

GLOSSARY

A

5' VNTR. A specific variation outside the insulin gene that is implicated in NIDDM and IDDM susceptibility.

AADC INHIBITORS. Drugs that block the amino acid decarboxylase; one type of enzyme that breaks down dopamine. Also called DC inhibitors, they include carbidopa and benserazide.

ABDOMINAL HERNIA. Bulging of an organ or tissue through the muscle of the stomach wall.

ABDUCENS NERVE. Cranial nerve VI; the nerve that extends from the midbrain to the lateral rectus muscle of the eye and controls movement of the eye toward the ear (abduction).

ABDUCTION. Turning away from the body.

ABSCESS. A localized collection of pus or infection that is walled off from the rest of the body.

ABSENCE SEIZURE. A brief seizure with an accompanying loss of awareness or alertness.

ACAMPROSATE. An anti-craving medication used in Europe to reduce the craving for alcohol. It is presently undergoing tests for approval in the United States.

ACANTHOCYTOSIS. The presence of acanthocytes in the blood. Acanthocytes are red blood cells that have the appearance of thorns on their outer surface.

ACANTHOSIS NIGRICANS. A skin condition characterized by darkly pigmented areas of velvety wart-like growths. Acanthosis nigricans usually affects the skin of the armpits, neck, and groin.

ACCOMMODATION. The ability of the lens to change its focus from distant to near objects. It is achieved through the action of the ciliary muscles that change the shape of the lens.

ACETYLCHOLINE. A neurotransmitter (chemical messenger) used to certain nerve cells to send messages to adjacent cells.

ACETYLCHOLINESTERASE (ACHE). An enzyme found in nerve tissue.

ACHROMATOPSIA. The inability to distinguish any colors.

ACID MALTASE. The enzyme that regulates the amount of glycogen stored in muscle cells. When too much glycogen is present, acid maltase is released to break it down into waste products.

ACIDOSIS. A condition of decreased alkalinity resulting from abnormally high acid levels (low pH) in the blood and tissues. Usually indicated by sickly sweet breath, headaches, nausea, vomiting, and visual impairments.

ACONDROPLASIA. An autosomal dominant form of dwarfism caused by a defect in the formation of cartilage at the ends of long bones. Affected individuals typically have short limbs, a large head with a prominent forehead and flattened profile, and a normal-sized trunk.

ACQUIRED ANGIONEUROTIC EDEMA. Abbreviated AANE, or AAE, this is a non-hereditary form of angio edema that generally begins to show symptoms in, or after, the fourth decade of life.

ACQUIRED IMMUNITY. Also called “specific immunity,” refers to immune reaction mediated by B-cells and/or T-cells. Includes humoral and cellular immunity.

ACROCENTRIC. A chromosome with the centromere positioned at the top end.

ACROCEPHALOPOLYSYNDACTYL SYNDROMES. A collection of genetic disorders characterized by cone shaped abnormality of the skull and partial fusing of adjacent fingers or toes.

ACROCEPHALY. An abnormal cone shape of the head.

ACROMELIC. The anatomical term used to denote the end of a limb (arm or leg). In the context of Robinow syndrome, it refers to bones of the hands and feet.

ACROOSTEOLYSIS. Loss of bone tissue at the ends of the fingers and/or toes.

ACROPARESTHESIAS. Painful burning sensation in hands and feet.

ACTION POTENTIAL. The wave-like change in the electrical properties of a cell membrane, resulting from the difference in electrical charge between the inside and outside of the membrane. The action potential acts as a signal for certain activities and processes in the body.

ACUPUNCTURE. An alternative health procedure based on ancient Chinese methods, involving insertion of thin needles at specific pressure points in the body.

ACUTE PHASE REACTANTS. Blood proteins whose concentrations increase or decrease in reaction to the inflammation process.

ACUTE PHASE. The initial phase of LHON where visual blurring begins in both eyes, and central vision is lost.

ADDUCTED THUMBS. Thumbs clasped across the palm.

ADDUCTION. Movement toward the body. In Duane retraction syndrome, turning the eye inward toward the nose.

ADENOCARCINOMA. A type of cancer which is in a gland-like form.

ADENOMATOUS. Derived from glandular structures

ADRENAL. A pair of glands located on top of the kidneys that secrete substances or hormones, like steroids and adrenaline, which regulate various functions, such as water balance and stress response.

ADRENAL GLAND. A triangle-shaped endocrine gland, located above each kidney, that synthesizes aldosterone, cortisol, and testosterone from cholesterol. The adrenal glands are responsible for salt and water levels in the body, as well as for protein, fat, and carbohydrate metabolism.

