Glossary

A

Ablation: elimination or removal

Absorption: When digested food moves from the digestive tract into the blood stream.

ACE (angiotensin-converting enzyme) inhibitor: a medication that lowers blood pressure.

Acquired genetic mutation: *See:* somatic cell genetic mutation

Acute: Sharp. Strong. A reaction or illness that occurs strongly but only lasts for a short time.

Additive genetic effects: When the combined effects of alleles at different loci are equal to the sum of their individual effects. *See also:* anticipation, complex trait

Adenine (A): A nitrogenous base, one member of the base pair AT (adenine-thymine). *See also:* base pair, nucleotide

Adduction: A movement of a body part towards the midline of the body.

Adrenaline: A hormone that activates in certain situations, usually when you are scared or in an emergency. It speeds up the reactions in your body.

Affected relative pair: Individuals related by blood, each of whom is affected with the same trait. Examples are affected sibling, cousin, and avuncular pairs. *See also:* avuncular relationship

Aggregation technique: A technique used in model organism studies in which embryos at the 8-cell stage of development are pushed together to yield a single embryo (used as an alternative to microinjection). *See also:* model organisms

Allele: Alternative form of a genetic locus; a single allele for each locus is inherited from each parent (e.g., at a locus for eye color the allele might result in blue or brown eyes). *See also:* locus, gene expression

Allergy: When your body is sensitive to something, like pollen, foods, animals, or drugs, you may have a reaction such as sneezing, itching, and skin rashes.

Allogeneic: Variation in alleles among members of the same species.

Alternative splicing: Different ways of combining a gene's exons to make variants of the complete protein

Alveoli: Tiny air sacs in the lungs.

Ambulatory: Able to get out of bed and walk around.

Anatomy: The study of the different parts of the body.

Amino acid: Any of a class of 20 molecules that are combined to form proteins in living things. The sequence of amino acids in a protein and hence protein function are determined by the genetic code.

Amplification: An increase in the number of copies of a specific DNA fragment; can be in vivo or in vitro. *See also:* cloning, polymerase chain reaction

Anemia: An illness in which there is a low blood count in the body. This is when you don’t have enough red blood cells.

Anesthesia: A drug that doctors may give patients before an operation. It may be an injection or a gas that you breathe in. It doesn't hurt and it may make you lose normal sensation or go to sleep so that you can't feel anything while the doctors do what they need to do.

Aneurysm: a sac-like protrusion from a blood vessel or the heart.

Angina or angina pectoris: chest pain.

Angiography: an x-ray that uses dye injected into the coronary arteries so that blood circulation can be studied.

Angioplasty: a procedure for treating diseased arteries non-surgically.

Animal model: *See:* model organisms

Annotation: Adding pertinent information such as gene coded for, amino acid sequence, or other commentary to the database entry of raw sequence of DNA bases. *See also:* bioinformatics

Antibiotic: medication to stop the spread of an infection.

Antibodies: proteins produced by your body to act specifically against an invading particle, or antigen. Antibodies are part of your immune system.

Anticipation: Each generation of offspring has increased severity of a genetic disorder; e.g., a grandchild may have earlier onset and more severe symptoms than the parent, who had earlier onset than the grandparent. *See also:* additive genetic effects, complex trait

Anticoagulant: a medication that keeps blood from clotting.

Antigen: Any outside substance that causes an immune response.

Antihypertensive: a medication or other therapy that lowers blood pressure.

Antisense: Nucleic acid that has a sequence exactly opposite to an mRNA molecule made by the body; binds to the mRNA molecule to prevent a protein from being made. *See also:* transcription

Aorta: The largest artery of the body that supplies oxygenated blood to all other arteries.

Aortic valve: the valve that regulates blood flow from the heart into the aorta.

Aphasia: the inability to speak or understand due to brain injury or disease.

Apoptosis: Programmed cell death, the body's normal method of disposing of damaged, unwanted, or unneeded cells. *See also:* cell

Arrayed library: Individual primary recombinant clones (hosted in phage, cosmid, YAC, or other vector) that are placed in two-dimensional arrays in microtiter dishes. Each primary clone can be identified by the identity of the plate and the clone location (row and column) on that plate. Arrayed libraries of clones can be used for many applications, including screening for a specific gene or genomic region of interest. *See also:* library, genomic library, gene chip technology

Arrhythmia (or dysrhythmia): an abnormal heartbeat.

Arterioles: small branches of arteries.

Arteriosclerosis: commonly called "hardening of the arteries;" a variety of conditions caused by fatty or calcium deposits in the artery causing the walls them to thicken.

Artery: a blood vessel that carries oxygen-rich blood away from the heart to the body.

Assembly: Putting sequenced fragments of DNA into their correct chromosomal positions.

Asthma: When a person with swollen or irritated airways has trouble breathing normally because not enough oxygen is going into their lungs.

Atherectomy: a non-surgical procedure for removing plaque from the walls of arteries with a rotating blade.

Atherosclerosis: the disease of accumulated plaque (built-up waxy substance) in the blood vessels.

Atrium (atria pl.): one of two upper chambers in the heart.

Atrioventricular block: an interruption of the electrical signal between the atria and the ventricles.

Atrioventricular (AV) node: a cluster of cells between the atria and ventricles that regulate the electrical current.

Auditory Nerve: The nerve that carries impulses from the inner ear to the brain.

Autoradiography: A technique that uses X-ray film to visualize radioactively labeled molecules or fragments of molecules; used in analyzing length and number of DNA fragments after they are separated by gel electrophoresis.

Autosomal dominant: A gene on one of the non-sex chromosomes that is always expressed, even if only one copy is present. The chance of passing the gene to offspring is 50% for each pregnancy. *See also:* autosome, dominant, gene

Autosome: A chromosome not involved in sex determination. The diploid human genome consists of a total of 46 chromosomes: 22 pairs of autosomes, and 1 pair of sex chromosomes (the X and Y chromosomes). *See also:* sex chromosome

Avuncular relationship: The genetic relationship between nieces and nephews and their aunts and uncles.

B

Backcross: A cross between an animal that is heterozygous for alleles obtained from two parental strains and a second animal from one of those parental strains. Also used to describe the breeding protocol of an outcross followed by a backcross. *See also:* model organisms

Bacteria: A tiny particle that can enter your body and make you sick. Bacteria lives on things that aren't clean and when you breathe, touch, or swallow them they can enter your body. You may use antibiotics to help your body fight them off.

Bacterial artificial chromosome (BAC): A vector used to clone DNA fragments (100- to 300-kb insert size; average, 150 kb) in *Escherichia coli* cells. Based on naturally occurring F-factor plasmid found in the bacterium *E. coli*. *See also:* cloning vector

Bacteriophage: *See:* phage

Base One of the molecules that form DNA and RNA molecules. *See also:* nucleotide, base pair, base sequence

Base pair (bp): Two nitrogenous bases (adenine and thymine or guanine and cytosine) held together by weak bonds. Two strands of DNA are held together in the shape of a double helix by the bonds between base pairs.

Base sequence: The order of nucleotide bases in a DNA molecule; determines structure of proteins encoded by that DNA.

Base sequence analysis: A method, sometimes automated, for determining the base sequence.

Behavioral genetics: The study of genes that may influence behavior.

Beta blocker: an antihypertensive medication that limits the activity of epinephrine (hormone that increases blood pressure).

Bile: A liquid made by the liver that helps the digestion of fats.

Bioinformatics: The science of managing and analyzing biological data using advanced computing techniques. Especially important in analyzing genomic research data.
See also: informatics

Biopsy: the procedure of taking a small tissue sample for examination.

Bioremediation: the use of biological organisms such as plants or microbes to aid in removing hazardous substances from an area.

Biotechnology: A set of biological techniques developed through basic research and now applied to research and product development. In particular, biotechnology refers to the use by industry of recombinant DNA, cell fusion, and new bioprocessing techniques.

Birth defect: Any harmful trait, physical or biochemical, present at birth, whether a result of a genetic mutation or some other nongenetic factor.
See also: congenital, gene, mutation, syndrome

BLAST: A computer program that identifies homologous (similar) genes in different organisms, such as human, fruit fly, or nematode.

Blood Gas: A blood test that measures the concentration of oxygen in the blood.

Blood clot: a gelled mass of blood tissue.

Blood Pressure: This is a fun painless test to check the pressure of your blood. A doctor or nurse will put a blood pressure band around your upper arm. This band will inflate like a balloon to squeeze your arm and create pressure to check if blood flow in the heart is working.

Blood pressure cuff: a device usually placed around the upper of the arm to measure blood pressure.

