

Glossary

antibiotic: A drug that can kill or inhibit the growth of bacteria. Antibiotics are not effective against viruses.

aorta: The largest artery in the human body. It originates in the left ventricle of the heart and extends down into the abdomen.

asthma: A chronic inflammatory disease of the airways. Its symptoms include wheezing, coughing, and shortness of breath.

biopsy: A medical procedure that involves removing of a small amount of tissue for examination. The tissue sample is often analyzed by a pathologist to determine the presence or extent of a disease.

bone marrow: Spongy tissue found in the hollow interiors of some large bones. Bone marrow contains stem cells that produce various types of blood cells.

cardiologist: A doctor with specialized training in the prevention, diagnosis, and treatment of diseases associated with the heart and circulatory system.

central nervous system: That part of the nervous system consisting of the brain and spinal cord.

chromosome: An organized package of DNA found in the nucleus of the cell. Humans have 23 pairs of chromosomes—22 pairs of numbered chromosomes, called autosomes, and one pair of sex chromosomes, X and Y.

clinical trial: A controlled study designed to measure the safety or effectiveness of a new drug or medical procedure.

common disease: A disease that affects a relatively large population. In contrast, a rare disease is defined as one affecting fewer than 200,000 people in the United States.

conjunctivitis: An inflammation of the outermost layer of the eye (also called pinkeye). The inflammation may be caused by a virus, a bacterium, or an allergic reaction.

connective tissue: A type of tissue that functions to connect other tissues to each other and hold organs in place. The protein collagen is an important structural component of connective tissues.

disease: A condition characterized by the improper functioning of one or more body parts or systems. The most common causes of diseases are genetic mutation, infection by a pathogen, and exposure to a harmful substance in the environment.

echocardiogram: A medical procedure that uses sound waves to create a moving picture of the heart. An echocardiogram provides a more detailed view of the heart than an X-ray does, and it doesn't involve exposure to radiation.

Ehlers-Danlos syndrome: A group of inherited diseases of the connective tissue. The syndrome is caused by defects in the collagen protein that result in a variety of symptoms affecting multiple body systems. The condition may be mild or life threatening.

genetics: The study of the inheritance patterns of traits. Genetic information is coded in the molecule DNA. Changes to DNA, called mutations, can cause disease.

heart murmur: An extra or unusual sound in the heartbeat. Some heart murmurs are harmless, while others indicate heart problems.

infectious agent: Usually a microscopic agent, such as a virus or bacterium, that can cause an infection and be spread from person to person.

institutional review board: A committee established to approve, monitor, and review medical research involving human subjects.

karyotype: A photograph showing the number and shape of an individual's chromosomes.

leukemia: A cancer of the blood-forming tissues in the bone marrow. Leukemia is often characterized by the production of large numbers of unhealthy white blood cells.

lumbar puncture: A medical procedure also known as a spinal tap. It involves obtaining a small sample of cerebrospinal fluid for analysis. It can be used to look for the presence of an infectious disease such as meningitis or for cancer cells.

Lyme disease: A disease transmitted by a tick bite. Caused by a bacterial infection, Lyme disease is characterized by headaches, fever, depression, and a circular skin rash.

lymph node: A small, spherical organ of the immune system. Lymph nodes are distributed throughout the body and are connected by lymph vessels. They trap bacteria and foreign particles.

Marfan syndrome: A genetic disorder of the connective tissue. Because connective tissue is found throughout the body, features of Marfan syndrome are observed in many different body systems.

medical geneticist: A doctor with specialized training in the diagnosis and treatment of genetic diseases.

methotrexate: A drug used to treat various forms of cancer as well as other diseases. It works against rapidly dividing cells by interfering with the synthesis of DNA, RNA, and protein molecules.

mitral valve prolapse: A heart problem resulting from a faulty valve that separates the upper and lower chambers of the left side of the heart. Symptoms of the condition include chest pain, fatigue, heart palpitations, cough, and shortness of breath after activity.

mutation: A change to the sequence of a DNA molecule. Mutations may be caused by radiation, chemicals, viruses, and mistakes that occur during DNA replication.

myopia: A condition in which the eye focuses incorrectly, making distant objects appear blurred (also called nearsightedness).

necrotizing fasciitis: A rare but very serious bacterial infection that can destroy skin, muscle, and underlying tissues (also called flesh-eating disease). If not treated immediately, it can be fatal.

ophthalmologist: A doctor with specialized training in the diagnosis and treatment of eye problems.

Orphan Drug Act: A piece of federal legislation passed in 1983. It gives pharmaceutical and healthcare companies financial incentives to develop drugs and other products aimed at the treatment of rare diseases.

orthopedist: A doctor with specialized surgical training in the treatment of disorders of the skeletal system.

osteogenesis imperfecta: A genetic disease characterized by brittle bones that easily break.

platelet: A type of blood cell produced by the bone marrow. Platelets help blood coagulate in response to damage to the blood vessels.

prednisone: A drug used to treat inflammatory diseases and some forms of cancer. In the treatment of leukemia, prednisone induces cancer cells to commit suicide.

pulmonologist: A doctor with specialized training in the diagnosis and treatment of respiratory diseases.

rare disease: In the United States, a rare disease is one that affects fewer than 200,000 people.

red blood cell: A type of blood cell produced in the bone marrow. Red blood cells are responsible for transporting oxygen to the cells of the body.

Rocky Mountain spotted fever: A disease transmitted by a tick bite. Caused by a bacterial infection, Rocky Mountain spotted fever is characterized by headaches, fever, muscle pain, and skin rash.

scleroderma: A disease of the connective tissue that involves an excessive accumulation of the protein collagen. Because connective tissue is found throughout the body, features of scleroderma are observed in many different body systems.

scoliosis: A medical condition in which a person's spine shows a curve from side to side. Scoliosis may be associated with other diseases, and its symptoms range from mild to severe.

stem cell: A cell with the potential to form many of the different cell types found in the body. When stem cells divide, they can form more stem cells or other cells that perform specialized functions.

stigma: In a social setting, a public disapproval of some personal characteristic or belief that is at odds with the cultural norms.

vincristine: A drug used to treat some forms of cancer. It targets rapidly dividing cells by interfering with the action of a protein needed for cell division.

white blood cell: A type of blood cell produced by the bone marrow. White blood cells are part of the immune system and help protect the body against foreign material and infection by pathogens.