ADRENAL INSUFFICIENCY. Problems with the adrenal glands that can be life threatening if not treated. Symptoms include sluggishness, weakness, weight loss, vomiting, darkening of the skin and mental changes.

ADRENOCORTICOTROPIN (CORTICOTROPHIN). A hormone that acts on cells of the adrenal cortex, causing them to produce male sex hormones and hormones that control water and mineral balance in the body.

ADVANCED BONE AGE. The bones, on x-ray, appear to be those of an older individual.

ADYSPLASIA. A term referring to the combination of renal agenesis (complete absence of one or both kidneys) and renal dysplasia (developmental anomaly of the kidney).

AFFECTIVE FLATTENING. A loss or lack of emotional expressiveness. It is sometimes called blunted or restricted affect.

AFLATOXIN. A substance produced by molds that grow on rice and peanuts. Exposure to aflatoxin is thought to explain the high rates of primary liver cancer in Africa and parts of Asia.

AGE-ASSOCIATED MEMORY IMPAIRMENT (AAMI). A condition in which an older person suffers some memory loss and takes longer to learn new information. AAMI is distinguished from dementia in that it is not progressive and does not represent a serious decline from the person's previous level of functioning.

AGENESIS. Failure of an organ, tissue or cell to develop or grow.

AGENESIS OF THE CORPUS CALLOSUM. Failure of the corpus callosum to form and develop. The corpus callosum is the band of nerve fibers located between the two sides, or hemispheres, of the brain.

AGNOSIA. Loss of the ability to recognize objects by use of the physical senses.

AGYRI. A lack of convolutions (gyri) or normal folds in the brain tissue.

AKATHISIA. Agitated or restless movement, usually affecting the legs and accompanied by a sense of discomfort. It is a common side effect of neuroleptic medications.

AKINESIA. A loss of the ability to move; freezing in place.

ALKALINE. Having a basic pH; not acidic.

ALKALINIZATION. The process of making a solution more basic, rather than more acidic, by raising the pH.

ALLELE. One of two or more alternate forms of a gene.

ALLELIC. Related to the same gene.

ALLELIC VARIANTS. A disease is said to have allelic variants when different mutations in the same allele result in identical, or nearly identical, symptoms. An allele is the combined locations of a gene on the two paired chromosomes that contain this gene.

ALLERGEN. A substance or organism foreign to the body; allergens stimulate the immune system to produce antibodies.

ALLERGIC RHINITIS. Hay fever.

ALLERGY. Condition in which immune system is hypersensitive to contact with allergens; an abnormal response by the immune system to contact with an allergen; condition in which contact with allergen produces symptoms such as inflammation of tissues and production of excess mucus in respiratory system.

ALLOSTERIC ACTIVATOR SITE. Site at which an enzyme is regulated separate from its site of catalytic enzymatic activity.

ALOPECIA AREATA. A nonscarring hair loss syndrome characterized by smooth round or oval hairless areas on the scalp.

ALOPECIA. Loss of hair or baldness.

ALPHA-FETOPROTEIN (AFP). A chemical substance produced by the fetus and found in the fetal circulation. AFP is also found in abnormally high concentrations in most patients with primary liver cancer.

ALPHA-L-IDURONIDASE. An enzyme that breaks down dermatan sulfate and heparan sulfate. People with Hurler syndrome do not make enough of this enzyme.

ALPHA-THALASSEMIA. Autosomal recessive disorder where no functional hemoglobin is produced. Leads to severe untreatable anemia.

ALTERATION. Change or mutation in a gene, specifically in the DNA that codes for the gene.

ALTERNATE COMPLEMENT PATHWAY. A cascade of enzymatic reactions that produce antibacterial proteins. This pathway helps to ward off infections.

ALZHEIMER DISEASE. A neurodegenerative disease marked by the loss of cognitive ability, generally over a period of 10–15 years, associated with the development of abnormal tissues and protein deposits in the brain.

AMASTIA. A birth defect involving absent breast(s).

AMBIGUOUS. Unclear or open to more than one interpretation.

AMELOGENESIS IMPERFECTA. A hereditary dental defect characterized by discoloration of the teeth.

AMINO ACID. Organic compounds that form the building blocks of protein. There are 20 types of amino acids (eight are “essential amino acids” which the body cannot make and must therefore be obtained from food).

AMNIOCENTESIS. A procedure performed between 16 and 18 weeks of pregnancy in which a needle is

inserted through a woman’s abdomen into the uterus to draw out a small sample of the amniotic fluid from around the fetus; either the fluid itself or cells from the fluid can be used for a variety of tests to obtain information about genetic disorders and other medical conditions in the fetus.

AMNION. The thin, protective sac that suspends the fetus in a protective amniotic fluid.

AMNION. Thin, tough innermost layer of the amniotic sac.