Blood Type: There are four major types, each with a different type of chemical marker that's attached to your red blood cells. The markers determine if we have type A blood, type B blood, and type O blood, and type AB. Each type can also be positive (+) or negative (-).

Blood Vessel: A tube through which blood flows

Bone Marrow: This is the gel like stuff that is in the middle of your bones. Marrow helps your body make different kinds of blood cells that help your body fight off bacteria and also helps carry oxygen to the whole body.

Bone Scan: To study your bones, doctors may give you a dose of a radioactive substance by injection, then they take a picture that lets them understand how the substance traveled through your body.

Brady...: suffix meaning slow.

Bradycardia: abnormally slow heartbeat.

Bronchitis: An infection of the tubes in the lungs.

Bundle-branch block: a condition in which the heart's electrical system is unable to normally conduct the electrical signal.

C

Calcium channel blocker (or calcium blocker): a medication that lowers blood pressure.

Cancer: Diseases in which abnormal cells divide and grow unchecked. Cancer can spread from its original site to other parts of the body and can be fatal. *See also:* hereditary cancer, sporadic cancer

Candidate gene: A gene located in a chromosome region suspected of being involved in a disease. *See also:* positional cloning, protein

Capillaries: tiny blood vessels between arteries and veins that distribute oxygen-rich blood to the body.

Capillary array: Gel-filled silica capillaries used to separate fragments for DNA sequencing. The small diameter of the capillaries permit the application of higher electric fields, providing high speed, high throughput separations that are significantly faster than traditional slab gels.

Carbohydrates: The group of compound that includes sugars and starches.

Carcinogen Something which causes cancer to occur by causing changes in a cell's DNA.s*ee also:* mutagene

Cardiac: pertaining to the heart.

Cardiac arrest: the stopping of heartbeat.

Cardiac catheterization: a diagnostic procedure in which a tiny, hollow tube (catheter) is inserted into an artery to go to the heart in order to image the heart and blood vessels.

Cardiac Muscle Tissue: Muscle tissue found only in the heart, which makes your heart beat.

Cardiac output: the amount of blood that goes through the circulatory system in one minute.

Cardiology: the clinical study and practice of treating the heart.

Cardiomyopathy: a disease of the heart muscle that causes it to lose its pumping strength.

Cardiovascular (CV): pertaining to the heart and blood vessel (circulatory) system.

Cardioversion: the procedure of applying electrical shock to the chest to change an abnormal heartbeat into a normal one.

Carotid artery: the major arteries in the neck that supply blood to the brain.

Carrier: An individual who possesses an unexpressed, recessive trait.

cDNA library: A collection of DNA sequences that code for genes. The sequences are generated in the laboratory from mRNA sequences. *See also:* messenger RNA

Catheter: A long, thin, hollow tube that is inserted by a doctor into a large blood vessel and lets them perform important tests in a way that’s not too disruptive.

Cell: The basic unit of any living organism that carries on the biochemical processes of life. *See also:* genome, nucleus

Centimorgan (cM): A unit of measure of recombination frequency. One centimorgan is equal to a 1% chance that a marker at one genetic locus will be separated from a marker at a second locus due to crossing over in a single generation. In human beings, one centimorgan is equivalent, on average, to one million base pairs. *See also:* megabase

Centromere:A specialized chromosome region to which spindle fibers attach during cell division.

Cerebral embolism: a blood clot from one part of the body that is carried by the bloodstream to the brain where it blocks an artery.

Cerebral hemorrhage: bleeding within the brain.

Cerebral thrombosis: formation of a blood clot in an artery that supplies blood to the brain.

Cerebrovascular: pertaining to blood vessels in the brain.

Cerebrovascular accident: apoplexy or stroke; an impeded blood supply to the brain.

Cerebrovascular occlusion: an obstruction in the blood vessel in the brain.

Cerebrum: The largest part of the brain that controls a person's thoughts. It is what makes up personality, the senses, and movement.

Chest pain: See *angina*.

Chimera (pl. chimaera): An organism that contains cells or tissues with a different genotype. These can be mutated cells of the host organism or cells from a different organism or species.

Chimeraplasty: An experimental targeted repair process in which a desirable sequence of DNA is combined with RNA to form a chimeraplast. These molecules bind selectively to the target DNA. Once bound, the chimeraplast activates a naturally occurring gene-correcting mechanism. Does not use viral or other conventional gene-delivery vectors.
See also: gene therapy, cloning vector

Cholesterol: a waxy substance that is produced in the human body, in animal fats, and in dairy products and is transported in the blood.

Chloroplast chromosome: Circular DNA found in the photosynthesizing organelle (chloroplast) of plants instead of the cell nucleus where most genetic material is located.

Chromomere: One of the serially aligned beads or granules of a eukaryotic chromosome, resulting from local coiling of a continuous DNA thread.

Chromosomal deletion: The loss of part of a chromosome's DNA.

Chromosomal inversion: Chromosome segments that have been turned 180 degrees. The gene sequence for the segment is reversed with respect to the rest of the chromosome.

Chromosome: The self-replicating genetic structure of cells containing the cellular DNA that bears in its nucleotide sequence the linear array of genes. In prokaryotes, chromosomal DNA is circular, and the entire genome is carried on one chromosome.

Eukaryotic genomes consist of a number of chromosomes whose DNA is associated with different kinds of proteins.

Chromosome painting: Attachment of certain fluorescent dyes to targeted parts of the chromosome. Used as a diagnositic for particular diseases, e.g. types of leukemia.

Chromosome region p: A designation for the short arm of a chromosome.

Chromosome region q: A designation for the long arm of a chromosome.

Chronic: An illness that is ongoing or that may keep going away and coming back.
Cineangiography: the procedure of taking moving pictures to show the passage of dye through blood vessels.

Circulatory system: pertaining to the heart and blood vessels, and the circulation of blood.

Claudication: pain or fatigue in arms and legs due to poor supply of oxygen to the muscles.

Clone: An exact copy made of biological material such as a DNA segment (e.g., a gene or other region), a whole cell, or a complete organism.

Clone bank: *See:* genomic library

Cloning Using specialized DNA technology to produce multiple, exact copies of a single gene or other segment of DNA to obtain enough material for further study. This process, used by researchers in the Human Genome Project, is referred to as cloning DNA. The resulting cloned (copied) collections of DNA molecules are called clone libraries. A second type of cloning exploits the natural process of cell division to make many copies of an entire cell. The genetic makeup of these cloned cells, called a cell line, is identical to the original cell. A third type of cloning produces complete, genetically identical animals such as the famous Scottish sheep, Dolly. *See also:* cloning vector

Cloning vector: DNA molecule originating from a virus, a plasmid, or the cell of a higher organism into which another DNA fragment of appropriate size can be integrated without loss of the vector's capacity for self-replication; vectors introduce foreign DNA into host cells, where the DNA can be reproduced in large quantities. Examples are plasmids, cosmids, and yeast artificial chromosomes; vectors are often recombinant molecules containing DNA sequences from several sources.

Code: *See:* genetic code

Codominance: Situation in which two different alleles for a genetic trait are both expressed. *See also:* autosomal dominant, recessive gene

Codon: *See:* genetic code

Coisogenic or congenic: Nearly identical strains of an organism; they vary at only a single locus.

Comparative genomics: The study of human genetics by comparisons with model organisms such as mice, the fruit fly, and the bacterium *E. coli*.

Complementary DNA (cDNA): DNA that is synthesized in the laboratory from a messenger RNA template.

Complementary sequence: Nucleic acid base sequence that can form a double-stranded structure with another DNA fragment by following base-pairing rules (A pairs with T and C with G). The complementary sequence to GTAC for example, is CATG.

Complete Blood Count (CBC): A doctor may do a test to measure what is in your blood. He or she will look for the different amounts of white blood cells, red blood cells and platelets.

Complex trait: Trait that has a genetic component that does not follow strict Mendelian inheritance. May involve the interaction of two or more genes or gene-environment interactions. *See also:* Mendelian inheritance, additive genetic effects

Computational biology: *See:* bioinformatics

Computed tomography (CT or CAT scan): an x-ray procedure that produces cross-sectional images of the body.

Confidentiality: In genetics, the expectation that genetic material and the information gained from testing that material will not be available without the donor's consent.

Congenital: Any trait present at birth, whether the result of a genetic or nongenetic factor. *See also:* birth defect

Congestive heart failure: a condition in which the heart cannot pump out all of the blood that enters it, which leads to an accumulation of blood in the vessels and fluid in the body tissues.

