
Appendix A: Glossary and List of Abbreviations

5'-nucleotidase test: A test that measures the levels of 5'-nucleotidase, an enzyme produced by the liver.

AA: *Aplastic anemia.* A condition that occurs when the bone marrow fails to produce the proper amount and type of blood cells.

ABR: *Auditory brainstem evoked response test.* Also referred to as BAER.

aCGH: *Array comparative genomic hybridization.* A microarray technique that can detect changes (loss or gains) in DNA.

Adenocarcinoma: A type of cancer that initiates in the mucus-producing glandular cells of the body.

Agensis: Failure of an organ to develop during embryonic development.

AK: *Actinic keratosis.* Precancerous skin lesion.

Alloimmunization: An immune response to foreign antigens after exposure to genetically different cells or tissue.

Allosensitization: A condition caused by exposure to an alloantigen that induces immunologic memory cells.

ALP: *Alkaline phosphatase.* An enzyme measured in the blood that is used to detect liver and bone disease.

Alpha-thalassemia minor: An inherited blood disorder that affects the alpha chain of hemoglobin. The minor indication reflects mild symptoms.

ALT: *Alanine aminotransferase.* An enzyme measured to assess liver function.

Amenorrhea: Absence of menstruation.

AMH: *Anti-Müllerian hormone.* A hormone used as a marker for ovarian reserve.

AML: *Acute myelogenous leukemia.* A cancer of the blood and bone marrow.

Amniocentesis: A medical procedure in which amniotic fluid is removed from the uterus for testing.

ANC: *Absolute neutrophil count.* The number of neutrophils in the blood. Neutrophils are immune cells that fight infection.

Androgens: Hormones produced in the body that stimulate the development of male sex characteristics, such as testes formation and sperm production.

Anorectal malformations: A spectrum of disorders involving the rectum and the anus. These malformations may include a blockage of the anus, a failure of the rectum to connect to the anus, or an abnormal passage between the rectum and another part of the body, such as the urinary tract or the reproductive system.

Anoscopy: A medical procedure used to identify abnormalities inside the anus and the rectum.

Anovulatory cycles: Menstrual cycles without ovulation.

Antegrade continence enema: A procedure that empties the bowel.

Antibodies: Proteins produced by the blood that attack foreign substances, such as bacteria, viruses, and foreign tissue that the body does not recognize as part of itself.

Aphthae: Ulcers of the oral mucosa.

Aphthous stomatitis: Recurrent aphthous ulcers or canker sores that occur in the oral mucosa.

Aseptic necrosis of bone: The loss of bone primarily in the hip, knee, and shoulder joints.

AST: *Aspartate aminotransferase*. An enzyme measured to detect liver damage.

ATG: *Antithymocyte globulin*. Animal-derived antibodies that attack a patient's immune cells. Treatment with ATG helps prevent the patient's immune system from rejecting a transplant. ATG is also used as a therapy for aplastic anemia.

ATR: *Ataxia telangiectasia and Rad3-related protein*. A serine-threonine protein kinase that responds to DNA damage and phosphorylates multiple Fanconi anemia proteins.

Audiometric threshold: The softest level of sound a person can detect.

Autosomal dominant condition: A genetic inheritance pattern in which an affected individual has one copy of a mutant gene and one normal gene on a pair of autosomal chromosomes.

Autosomal recessive condition: A genetic inheritance pattern in which an affected individual has two copies of a mutant gene on a pair of autosomal chromosomes.

B cells: A type of white blood cell that is responsible for antibody production.

BA: *Bone age*. A test used to assess the degree of bone maturation in children.

Basophil: A type of white blood cell that is involved in allergic reactions.

BCC: *Basal cell carcinoma*. The most common type of skin cancer in the general public.

Beta-thalassemia minor: An inherited blood disorder that affects the beta chain of hemoglobin. The minor indication reflects mild signs and symptoms.

Biallelic variants: Genetic variants that are found in both copies (alleles) of the same gene.

Binucleated erythroid cells: Erythrocytes (red blood cells) that contain two nuclei.

Biopsy: A medical procedure in which a small piece of tissue is removed surgically, which is then examined under a microscope to determine whether dysplasia (pre-cancer) or cancer is present.

