

# Glossary

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**Ablation:** To remove or destroy, especially by cutting or abrading diseased tissue.<sup>1</sup>

**Absolute neutrophil count (ANC):** This number is important in determining the body's capacity to fight a bacterial infection. To determine the ANC, multiply the percentage of neutrophils (found in the "differential" section of the CBC) by the total number of white blood cells. Include both mature neutrophils (usually designated as "segs") and more immature forms (often called "bands").<sup>2</sup>

**Acute lymphocytic leukemia (ALL):** An aggressive (fast-growing) type of leukemia (blood cancer) in which too many lymphoblasts (immature white blood cells) are found in the blood and bone marrow. Common symptoms are weakness and fatigue, easy bruising and petechiae, and sometimes frequent infections.<sup>4</sup> ALL is uncommon in FA patients.

**Acute myeloid leukemia (AML):** A quickly progressing disease which frequently develops in FA patients, in which too many immature white blood cells (not lymphocytes) are found in the blood and bone marrow. The cells that dominate the bone marrow of patients with AML are known as "blasts." Anemia, low platelet counts, and variable white blood cell counts characterize this disease. Common symptoms are weakness and fatigue, easy bruising and petechiae, and sometimes frequent infections. Also called acute myeloblastic leukemia, acute myelogenous leukemia, acute nonlymphocytic leukemia, AML, and ANLL.<sup>2,4</sup>

**Adenocarcinoma:** Cancer that begins in cells that line certain internal organs, such as the liver, stomach, and lungs, and that have gland-like (secretory) properties.<sup>4,5</sup>

**Adenoma:** An ordinarily benign neoplasm of epithelial tissue, such as in the liver, in which the tumor cells form glands or gland-like structures in the stroma.<sup>6</sup>

**Adenopathy:** Any enlargement involving lymph nodes.

**Adrenal insufficiency:** An endocrine or hormonal disorder characterized by weight loss, muscle weakness, fatigue, low blood pressure, and sometimes darkening of the skin in both exposed and nonexposed parts of the body. Occurs when the adrenal glands do not produce enough of the hormone cortisol and, in some cases, the hormone aldosterone.<sup>7</sup> Adrenal insufficiency also occurs if there is ACTH deficiency. In FA patients, this is most often an acquired abnormality due to prolonged use of steroids necessitating slow withdrawal. The disease is also called Addison's Disease or hypocortisolism.

**Adrenocorticotrophic hormone (ACTH):** An ACTH test measures the adrenocorticotrophic hormone, a hormone released from the anterior pituitary gland in the brain. ACTH levels in the blood are measured to help detect, diagnose, and monitor conditions associated with excessive or deficient cortisol in the body.<sup>3</sup>

**Alanine aminotransferase (ALT):** An enzyme found mostly in the liver; smaller amounts of it are also in the kidneys, heart, and muscles. A blood test can be done to measure the level of ALT.<sup>5</sup> When the liver is damaged, such as by some drugs or viruses, ALT is released into the blood stream, usually before more obvious symptoms of liver damage occur, such as jaundice (yellowing of the eyes and skin).<sup>3</sup>

**Alkaline phosphatase (Alk Phos or ALP):** A protein found in all body tissues. Tissues with particularly high amounts of ALP include the liver, bile ducts, and bone. A blood test can be done to measure the level of ALP.<sup>5</sup>

**Amniocentesis:** A prenatal test usually performed in the 15th to 17th week of pregnancy. A needle is inserted through the abdomen or through the cervix into the uterus, and amniotic fluid is extracted. Cells are studied for the detection of chromosome abnormalities, either abnormal numbers of chromosomes (as in Down syndrome, in which there are three chromosome 21s) or hypersensitivity to DEB (as in patients with FA). These fetal cells can also be tested for HLA matching.<sup>3</sup>

**Anastomosis:** The surgical union of parts and especially hollow tubular parts, such as the anastomosis of the ureter and colon.<sup>1</sup>

**Androgens:** Artificial male hormones that may stimulate production of one or more types of blood cells for extended periods of time in FA patients.<sup>2</sup> Androgens are also normally made in boys during puberty and in adult men.

**Anemia:** Decrease in the oxygen-carrying capacity of the blood; indicated by a low red blood cell count, low hemoglobin, low hematocrit.<sup>2</sup>

**Angiography:** The radiographic visualization of the blood vessels after injection of a radiopaque substance (anything that does not let x-rays or other types of radiation penetrate).<sup>1</sup>

**Antibody:** A complex molecule produced by certain blood cells in response to stimulation by an antigen. Antibodies bind to antigens, thus marking them for removal or destruction. The marked antigens are then destroyed by other blood cells.<sup>2</sup>

**Antigens:** Proteins present on the surface of all cells, bacteria, and viruses. Bodies are accustomed to their own antigens and usually don't attack them, but the body considers foreign antigens (such as bacteria, viruses, or grains of pollen) dangerous and will attack them. Bone marrow transplant specialists look for "matching" HLA antigens on the white cells. These antigens can help predict the likely success of a marrow transplant.<sup>2</sup>

**Anti-thymocyte globulin (ATG):** A purified gamma immunoglobulin (IgG) with immunosuppressive activity which specifically recognizes and destroys T lymphocytes. Administering antithymocyte globulin with chemotherapy prior to stem cell transplantation may reduce the risk of graft-versus-host disease.<sup>8</sup>

**Aperistalsis:** Absence of peristalsis, which is successive waves of involuntary contraction passing along the walls of the esophagus or intestine and forcing the contents onward.<sup>6</sup> Common but transient complication during BMT or after surgery.

**Apheresis:** Withdrawal of blood from a donor's body, removal of one or more components (such as plasma, blood platelets, or white blood cells) from the blood, and transfusion of the remaining blood back into the donor; also called pheresis.<sup>1</sup>

**Aplasia:** Lack of development of an organ or tissue, or of the cellular products from an organ or tissue. In the case of FA, this term refers to lack of adequate blood cell production from the bone marrow. Also refers to the lack of thumb and radius in some FA patients.<sup>2</sup>

**Aplastic anemia:** Failure of the bone marrow (aplasia) to produce one or more of the three blood cell types (red blood cells, white blood cells, or platelets). Anemia

typically refers to decreased hemoglobin in red blood cells but, when used in this context, refers to any new blood cells. Bone marrow biopsy results reveal a lower number of blood cells than normal.<sup>9</sup>

**Aspartate aminotransferase (AST):** An enzyme found in liver cells. Testing for AST is usually done to detect liver damage. AST levels are also often compared with levels of other liver enzymes, ALP, and ALT, to determine which form of liver disease is present.<sup>3</sup> A blood test can be done to measure the level of AST.<sup>5</sup>

**Atresia:** Absence or closure of a natural passage of the body, such as of the small intestine or absence or disappearance of an anatomical part (such as an ovarian follicle) by degeneration.<sup>1</sup>

**Audiogram:** A graphic representation of the relation of sound or acoustic frequency and the minimum sound intensity for a hearing test to determine hearing loss.<sup>1</sup>

**Autoimmune hemolytic anemia:** A drop in the number of red blood cells due to increased destruction by the body's defense (immune) system.<sup>5</sup>