Conjunctivitis: When the eye gets infected with bacteria; also known as "pink eye." It is pretty contagious so if you have it you want to make sure you don't touch other people.

Consciousness: When you are fully awake and your brain is working normally.

Conserved sequence: A base sequence in a DNA molecule (or an amino acid sequence in a protein) that has remained essentially unchanged throughout evolution.

Constitutive ablation: Gene expression that results in cell death.

Contagious: When a person can catch a sickness from another, like the common cold.

Contig: Group of cloned (copied) pieces of DNA representing overlapping regions of a particular chromosome.

Contig map: A map depicting the relative order of a linked library of overlapping clones representing a complete chromosomal segment.

Cornea: The clear layer that protects the front of the eye.

Coronary arteries: two arteries that come from the aorta to provide blood to the heart muscle.

Coronary artery bypass graft (CAB or CABG): a surgical procedure in which a healthy blood vessel is transplanted from another part of the body into the heart to replace or bypass a diseased vessel.

Coronary artery spasm: a sudden closing of an artery, which cuts off blood flow to the heart and causes symptom of angina or heart attack.

Coronary heart disease: a condition in which the coronary arteries narrow from an accumulation of plaque (atherosclerosis) and cause a decrease in blood flow.

Coronary occlusion: an obstruction of one of the coronary arteries that decreases flow to the heart muscle.

Coronary thrombosis: the formation of a clot in one of the arteries that carry blood to the heart muscle.

Cosmid: Artificially constructed cloning vector containing the cos gene of phage lambda. Cosmids can be packaged in lambda phage particles for infection into *E. coli*; this permits cloning of larger DNA fragments (up to 45kb) than can be introduced into bacterial hosts in plasmid vectors.

Crossing over: The breaking during meiosis of one maternal and one paternal chromosome, the exchange of corresponding sections of DNA, and the rejoining of the chromosomes. This process can result in an exchange of alleles between chromosomes. *See also:* recombination

CT Scan: Cat Scans. (Computed Axial Tomography Scan) This is a painless X-rays that uses a computer to make images of your body. This test can produce images of bone, blood and soft tissues so it is often used for patients who have had an injury like a car collision or sports injury.

Cyanosis: insufficient oxygen in the blood.

Cytogenetics: The study of the physical appearance of chromosomes. *See also:* karyotype

Cytological band: An area of the chromosome that stains differently from areas around it. *See also:* cytological map

Cytological map: A type of chromosome map whereby genes are located on the basis of cytological findings obtained with the aid of chromosome mutations.

Cytoplasmic (uniparental) inheritance: *See:* cytoplasmic trait

Cytoplasmic trait: A genetic characteristic in which the genes are found outside the nucleus, in chloroplasts or mitochondria. Results in offspring inheriting genetic material from only one parent.

Cytosine (C): A nitrogenous base, one member of the base pair GC (guanine and cytosine) in DNA. *See also:* base pair, nucleotide

D

Data warehouse: A collection of databases, data tables, and mechanisms to access the data on a single subject.

Defibrillator: an electronic device used to establish normal heartbeat.

Dehydration: You may or may not feel very thirsty. This happens when your body has less than the normal amount of fluids.

Deletion: A loss of part of the DNA from a chromosome; can lead to a disease or abnormality. *See also:* chromosome, mutation

Deletion map: A description of a specific chromosome that uses defined mutations -- specific deleted areas in the genome-- as 'biochemical signposts,' or markers for specific areas.

Deoxyribonucleotide: *See:* nucleotide

Deoxyribose: A type of sugar that is one component of DNA (deoxyribonucleic acid).

Department of Emergency Medicine (DEM): You may be taken here when there is an emergency that has to be treated right away.

Depressant: A drug that slows down (depresses) the actions of the body.

Dermis: The inner layer of your skin.

Diastolic blood pressure: the lowest blood pressure measure in the arteries, which occurs between heartbeats.

Diploid: A full set of genetic material consisting of paired chromosomes, one from each parental set. Most animal cells except the gametes have a diploid set of chromosomes. The diploid human genome has 46 chromosomes. *See also:* haploid

Directed evolution: A laboratory process used on isolated molecules or microbes to cause mutations and identify subsequent adaptations to novel environments.

Directed mutagenesis: Alteration of DNA at a specific site and its reinsertion into an organism to study any effects of the change.

Directed sequencing: Successively sequencing DNA from adjacent stretches of chromosome.

Disease-associated genes: Alleles carrying particular DNA sequences associated with the presence of disease.

Diuretic: a medication that lowers blood pressure.

DNA (deoxyribonucleic acid): The molecule that encodes genetic information. DNA is a double-stranded molecule held together by weak bonds between base pairs of nucleotides. The four nucleotides in DNA contain the bases adenine (A), guanine (G), cytosine (C), and thymine (T). In nature, base pairs form only between A and T and between G and C; thus the base sequence of each single strand can be deduced from that of its partner.

DNA bank: A service that stores DNA extracted from blood samples or other human tissue.

DNA probe: *See:* probe

DNA repair genes: Genes encoding proteins that correct errors in DNA sequencing.

DNA replication: The use of existing DNA as a template for the synthesis of new DNA strands. In humans and other eukaryotes, replication occurs in the cell nucleus.

DNA sequence: The relative order of base pairs, whether in a DNA fragment, gene, chromosome, or an entire genome. *See also:* base sequence analysis

Domain: A discrete portion of a protein with its own function. The combination of domains in a single protein determines its overall function.

Dominant: An allele that is almost always expressed, even if only one copy is present. *See also:* gene, genome

Doppler ultrasound: A procedure that uses sound waves to evaluate heart, blood vessels, and valves.

Double helix: The twisted-ladder shape that two linear strands of DNA assume when complementary nucleotides on opposing strands bond together.

Draft sequence: The sequence generated by the HGP as of June 2000 that, while incomplete, offers a virtual road map to an estimated 95% of all human genes. Draft

sequence data are mostly in the form of 10,000 base pair-sized fragments whose approximate chromosomal locations are known.
See also: sequencing, finished DNA sequence, working draft DNA sequence

Dyspnea: shortness of breath.

Dysrhythmia (or arrythmia): an abnormal heart rhythm.

E

Edema: swelling.

Ejection fraction: the measurement of the blood pumped out of the ventricles.

Electrocardiogram (ECG or EKG): a procedure that measures the electrical activity in a heartbeat.

Electrophysiological study (EPS): a cardiac catheterization to study electrical current in patients who have arrhythmias.

Embryonic stem (ES) cells: An embryonic cell that can replicate indefinitely, transform into other types of cells, and serve as a continuous source of new cells.

Endarterectomy: the surgical removal of plaque or blood clots in an artery.

Endocardium: the membrane that covers the inside surface of the heart.

Endocarditis: a bacterial infections of the heart lining.

Endonuclease: *See:* restriction enzyme

Enlarged heart: a condition of the heart in which it is abnormally larger than normal.

Enzyme: A protein that acts as a catalyst, speeding the rate at which a biochemical reaction proceeds but not altering the direction or nature of the reaction.

Epicardium: the membrane that covers the outside of the heart.

Epistasis: One gene interfers with or prevents the expression of another gene located at a different locus.

Escherichia coli: Common bacterium that has been studied intensively by geneticists because of its small genome size, normal lack of pathogenicity, and ease of growth in the laboratory.

Estrogen: a hormone produced by the ovaries.

Eugenics: The study of improving a species by artificial selection; usually refers to the selective breeding of humans.

Eukaryote: Cell or organism with membrane-bound, structurally discrete nucleus and other well-developed subcellular compartments. Eukaryotes include all organisms except viruses, bacteria, and bluegreen algae. *See also:* prokaryote, chromosome

Evolutionarily conserved: *See:* conserved sequence

Exercise stress test: a test, usually on a treadmill, for diagnosing coronary artery disease.

Exogenous DNA: DNA originating outside an organism that has been introducted into the organism.

Exon: The protein-coding DNA sequence of a gene. *See also:* intron

Exonuclease: An enzyme that cleaves nucleotides sequentially from free ends of a linear nucleic acid substrate.

Expressed gene: *See:* gene expression

Expressed sequence tag (EST): A short strand of DNA that is a part of a cDNA molecule and can act as identifier of a gene. Used in locating and mapping genes. *See also:* cDNA, sequence tagged site

F

Familial generation (F1, F2): Each generation of offspring in a breeding program, designated F1, F2, etc.

Fibrillation: rapid contractions of the heart muscles.