AMNIOTIC FLUID. The liquid in the amniotic sac that cushions the fetus and regulates temperature in the placental environment. Amniotic fluid also contains fetal cells.

AMNIOTIC SAC. The membranous sac that contains the fetus and the amniotic fluid during pregnancy.

AMNIOTIC SAMPLE. Sample of amniotic fluid, the protective fluid surrounding a fetus in the womb.

AMPLIFICATION. A process by which something is made larger. In clotting, only a very few chemicals are released by the initial injury; they result in a cascade of chemical reactions which produces increasingly larger quantities of different chemicals, resulting in an appropriately-sized, strong fibrin clot.

AMPUTATION. Surgical removal of any portion of the body.

AMYLASE. A digestive enzyme found in saliva or pancreatic fluid that breaks down starch and sugars.

AMYLOID. A waxy translucent substance composed mostly of protein, that forms plaques (abnormal deposits) in the brain.

AMYLOIDOSIS. Accumulation of amyloid deposits in various organs and tissues in the body such that normal functioning of an organ is compromised.

AMYOPLASIA. The mildest form of arthrogryposis multiplex congenita, characterized by sporadic and recurrent contractures of the wrists, elbows, and knees; club feet, and an abnormal internal rotation of the shoulders.

AMYOTROPHIC LATERAL SCLEROSIS (ALS). A neurodegenerative disease that is caused by degeneration of the motor fibers and neurons in the brain, brainstem, and spinal cord, leading to stiffness, weak muscles, and respiratory failure. Also called Lou Gehrig’s disease.

ANAGEN. The growth phase of the human hair growth cycle.

ANALYTE. A chemical substance such as an enzyme, hormone, or protein.

ANDROGENS. A group of steroid hormones that stimulate the development of male sex organs and male secondary sexual characteristics.

ANEMIA. A condition in which there is an abnormally low number of red blood cells in the bloodstream. It may be due to loss of blood, an increase in red blood cell destruction, or a decrease in red blood cell production. Major symptoms are paleness, shortness of breath, unusually fast or strong heart beats, and tiredness.

ANESTHESIA. Lack of normal sensation (especially to pain) brought on by medications just prior to surgery or other medical procedures.

ANESTHETIC. Drug used to temporarily cause loss of sensation in an area of the body. An anesthetic may either be general, associated with a loss of consciousness, or local, affecting one area only without loss of consciousness. Anesthetics are administered either via inhalation or needle injection.

ANEUPLOIDY. Having too many or too few copies of a specific chromosome; the most common forms are trisomy (three) and monosomy (one); two copies of a chromosome is normal.

ANEURYSM. Widening of an artery, which could eventually bleed.

ANGELMAN SYNDROME. A syndrome caused by a deletion in the maternally inherited chromosome 15 or uniparental disomy of the paternal chromosome 15.

ANGIOGRAPHY. Radiographic examination of blood vessels after injection with a radiopaque contrast substance or dye.

ANGIOID STREAKS. Gray, orange, or red wavy branching lines in Bruch's membrane.

ANGIOKERATOMA. Skin rash comprised of red bumps. Rash most commonly occurs between the belly button and the knees.

ANGIOMA. A benign tumor composed of blood vessels or lymph vessels.

ANGIONEUROTIC EDEMA. Recurrent episodes of swelling of the tissues of the body caused by an overactive immune system. This is also called angioedema.

ANGIOTENSINOGEN. A plasma globulin (protein) formed in the liver and directly involved in the regulation of blood pressure.

ANKYLOSIS. Immobility of a joint due to the formation of new bone at the site of inflammation.

ANOMALOUS VENOUS RETURN. Normally, the veins that bring blood containing oxygen from the lungs to the heart (called pulmonary veins) are connected to the left

atrium. In this situation, the pulmonary veins are connected to the right atrium.

ANOMALOUS. Irregular or different from normal.

ANOMALY. Generally, something that is different from what is normal or expected. More specifically in medicine, a malformation or abnormality in any part of the body.

ANOPHTALMIA. A medical condition in which one eye is missing.

ANOTIA. Absence of an ear.

ANTERIOR FONTANELLE. The soft-spot on the skull of an infant that is located in the center of the head just behind the hairline.

ANTERIOR HORN CELLS. Subset of motor neurons within the spinal cord.

ANTI-ANDROGEN DRUGS. Drugs that block the activity of the male hormone.

ANTIBIOTICS. A group of medications that kill or slow the growth of bacteria.

ANTIBODY. A special protein made by the body's immune system as a defense against foreign material (bacteria, viruses, etc.) that enters the body. It is uniquely designed to attack and neutralize the specific antigen that triggered the immune response.

ANTICIPATION. The apparent tendency of certain diseases to appear at earlier ages and with increasing severity in successive generations.