**Autologous stem cells:** Bone marrow stem cells derived from the patient.

**Autosomal recessive:** One of several ways that a trait, disorder or disease can be inherited. An autosomal recessive disorder means that two copies of an abnormal gene must be present in order for the disease or trait to appear. Genes are found in pairs, one from the mother and one from the father. Recessive inheritance means both genes in a pair must be defective to cause disease. People with only one gene that is not working in the pair do not have the disease but are carriers. They can pass the non-working gene to their children.<sup>5</sup>

**Avascular necrosis (AVN):** Avascular necrosis occurs when part of the bone does not get blood and dies. If this condition is not treated, bone damage gets worse. Eventually, the healthy part of the bone may collapse.<sup>5</sup>

**Azospemia:** Lack of sperm.<sup>1</sup>

**B cells:** Type of lymphocytes responsible for antibody production.

**Baseline test:** Test which measures an organ's normal level of functioning. Used to determine if any changes in organ function occur following treatment.<sup>2</sup>

**Basophil:** Type of white blood cell; a type of granulocyte, involved in allergic reactions.<sup>2</sup>

**Bicornuate uterus:** Commonly referred to as a heart-shaped uterus, is a type of a uterine malformation where two horns form at the upper part of the uterus.<sup>1</sup> This is one example of a congenital uterine malformation. These malformations do not cause infertility. The extent and location of the malformation can affect the likelihood of a pregnancy reaching full-term. Sometimes called hemi-uterus.

**Bifid:** Separated or cleft into two parts. In FA patients, most commonly refers to a thumb abnormality.

**Biliary:** Of, relating to, or conveying bile.<sup>1</sup>

**Biliary ducts:** Ducts by which bile passes from the liver or gallbladder to the duodenum.<sup>1</sup>

**Bilirubin:** A product that results from the breakdown of hemoglobin. Total and direct bilirubin are usually measured to screen for or to monitor liver or gallbladder problems.<sup>5</sup>

**Blast cell:** An immature cell. Too many blast cells in the bone marrow or blood may indicate the onset of leukemia.<sup>2</sup>

**Blind loop syndrome:** Occurs when part of the intestine becomes blocked, so that digested food slows or stops moving through the intestines. This causes bacteria to overgrow in the intestines and causes problems in absorbing nutrients.<sup>5</sup>

**Blood urea nitrogen (BUN):** Urea nitrogen is what forms when protein breaks down. A test can be done to measure the amount of urea nitrogen in the blood.<sup>5</sup>

**Bone marrow:** Soft tissue within the bones where blood cells are manufactured.<sup>2</sup>

**Bone marrow aspiration:** Test in which a sample of bone marrow cells is removed with a sturdy needle and examined under a microscope. Aspirates are used to examine more specifically the types of cells in the bone marrow, and the chromosomal pattern.<sup>2</sup>

**Bone marrow biopsy:** Procedure in which a special type of needle is inserted into the bone, and a piece of bone (a plug) with marrow is removed. This test is very helpful in assessing the architecture and arrangement of cells within the bone marrow. Commonly used to test for cellularity of the bone marrow.

**Bone mineral density (BMD) test:** Used to assess for osteopenia or osteoporosis.<sup>5</sup>

**Brainstem-evoked auditory response (BAER):** A test to measure the brain wave activity that occurs in response to clicks or certain tones. The test is done to help diagnose nervous system problems and hearing losses (especially in low birth weight newborns), and to assess neurological functions.<sup>5</sup>

**Bullae:** Bullae are blisters wider than 1 centimeter. Bullae that are filled with clear fluid may occur on the skin.<sup>5</sup>

**Café au lait spot:** A birthmark that is light tan, the color of coffee with milk.<sup>5</sup>

**Cardiomyopathy:** A weakening of the heart muscle or a change in heart muscle structure, often associated with inadequate heart pumping or other heart function problems.<sup>5</sup>

**Carpectomy:** Removal of a carpal bone(s).

**Carpus:** The group of bones supporting the wrist.<sup>1</sup>

**Central line:** See **Hyperalimentation**.

**Chelation:** The use of a chelator (an organic chemical that bonds with and removes free metal ions) to bind with a metal (such as iron) in the body. Chelation may inactivate and/or facilitate excretion of a toxic metal. In FA patients, most often refers to a method for getting rid of excess iron.

**Cholestasis:** Any condition in which the flow of bile from the liver is blocked. Blood tests may show higher than normal levels of bilirubin and alkaline phosphatase. Imaging tests are used to diagnose this condition.<sup>5</sup>

**Chorionic villus sampling (CVS):** An early prenatal diagnostic test. In the first trimester of pregnancy, an instrument is inserted vaginally or through the abdomen into the uterus under ultrasound guidance to identify the placenta and the fetus. Villus cells, which later form part of the placenta, are removed. These cells are then studied for chromosome abnormalities, either for abnormal numbers of chromosomes (as in Down syndrome, where there are three chromosome 21s) or

hypersensitivity to DEB (as in patients with FA). These cells may also be tested for HLA matching.

**Chromosomes:** Structures in the cell nucleus which contain the genes responsible for heredity. Normal human cells contain twenty-three pairs of chromosomes. One of each pair is inherited separately from a person's father and mother.<sup>2</sup>

**Cirrhosis:** Scarring of the liver and poor liver function as a result of chronic liver disease.<sup>5</sup>

**Clastogen:** An agent that causes breaks in chromosomes.<sup>6</sup>

**Colony stimulating factors (also known as hematopoietic growth factors or cytokines):** Substances produced naturally by the body (and also synthetically) which stimulate the production of certain blood cells. Examples are G-CSF (Neupogen), GM-CSF, various "interleukins," stem cell factor (or steel factor), erythropoietin (EPO, Epogen), etc.<sup>2</sup>

**Colposcopy:** An examination by means of a colposcope, a magnifying instrument designed to facilitate visual inspection of the vagina and cervix. The instrument usually contains a green filter which enables the clinician to see abnormal vessels related to any lesions.

**Comparative genomic hybridization (CGH):** A fluorescent molecular cytogenetic technique that identifies DNA gains, losses, and amplifications, mapping these variations to normal chromosomes. It is a powerful tool for screening chromosomal copy number changes in tumor genomes and has the advantage of analyzing entire genomes within a single experiment.<sup>10</sup>

**Complementation groups:** When a mutant (or defective) cell is able to restore normal function to (or complement) another defective cell, the mutations in those cells are said to be in different complementation groups. That means the mutations are in different genes. If a mutant or defective cell is not able to restore normal function to another defective cell, the mutations are said to be in the same complementation group (in other words, in the same gene).<sup>2</sup>

**Complete blood count (CBC):** Gives the number, and/or percentage, and/or characteristics of certain blood cells, primarily white cells, red cells, and platelets.

**Computed tomography (CT, aka CT scan):** An imaging method that uses x-rays to create cross-sectional pictures of the body.<sup>5</sup>

**Consanguinity:** Relationship by blood via descent from the same ancestor, and not by marriage or affinity.

**Cortisol level:** A blood test that measures the amount of cortisol, a steroid hormone produced by the adrenal cortex in response to a hormone called ACTH (produced by the pituitary gland). Cortisol levels are often measured to evaluate how well the pituitary and adrenal glands are working.<sup>5</sup>

**Creatinine:** The creatinine blood test is usually ordered along with a BUN (blood urea nitrogen) test to assess or monitor kidney function.