Fingerprinting: In genetics, the identification of multiple specific alleles on a person's DNA to produce a unique identifier for that person. *See also:* forensics

Finished DNA Sequence: High-quality, low error, gap-free DNA sequence of the human genome. Achieving this ultimate 2003 HGP goal requires additional sequencing to close gaps, reduce ambiguities, and allow for only a single error every 10,000 bases, the agreed-upon standard for HGP finished sequence. *See also:* sequencing, draft sequence

Flow cytometry: Analysis of biological material by detection of the light-absorbing or fluorescing properties of cells or subcellular fractions (i.e., chromosomes) passing in a narrow stream through a laser beam. An absorbance or fluorescence profile of the sample is produced. Automated sorting devices, used to fractionate samples, sort successive droplets of the analyzed stream into different fractions depending on the fluorescence emitted by each droplet.

Flow karyotyping: Use of flow cytometry to analyze and separate chromosomes according to their DNA content.

Fluorescence in situ hybridization (FISH): A physical mapping approach that uses fluorescein tags to detect hybridization of probes with metaphase chromosomes and with the less-condensed somatic interphase chromatin.

Flutter: ineffective contractions of the heart muscles.

Forensics: The use of DNA for identification. Some examples of DNA use are to establish paternity in child support cases; establish the presence of a suspect at a crime scene, and identify accident victims.

Fraternal twin: Siblings born at the same time as the result of fertilization of two ova by two sperm. They share the same genetic relationship to each other as any other siblings. *See also:* identical twin

Full gene sequence: The complete order of bases in a gene. This order determines which protein a gene will produce.

Functional genomics: The study of genes, their resulting proteins, and the role played by the proteins the body's biochemical processes.

G

Gamete: Mature male or female reproductive cell (sperm or ovum) with a haploid set of chromosomes (23 for humans).

Gated blood pool scan: a nuclear x-ray evaluation of how blood pools in the heart while the patient is exercising or at rest.

GC-rich area: Many DNA sequences carry long stretches of repeated G and C which often indicate a gene-rich region.

Gel electrophoresis: *See:* electrophoresis

Gene: The fundamental physical and functional unit of heredity. A gene is an ordered sequence of nucleotides located in a particular position on a particular chromosome that encodes a specific functional product (i.e., a protein or RNA molecule). *See also:* gene expression

Gene amplification: Repeated copying of a piece of DNA; a characteristic of tumor cells. *See also:* gene, oncogene

Gene chip technology: Development of cDNA microarrays from a large number of genes. Used to monitor and measure changes in gene expression for each gene represented on the chip.

Gene expression: The process by which a gene's coded information is converted into the structures present and operating in the cell. Expressed genes include those that are transcribed into mRNA and then translated into protein and those that are transcribed into RNA but not translated into protein (e.g., transfer and ribosomal RNAs).

Gene family: Group of closely related genes that make similar products.

Gene library: *See:* genomic library

Gene mapping Determination of the relative positions of genes on a DNA molecule (chromosome or plasmid) and of the distance, in linkage units or physical units, between them.

Gene pool: All the variations of genes in a species. *See also:* allele, gene, polymorphism

Gene prediction: Predictions of possible genes made by a computer program based on how well a stretch of DNA sequence matches known gene sequences

Gene product: The biochemical material, either RNA or protein, resulting from expression of a gene. The amount of gene product is used to measure how active a gene is; abnormal amounts can be correlated with disease-causing alleles.

Gene testing: *See:* genetic testing, genetic screening

Gene therapy: An experimental procedure aimed at replacing, manipulating, or supplementing nonfunctional or misfunctioning genes with healthy genes. *See also:* gene, inherit, somatic cell gene therapy, germ line gene therapy

Gene transfer: Incorporation of new DNA into and organism's cells, usually by a vector such as a modified virus. Used in gene therapy. *See also:* mutation, gene therapy, vector

Genetic code: The sequence of nucleotides, coded in triplets (codons) along the mRNA, that determines the sequence of amino acids in protein synthesis. A gene's DNA sequence can be used to predict the mRNA sequence, and the genetic code can in turn be used to predict the amino acid sequence.

Genetic counseling: Provides patients and their families with education and information about genetic-related conditions and helps them make informed decisions.

Genetic discrimination: Prejudice against those who have or are likely to develop an inherited disorder.

Genetic engineering: Altering the genetic material of cells or organisms to enable them to make new substances or perform new functions.

Genetic engineering technology: *See:* recombinant DNA technology

Genetic illness: Sickness, physical disability, or other disorder resulting from the inheritance of one or more deleterious alleles.

Genetic informatics: *See:* bioinformatics

Genetic map: *See:* linkage map

Genetic marker: A gene or other identifiable portion of DNA whose inheritance can be followed. *See also:* chromosome, DNA, gene, inherit

Genetic material: *See:* genome

Genetic mosaic: An organism in which different cells contain different genetic sequence. This can be the result of a mutation during development or fusion of embryos at an early developmental stage.

Genetic polymorphism: Difference in DNA sequence among individuals, groups, or populations (e.g., genes for blue eyes versus brown eyes).

Genetic predisposition: Susceptibility to a genetic disease. May or may not result in actual development of the disease.

Genetic screening: Testing a group of people to identify individuals at high risk of having or passing on a specific genetic disorder.

Genetic testing: Analyzing an individual's genetic material to determine predisposition to a particular health condition or to confirm a diagnosis of genetic disease.

Genetics: The study of inheritance patterns of specific traits.

Genome: All the genetic material in the chromosomes of a particular organism; its size is generally given as its total number of base pairs.

Genome project: Research and technology-development effort aimed at mapping and sequencing the genome of human beings and certain model organisms. *See also:* Human Genome Initiative

Genomic library: A collection of clones made from a set of randomly generated overlapping DNA fragments that represent the entire genome of an organism. *See also:* library, arrayed library

Genomic sequence: *See:* DNA

Genomics: The study of genes and their function.

Genotype: The genetic constitution of an organism, as distinguished from its physical appearance (its phenotype).

Germ cell: Sperm and egg cells and their precursors. Germ cells are haploid and have only one set of chromosomes (23 in all), while all other cells have two copies (46 in all).

Germ line The continuation of a set of genetic information from one generation to the next. *See also:* inherit

Germ line gene therapy: An experimental process of inserting genes into germ cells or fertilized eggs to cause a genetic change that can be passed on to offspring. May be

used to alleviate effects associated with a genetic disease. *See also:* genomics, somatic cell gene therapy.

Germ line genetic mutation: *See:* mutation

Guanine (G): A nitrogenous base, one member of the base pair GC (guanine and cytosine) in DNA. *See also:* base pair, nucleotide

Gyandromorph: Organisms that have both male and female cells and therefore express both male and female characteristics.

H

Haploid:A single set of chromosomes (half the full set of genetic material) present in the egg and sperm cells of animals and in the egg and pollen cells of plants. Human beings have 23 chromosomes in their reproductive cells. *See also:* diploid

Haplotype: A way of denoting the collective genotype of a number of closely linked loci on a chromosome.

Heart attack: also called myocardial infarction: Damage to the heart muscle due to insufficient blood supply.

Heart block: interrupted electrical impulse to heart muscles.

Heart-lung machine: a machine that pumps blood for the heart during open heart surgery.

Heart valve prolapse: a condition of the heart valve in which it is partially open when it should be closed.

Hemizygous: Having only one copy of a particular gene. For example, in humans, males are hemizygous for genes found on the Y chromosome.

Hereditary cancer: Cancer that occurs due to the inheritance of an altered gene within a family. *See also:* sporadic cancer

Heterozygosity: The presence of different alleles at one or more loci on homologous chromosomes.

Heterozygote: *See:* heterozygosity

High blood pressure: blood pressure that is above the normal range.

High density lipoprotein (HDL): the "good" cholesterol that promotes the breakdown, and, removal from the body of its cholesterol.

Highly conserved sequence: DNA sequence that is very similar across several different types of organisms. *See also:* gene, mutation

High-throughput sequencing: A fast method of determining the order of bases in DNA. *See also:* sequencing

Holter monitor: A portable ECG machine.

Homeobox: A short stretch of nucleotides whose base sequence is virtually identical in all the genes that contain it. Homeoboxes have been found in many organisms from fruit flies to human beings. In the fruit fly, a homeobox appears to determine when particular groups of genes are expressed during development.

Homolog: A member of a chromosome pair in diploid organisms or a gene that has the same origin and functions in two or more species.

Homologous chromosome: Chromosome containing the same linear gene sequences as another, each derived from one parent.

Homologous recombination: Swapping of DNA fragments between paired chromosomes.