ANTICOAGULANT. Drugs used to prevent blood clots.

ANTIDIURETIC HORMONE (VASOPRESSIN). A hormone that acts on the kidneys to regulate water balance.

ANTIGEN. A substance or organism that is foreign to the body and stimulates a response from the immune system.

ANTIGEN PRESENTING CELL. Cells that are able to present foreign antigen in conjunction with MHC proteins to the immune system.

ANUS. The opening at the end of the intestine that carries waste out of the body

AORTA. The main artery located above the heart which pumps oxygenated blood out into the body. Many congenital heart defects affect the aorta.

AORTIC REGURGITATION. A condition in which the aortic valve does not close tightly, allowing blood to flow backwards from the aorta into the heart.

AORTIC ROOT. The location where the aorta (main heart blood vessel) inserts in the heart. Enlargement of the aortic root can cause it to rupture.

AORTIC STENOSIS. A condition in which the aortic valve does not open properly, making it difficult for blood to leave the heart.

APHASIA. The loss of the ability to speak or to understand written or spoken language. A person who cannot speak or understand language is said to be *aphasic*.

APLASIA. Defective development resulting in absence of all or part of an organ or tissue.

APLASTIC ANEMIA. A form of anemia characterized by a greatly decreased formation of red and white blood cells as a result of abnormal bone marrow.

APNEA. An irregular breathing pattern characterized by abnormally long periods of the complete cessation of breathing.

APOENZYME. An enzyme that cannot function without assistance from other chemicals called cofactors.

APOPTOSIS. The normally programmed cell death process in which cells die in order to be replaced with new cells.

APPENDECTOMY. The procedure to surgically remove an appendix.

APPENDICITIS. Inflammation of the appendix.

APPENDIX. A portion of intestine attached to the cecum.

APRAXIA. Impairment of the ability to make purposeful movements, but not paralysis or loss of sensation.

AQUEOUS HUMOR. A fluid produced by the ciliary body and contained within the front chamber of the eye.

ARACHNODACTYLY. A condition characterized by abnormally long and slender fingers and toes.

ARNOLD-CHIARI MALFORMATION. A congenital anomaly in which parts of the brain protrude through the opening in the base of the skull into the spinal column.

ARRHYTHMIA. Abnormal heart rhythm, examples are a slow, fast, or irregular heart rate.

ARTERIAL. Term used to describe an artery or the entire system of arteries.

ARTERIOLE. The smallest type of artery.

ARTERIOPATHY. Damage to blood vessels.

ARTERIOSCLEROSIS. Hardening of the arteries that often results in decreased ability of blood to flow smoothly.

ARTERIOVENOUS MALFORMATION (AVM). Abnormal, direct connection between the arteries and veins (blood vessels). Can range from very small to large in size. Bleeding or an aneurysm may result.

ARTERY. A blood vessel that carries blood away from the heart to peripheral tissues.

ARTHROCHALASIA. Excessive looseness of the joints.

ARTHROGRYPOSIS. Abnormal joint contracture.

ASPERGER SYNDROME. A term used to describe high-functioning individuals with autism. These individuals usually have normal IQ and some language skills.

ASPHYXIA. Lack of oxygen. In the case of cerebral palsy, lack of oxygen to the brain.

ASPIRATION PNEUMONIA. Lung infection due to food or liquids accidentally getting into lungs.

ASPIRATION. Inhalation of food or saliva.

ASPLENIA. The absence of the spleen in the body.

ASSOCIATION. A non-random occurrence in two or more individuals of the same group of anomalies that are not otherwise known to be a sequence or syndrome.

ASTIGMATISM. A cause of poor eyesight, usually due to an error in the refraction of light within the eye.

ASTROCYTOMA. Tumor of the central nervous system derived from astrocytes

ASYMMETRIC SEPTAL HYPERTROPHY. A condition in which the septum (the wall that separates the atria of the heart) is abnormally excessively thickened. In microscopic examination, normal alignment of muscle cells is absent (myocardial disarray).

ASYMMETRY. Without symmetry, as when a disease, condition, or symptom is not manifested equally on both sides of the body or body part.

ASYMPTOMATIC. Without symptoms.

ASYMPTOMATIC CARRIER. A person who carries a recessive trait but does not show any characteristics of the trait.

ATAXIA. A condition marked by impaired muscular coordination, most frequently resulting from disorders in the brain or spinal cord.

ATHEROSCLEROSIS. Hardening of the arteries caused by cholesterol and fat deposits. Increases risk of heart disease, stroke, and other complications.

ATHETOSIS. A condition marked by slow, writhing, involuntary muscle movements.

ATOPIC. A condition or disease that is the result of an allergic reaction.