**Cryptorchidism:** The condition that occurs when one or both testicles fail to descend into the scrotum before birth.<sup>5</sup>

**Culture:** A specimen of blood, urine, sputum or stool which is taken and grown in the laboratory. This culture

is then tested to determine whether infection is present and which antibiotic to use.<sup>2</sup>

**Cytokines:** See **Colony stimulating factors**.<sup>2</sup>

**Cytomegalovirus (CMV):** A condition caused by a member of the herpesvirus family which can cause disease in different parts of the body in people with weakened immune systems, such as during a bone marrow transplant.<sup>5</sup>

**Cytopenia:** A deficiency of cellular elements of the blood, especially deficiency of one or more specific elements (as granulocytes in granulocytopenia).<sup>1</sup>

**Dermatofibroma:** A benign, chiefly fibroblastic, nodule of the skin, found especially on the extremities of adults.<sup>1</sup>

**Desquamation:** Shedding of the outer layers of the skin; peeling off in the form of scales.<sup>1</sup>

**Diepoxybutane (DEB):** A chemical agent that damages DNA in cell culture and is used in a diagnostic test for FA, either before or after birth.<sup>2</sup>

**Differential:** Percent of different types of white blood cells in the blood.<sup>2</sup>

**DNA:** This abbreviation stands for deoxyribonucleic acid. DNA is the component of the chromosomes that carries the genetic code.<sup>2</sup>

**DNA repair:** A collection of processes by which a cell identifies and corrects damage to the DNA molecules that encode its genome. The DNA repair ability of a cell is vital to the integrity of its genome and thus to its normal functioning and that of the organism.<sup>11</sup> This is abnormal in patients with FA.

**Dorsal:** Being or located near, on, or toward the back or posterior part of the human body.<sup>1</sup>

**Dual energy absorptiometry (DXA or DXA-scan):** The primary test used to identify osteoporosis and low bone mass. It uses a low energy x-ray to evaluate bone density in the hip and/or spine and sometimes the wrist.

**Dyslipidemia:** A disorder characterized by high blood cholesterol and triglycerides in the blood. A lipid disorder increases the risk for atherosclerosis and heart disease.<sup>5</sup>

**Dysphagia:** Difficulty swallowing.<sup>5</sup>

**Dysplasia:** A deleterious change in the microscopic appearance of cells. Can suggest early stages of progression to cancer.

**Ectopic:** Occurring in an abnormal position, such as a pregnancy occurring in a Fallopian tube instead of in the uterus.<sup>1</sup>

**Encopresis:** The voluntary or involuntary passage of stools in a child over age four, which causes the soiling of clothes.<sup>5</sup>

**Endoscope:** A medical device consisting of a camera mounted on a flexible tube. Small instruments can be used to take samples of suspicious tissues through the endoscope. In gastrointestinal (digestive tract) endoscopy, this device is inserted through the mouth or anus.<sup>5</sup>

**Enteral alimentation:** Tube feeding whereby liquid food is given through a tube into the stomach or small bowel.<sup>12</sup>

**Eosinophil (EOS):** A type of white blood cell; a type of granulocyte.<sup>2</sup>

**Epicanthal folds:** Skin of the upper eyelid, from the nose to the inner side of the eyebrow, that covers the inner corner (canthus) of the eye. The presence of an epicanthal fold is normal in people of Asiatic descent. However, it may also be due to certain medical conditions.<sup>5</sup>

**Epiphysis:** The growth area near the end of long bones. A part or process of a bone that ossifies separately and, at the end of childhood growth, becomes fused to the main part of the bone, especially an end of a long bone. This signals the end of linear growth.

**Epithelium:** Cells that line hollow organs and glands and those that make up the outer surface of the body to protect or enclose organs. Most produce mucus or other secretions. Certain types of epithelial cells have tiny hairs called cilia, which help remove foreign substances, for example, from the respiratory tract. Epithelial cells are arranged in single or multiple layers, depending on the organ and location.<sup>5</sup>

**Epstein-Barr virus (EBV):** A herpesvirus that causes infectious mononucleosis and is associated with Burkitt's lymphoma and nasopharyngeal carcinoma.<sup>1</sup> Reactivation of the virus after organ or bone marrow transplant can result in post-transplant lympho-proliferative disease (PTLD) or lymphoma.

**Erythroblast:** An immature red blood cell.<sup>2</sup>

**Erythrocyte:** Red blood cell; red blood cells go through various stages, starting out as erythroblasts, changing to reticulocytes, and finally becoming erythrocytes.<sup>2</sup>

**Erythrocyte sedimentation rate:** Used to detect and monitor the activity of inflammation as an aid in the diagnosis and activity of the underlying cause.<sup>3</sup>

**Erythroderma:** Generalized skin redness.<sup>5</sup>

**Erythroplasia (Erythroplakia):** A reddened patch with a velvety surface on the oral or genital mucosa that is considered to be a precancerous lesion.<sup>1</sup>

**Erythropoietin (EPO):** A colony-stimulating factor which influences red cell production in some conditions.<sup>2</sup>

**Esophageal atresia (EA):** A disorder of the digestive system in which the esophagus does not develop properly. There are several types. In most cases, the upper esophagus ends and does not connect with the lower esophagus and stomach. The top end of the lower esophagus connects to the windpipe. This connection is called a tracheoesophageal fistula (TEF). Other types of esophageal atresia involve narrowing of the esophagus, and may also be associated with other birth defects.<sup>5</sup>

**Esophagoscopy:** Examination of the esophagus by means of an esophagoscope, a thin, tube-like instrument with a light and a lens for viewing. It may also have a tool to remove tissue to be checked under a microscope for signs of disease.<sup>8</sup>

**Extramedullary leukemia:** The occurrence of myelogenous leukemia outside the bone marrow, such as in the spinal fluid, testes or ovaries, or skin.<sup>13</sup>

**Ferritin:** The form in which iron is stored in the body.

**5'-nucleotidase (5'-NT):** 5'-nucleotidase is a protein produced by the liver; the test measures the amount of this protein in the blood.<sup>5</sup>

**Fluorescent *in situ* hybridization (FISH):** A research method to visualize and map the genetic material in an individual's cells, including specific genes or portions of genes. This is important for understanding a variety of chromosomal abnormalities and other genetic mutations. Unlike most other techniques used to study chromosomes, FISH does not have to be performed on cells that are actively dividing.<sup>14</sup> This makes it a very sensitive procedure for detecting chromosomal abnormalities.

**Folate deficiency (aka folic acid):** A type of B vitamin. Folic acid is found naturally in dark-green leafy vegetables, citrus fruits, beans, and whole grains. Not getting enough folate results in a form of megaloblastic anemia.<sup>5</sup>

**Follicle stimulating hormone (FSH):** A hormone from the anterior lobe of the pituitary gland that stimulates the growth of the ovum-containing follicles in the ovary in women and that activates sperm-forming cells in men. At menopause in women, the level stays persistently high.