BMD: *Bone mineral density.* A measurement of the mineral content of bones.

BMF: *Bone marrow failure.* A condition that occurs when bone marrow fails to produce an adequate number of blood cells.

BMI: *Body mass index.* A measure of physical fitness that accounts for height and body weight.

Bone marrow: The spongy tissue inside bones that produces blood cells.

Bronchoscopy: An endoscopic procedure that allows internal visualization of the lungs.

BU: *Busulfan.* An alkylating agent used to treat chronic myelocytic leukemia.

Café au lait macules: Flat, light brown birthmarks.

Carrier: An individual who inherits a single copy of an abnormal gene for an autosomal recessive disorder. Carriers usually do not develop the disorder but can pass on a copy of the abnormal gene to their offspring.

Carrier frequency: The proportion of carriers in a population.

CBC: *Complete blood count.* A laboratory test that provides the number, and/or percentage, and/or characteristics of certain blood cells, primarily white cells, red cells, and platelets.

Centralization: A surgical procedure that moves and centers the wrist over the end of the ulna.

Cetuximab: An epidermal growth factor receptor inhibitor used to treat some cancers.

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

Cholestatic jaundice: Yellowing of the skin and eyes due to obstructed bile flow in the liver.

Cholesterol: A sterol compound found in most tissues of the human body that is necessary for cell membranes and precursors of other steroid compounds.

Chromosome: The structure of nucleic acids and proteins that carries genetic information that is found in the nucleus of most living cells. Most humans have 23 pairs of chromosomes, including 1 pair of sex chromosomes (females have two 'X' sex chromosomes; males have one 'X' and one 'Y' sex chromosome).

Chromosome breakage test: The gold standard test for diagnosing Fanconi anemia. This test measures the types and rates of breakages and rearrangements found in the chromosomes of cells after treatment with DNA damaging agents. It also reveals how well the chromosomes can repair themselves after injury.

Cirrhosis: Abnormal liver function resulting from long-term damage.

Cisplatin: An alkylating agent used to treat many cancers.

Clastogen: An agent that induces breaks in chromosomes.

Clonal abnormalities: Changes in the structure or number of chromosomes in certain cells of the bone marrow.

Clonal evolution: A process by which cells acquire new abnormalities.

Clonal expansion: An increase in the percentage of cells with identical abnormalities.

Clone: A population of cells.

CMC: *Carpometacarpal joints*. Five joints in the wrist that articulate the distal row of carpal bones and the proximal bases of the five metacarpal bones.

CMM: *Congenital mirror movements*. Intentional movements of one side of the body are mirrored by involuntary movements of the other side.

CMV: *Cytomegalovirus*. A relatively common virus in the herpes family that causes mild symptoms in healthy people but can pose a serious health risk to immune-compromised individuals.

CNA: *Copy number aberrations*. Deletions or amplifications in chromosomes that occur in cancer cells.

Colposcopy: A medical procedure that examines the vulva, vagina, and cervix.

Complementation group: A group of genes that works together to produce a person's physical characteristics. Prior to the identification of the genes and genetic mutations that cause Fanconi anemia (FA), patients with the disease were classified into sub-categories known as complementation groups based on the patient's cellular features. These complementation groups correspond to the various FA genes (e.g., individuals who belong to complementation group A have mutations in the *FANCA* gene, whereas individuals who belong to complementation group B have mutations in the *FANCB* gene).

Cortisol: A steroid hormone produced in the adrenal glands that plays important roles in the body's stress response, immunity, metabolism of nutrients, and other processes.

C-reactive protein test: A liver function test that measures C-reactive protein, a protein produced by the liver.

CRISPR/Cas9: *Clustered regularly interspaced palindromic repeats/Cas9*. CRISPR is a gene editing technique based on the bacterial CRISPR-Cas9 antiviral defense system that can be used to edit the genomes of living organisms.

Cryopreservation: The use of very low temperatures to preserve living cells and tissues.

CsA: *Cyclosporine A*. A drug that suppresses the immune system and is used to prevent transplant rejection.

CVS: *Chorionic villus sampling*. A prenatal procedure in which a sample of the chorionic villus from the placenta is removed and tested.

CY: *Cyclophosphamide*. A drug that is used to suppress the immune system and treat cancer.