Homology: Similarity in DNA or protein sequences between individuals of the same species or among different species.

Homozygote: An organism that has two identical alleles of a gene. *See also:* heterozygote

Homozygous: *See:* homozygote

Human artificial chromosome (HAC): A vector used to hold large DNA fragments. *See also:* chromosome, DNA

Human gene therapy: *See:* gene therapy

Human Genome Initiative: Collective name for several projects begun in 1986 by DOE to create an ordered set of DNA segments from known chromosomal locations, develop new computational methods for analyzing genetic map and DNA sequence data, and develop new techniques and instruments for detecting and analyzing DNA. This DOE initiative is now known as the Human Genome Program. The joint national effort, led by DOE and NIH, is known as the Human Genome Project.

Human Genome Project (HGP): Formerly titled Human Genome Initiative. *See also:* Human Genome Initiative

Hybrid: The offspring of genetically different parents. *See also:* heterozygote

Hybridization: The process of joining two complementary strands of DNA or one each of DNA and RNA to form a double-stranded molecule.

Hypertension: high blood pressure.

Hypertrophic obstructive cardiomyopathy (HOCM): a bulge in the ventricle that hampers blood flow.

Hypoglycemia: low levels of blood sugar.

Hypoxia: abnormal oxygen content in the organs and tissues of the body.

I

Identical twin: Twins produced by the division of a single zygote; both have identical genotypes. *See also:* fraternal twin

Immunosuppresive medications: medications that suppress the body's immune system; used to minimize rejection of transplanted organs.

Immunotherapy: Using the immune system to treat disease, for example, in the development of vaccines. May also refer to the therapy of diseases caused by the immune system. *See also:* cancer

Impedance plethysmography: a test to evaluate blood flow through the leg.

Imprinting A phenomenon in which the disease phenotype depends on which parent passed on the disease gene. For instance, both Prader-Willi and Angelman syndromes are inherited when the same part of chromosome 15 is missing. When the father's complement of 15 is missing, the child has Prader-Willi, but when the mother's complement of 15 is missing, the child has Angelman syndrome.

In situ hybridization: Use of a DNA or RNA probe to detect the presence of the complementary DNA sequence in cloned bacterial or cultured eukaryotic cells.

In vitro: Studies performed outside a living organism such as in a laboratory.

In vivo: Studies carried out in living organisms.

Independent assortment: During meiosis each of the two copies of a gene is distributed to the germ cells independently of the distribution of other genes. *See also:* linkage

Inferior vena cava: the large blood vessel (vein) that returns blood from the legs and abdomen to the heart.

Informatics: *See:* bioinformatics

Informed consent: An individual willingly agrees to participate in an activity after first being advised of the risks and benefits. *See also:* privacy

Inherit: In genetics, to receive genetic material from parents through biological processes.

Inherited: *See:* inherit

Inotropic medications: medications that increase strength of the contractions in the heart.

Insertion: A chromosome abnormality in which a piece of DNA is incorporated into a gene and thereby disrupts the gene's normal function. *See also:* chromosome, DNA, gene, mutation

Insertional mutation: *See:* insertion

Intellectual property rights: Patents, copyrights, and trademarks. *See also:* patent

Interference: One crossover event inhibits the chances of another crossover event. Also known as positive interference. Negative interference increases the chance of a second crossover. *See also:* crossing over

Interphase: The period in the cell cycle when DNA is replicated in the nucleus; followed by mitosis.

Intravascular echocardiography: echocardiography and cardiac catheterization.

Intron: DNA sequence that interrupts the protein-coding sequence of a gene; an intron is transcribed into RNA but is cut out of the message before it is translated into protein. *See also:* exon

Ischemia: decreased flow of oxygenated blood to an organ due to obstruction in an artery.

Ischemic heart disease: coronary artery disease or coronary heart disease caused by narrowing of the coronary arteries and decreased blood flow to the heart.

Isoenzyme: An enzyme performing the same function as another enzyme but having a different set of amino acids. The two enzymes may function at different speeds.

J

Jugular veins: veins that carry blood from the head back to the heart.

Junk DNA: Stretches of DNA that do not code for genes; most of the genome consists of so-called junk DNA which may have regulatory and other functions. Also called non-coding DNA.

K

Karyotype :A photomicrograph of an individual's chromosomes arranged in a standard format showing the number, size, and shape of each chromosome type; used in low-resolution physical mapping to correlate gross chromosomal abnormalities with the characteristics of specific diseases.

Kilobase (kb): Unit of length for DNA fragments equal to 1000 nucleotides.

Knockout: Deactivation of specific genes; used in laboratory organisms to study gene function. *See also:* gene, locus, model organisms

L

Lesion: an injury or wound.

Library: An unordered collection of clones (i.e., cloned DNA from a particular organism) whose relationship to each other can be established by physical mapping. *See also:* genomic library, arrayed library

Linkage: The proximity of two or more markers (e.g., genes, RFLP markers) on a chromosome; the closer the markers, the lower the probability that they will be separated during DNA repair or replication processes (binary fission in prokaryotes, mitosis or meiosis in eukaryotes), and hence the greater the probability that they will be inherited together.

Linkage disequilibrium: Where alleles occur together more often than can be accounted for by chance. Indicates that the two alleles are physically close on the DNA strand. *See also:* Mendelian inheritance

Linkage map: A map of the relative positions of genetic loci on a chromosome, determined on the basis of how often the loci are inherited together. Distance is measured in centimorgans (cM).

Lipid: a fatty substance in the blood.

Lipoproteins: transporters of fatty substances in the blood.

Localize: Determination of the original position (locus) of a gene or other marker on a chromosome.

Locus (pl. loci): The position on a chromosome of a gene or other chromosome marker; also, the DNA at that position. The use of locus is sometimes restricted to mean expressed DNA regions. *See also:* gene expression

Long-Range Restriction Mapping: Restriction enzymes are proteins that cut DNA at precise locations. Restriction maps depict the chromosomal positions of restriction-enzyme cutting sites. These are used as biochemical "signposts," or markers of specific areas along the chromosomes. The map will detail the positions where the DNA molecule is cut by particular restriction enzymes.

Low density lipoprotein (LDL): the primary cholesterol carrying.

Lumen: the hollow area within a tube.

M

Macrorestriction map: Map depicting the order of and distance between sites at which restriction enzymes cleave chromosomes.

Magnetic resonance imaging (MRI): a non-nuclear imaging procedure much like an x-ray.

Mapping: *See:* gene mapping, linkage map, physical map

Mapping population: The group of related organisms used in constructing a genetic map.

Marker: *See:* genetic marker

Mass spectrometry: An instrument used to identify chemicals in a substance by their mass and charge.

Megabase (Mb): Unit of length for DNA fragments equal to 1 million nucleotides and roughly equal to 1 cM. *See also:* centimorgan

Meiosis The process of two consecutive cell divisions in the diploid progenitors of sex cells. Meiosis results in four rather than two daughter cells, each with a haploid set of chromosomes. *See also:* mitosis

Mendelian inheritance: One method in which genetic traits are passed from parents to offspring. Named for Gregor Mendel, who first studied and recognized the existence of genes and this method of inheritance. *See also:* autosomal dominant, recessive gene, sex-linked

Messenger RNA (mRNA): RNA that serves as a template for protein synthesis. *See also:* genetic code

Metaphase: A stage in mitosis or meiosis during which the chromosomes are aligned along the equatorial plane of the cell.

Microarray: Sets of miniaturized chemical reaction areas that may also be used to test DNA fragments, antibodies, or proteins.

Microbial genetics: The study of genes and gene function in bacteria, archaea, and other microorganisms. Often used in research in the fields of bioremediation, alternative energy, and disease prevention. *See also:* model organisms, biotechnology, bioremediation

Microinjection: A technique for introducing a solution of DNA into a cell using a fine microcapillary pipet.

Micronuclei: Chromosome fragments that are not incorporated into the nucleus at cell division.

Mitochondrial DNA: The genetic material found in mitochondria, the organelles that generate energy for the cell. Not inherited in the same fashion as nucleic DNA. *See also:* cell, DNA, genome, nucleus

Mitosis: The process of nuclear division in cells that produces daughter cells that are genetically identical to each other and to the parent cell. *See also:* meiosis

Mitral valve: the valve that controls blood flow between the left atrium and left ventricle in the heart.

Mitral valve prolapse: a bulge in the valve between the left atrium and left ventricle of the heart that causes backward flow of blood into the atrium.

Model organisms: A laboratory animal or other organism useful for research.