**Forced Expiratory Volume (FEV1):** The volume of air that can be forced out in one second after taking a deep breath, an important measure of pulmonary function.<sup>15</sup>

**Founder effect:** The effect on the resulting gene pool that occurs when a new isolated population is founded by a small number of individuals possessing limited genetic variation relative to the larger population from which they have migrated.<sup>16</sup> If one or more members of the founder group were carriers of an FA gene mutation, the descendants would be more likely to contain carriers of that mutated gene.

**Fructosamine:** A protein that attaches to glucose in the bloodstream. If a patient's fructosamine is elevated, then the patient's average glucose level over the previous 2 to 3 weeks has also been elevated.<sup>17</sup>

**Gamma-glutamyl transpeptidase (GGT):** A test to measure the amount of the enzyme GGT in the blood to detect diseases of the liver, bile ducts, and kidney. It is also used to differentiate liver or bile duct disorders from bone disease. High concentrations of GGT are found in the liver, bile ducts, and the kidney. GGT is measured in combination with other tests. In particular, the enzyme ALP is increased in liver and bile duct disease as well as in bone disease. GGT is elevated in liver and bile duct disease, but not in bone disease. So, a patient with an elevated ALP and a normal GGT probably has bone disease, not liver or bile duct disease.<sup>5</sup>

**Gastritis:** Gastritis is an inflammation of the lining of the stomach.<sup>5</sup> It may be accompanied by pain and bleeding.

**Gastroesophageal reflux (GER):** A condition in which food or liquid travels backwards from the stomach to the esophagus.<sup>5</sup>

**Gastroesophageal reflux disease (GERD):** Heartburn and other disease occurring as a result of irritation of the esophagus.

**Gastroparesis:** Delayed gastric emptying; a condition in which the stomach's ability to empty its contents is impaired, unrelated to obstruction.<sup>5</sup>

**Gastrostomy:** The surgical formation of an opening through the abdominal wall into the stomach, often used to provide nutrition.<sup>1</sup>

**Giardia:** A microscopic organism, *Giardia lamblia*, that causes Giardiasis, an infection of the small intestine.<sup>5</sup>

**Gingival atrophy:** Recession of the gums around the teeth.

**Glomerular filtration rate (GFR):** A test used to check how well the kidneys are working. Specifically, it estimates how much blood passes through the tiny filters in the kidneys, called glomeruli, each minute. The GFR test measures how well kidneys are filtering a waste called creatinine, which is produced by the muscles. When the kidneys aren't working as well as they should, creatinine builds up in the blood.<sup>5</sup>

**Glycosylated hemoglobin (HbA1c):** The test represents average blood sugar over the life span of the red cell by measuring glycated hemoglobin in the blood. Glycated hemoglobin is a substance in red blood cells formed when blood sugar (glucose) attaches to hemoglobin. It can measure blood sugar control over several months and can give a good estimate of how well a patient has managed diabetes over the last 2 or 3 months.<sup>5</sup>

**GM-CSF:** Drug that stimulates the marrow to make more white blood cells. See **Colony-stimulating factors**.

**Gonadotropin releasing hormone (GnRH):** Normally, the hypothalamus in the brain releases GnRH, which stimulates the pituitary gland to release other hormones, including FSH and LH. These hormones then stimulate the female ovaries and male testes to secrete hormones that are responsible for normal sexual development in puberty and are important to the process of ovulation in females. A disruption in this chain of events causes a

deficiency of the sex hormones (estrogen and testosterone) and halts normal sexual maturation.<sup>5</sup>

**Graft-versus-host disease (GvHD):** A complication of bone marrow transplantation which occurs when donor T-cells attack the patient's cells. GvHD is more likely to occur when there is HLA mismatching. GvHD is classified in stages from Grade I (minor) to Grade IV (extremely serious).<sup>2</sup>

**Granulocyte:** Type of white blood cell. It is also called neutrophil or polymorphonuclear leukocyte (poly), which is the infection-fighting cell.

**Gynecomastia:** The development of large breasts in males. Gynecomastia during puberty is not uncommon and usually goes away over a period of months.<sup>5</sup>

**H2-antagonists:** Medicines that help decrease stomach acid.<sup>5</sup>

**Hematocrit:** Ratio of red blood cells to plasma in the blood; portion of the blood's total volume that is made up of red blood cells.<sup>2</sup>

**Hematopoiesis:** The formation and development of blood cells.<sup>2</sup>

**Hematopoietic growth factors:** See **Colony stimulating factors**.

**Hemochromatosis (aka iron overload):** Occurs when too much iron builds up in the liver. This leads to liver enlargement. In Fanconi anemia, this is usually caused from ineffective blood production, abnormal iron absorption, or receiving a large number of blood transfusions, which boost iron levels.

**Hemodynamic:** Relating to or functioning in the mechanics of blood circulation.<sup>1</sup>

**Hemoglobin:** The oxygen-carrying pigment of the red blood cells; combines with oxygen from the lungs and carries it to the body's cells.<sup>2</sup>

**Hemoglobinopathy:** A blood disorder (such as sickle-cell anemia) caused by a genetically determined change in the molecular structure of hemoglobin.<sup>1</sup>

**Hepatic sinusoids:** A minute endothelium-lined space or passage for blood in the tissues of the liver.<sup>1</sup>

**Hepatitis:** Inflammation of the liver.<sup>5</sup>

**Hepatocellular carcinoma:** Cancer of the liver.<sup>5</sup>

**Hepatomegaly:** Enlarged liver.<sup>5</sup>

**Herpes simplex (HSV):** An infection that mainly affects the mouth or genital area. There are two strains of HSV: **Type 1 (HSV-1)** is usually associated with infections of the lips, mouth, and face. It is transmitted by contact with infected saliva. By adulthood, up to 90% of people will have antibodies to HSV-1. **Type 2 (HSV-2)** is sexually transmitted. Symptoms include genital ulcers or sores. However, some people have HSV-2 but do not show symptoms. Up to 30% of U.S. adults have antibodies against HSV-2. Cross-infection of type 1 and 2 viruses may occur from oral-genital contact.<sup>5</sup> Tested prior to transplant because it can reactivate during periods of immune suppression.

**Heterozygotes:** Everyone has two copies of nearly all of his genes. Heterozygous means that one of the copies of a gene is slightly different from the other copy of the gene. One gene may have an FA mutation and the other may not (i.e., a carrier is heterozygous). An individual with FA may be heterozygous if he has two different mutations in his FA genes.

**Homozygous:** Both copies of a gene are exactly the same. An individual with FA is homozygous if he has the same gene mutation in both copies of his FA genes.

**Human leukocyte antigen (HLA) tissue typing:** The tissue-typing test done on white cells to determine if a bone marrow donor and recipient are compatible.<sup>2</sup>

**Human papillomavirus (HPV):** There are more than 100 different types of HPV. Some types are associated with benign, common or flat warts on the skin. Some HPV types are spread primarily through sexual contact and can cause genital warts or cancer of the oropharynx, cervix, vulva, vagina, anus, and (rarely) penis.