Cytogenetic evaluation: A laboratory test that examines parts of the patient's cells, including chromosomes.

Cytopenia: An abnormally low number of blood cells.

DEB: *Diepoxybutane*. A DNA damaging agent used in the chromosome breakage test.

Diabetes mellitus: A metabolic disease in which the body's ability to produce or respond to the hormone insulin is impaired.

DNA ICLs: *DNA interstrand crosslinks*. Crosslinked DNA that occurs when exogenous or endogenous agents react with two nucleotides of DNA, forming a covalent linkage between them.

Duodenal atresia: A condition in which the duodenum is incomplete or blocked and does not allow the contents of the stomach to enter the intestines.

DXA: *Dual energy absorptiometry*. 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: Unhealthy levels of cholesterol and triglycerides.

Dysmorphology: The study of human congenital malformations and syndromes.

Dyspareunia: Pain during sexual intercourse or other sexual activity that involves vaginal penetration.

Dysphagia: Difficulty swallowing.

Dysuria: Painful urination.

EA: *Esophageal atresia*. A congenital medical condition in which the esophagus does not develop properly; frequently, the lower end of the esophagus is incomplete or blocked and does not allow food to pass into the stomach.

EBV: *Epstein-Barr virus*. A herpes virus that can be reactivated after bone marrow transplant, resulting in post-transplant lympho-proliferative disease or lymphoma.

Echocardiogram: A non-invasive imaging procedure used to assess heart function.

EGFR: *Epidermal growth factor receptor*. A protein that is a receptor for members of the epidermal growth factor family of ligands.

Electrocardiogram: A test that records the electrical signals of the heart.

Encopresis: Involuntary leakage of stool.

Endocarditis: Infection of the endocardium, which is the inner lining of the heart chambers and valves.

Endocrine: Relating to the body system that produces hormones.

Endoscopy: Insertion of a long, thin tube that is used to observe an internal organ.

Enteral supplementation: Supplemental feeding via feeding tube.

Erythrocytes: Red blood cells that carry oxygen to the body's tissues.

Erythroplakia: Also known as *erythroplasia*. A reddened patch in the oral or genital mucosa that is considered a precancerous lesion.

Erythropoietin: A hormone that plays a key role in the production of red blood cells.

Esophageal stenosis: Narrowing of the esophagus.

Esophagitis: Inflammation or irritation of the esophagus.

Esophagoscopy: Examination of the esophagus by means of a flexible endoscope.

Estrogens: Steroid hormones that promote the development and maintenance of female characteristics of the body.

Exons: Segments of DNA that contain information needed to make proteins.

Extracorporeal photopheresis: A procedure used to treat chronic graft-versus-host disease in which the patient's blood is treated with drugs that become active when they are exposed to ultraviolet (UV) light.

FA: *Fanconi anemia*. An inherited disease that affects the ability of cells in the body to repair DNA. Fanconi anemia can lead to bone marrow failure and cancer.

FAAP: *Fanconi anemia core complex associated proteins*. Proteins that play a role in the FA pathway that have not been ascribed to an FA disease phenotype.

Ferritin: A blood protein that binds and stores iron. The levels of ferritin in the blood increase as the amount of iron in the body increases.

FISH: *Fluorescence in situ hybridization*. A laboratory technique that allows visualization of the chromosomal abnormalities in cells.

Flow cytometry: A laboratory technique to separate, count, and evaluate cells with distinct characteristics; used to diagnose blood cancers and other conditions.

FLU: *Fludarabine*. A drug used to suppress the immune system before hematopoietic cell transplant and for treating some cancers.

Fluoroscopy: A type of medical imaging that uses continuous X-ray images.

Fructosamine test: A laboratory test that measures the total amount of fructosamine, a glycosylated protein, in the blood.

FSH: *Follicle stimulating hormone*. A hormone produced by the pituitary gland that stimulates the growth of ovarian follicles in females and sperm-producing cells in males.

FT4: *Free thyroxine*, also called *Free T4*. Thyroxine is a hormone produced by the thyroid that plays a role in several bodily functions, including growth and metabolism. It exists in two forms in the blood: T4 that is bonded to protein in the blood and free T4. Free T4 is the type available for use by the body's tissues.