Modeling: The use of statistical analysis, computer analysis, or model organisms to predict outcomes of research.

Molecular biology: The study of the structure, function, and makeup of biologically important molecules.

Molecular farming: The development of transgenic animals to produce human proteins for medical use.

Molecular genetics: The study of macromolecules important in biological inheritance.

Molecular medicine: The treatment of injury or disease at the molecular level. Examples include the use of DNA-based diagnostic tests or medicine derived from DNA sequence information.

Monogenic disorder: A disorder caused by mutation of a single gene. *See also:* mutation, polygenic disorder

Monogenic inheritance: *See:* monogenic disorder

Monounsaturated fats: dietary fats, such as olive oil or canola oil, that don't seem to have any affect on blood cholesterol.

Monosomy: Possessing only one copy of a particular chromosome instead of the normal two copies. *See also:* cell, chromosome, gene expression, trisomy

Morbid map: A diagram showing the chromosomal location of genes associated with disease.

Mouse model: *See:* model organisms

Multifactorial or multigenic disorder: *See:* polygenic disorder

Multiplexing: A laboratory approach that performs multiple sets of reactions in parallel (simultaneously); greatly increasing speed and throughput.

Murine:Organism in the genus Mus. A rat or mouse.

Murmur: a blowing or rasping sound heard while listening to the heart that may or may not indicate problems within the heart or circulatory system.

Mutagen: An agent that causes a permanent genetic change in a cell. Does not include changes occurring during normal genetic recombination.

Mutagenicity: The capacity of a chemical or physical agent to cause permanent genetic alterations. *See also:* somatic cell genetic mutation

Mutation: Any heritable change in DNA sequence. *See also:* polymorphism

Myocardial infarction (heart attack): damage to or death of heart muscle tissue due to insufficient blood supply.

Myocardial ischemia: insufficient blood flow to part of the heart.

Myocardium: the muscle wall of the heart.

N

Necrosis: pertaining to the death of tissue.

Nitrogenous base: A nitrogen-containing molecule having the chemical properties of a base. DNA contains the nitrogenous bases adenine (A), guanine (G), cytosine (C), and thymine (T). *See also:* DNA

Nitroglycerin: a medication used to relax or dilate arteries.

Noninvasive procedure: a diagnostic effort or treatment that does not require entering the body or puncturing the skin.

Northern blot: A gel-based laboratory procedure that locates mRNA sequences on a gel that are complementary to a piece of DNA used as a probe. *See also:* DNA, library

Nuclear transfer: A laboratory procedure in which a cell's nucleus is removed and placed into an oocyte with its own nucleus removed so the genetic information from the donor nucleus controls the resulting cell. Such cells can be induced to form embryos. This process was used to create the cloned sheep "Dolly". *See also:* cloning

Nucleic acid: A large molecule composed of nucleotide subunits. *See also:* DNA

Nucleolar organizing region: A part of the chromosome containing rRNA genes.

Nucleotide: A subunit of DNA or RNA consisting of a nitrogenous base (adenine, guanine, thymine, or cytosine in DNA; adenine, guanine, uracil, or cytosine in RNA), a phosphate molecule, and a sugar molecule (deoxyribose in DNA and ribose in RNA). Thousands of nucleotides are linked to form a DNA or RNA molecule. *See also:* DNA, base pair, RNA

Nucleus: The cellular organelle in eukaryotes that contains most of the genetic material.

O

Obesity: overweight by 30 percent of the ideal body weight.

Occluded artery: an artery that is narrowed by plaque that impedes blood flow.

Oligo: *See:* oligonucleotide

Oligogenic: A phenotypic trait produced by two or more genes working together. *See also:* polygenic disorder

Oligonucleotide: A molecule usually composed of 25 or fewer nucleotides; used as a DNA synthesis primer. *See also:* nucleotide

Oncogene: A gene, one or more forms of which is associated with cancer. Many oncogenes are involved, directly or indirectly, in controlling the rate of cell growth.

Open heart surgery: surgery that involves opening the chest and heart while a heart-lung machine pumps blood for the heart.

Open reading frame (ORF): The sequence of DNA or RNA located between the start-code sequence (initiation codon) and the stop-code sequence (termination codon).

Operon: A set of genes transcribed under the control of an operator gene.

Overlapping clones: *See:* genomic library

P

P1-derived artificial chromosome (PAC): One type of vector used to clone DNA fragments (100- to 300-kb insert size; average, 150 kb) in *Escherichia coli* cells. Based on bacteriophage (a virus) P1 genome. *See also:* cloning vector

Pacemaker: an electronic device that is surgically implanted into the patient's heart and chest to regulate heartbeat.

Palpitation: irregular heartbeat.

Patent: In genetics, conferring the right or title to genes, gene variations, or identifiable portions of sequenced genetic material to an individual or organization. *See also:* gene

Pedigree: A family tree diagram that shows how a particular genetic trait or disease has been inherited. *See also:* inherit

Penetrance: The probability of a gene or genetic trait being expressed. "Complete" penetrance means the gene or genes for a trait are expressed in all the population who have the genes. "Incomplete" penetrance means the genetic trait is expressed in only part of the population. The percent penetrance also may change with the age range of the population.

Peptide: Two or more amino acids joined by a bond called a "peptide bond." *See also:* polypeptide

Percutaneous transluminal coronary angioplasty (PTCA): angioplasty.

Pericarditis: inflammation of the membrane that surrounds the heart.

Pericardiocentesis: a diagnostic procedure in which a needle is used to draw fluid from the pericardium.

Pericardium: the membrane that surrounds the heart.

Plaque: deposits of fat or other substances attached to the artery wall.

Phage: A virus for which the natural host is a bacterial cell.

Pharmacogenomics: The study of the interaction of an individual's genetic makeup and response to a drug.

Phenocopy: A trait not caused by inheritance of a gene but appears to be identical to a genetic trait.

Phenotype: The physical characteristics of an organism or the presence of a disease that may or may not be genetic. *See also:* genotype

Physical map: A map of the locations of identifiable landmarks on DNA (e.g., restriction-enzyme cutting sites, genes), regardless of inheritance. Distance is measured in base pairs. For the human genome, the lowest-resolution physical map is the banding patterns on the 24 different chromosomes; the highest-resolution map is the complete nucleotide sequence of the chromosomes.

Plasmid: Autonomously replicating extra-chromosomal circular DNA molecules, distinct from the normal bacterial genome and nonessential for cell survival under nonselective conditions. Some plasmids are capable of integrating into the host genome. A number of artificially constructed plasmids are used as cloning vectors.

Platelets: cells found in the blood.

Pleiotropy: One gene that causes many different physical traits such as multiple disease symptoms.

Pluripotency: The potential of a cell to develop into more than one type of mature cell, depending on environment.

Polygenic disorder: Genetic disorder resulting from the combined action of alleles of more than one gene (e.g., heart disease, diabetes, and some cancers). Although such disorders are inherited, they depend on the simultaneous presence of several alleles; thus the hereditary patterns usually are more complex than those of single-gene disorders. *See also:* single-gene disorder

Polymerase chain reaction (PCR): A method for amplifying a DNA base sequence using a heat-stable polymerase and two 20-base primers, one complementary to the (+)

strand at one end of the sequence to be amplified and one complementary to the (-) strand at the other end. Because the newly synthesized DNA strands can subsequently serve as additional templates for the same primer sequences, successive rounds of primer annealing, strand elongation, and dissociation produce rapid and highly specific amplification of the desired sequence. PCR also can be used to detect the existence of the defined sequence in a DNA sample.

Polymerase, DNA or RNA: Enzyme that catalyzes the synthesis of nucleic acids on preexisting nucleic acid templates, assembling RNA from ribonucleotides or DNA from deoxyribonucleotides.

Polymorphism: Difference in DNA sequence among individuals that may underlie differences in health. Genetic variations occurring in more than 1% of a population would be considered useful polymorphisms for genetic linkage analysis. *See also:* mutation

Polypeptide: A protein or part of a protein made of a chain of amino acids joined by a peptide bond.

Polyunsaturated fat: a type of fat found in vegetable oils and margarines that doesn't appear to raise blood cholesterol levels.

Population genetics: The study of variation in genes among a group of individuals.

Positional cloning: A technique used to identify genes, usually those that are associated with diseases, based on their location on a chromosome.

Positron emission tomography (PET): a nuclear scanning device that gives a three-dimensional picture of the heart to provide information about the flow of blood through the coronary arteries to the heart muscle.

Premature chromosome condensation (PCC): A method of studying chromosomes in the interphase stage of the cell cycle.