**Hydronephrosis:** Bilateral hydronephrosis is the enlargement (distention) of the pelvis and urine collecting structures of both kidneys which occurs when urine is unable to drain from the kidney down the ureters into the bladder. It is not itself a disease, but rather a physical result of whatever disease is keeping urine from draining out of the kidneys, ureters, and bladder. Unilateral hydronephrosis is swelling of one kidney due to a backup of urine.<sup>5</sup>

**Hydroureter:** Abnormal distension of the ureter with urine or watery fluid, due to obstruction.<sup>1</sup>

**Hyperalimentation:** The administration of nutrients by intravenous feeding, especially to patients who cannot ingest food through the alimentary tract. Total parenteral nutrition (TPN) is a method of feeding that bypasses the gastrointestinal tract. Fluids are given into a vein to provide most of the necessary nutrients the body needs. An IV line is often placed into a vein in the hand, foot, or scalp. The belly button also has a large vein (umbilical vein) that may be used. Sometimes a longer IV, called a central line or peripherally-inserted

central catheter (PICC) line, is used to provide long-term IV fluids or feedings. This type of IV can deliver nutrients of higher concentration to larger veins located centrally in the body.<sup>1,5</sup>

**Hyperbilirubinemia:** The presence of an excess of bilirubin in the blood.<sup>1</sup>

**Hyperglycemia:** Abnormally high blood sugar.<sup>5</sup>

**Hyperlipidemia:** High blood cholesterol and/or triglycerides.<sup>5</sup>

**Hyperpigmentation:** Excess pigmentation in a body part or tissue (such as the skin).<sup>1</sup>

**Hyperplasia:** Hyperplasia is a thickening of normal tissue. Specifically, it is the increased cell production in a normal tissue or an organ. Hyperplasia may be a sign of precancerous changes.<sup>5</sup>

**Hypogonadism:** Occurs when the gonads produce little or no hormones. In men, the gonads are the testes; in women, they are the ovaries. In girls, hypogonadism during childhood will result in lack of menstruation and breast development and short height. If hypogonadism occurs after puberty in females, symptoms include loss of menstruation, low libido, hot flashes, and loss of body hair. In boys, hypogonadism in childhood results in lack of muscle and beard development and growth problems. In men the usual complaints are sexual dysfunction, decreased beard and body hair, breast enlargement, and muscle loss.<sup>5</sup>

**Hypoparathyroidism:** A condition in which the body produces too little parathyroid hormone. Calcium and phosphorus form the mineral component of bones. Parathyroid hormone (PTH) regulates the amount of calcium and phosphorus in bone and blood. PTH is

made by four small parathyroid glands located in the neck behind the thyroid gland. Hypoparathyroidism occurs when there is too little PTH. Blood calcium levels fall, and phosphorus levels rise.<sup>5</sup>

**Hypopharynx:** The lower part of the pharynx and the part of the throat that connects to the esophagus.

**Hypopigmentation:** Diminished pigmentation in a body part or tissue (such as the skin).<sup>1</sup>

**Hypoplastic:** A condition of arrested development in which an organ or part remains below the normal size or in an immature state.<sup>1</sup>

**Hypospadias:** A relatively common congenital defect in which the opening of the urethra is on the underside, rather than at the end, of the penis. Infants with hypospadias should not be circumcised; the foreskin should be preserved for use in later surgical repair.<sup>5</sup>

**Hypothalamus:** An area of the brain that produces chemical messages that control body temperature, hunger, moods, release of hormones from many glands, especially the pituitary gland. These messages also influence sex drive, sleep, and thirst.<sup>5</sup>

**Hypothyroidism:** A condition in which the thyroid gland fails to produce enough thyroid hormone.<sup>5</sup>

**Ileus:** Obstruction of the bowel; the condition is commonly marked by a painful distended abdomen, vomiting of dark or fecal matter, toxemia, and dehydration that results when the intestinal contents back up because peristalsis fails, although the lumen (the inside space of a tubular structure, in this case the bowel) is not occluded.<sup>1,11</sup>

**IM:** Refers to giving an injection intramuscularly.

**Immune response:** The body's defense against disease and foreign substances, including transplanted bone marrow; substances may be recognized as "foreign" and then killed by other cells.<sup>2</sup>

**Immunoglobulin:** A protein produced by plasma cells and lymphocytes and characteristic of these types of cells. Immunoglobulins play an essential role in the body's immune system. They attach to foreign substances, such as bacteria, and assist in destroying them. Immunoglobulin is abbreviated Ig. The classes of immunoglobulins are termed Immunoglobulin A (IgA), Immunoglobulin D (IgD), Immunoglobulin E (IgE), Immunoglobulin G (IgG), and Immunoglobulin M (IgM).<sup>15</sup>

**Immunosuppression:** Suppression (as by drugs) of natural immune responses necessary to fight disease or a harmful substance.<sup>1,5</sup>

**Imperforate anus:** A congenital defect in which the opening to the anus is missing or blocked. It may occur in several forms. The rectum may end in a blind pouch that does not connect with the colon. Or, it may have openings to the urethra, bladder, base of penis or scrotum in boys, or vagina in girls. In girls, it is associated with malformations of the uterus and vagina which can affect sexual function and fertility. A condition of stenosis (narrowing) of the anus or absence of the anus may be present.<sup>5</sup>

**Insulin-like growth factor 1 (IGF-1):** The hormone produced by the liver, bones, and other tissues in response to growth hormone, with production declining after puberty.

**Intravenous (IV):** Injection directly into the vein.<sup>2</sup>

***In vitro* fertilization (IVF):** Fertilization of an egg in a laboratory dish or test tube via a mixture of sperm with eggs. The eggs are surgically removed from an ovary, mixed with the sperm, and then one or more of the resulting fertilized eggs are implanted into a female's uterus.<sup>1</sup>

**Jejunum:** The section of the small intestine that comprises the first two-fifths beyond the duodenum and that is larger, thicker-walled, more vascular and with more circular folds and fewer Peyer's patches than the ileum.<sup>1</sup>

**Karnofsky Performance Status (KPS):** A standard way of measuring the ability of cancer or bone marrow transplant patients to perform ordinary tasks. The Karnofsky Performance Status scores range from 0 to 100. A higher score means the patient is better able to carry out daily activities. KPS may be used to determine a patient's prognosis, to measure changes in a patient's ability to function, or to decide if a patient could be included in a clinical trial.<sup>4</sup>

**Keratoconjunctivitis sicca syndrome:** Dry eye syndrome in which the tear glands produce fewer tears than normal.<sup>5</sup>

**Klippel-Feil Syndrome:** A rare disorder characterized by the congenital fusion of any 2 of the 7 cervical (neck) vertebrae. The most common signs of the disorder are short neck, low hairline at the back of the head, and restricted mobility of the upper spine.<sup>18</sup>

**Lagophthalmos:** Pathological incomplete closure of the eyelids; inability to close the eyelids fully.<sup>1</sup>

**Lansky status:** A performance test to quantify cancer or bone marrow transplant patients' general wellbeing to determine whether they can receive chemotherapy,

whether dose adjustment is necessary, and as a measure for the required intensity of palliative care. It is also used in cancer randomized controlled trials as a measure of quality of life. Children, who might have more trouble expressing their experienced quality of life, are evaluated according to the Lansky status criteria, using a somewhat more observational scoring system, such as “fully active, normal” to “doesn’t play, does not get out of bed” to “unresponsive.”<sup>19</sup>

**Larynx:** The upper part of the respiratory passage that is bounded above by the glottis, is continuous below with the trachea, has a complex cartilaginous or bony skeleton capable of limited motion through the action of associated muscles, and has a set of elastic vocal cords that play a major role in sound production and speech; also called “voice box.”<sup>1</sup>

**Leukemia:** Leukemia is a group of bone marrow diseases involving an uncontrolled increase in white blood cells (leukocytes). The leukemia most commonly acquired by FA patients is acute myelogenous leukemia.<sup>5</sup>

**Leukocytes:** White blood cells.<sup>2</sup>

**Leukopenia:** Low white cell count.<sup>2</sup>

**Leukoplakia:** A condition commonly considered precancerous in which thickened white patches of epithelium occur on the mucous membranes especially of the mouth, vulva, and renal pelvis.<sup>1</sup>

**Lichen planus:** A condition of the genitals, skin or mouth that results in an itchy, swollen rash on the genitals (typically on the vulva); tender, painful, bluish-white lesions in the mouth; and/or shiny, scaly, flat-topped, purplish-pink bumps on the skin.