Gastric accommodation: The gastric accommodation reflex allows the proximal stomach to have an appropriate gastric volume to accommodate an ingested meal.

Gastrojejunostomy: A surgical procedure to create an anastomosis from the stomach to the middle part of the intestine.

Gastrostomy tubes: A feeding tube inserted through the abdomen that delivers nutrition directly to the stomach.

G-Banding: A laboratory technique used to stain and visualize chromosomes for analysis.

G-CSF: *Granulocyte colony-stimulating factor*. A growth factor drug that stimulates the bone marrow to release stem cells.

GERD: *Gastroesophageal reflux disease*. A chronic digestive disorder of persistent acid reflux that occurs when the lower esophageal sphincter is weak or relaxes inappropriately, allowing stomach acid to flow up into the esophagus.

GGT: *Gamma-glutamyl transpeptidase*. An enzyme that is found in many organs throughout the body. A GGT blood test can indicate liver damage.

GHD: *Growth hormone deficiency*. A metabolic condition caused by insufficient levels of growth hormone in the body.

Gingivitis: Inflammation of the gums, or gingiva.

Glucose: A sugar that provides fuel for human cells to function.

GM-CSF: *Granulocyte-macrophage colony-stimulating factor.* A hematopoietic growth factor and immune modulator that has profound effects on the functional activities of circulating leukocytes and stimulates multipotent progenitor cells. It is used clinically to treat neutropenia in patients undergoing chemotherapy as well as after bone marrow transplant

GnRH: *Gonadotropin releasing hormone.* A hormone regulator of the secretion of follicle stimulating hormone (FSH) and luteinizing hormone from the anterior pituitary.

Gonadotoxic therapy: Treatments, such as chemotherapy and radiation, that impair reproductive function.

Granulocyte: White blood cell (neutrophil, basophil, or eosinophil).

Growth curves: Charts that allow physicians to monitor a child's physical growth over time in comparison with other children of the same age and gender.

GvHD: *Graft-versus-host disease.* Complication of allogeneic stem cell transplantation where donated bone marrow or peripheral blood stem cells interpret the recipient's body as foreign and attack the body.

Halitosis: Unpleasant breath odor.

Haploidentical transplant: A half matched transplant from a biological parent or sibling donor.

HAT: *Hearing assistive technology.* Technology systems and/or devices (frequently digital or wireless) that help people with hearing, voice, speech, or language disorders to communicate more effectively in their daily lives.

HbA1c: *Glycosylated hemoglobin.* Hemoglobin bound glucose that is measured to monitor control of diabetes over time.

HbF: *Fetal hemoglobin.* The main blood protein that carries oxygen in the fetus.

HCC: *Hepatocellular carcinoma.* Liver cancer.

HCT: *Hematopoietic cell transplantation.* An allogeneic HCT is a procedure in which a donor's bone marrow stem cells or umbilical cord blood are used to replace diseased bone marrow stem cells of a recipient.

HDL: *High-density lipoprotein.* A lipoprotein that removes cholesterol from the blood and carries it back to the liver to be flushed from the body. Commonly known as the "good" cholesterol because higher levels of HDL are associated with reduced risk of atherosclerosis and heart disease.

Hemizygous variant: Having only one copy of a gene present in diploid cells.

Hepatic fibrosis: Imbalance between production and dissolution of extracellular matrix in the liver caused by injury that leads to build up of scar tissue.

Hepatic transaminases: Enzymes measured on a liver function test. Elevated levels may indicate liver damage.

Hepatitis: Inflammation of the liver.

Hepatocellular adenoma: Benign liver tumor.

Heterozygous: Having two different alleles of a particular gene or genes.

HgB: *Hemoglobin*. A red blood cell protein that transports oxygen throughout the body via the bloodstream.

HLA: *Human leukocyte antigen*. A protein on the surface of cells that helps the body to determine what is "self" and what is "foreign." An HLA-matched donor increases the chances that the patient's body will accept the transplant as "self."

HNSCC: *Head and neck squamous cell carcinoma*. Cancers that develop in the mucous membranes of the oral cavity, oropharynx, hypopharynx, and larynx.

Homozygous: Having two identical alleles in a particular gene or genes. An individual with FA is homozygous if he or she has the same gene mutation in both copies of the FA gene.

