and scarred over time. This tissue becomes thick and stiff, making it difficult to breathe properly and preventing the blood from receiving adequate oxygen.

Pulmonary Function Test (PFT): Noninvasive tests that show how well the lungs are working. The tests measure lung volume, capacity, rates of flow, and gas exchange. This information can help diagnose and decide the treatment of certain lung disorders.

Purpura: This purplish or brownish red discoloration is caused by hemorrhage into the tissues. It is easily visible through the epidermis.

Rejection: An immune response in which foreign tissue (as of a skin graft or transplanted organ) is attacked by immune system components of the recipient organism.

Reticulated Skin Pigmentation: Skin pigmentation, or coloring, resembling a net. <hyperpigmentation>

Retinopathy: Any of various noninflammatory disorders of the retina including some that cause blindness.

Revesz Syndrome (RS): A variant of dyskeratosis congenita involving abnormalities in the light-sensitive tissue at the back of the eye (retina).

Schatzki's Rings: A local narrowing in the lower part of the esophagus that may cause dysphagia, or difficulty in swallowing.

Severe Combined Immunodeficiency (SCID): A rare congenital disorder of the immune system that is characterized by inability to produce a normal complement of antibodies and T cells and that usually results in early death.

Senescence: The process of growing old. In biology, senescence is a process by which a cell ages and permanently stops dividing but does not die.

Shelterin: A six protein complex known to protect chromosome ends and regulate telomerase activity.

Somatic Variant: An alteration in DNA that occurs after conception and is not present within the germline. Somatic variants can occur in any of the cells of the body except the germ cells (sperm and egg) and therefore are not passed on to children. Somatic variants can (but do not always) cause cancer or other diseases.

Squamous Cell Cancer (SCC): Cancer of a kind of epithelial cell. Is one of the major forms of skin cancer but can also occur in the digestive tract, lungs, and other areas of the body.

Strictures: Abnormal narrowing of bodily passages (as from inflammation, cancer, or the formation of scar tissue).

Syndrome: A group of signs and symptoms that occur together and characterize a particular abnormality or condition.

Taurodontism: A dental condition marked by the enlargement of the pulp cavities and the reduction of the roots.

Telangiectasias: A permanent dilation of preexisting blood vessels (capillaries, arterioles, venules) creating small focal red lesions, most commonly in the skin or mucous membranes.

Telomerase: A ribonucleoprotein (RNA) that is an enzyme that adds DNA sequence repeats to the end of DNA strands in the telomere regions, which are found at the end of chromosomes.

Telomere: The end of a chromosome. Telomeres are made of repetitive sequences of non-coding DNA that protect the chromosome from damage. Each time the cell divides, the telomeres become shorter. Eventually, the telomeres become so short that the cell can no longer divide and it dies.

Telomere Biology Disorder (TBD): Telomere biology disorders are a complex set of illnesses defined by the presence of very short telomeres.

Thelarche: The beginning of breast development at the onset of puberty.

Thrombocytopenia: Persistent decrease in the number of blood platelets.

Trichiasis: A turning inward of the eyelashes often causing irritation of the eyeball.

Urethral Stenosis: A narrowing of the diameter of the urethra.

Urogenital: Of, relating to, affecting, treating or being the organs or functions of excretion and reproduction.

Varices: An abnormally dilated or swollen blood or lymph vessel, especially a vein.

Veno-Occlusive Disease: Disorder in which veins are partially or completely obstructed or the blood flow through the veins is less than optimal.

X chromosome inactivation: Refers to the silencing of one X chromosome in female mammalian cells to equalize the gene products from the X chromosome between females and males. Skewed X-chromosome inactivation can occur when the X-inactivation of one X chromosome is favored over the other, leading to an uneven number of cells with each chromosome inactivated.

X-linked recessive (XLR): Refers to genetic conditions associated with pathogenic variants in genes on the X chromosome. A male carrying such a variant will be affected, because they carry only one X chromosome. A female carrying a variant in one gene, with a normal gene on the other X chromosome, is *generally* unaffected.