**Luteinizing hormone (LH):** A glycoprotein hormone that in the female stimulates ovulation and, together with follicle-stimulating hormone, secretion of estrogen from developing ovarian follicles. In the male stimulates the development of interstitial tissue in the testis and the secretion of testosterone.<sup>1</sup>

**Lymphadenopathy:** Abnormal enlargement of the lymph nodes.<sup>1</sup>

**Lymphocyte:** Type of white blood cell that fights infection by producing antibodies and other protective substances; occurs in two forms: B cells that recognize specific antigens and produce antibodies against them, and T-cells that recognize specific antigens, release factors that attract other T-cells, natural killer cells and macrophages to remove foreign cells and microbial pathogens. Lymphocytes are produced in the lymph system, not in the bone marrow.<sup>2</sup>

**Lymphoid:** Referring to lymphocytes, a type of white blood cell. Also refers to tissue in which lymphocytes develop.<sup>4</sup>

**Lymphoma:** Cancer that begins in cells of the immune system. There are two basic categories of lymphomas. One is Hodgkin lymphoma, which is marked by the presence of a type of cell called the Reed-Sternberg cell. The other category is non-Hodgkin lymphomas, which includes a large, diverse group of cancers of immune system cells; some of these cancers have a slow-growing course and some have a fast-growing course. They behave and respond to treatment differently. Both Hodgkin and non-Hodgkin lymphomas can occur in children and adults, and prognosis and treatment depend on the stage and the type of cancer.<sup>4</sup>

**Macrocyte:** An abnormally large erythrocyte (red blood cell).<sup>2</sup>

**Macrophage:** A type of white blood cell that assists in the body's fight against bacteria and infection by engulfing and destroying invading organisms.<sup>2</sup>

**Matched platelet transfusions:** Transfusions from a donor who has been HLA-matched to a particular patient.<sup>2</sup>

**Medulloblastoma:** A malignant brain tumor that begins in the lower part of the brain and that can spread to the spine or to other parts of the body.<sup>4</sup>

**Megakaryocyte:** Large cell in the bone marrow from which pieces break off to form platelets.<sup>2</sup>

**Menopause:** The transition period in a woman's life when her ovaries stop producing eggs, her body produces less estrogen and progesterone, and menstruation becomes less frequent, eventually stopping altogether.<sup>5</sup>

**Mesenchymal stromal cells (MSC):** Multipotent stem cells that can differentiate into a variety of cell types, such as bone cells, fat cells, and cartilage cells. MSC are typically isolated from bone marrow.

**Metacarpus:** The part of the hand (metacarpal) or foot (metatarsal) between the carpus and the phalanges that contains five elongated bones when all the digits are present.

**Microcephaly:** A condition of abnormal smallness of the head.<sup>1</sup>

**Micrognathia:** Abnormal smallness of one or both jaws.<sup>1</sup>

**Micropenis:** Smallness of the penis, especially to an abnormal degree.<sup>1</sup>

**Microphthalmia:** Abnormal smallness of the eye, usually occurring as a congenital anomaly.<sup>1</sup>

**Mitomycin C (MMC):** A chemical which, in sufficient doses, causes destruction and rearrangement of the chromosomes in cells. Because Fanconi anemia cells are unusually sensitive to MMC, it is used to diagnose this condition.<sup>2</sup>

**Motile or motility:** Exhibiting or capable of movement.<sup>1</sup>

**Murine model:** A model of disease for use in research, using mice as the model animal for preclinical experimentation.<sup>1</sup>

**Mutation:** A mutation is a change in the DNA of a gene that causes the gene either not to make any protein or to change the protein so that it does not work correctly. DNA is made up of the chemical letters A, C, T, and G. A mutation can be the addition, deletion, or simply a change in the chemical letters of DNA that makes up the gene.

**Myelodysplasia (MDS or myelodysplastic syndrome):** Abnormal production, maturation, and appearance of blood cells; often leading to deficiency of red cells, white cells and platelets; sometimes leading to bone marrow failure or leukemia.<sup>2</sup>

**Myelosuppressive:** Suppression of the bone marrow's production of blood cells and platelets.<sup>1</sup>

**Myocardium:** The middle muscular layer of the heart wall.<sup>1</sup>

**Myofasciitis:** Inflammation of a muscle and its fascia.<sup>20</sup>

**Nasogastric tube:** A tube inserted through the nasal passages into the stomach, often used to provide nutrition.

**Nasojejunal tube:** A soft feeding tube inserted through the nasal passages into the small bowel. It is placed by using an x-ray monitor to guide the tube.

**Nasopharynx:** The upper part of the pharynx continuous with the nasal passages.<sup>1</sup>

**Neck dissection:** Surgery to remove lymph nodes and other tissues in the neck.<sup>8</sup>

**Neoplasia:** Abnormal cell growth resulting in formation of either benign, precancerous or cancerous tumors.

**Neuroblastoma:** Cancer that arises in immature nerve cells and affects mostly infants and children.<sup>4</sup>

**Neutropenia:** Low neutrophil (poly) count.<sup>2</sup>

**Neutrophil:** Type of white blood cell; also called a poly; granulocyte; the body's primary defense against harmful bacteria.<sup>2</sup>

**Nevi:** A congenital or acquired usually highly pigmented area on the skin that is either flat or raised, such as a mole.<sup>1</sup>

**Non-alcoholic steato hepatitis (NASH):** A common, often silent, liver disease. It resembles alcoholic liver disease, but occurs in people who drink little or no alcohol. The major feature is fat in the liver, along with inflammation and damage. NASH can be severe and can lead to cirrhosis, in which the liver is permanently damaged, scarred, and no longer able to work properly.<sup>7</sup>

**Odynophagia:** Pain produced by swallowing.<sup>1</sup>

**Oral glucose tolerance test (OGTT):** An OGTT is a series of blood glucose tests. Blood samples for fasting glucose and insulin are collected; then the patient drinks a standard amount of a glucose solution to challenge his

or her system. This is followed by one or more additional glucose and insulin tests performed at specific intervals to track glucose and insulin levels over time. The OGTT may be ordered to help diagnose diabetes and as a follow-up test to an elevated blood glucose.<sup>3</sup>