HPV: *Human papilloma virus*. A virus that can cause warts and cancer.

HR: *Homologous recombination proteins*. Proteins that participate in homologous repair of DNA.

HSC: *Hematopoietic stem cells*. Rare blood cells in bone marrow that give rise to all other blood cells during a process called hematopoiesis.

Hydronephrosis: Swelling of the kidneys; occurs when urine accumulates and is unable to make its way out of the kidneys.

Hyperestrogenism: Higher than normal levels of estrogen.

Hypergonadotropic hypogonadism: Failure of the testes to produce sufficient quantities of testosterone.

Hyperpigmentation: A condition in which patches of skin are darker in color than normal surrounding skin.

Hypertransaminasemia: Elevated levels of the liver enzymes alanine transaminase (ALT) and aspartate transaminase (AST).

Hypoestrogenism: Lower than normal levels of estrogen.

Hypogonitalism: Diminished functional activity of the genitals.

Hypomagnesemia: Lower than normal blood magnesium levels.

Hypoparathyroidism: A state of decreased secretion or activity of parathyroid hormone.

Hypopharyngeal cancer: Disease in which malignant cells form in the tissues of the hypopharynx.

Hypopigmentation: A condition in which patches of skin are lighter in color than normal surrounding skin.

Hypopituitarism: Diminished hormone secretion by the pituitary gland.

Hypoplasia: Under- or incomplete development of an organ or tissue in the body.

Hypothyroidism: A condition caused by low levels of the thyroid hormone.

IFAR: *International Fanconi Anemia Registry*. A registry that serves as the central repository for clinical, hematologic, and genetic information on patients with Fanconi Anemia, and cellular material from patients and their families. This growing clinical database supports the study of the full spectrum of the diverse features of the disease. Established at The Rockefeller University in 1982.

IGF-1: *Insulin-like growth factor 1*. The hormone that mediates the growth promoting effect of growth hormone (GH). Elevated levels indicate sustained production of GH.

IGFBP-3: *IGF-binding protein 3*. The protein that binds IGF-1. Elevated levels indicate high levels of growth hormone.

Impaired glucose tolerance: An abnormal state when blood glucose is raised beyond normal levels, but not high enough to warrant a diabetes diagnosis.

Imperforate anus: A physical defect in which the opening to the anus is missing or blocked.

In cis variant: Variants located together on the same copy of a given gene.

In trans variant: Variants situated on opposite copies of the same gene.

IVF: *In vitro fertilization*. A treatment for infertility, in which eggs are removed from a woman's ovary and are fertilized by male sperm in a laboratory setting. The fertilized eggs are then prodded to implant in the woman's uterus.

Jejunal feeds: Feeding directly into the small intestine.

Jejunostomy: An operative procedure in which a feeding tube is placed into the proximal jejunum.

Laryngeal cancer: Disease in which malignant cells form in the tissues of the larynx.

Laryngeal edema: Swelling of the larynx.

LDL: *Low-density lipoprotein*. A lipoprotein that delivers fat molecules to cells throughout the body. Commonly known as the "bad" cholesterol because higher levels of LDL have been associated with the progression of atherosclerosis and blockage of the arteries.

Leukemia: A bone marrow cancer characterized by an uncontrolled increase in white blood cells (leukocytes).

Leukoplakia: White patches of epithelium in the oral cavity.

LFT: *Liver function tests.* A set of blood tests used to help diagnose and monitor liver function, infection, damage, or disease by measuring the levels of certain enzymes and proteins in the blood. Common LFTs include: alanine transaminase (ALT); aspartate transaminase (AST); alkaline phosphatase (ALP); albumin and total protein; bilirubin; gamma-glutamyltransferase (GGT); L-lactate dehydrogenase (LD); and prothrombin time (PT).

Lichen planus: A chronic inflammatory skin condition affecting the skin and mucosal surfaces.

Lichen sclerosis: A chronic condition that affects the skin of the genital and anal areas. May increase the risk of cancer.

Lymphocyte: A type of white blood cell that fights infection by producing antibodies and other protective substances. There are two types of lymphocytes: B-cells and T-cells.

Macrocytosis: Term used to describe red blood cells that are larger than normal.