Primer: Short preexisting polynucleotide chain to which new deoxyribonucleotides can be added by DNA polymerase.

Privacy: In genetics, the right of people to restrict access to their genetic information.

Probe: Single-stranded DNA or RNA molecules of specific base sequence, labeled either radioactively or immunologically, that are used to detect the complementary base sequence by hybridization.

Prokaryote: Cell or organism lacking a membrane-bound, structurally discrete nucleus and other subcellular compartments. Bacteria are examples of prokaryotes. *See also:* chromosome, eukaryote

Promoter: A DNA site to which RNA polymerase will bind and initiate transcription.

Pronucleus: The nucleus of a sperm or egg prior to fertilization. *See also:* nucleus, transgenic

Protein: A large molecule composed of one or more chains of amino acids in a specific order; the order is determined by the base sequence of nucleotides in the gene that codes for the protein. Proteins are required for the structure, function, and regulation of the body's cells, tissues, and organs; and each protein has unique functions. Examples are hormones, enzymes, and antibodies.

Proteome: Proteins expressed by a cell or organ at a particular time and under specific conditions.

Proteomics: The study of the full set of proteins encoded by a genome.

Pseudogene: A sequence of DNA similar to a gene but nonfunctional; probably the remnant of a once-functional gene that accumulated mutations.

Pulmonary: pertains to lungs and respiratory system.

Pulmonary edema: a condition in which there is a fluid accumulation in the lungs caused by an incorrectly functioning heart.

Pulmonary valve: the heart valve located between the right ventricle and the pulmonary artery that controls blood flow to the lungs.

Pulmonary vein: the vessel that carries newly oxygenated blood to the heart from the lungs.

Pulse oximeter: a device that measures the amount of oxygen in the blood.

Purine: A nitrogen-containing, double-ring, basic compound that occurs in nucleic acids. The purines in DNA and RNA are adenine and guanine. *See also:* base pair

Pyrimidine: A nitrogen-containing, single-ring, basic compound that occurs in nucleic acids. The pyrimidines in DNA are cytosine and thymine; in RNA, cytosine and uracil. *See also:* base pair

R

Radiation hybrid: A hybrid cell containing small fragments of irradiated human chromosomes. Maps of irradiation sites on chromosomes for the human, rat, mouse, and other genomes provide important markers, allowing the construction of very precise STS maps indispensable to studying multifactorial diseases. *See also:* sequence tagged site

Radioisotope: a radioactive material injected into the body so that a nuclear scanner can make pictures.

Radionuclide ventriculography: a diagnostic procedure used to determine the shape and size of the heart's chambers.

Rare-cutter enzyme: *See:* restriction-enzyme cutting site

Recessive gene: A gene which will be expressed only if there are 2 identical copies or, for a male, if one copy is present on the X chromosome.

Reciprocal translocation: When a pair of chromosomes exchange exactly the same length and area of DNA. Results in a shuffling of genes.

Recombinant clone: Clone containing recombinant DNA molecules. *See also:* recombinant DNA technology

Recombinant DNA molecules: A combination of DNA molecules of different origin that are joined using recombinant DNA technologies.

Recombinant DNA technology: Procedure used to join together DNA segments in a cell-free system (an environment outside a cell or organism). Under appropriate conditions, a recombinant DNA molecule can enter a cell and replicate there, either autonomously or after it has become integrated into a cellular chromosome.

Recombination: The process by which progeny derive a combination of genes different from that of either parent. In higher organisms, this can occur by crossing over. *See also:* crossing over, mutation

Regulatory region or sequence: A DNA base sequence that controls gene expression.

Regurgitation: backward flow of blood caused by a defective heart valve.

Renal: pertains to kidneys.

Repetitive DNA: Sequences of varying lengths that occur in multiple copies in the genome; it represents much of the human genome.

Reporter gene:*See:* marker

Resolution: Degree of molecular detail on a physical map of DNA, ranging from low to high.

Restriction enzyme, endonuclease: A protein that recognizes specific, short nucleotide sequences and cuts DNA at those sites. Bacteria contain over 400 such enzymes that recognize and cut more than 100 different DNA sequences. *See also:* restriction enzyme cutting site

Restriction fragment length polymorphism (RFLP): Variation between individuals in DNA fragment sizes cut by specific restriction enzymes; polymorphic sequences that result in RFLPs are used as markers on both physical maps and genetic linkage maps. RFLPs usually are caused by mutation at a cutting site. *See also:* marker, polymorphism

Restriction-enzyme cutting site A specific nucleotide sequence of DNA at which a particular restriction enzyme cuts the DNA. Some sites occur frequently in DNA (e.g., every several hundred base pairs); others much less frequently (rare-cutter; e.g., every 10,000 base pairs).

Retroviral infection:The presence of retroviral vectors, such as some viruses, which use their recombinant DNA to insert their genetic material into the chromosomes of the host's cells. The virus is then propogated by the host cell.

Reverse transcriptase: An enzyme used by retroviruses to form a complementary DNA sequence (cDNA) from their RNA. The resulting DNA is then inserted into the chromosome of the host cell.

Rheumatic fever: a childhood disease that may damage the heart valves or the outer lining of the heart.

Ribonucleotide: *See:* nucleotide

Ribose The five-carbon sugar that serves as a component of RNA. *See also:* ribonucleic acid, deoxyribose

Ribosomal RNA (rRNA): A class of RNA found in the ribosomes of cells.

Ribosomes: Small cellular components composed of specialized ribosomal RNA and protein; site of protein synthesis. *See also:* RNA

Risk communication: In genetics, a process in which a genetic counselor or other medical professional interprets genetic test results and advises patients of the consequences for them and their offspring.

Risk factor: a condition, element, or activity that may adversely affect the heart.

RNA (Ribonucleic acid): A chemical found in the nucleus and cytoplasm of cells; it plays an important role in protein synthesis and other chemical activities of the cell. The structure of RNA is similar to that of DNA. There are several classes of RNA molecules, including messenger RNA, transfer RNA, ribosomal RNA, and other small RNAs, each serving a different purpose.

S

Sanger sequencing: A widely used method of determining the order of bases in DNA. *See also:* sequencing, shotgun sequencing

Satellite: A chromosomal segment that branches off from the rest of the chromosome but is still connected by a thin filament or stalk.

Saturated fat: fat that is found in foods from animal meats and skin, dairy products and some vegetables.

Scaffold: In genomic mapping, a series of contigs that are in the right order but not necessarily connected in one continuous stretch of sequence.

Segregation: The normal biological process whereby the two pieces of a chromosome pair are separated during meiosis and randomly distributed to the germ cells.

Sequence: *See:* base sequence

Sequence assembly: A process whereby the order of multiple sequenced DNA fragments is determined.

Sequence tagged site (STS): Short (200 to 500 base pairs) DNA sequence that has a single occurrence in the human genome and whose location and base sequence are known. Detectable by polymerase chain reaction, STSs are useful for localizing and orienting the mapping and sequence data reported from many different laboratories and serve as landmarks on the developing physical map of the human genome. Expressed sequence tags (ESTs) are STSs derived from cDNAs.

Sequencing: Determination of the order of nucleotides (base sequences) in a DNA or RNA molecule or the order of amino acids in a protein.

Sequencing technology: The instrumentation and procedures used to determine the order of nucleotides in DNA.

Septal defect: a hole in the wall of the heart.

Septum: the muscle wall that divides the heart chambers.

Sex chromosome:The X or Y chromosome in human beings that determines the sex of an individual. Females have two X chromosomes in diploid cells; males have an X and a Y chromosome. The sex chromosomes comprise the 23rd chromosome pair in a karyotype. *See also:* autosome

Sex-linked: Traits or diseases associated with the X or Y chromosome; generally seen in males. *See also:* gene, mutation, sex chromosome

Shock: impaired body function due to blood loss or a disturbance in the circulatory system.

Shotgun method: Sequencing method that involves randomly sequenced cloned pieces of the genome, with no foreknowledge of where the piece originally came from. This can be contrasted with "directed" strategies, in which pieces of DNA from known

chromosomal locations are sequenced. Because there are advantages to both strategies, researchers use both random (or shotgun) and directed strategies in combination to sequence the human genome. *See also:* library, genomic library

Shunt: a connector to allow blood flow between two locations.

Silent ischemia: ischemia not accompanied by chest pain.

Single nucleotide polymorphism (SNP): DNA sequence variations that occur when a single nucleotide (A, T, C, or G) in the genome sequence is altered. *See also:* mutation, polymorphism, single-gene disorder

Single-gene disorder:Hereditary disorder caused by a mutant allele of a single gene (e.g., Duchenne muscular dystrophy, retinoblastoma, sickle cell disease). *See also:* polygenic disorders

Sinus node: the cells that produce the electrical impulses that cause the heart to contract.