**Oropharynx:** The part of the pharynx that is below the soft palate and above the epiglottis and is continuous with the mouth.<sup>1</sup>

**Osteosarcoma:** A malignant tumor derived from bone or containing bone tissue; also called osteogenic sarcoma.<sup>1</sup>

**Osteotomy:** A surgical operation in which a bone is divided or a piece of bone is excised (as to correct a deformity).<sup>1</sup>

**Otoacoustic emission test (OAE):** A test that can show whether parts of the ear respond properly to sound. During this test, a sponge earphone is placed into the ear canal. The ear is stimulated with sound, and the echo is measured. The echo is found in everyone who hears normally. If there is no echo, it could indicate a hearing loss.<sup>21</sup>

**Ototoxic:** Having an adverse effect on organs or nerves involved in hearing or balance.<sup>5</sup>

**Oxidative stress:** Physiological stress on the body that is caused by the cumulative damage done by free radicals inadequately neutralized by antioxidants and that is held to be associated with aging and several congenital conditions, such as Fanconi anemia.<sup>1</sup>

**Pancytopenia:** Abnormally low number of red and white cells and platelets.<sup>2</sup>

**Panorex:** A dental x-ray taken outside of the mouth that shows all of the teeth on one view, in a panoramic fashion.<sup>22</sup>

**Parenteral infusion:** Situated or occurring outside the intestine, such as parenteral drug administration by intravenous, intramuscular, or subcutaneous injection; especially introduced otherwise than by way of the intestines.<sup>1</sup>

**Peliosis hepatis:** An abnormal condition characterized by the occurrence of numerous small blood-filled cystic lesions throughout the liver.<sup>1</sup>

**Peptic:** Refers to pepsin, a stomach enzyme that breaks down proteins. A peptic ulcer is erosion in the lining of the stomach or duodenum (the first part of the small intestine). If a peptic ulcer is located in the stomach, it is called a gastric ulcer.<sup>5</sup>

**Peripheral blood:** The blood in the bloodstream.<sup>2</sup>

**Peripherally inserted central catheter (PICC line):** A line inserted into a major vein to allow for administration of IV fluids or hyperalimentation.

**Peritonitis:** An inflammation (irritation) of the peritoneum, the tissue that lines the wall of the abdomen and covers the abdominal organs.<sup>5</sup>

**Petechiae:** Tiny red dots on the skin due to bleeding under the skin caused by low platelet count.<sup>2</sup>

**Phagocytosis:** Cell-eating. The engulfment and destruction of dangerous microorganisms or cells by certain white blood cells, including neutrophils (see **Absolute neutrophil count**).<sup>2</sup>

**Phalanx:** Any of the digital bones of the hand or foot distal to the metacarpals or metatarsals. Humans have

three digital bones in each finger and toe with the exception of the thumb and big toe, which have only two each.<sup>1</sup>

**Phlebotomy:** The letting of blood for transfusion, apheresis, diagnostic testing, or experimental procedures. In the past, phlebotomy was widely used to treat many types of disease but is now limited to the treatment of only a few specific conditions (such as hemochromatosis).<sup>1</sup>

**Photophobia:** Eye discomfort in bright light. Severe photophobia may be associated with eye problems and cause severe eye pain, even in relatively low light.<sup>5</sup>

**Pituitary:** A small gland joined to the hypothalamus (part of the brain). The pituitary produces many of the hormones that indirectly or directly affect basic bodily functions and include substances exerting a controlling and regulating influence on other endocrine organs, controlling growth and development, the body's response to stress, or modifying the contraction of smooth muscle, renal function, and reproduction.<sup>1,5</sup>

**Plasma:** A colorless fluid which contains water and other components in which red cells, white cells, and platelets are suspended.<sup>2</sup>

**Platelets:** Blood cell fragments containing clotting factors which prevent bleeding and bruising.<sup>2</sup>

**Pneumocystis:** A genus of microorganisms of uncertain affiliation that are usually considered protozoans or sometimes fungi and that include one (*P. carinii*) causing pneumonia, especially in immunocompromised individuals. This organism is common in the environment and does not cause illness in healthy people.<sup>1</sup> Transplant patients are typically given antibiotics to prevent pneumocystis.

**Polypharmacy:** The practice of administering many different medicines, especially concurrently for the treatment of the same disease.<sup>1</sup>

**Portal hypertension:** Hypertension in the hepatic portal system caused by venous obstruction or occlusion that produces splenomegaly (abnormal enlargement of the spleen) and ascites (abnormal accumulation of serous fluid in the abdominal cavity) in its later stages.<sup>1</sup>

**Post-prandial:** Occurring after a meal.<sup>1</sup>

**Prenatal diagnosis:** Many diseases that involve a single gene defect can now be diagnosed very early in pregnancy. Prenatal diagnosis looks at fetal cells in the mother's blood, amniotic fluid, or chorionic villi. This may detect problems while the baby is still growing or after birth. In late pregnancy, tests may examine blood from the umbilical cord.<sup>5</sup>

**Proband:** An individual being studied (as in a genetic investigation). Often the proband is the first affected family member who seeks medical attention.<sup>1</sup>

**Puberty:** The time during which sexual and physical characteristics mature due to hormonal changes. The exact age a child enters puberty depends on many factors, such as a person's genes, nutrition, and gender. During puberty, various endocrine glands produce hormones that cause body changes and the development of secondary sex characteristics. In girls, the ovaries begin to increase production of estrogen and other female hormones. In boys, the testicles increase production of testosterone. Breast development is the main sign that a girl is entering puberty. The first menstrual period (menarche) usually follows within about two years. The first sign of puberty in boys is enlargement of both testicles.<sup>5</sup>

**Radial ray deficiency:** The underdevelopment or total loss of the radius bone in the forearm. This condition often affects the development of the thumb. Some children with FA are born with no thumbs or thumbs not normally developed.<sup>5</sup>

**Radius:** The bone on the thumb side of the forearm.<sup>1</sup>

**Reactive airway disease:** The terms “reactive airway disease” and “asthma” are often used interchangeably; however, they’re not necessarily the same thing. Reactive airway disease is a general term that doesn’t indicate a specific diagnosis. It may be used to describe a history of coughing, wheezing or shortness of breath of unknown cause. These signs and symptoms may or may not be caused by asthma.<sup>9</sup>

**Recessive:** A mutation is said to be recessive if an individual must inherit two copies of the mutant gene, one from each parent, to show the mutant trait. Individuals with one mutant and one normal gene appear normal. They are called “carriers.”<sup>2</sup>

**Red blood cell (erythrocyte):** Oxygen-carrying cell in the blood which contains the pigment hemoglobin; produced in the bone marrow.<sup>2</sup>

**Refractory:** Resistant to treatment or cure.<sup>1</sup>

**Reticulocyte:** An immature red blood cell.<sup>2</sup>

**Retinoblastoma:** A cancer of the retina that generally affects children under the age of 6. It is most commonly diagnosed in children aged 1-2 years.<sup>5</sup>

**Schirmers’ test:** This test determines whether the eye produces enough tears to keep it moist.<sup>5</sup>

**Scleroderma:** A widespread connective tissue disease that involves changes in the skin, blood vessels,

muscles, and internal organs. Causes a build-up of collagen in the skin and other organs, which leads to the symptoms associated with the disease.<sup>5</sup>