Macrophage: A type of white blood cell that helps to destroy invading micro-organisms.

MDS: *Myelodysplastic syndrome.* The presentation of a set of health conditions that develop when the myeloid class of blood cells are not present in sufficient numbers in the bone marrow.

Megaduodenum: Congenital or acquired dilation and elongation of the duodenum.

Melanocytic nevi: Moles.

Melanoma: An aggressive form of skin cancer.

Menarche: The first occurrence of menstruation.

Menopause: The time that marks the end of menstrual cycles; diagnosed after 12 months without a menstrual period.

Menorrhagia: Heavy menstrual bleeding.

Metabolic syndrome: A cluster of conditions, including high blood pressure, high blood sugar, high triglycerides, and low HDL cholesterol, that occur together and increase the risk of heart disease, stroke, and diabetes.

Microcephaly: Smaller than normal head circumference.

Microdentia: Small teeth.

Micrognathia: Undersized lower jaw.

MLPA: *Multiplex ligation-dependent probe amplification.* An efficient and sensitive genomic testing technique for identifying large deletions of DNA sequence as part of the FA testing algorithm.

MMC: *Mitomycin C.* A chemotherapy agent used in the chromosome breakage test.

MMF: *Mycophenolate mofetil.* A drug used to suppress the immune system in patients who receive transplants.

MRI: *Magnetic resonance imaging.* An imaging technique used for visualizing internal organs.

MTX: *Methotrexate.* A chemotherapy drug used to treat leukemia and certain types of cancer of the breast, skin, head and neck, or lungs.

Mucositis: A condition that causes pain and inflammation on the surface of the mucous membrane.

Myocardium: The muscular middle layer of the wall of the heart.

Nasogastric tube: A flexible tube that is passed through the nose and down through the nasopharynx and esophagus into the stomach.

Nasojejunal tube: A flexible tube that is passed through the nose and into the jejunum.

Nasopharyngeal carcinoma: Disease in which malignant cells develop in the tissue of the nasopharynx.

Neutropenia: A health condition characterized by abnormally low levels of neutrophils in the blood.

Neutropenic ulcers: Lesions of the oral mucosa commonly encountered in patients receiving intensive myelosuppressive chemotherapy for diseases such as acute leukemia.

Neutrophils: A type of white blood cell that fights infection and helps heal damaged tissue.

Odynophagia: Painful swallowing.

OGTT: *Oral glucose tolerance test.* A blood test that measures the body's response to sugar. Variations of the test are commonly used to screen for type 2 diabetes and gestational diabetes.

Oligomenorrhea: Infrequent menstrual periods.

Opportunistic infection: A type of infection common in immune-compromised patients who are unable to fight off microbes that do not normally cause disease in humans.

Oropharyngeal cancer: Disease in which malignant cells develop in the tissues of the oropharynx.

Osteopenia: Lower-than-normal bone density. Often a precursor to osteoporosis.

Osteoporosis: A disease characterized by mineral and protein depletion in bones that leads to thinning and brittle bones that break easily.

Oxidative stress: Occurs when the levels of oxygen and its breakdown products, reactive oxygen species, are too high in cells. Oxidative stress may lead to DNA and other cellular damage.

Pap test: A gynecological test, also known as cervical cytology testing, used to detect cervical cancer and precancerous lesions.

Parenteral nutrition: Supplemental feeding via intravenous infusion.

PDT: *Photodynamic therapy.* Treatment that combines light and a photosensitizing drug to destroy precancerous and cancerous cells.

Periodontitis: A severe gum infection (gum disease) that can lead to tooth loss and other serious health conditions.

Petechiae: Small areas of bruising.

PGD: *Preimplantation genetic diagnosis.* A technology for examining the genetic profiles of in vitro-derived embryos before they are implanted in a woman's uterus.

PH: *Peliosis hepatis.* A condition that occurs when blood vessels in the liver called sinusoids become excessively dilated and form large blood-filled spaces, like cysts, that are scattered throughout the liver.

PHENOS: The acronym for the grouping of major phenotypic features common to individuals with FA, including skin Pigmentation, small Head, small Eyes, central Nervous system, Otology, and Short stature.

PLT: *Platelets.* Disc-shaped fragments of cells that circulate in the bloodstream and help promote clotting to stop or prevent bleeding.