Somatic cell: Any cell in the body except gametes and their precursors. *See also:* gamete

Somatic cell gene therapy: Incorporating new genetic material into cells for therapeutic purposes. The new genetic material cannot be passed to offspring. *See also:* gene therapy

Somatic cell genetic mutation: A change in the genetic structure that is neither inherited nor passed to offspring. Also called acquired mutations. *See also:* germ line genetic mutation

Southern blotting: Transfer by absorption of DNA fragments separated in electrophoretic gels to membrane filters for detection of specific base sequences by radio-labeled complementary probes.

Spectral karyotype (SKY): A graphic of all an organism's chromosomes, each labeled with a different color. Useful for identifying chromosomal abnormalities. *See also:* chromosome

Sphygmomanometer: the instrument used to measure blood pressure.

Splice site: Location in the DNA sequence where RNA removes the noncoding areas to form a continuous gene transcript for translation into a protein.

Sporadic cancer: Cancer that occurs randomly and is not inherited from parents. Caused by DNA changes in one cell that grows and divides, spreading throughout the body. *See also:* hereditary cancer

Stem cell: Undifferentiated, primitive cells in the bone marrow that have the ability both to multiply and to differentiate into specific blood cells.

Stent: a device implanted in a vessel used to help keep it open.

Stenosis: the narrowing or constriction of a blood vessel or valve in the heart.

Stethoscope: the instrument used to listen to the heart and other sounds in the body.

Streptokinase: a clot-dissolving medication.

Sternum: the breastbone.

Stress: mental or physical tension that results from physical, emotional, or chemical causes.

Stroke: the sudden disruption of blood flow to the brain.

Structural genomics: The effort to determine the 3D structures of large numbers of proteins using both experimental techniques and computer simulation

Subarachnoid hemorrhage: bleeding on the surface of the brain.

Substitution: In genetics, a type of mutation due to replacement of one nucleotide in a DNA sequence by another nucleotide or replacement of one amino acid in a protein by another amino acid. *See also:* mutation

Sudden death: death that occurs unexpectedly or immediately after onset of symptoms.

Superior vena cava: the large vein that returns blood to the heart from the head and arms.

Suppressor gene: A gene that can suppress the action of another gene.

Syndrome: The group or recognizable pattern of symptoms or abnormalities that indicate a particular trait or disease.

Syngeneic: Genetically identical members of the same species.

Syncope: light-headedness or fainting caused by insufficient blood supply to the brain.

Synteny: Genes occurring in the same order on chromosomes of different species. *See also:* linkage, conserved sequence

Systolic blood pressure: the highest blood pressure measured in the arteries.

T

Tachycardia: rapid heart beat.

Tachypnea: rapid breathing.

Tandem repeat sequences:Multiple copies of the same base sequence on a chromosome; used as markers in physical mapping. *See also:* physical map

Targeted mutagenesis: Deliberate change in the genetic structure directed at a specific site on the chromosome. Used in research to determine the targeted region's function. *See also:* mutation, polymorphism

Technology transfer: The process of transferring scientific findings from research laboratories to the commercial sector.

Telemetry unit: a small transmitter with wires that attach ECG patches to the chest that is used to send information about the heart via radio transmission to healthcare professionals for evaluation.

Telomerase: The enzyme that directs the replication of telomeres.

Telomere: The end of a chromosome. This specialized structure is involved in the replication and stability of linear DNA molecules. *See also:* DNA replication

Teratogenic: Substances such as chemicals or radiation that cause abnormal development of a embryo. *See also:* mutatgen

Thallium stress test: a study in which radioactive potassium is carried by the blood and the progress is followed by x-ray pictures.

Thrombolysis: the breaking up of a blood clot.

Thrombosis: a blood clot formed in the blood vessel or in the heart.

Thrombolytic therapy: a medication that dissolves blood clots.

Thymine (T): A nitrogenous base, one member of the base pair AT (adenine-thymine). *See also:* base pair, nucleotide

Tissue plasminogen activator (TPA): a medication used to dissolves blood clots.

Toxicogenomics: The study of how genomes respond to environmental stressors or toxicants. Combines genome-wide mRNA expression profiling with protein expression patterns using bioinformatics to understand the role of gene-environment interactions in disease and dysfunction.

Trans fat: vegetable oil that has been treated with hydrogen in order to make it more solid and give it a longer shelf life.

Transcription: The synthesis of an RNA copy from a sequence of DNA (a gene); the first step in gene expression. *See also:* translation

Transcription factor: A protein that binds to regulatory regions and helps control gene expression.

Transcriptome: The full complement of activated genes, mRNAs, or transcripts in a particular tissue at a particular time

Transesophageal echocardiography (TEE): a diagnostic test that is used to measure the sound waves that bounce off of the heart.

Transfection: The introduction of foreign DNA into a host cell. *See also:* cloning vector, gene therapy

Transfer RNA (tRNA): A class of RNA having structures with triplet nucleotide sequences that are complementary to the triplet nucleotide coding sequences of mRNA. The role of tRNAs in protein synthesis is to bond with amino acids and transfer them to the ribosomes, where proteins are assembled according to the genetic code carried by mRNA.

Transformation: A process by which the genetic material carried by an individual cell is altered by incorporation of exogenous DNA into its genome.

Transgenic: An experimentally produced organism in which DNA has been artificially introduced and incorporated into the organism's germ line. *See also:* cell, DNA, gene, nucleus, germ line

Transient ischemic attack (TIA): a stroke-like event that lasts for a short period of time and is caused by a blocked blood vessel.

Translation: The process in which the genetic code carried by mRNA directs the synthesis of proteins from amino acids. *See also:* transcription

Translocation: A mutation in which a large segment of one chromosome breaks off and attaches to another chromosome. *See also:* mutation

Transposable element: A class of DNA sequences that can move from one chromosomal site to another.

Tricuspid valve: the heart valve that controls blood flow from the right atrium into the right ventricle.

Triglyceride: a fat-like substance found in the blood.

Trisomy: Possessing three copies of a particular chromosome instead of the normal two copies. *See also:* cell, gene, gene expression, chromosome

U

Ultrasound: a diagnostic tool used to get a picture of the heart or other structures within the body.

Uracil: A nitrogenous base normally found in RNA but not DNA; uracil is capable of forming a base pair with adenine. *See also:* base pair, nucleotide

V

Valves: (the heart valves are tricuspid, pulmonic, mitral, and aortic): the "doors" between the chambers of the heart.

Valvuloplasty: repairing a heart valve.

Varicose vein: an abnormally dilated vein.

Vascular: pertaining to blood vessels.

Vasodilator: a medication that dilates or widens the opening in a blood vessel.

Vasodepressors: a medication that raises blood pressure.

Vector: *See:* cloning vector

Vein: a blood vessel that carries blood from the body back into the heart.

Ventricle: one of the two lower chambers of the heart.

Ventricular fibrillation: a condition in which the ventricles contract in rapid and unsynchronized rhythms and cannot pump blood into the body.

Ventricular tachycardia: a condition in which the ventricles cause a very fast heartbeat.

Vertigo: dizziness.

Virus: A noncellular biological entity that can reproduce only within a host cell. Viruses consist of nucleic acid covered by protein; some animal viruses are also surrounded by membrane. Inside the infected cell, the virus uses the synthetic capability of the host to produce progeny virus. *See also:* cloning vector

W

Western blot: A technique used to identify and locate proteins based on their ability to bind to specific antibodies. *See also:* DNA, Northern blot, protein, RNA, Southern blotting

Wild type: The form of an organism that occurs most frequently in nature.

Working Draft DNA Sequence: *See:* Draft DNA Sequence

X

X chromosome: One of the two sex chromosomes, X and Y. *See also:* Y chromosome, sex chromosome

Xenograft: Tissue or organs from an individual of one species transplanted into or grafted onto an organism of another species, genus, or family. A common example is the use of pig heart valves in humans.

X-ray: a machine that used radiation to produce pictures of the inside of the body.

Y

Y chromosome: One of the two sex chromosomes, X and Y. *See also:* X chromosome, sex chromosome

Yeast artificial chromosome (YAC): Constructed from yeast DNA, it is a vector used to clone large DNA fragments. *See also:* cloning vecter, cosmid

Z

Zinc-finger protein: A secondary feature of some proteins containing a zinc atom; a DNA-binding protein.