**Sertoli cell mass:** Any of the elongated striated cells in the testis to which the spermatids become attached and from which they apparently derive nourishment.<sup>1</sup>

**Serum glutamic oxaloacetic transaminase (SGOT) and serum glutamic pyruvic transaminase (SGPT):** Enzymes that are normally present in liver and heart cells. SGOT and SGPT are released into blood when the liver or heart is damaged. The blood SGOT and SGPT levels are thus elevated with liver damage (such as from viral hepatitis) or with an insult to the heart (such as a heart attack). Some medications can also raise SGOT and SGPT levels. SGOT is also called *aspartate aminotransferase* (AST). SGPT is also called *alanine aminotransferase* (ALT).<sup>15</sup>

**SGA:** Acronym for birth weight that was low or small for gestational age.

**Sicca syndrome:** A chronic inflammatory autoimmune disease that is characterized by dryness of mucous membranes, especially of the eyes and mouth, and by infiltration of the affected tissues by lymphocytes. Also called Sjögren's disease.<sup>1</sup>

**Siderosis:** The deposition of iron in tissue.<sup>6</sup>

**Situs inversus:** A congenital abnormality characterized by lateral transposition of the viscera (as of the heart or the liver).<sup>1</sup>

**Spectral karyotyping (SKY):** A molecular cytogenetic technique that permits the simultaneous visualization of all human (or mouse) chromosomes in different colors, considerably facilitating karyotype analysis.<sup>10</sup>

**Spina bifida:** A birth defect in which the backbone and spinal canal do not close before birth. Spina bifida includes any birth defect involving insufficient closure of the spine.<sup>5</sup>

**Sprenghel:** A condition in which the shoulder blade on one or both sides is underdeveloped and abnormally high.<sup>1</sup>

**SQ infusions:** Acronym for subcutaneous infusions.

**Stenosis:** A narrowing or constriction of the diameter of a bodily passage or orifice.<sup>1</sup>

**Stroma:** The supporting tissue of the bone marrow. This tissue provides the growth environment for blood cells.<sup>2</sup>

**Surveillance, Epidemiology and End Results Program (SEER):** A premier source for cancer statistics in the United States; sponsored by the National Cancer Institute, NIH. SEER collects information on incidence, survival, and prevalence from specific geographic areas representing 26 percent of the country's population and compiles reports on all of these, plus cancer mortality for the entire U.S.<sup>8</sup>

**Syndactyly:** A union of two or more fingers or toes that occurs in humans, often as a hereditary disorder marked by the joining or webbing of two or more fingers or toes.<sup>1</sup>

**T-cells:** Lymphocytes responsible for "cell-mediated" immune reactions; critical for immune resistance to viruses, fungi, parasites and certain bacteria; important cells in transplant (graft rejection and GvHD) reactions.<sup>2</sup>

**T-score:** A T-score compares your bone density to the optimal peak bone density for your gender. It is reported as number of standard deviations below the average. A T-score above minus-1 is considered normal. A T-score of minus-1 to minus-2.5 is considered osteopenia, and a risk for developing osteoporosis. A T-score below minus-2.5 is diagnostic of osteoporosis.<sup>23</sup>

**Teratoma:** A neoplasm composed of multiple tissues, such as skin, hair, and muscle, including tissues not normally found in the organ in which it arises, caused by the development of independent stem cells. Also called teratoblastoma or teratoid tumor.<sup>6</sup>

**Thalassemia:** Inherited disorders characterized by abnormal production of hemoglobin. They result in low hemoglobin production, and excessive destruction of red blood cells.<sup>5</sup>

**Thenar eminence:** The muscles at the base of the thumb.

**Thenar muscles:** Any of the muscles that comprise the intrinsic musculature of the thumb within the thenar eminence and include the abductor pollicis brevis, adductor pollicis, flexor pollicis brevis, and opponens pollicis.

**Thrombocyte (platelet):** Cell fragment which releases clotting factors in the blood.<sup>2</sup>

**Thrombocytopenia:** Low platelet count.<sup>2</sup>

**Thromboembolism:** The blocking of a blood vessel by a particle that has broken away from a blood clot at its site of formation.<sup>1</sup>

**Thymocytes:** T-cells.<sup>2</sup>

**Thyroid:** A gland located in the front of the neck below the larynx, or voice box, and comprises two lobes, one on either side of the windpipe. The thyroid is one of a group of glands that is part of the endocrine system. The endocrine glands produce, store, and release hormones into the bloodstream that travel through the body and direct the activity of the body's cells. Thyroid hormones regulate metabolism, which is the way the body uses energy, and affect nearly every organ in the body. The thyroid gland makes two thyroid hormones, triiodothyronine ( $T_3$ ) and thyroxine ( $T_4$ ). Thyroid hormones affect metabolism, brain development, breathing, heart and nervous system functions, body temperature, muscle strength, skin dryness, menstrual cycles, weight, and cholesterol levels. Thyroid hormone production is regulated by **thyroid-stimulating hormone (TSH)**, which is made by the pituitary gland. Located in the brain, the pituitary gland is the "master gland" of the endocrine system. The thyroid gland's production of **thyroid hormones ( $T_3$  and  $T_4$ )** is triggered by thyroid-stimulating hormone (TSH), which is made by the pituitary gland.<sup>5</sup>

**Thyrotropin:** Another term for thyroid-stimulating hormone (TSH).

**Thyroxine-binding globulin (TBG):** Serum TBG level is a blood test to measure the level of the protein, thyroxine-binding globulin, that moves thyroid hormone throughout the body.<sup>5</sup>

**Total body irradiation (TBI):** Radiation therapy to the entire body, usually followed by umbilical cord blood, bone marrow or peripheral stem cell transplantation.

**Total parenteral nutrition:** See **Hyperalimentation**.

**Tracheoesophageal fistula (TEF):** See **Esophageal atresia**.<sup>5</sup>

**Tracheomalacia:** Congenital tracheomalacia is a weakness and floppiness of the walls of the windpipe (trachea). Because the windpipe is the main airway, breathing difficulties begin soon after birth.<sup>5</sup>

**Transvaginal ultrasound:** A method to look at a woman's reproductive organs, including the uterus, ovaries, cervix, and vagina. Transvaginal means across or through the vagina.<sup>5</sup>

**25-OH vitamin D:** This test is the most accurate measure of the amount of vitamin D in the body.<sup>5</sup>

**Umbilical cord blood:** Blood left in the placenta and cord after a baby is born. Blood contains hematopoietic stem cells.

**Unicornuate uterus:** One half of a uterus that forms with a cervix and is usually connected to the vagina. This uterus will function normally, although the woman is at risk for premature delivery of a fetus and for breech presentation which may necessitate a cesarean section. Sometimes called hemi-uterus.<sup>24</sup>

**White blood cells:** Blood cells which fight infection.<sup>2</sup>

**Wilms tumor:** A cancerous tumor of the kidney that occurs in children.<sup>5</sup>

**Z-score:** A Z-score is used to compare a patient's results to results in healthy persons of the same age, weight, ethnicity, and gender. This is useful to determine if there is something unusual contributing to the patient's bone loss.<sup>23</sup>

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