POI: *Primary ovarian insufficiency.* Premature ovarian failure.

Pollicization: A surgical procedure that creates a functional thumb by moving the index finger and its nerves, arteries, tendons, and muscles to the thumb position.

Polypharmacy: The administration of many different medicines during the treatment of a single disease.

Pouce flottant: A so-called "floating" thumb or residual digit that lacks bones and is composed of skin and soft tissue.

Pre-axial polydactyly: A hand with more than one thumb. The thumbs may be fused together or may be separate digits.

Progesterone: A female sex hormone.

PSIS: *Pituitary stalk interruption syndrome*. A rare congenital anatomical defect of the pituitary gland characterized by a very thin or “interrupted” pituitary stalk; an ectopic or absent posterior pituitary; and aplasia or hypoplasia anterior pituitary, with permanent deficit of growth hormone (GH).

PT-Cy: *Post-transplant cyclophosphamide*. A treatment strategy following hematopoietic cell transplantation to reduce the occurrence of complications, such as graft-versus-host-disease and graft rejection, particularly in patients whose donors are not fully HLA-matched.

Purpura: Large areas of bruising.

Radialization: A surgical procedure to realign the bones of the wrist.

Radiosensitivity: Relative susceptibility of cells, tissues, organs, and organisms to ionizing radiation.

Radius: One of the two long bones in the forearm. The radial bone lies laterally and parallel to the ulna; extends from the lateral side of the elbow to the thumb side of the wrist; and pivots around the ulna to produce movement at the proximal and distal radio-ulnar joints.

RB: *Retinoblastoma gene*. The gene that encodes for the tumor suppressor protein, pRB.

Recessive: A genetic mutation is recessive if an individual must inherit two copies of the mutant gene to express the disease. Individuals with one mutant and one normal gene appear normal. They are called “heterozygotes” or “carriers.”

Recto-perineal fistula: A type of anorectal malformation in which the anus is not present and the rectum, instead, connects to the perineum.

Renal dysplasia: Abnormal formation of the kidney, along with irregular cysts.

ROS: *Reactive oxygen species*. Oxygen containing radicals that can cause tissue damage.

SCC: *Squamous cell carcinoma*. A type of cancer that is derived from squamous cells. Commonly found on the skin, oral cavity, and the anogenital region.

Serostatus: The presence or absence of a serological marker in the blood.

SGA: *Small for gestational age*. A term used to describe babies who measure smaller than usual in weight for the number of weeks of pregnancy, typically with birthweights below the 10th percentile for infants of the same gestational age.

Short bowel syndrome: A condition that occurs when a large segment of the small intestine is non-functional or has been surgically removed causing malabsorption of nutrients.

SIL: *Squamous intraepithelial lesion*. Abnormal growth of squamous cells on the cervix.

SNP arrays: *Single nucleotide polymorphism arrays.* A type of DNA microarray used to detect polymorphisms within a population.

Somatic stem cell mosaicism: Spontaneous correction or reversion of an inherited variant to a normal genetic status in a stem cell that then repopulates the bone marrow with unaffected cells.

SS: *Sweet's syndrome;* also called *acute neutrophilic dermatosis.* A rare skin condition which presents as painful red plaques or nodules.

Stadiometer: A piece of medical equipment used for measuring human height.

Stem cells: Cells that can develop into one of many types of specialized cells in the body.

Stem cell gene therapy: A novel treatment that uses gene therapy to correct a faulty gene in the stem cells of the recipient. Stem cells are obtained from the patient, grown and "corrected" in a laboratory, and then returned to the patient.

STI: *Sexually transmitted infection.* An infection transmitted predominantly through intimate skin-to-skin or sexual contact, though some also can be spread through non-sexual means such as via blood or blood products, or from mother to child during pregnancy and childbirth. More than 30 different bacteria, viruses, and parasites are known to be transmitted through sexual contact. Some can be treated and/or cured, some cannot.

Supernumerary teeth: Teeth that appear in addition to the normal number of teeth. Children typically have a full set of 20 baby teeth by three years of age. Most adults have a full set of 32 adult teeth by age 21.

T4: *Thyroxine.* A hormone secreted by the thyroid gland.

T cells: White blood cells that play a key role in the immune response by searching out and destroying material that is considered "foreign."

TALENs: *Transcription activator-like effector nucleases.* Restriction enzymes engineered to cleave specific regions of DNA.

TAR: *Thrombocytopenia absent radius syndrome.* A disorder characterized by absence of a radius in each forearm, short stature, and thrombocytopenia.

Tardive dyskinesia: A neurological disorder characterized by involuntary and abnormal movements of the jaw, lips and tongue, including facial grimacing, sticking out the tongue, and sucking or fish-like movements of the mouth.

TBG: *Thyroid hormone binding globulin.* A binding protein that transports thyroid hormones.

TBI: *Total body irradiation:* Radiation therapy to the entire body used in some hematopoietic cell transplant procedures.

TEF: *Tracheoesophageal fistula*. An abnormal passage between the esophagus and the trachea that may result in food from the esophagus crossing into the airways or air entering the esophagus.

Thrombocytopenia: Low platelet count.

Transaminitis: Also called *hypertransaminasemia*. A condition characterized by high levels of liver enzymes called transaminases.

Transferrin: A binding protein that transports iron in the blood.

Transferrin saturation: The amount of iron carried by the transferrin protein in the blood. Saturation increases as the amount of iron in the body increases.

Triglycerides: The building blocks of fats and oils.

Triphalangeal thumb: A thumb that has an extra bone (called a phalanx) that can vary in size and shape.

Trismus: Reduced opening of the mouth due to spasm of the jaw muscles.

TSH: *Thyroid stimulating hormone*. A hormone produced by the anterior pituitary and primary stimulus for thyroid hormone production.

UCB: *Umbilical cord blood*. Blood present in the placenta and umbilical cord of an infant after birth. This blood contains high numbers of stem cells that can be used in transplants.

UCL: *Ulnar collateral ligament*. A ligament on the inside of the elbow.

USP1: *Ubiquitin specific peptidase 1*. A protein that regulates proteins by removing ubiquitin substrates.

UV: *Ultraviolet light*. A type of electromagnetic radiation that covers the wavelength range 100-400 nm, which is a higher frequency and lower wavelength than visible light. Ultraviolet light is divided into three bands: UVA (315-400 nm), UVB (280-315 nm), and UVC (100-280 nm). In humans, increased exposure, particularly to high-frequency UVA, can damage living tissue and cause skin cancers, cataracts, and immune system damage.

UVA: *Ultraviolet A*. A subtype of ultraviolet radiation that causes premature aging and wrinkling of the skin.

UVB: *Ultraviolet B*. A subtype of ultraviolet radiation that induces DNA damage and is the major cause of skin cancer.

VACTERL: The acronym for a group of birth anomalies that are not necessarily related to each other but tend to occur together. These include Vertebral defects, Anorectal malformations, Cardiac abnormalities, Tracheo-Esophageal abnormalities, Renal defects, and Limb defects, such as extra fingers or toes, or abnormally formed forearms.

VACTERL-H: The acronym for a group of classical congenital abnormalities including Vertebral, Anal, Cardiac, Tracheo-esophageal fistula, Esophageal atresia, Renal defects, upper Limb defects, and Hydrocephalus.

Vaginal stenosis: Narrowing and shortening of the vagina.

Venous thromboembolism: A condition in which a blood clot forms in the leg, groin or arm.

Verruca vulgaris: Warts.

VUS: *Variants of unknown significance.* A form of a gene identified through sequencing in which the significance on health and function is not known.

WES: *Whole exome sequencing.* A genome sequencing technique that analyzes all protein-coding regions of the genome.

WGS: *Whole genome sequencing.* A genome sequencing technique that analyzes the entire genome.

WRN: *Werner syndrome ATP-dependent helicase complex.* Helicase involved in DNA repair that also has exonuclease activity.

X-Linked recessive inheritance: Genes that are inherited on the "X" sex chromosome. Males have one "X" chromosome; females have two. If a disorder is "X"-linked recessive, it means that females must inherit two copies of an abnormal gene for the disease to develop, whereas males need only inherit one.

Xerostomia: Dry mouth syndrome.

ZFN: *Zinc finger nucleases.* Restriction enzymes used in targeted gene editing of DNA.