GLOSSARY

allele: One of two or more alternative forms of a gene.

amplify: To make many copies of a specific region of DNA.

angina: Chest pain caused by reduced blood flow to the heart, often from narrowed, nearby arteries due to the buildup of plaque. See also: unstable angina.

anneal: To attach, for example, as when primers form hydrogen bonds with single-stranded template DNA in PCR.

antiplatelet therapy: Medication that reduces the blood's ability to clot in order to prevent dangerous clots after a medical or surgical procedure.

bacteriophage: A virus that infects bacteria by injecting its DNA into the cell and commandeering the host cell's molecular processes to make more bacteriophages.

base-calling software: Computer code used to analyze the output of automated DNA sequencing. It compiles laser excitation output from fluorescent nucleotides in DNA fragments, sorts the fragments into the correct order, and generates an output called a DNA chromatogram. See also: DNA chromatogram, Sanger sequencing.

capillary electrophoresis sequencing: A form of Sanger sequencing; nucleic acid fragments are loaded into a polymer-filled capillary tube to which an electric field is applied for size separation. See also: Sanger sequencing.

chelate: To bind chemically.

cofactors: Metallic ions required for the catalytic activity of enzymes such as DNases.

confidence interval: A measurement of confidence in statistics. In the case of DNA sequencing, it is an estimate of the probability that a particular computer-assigned nucleotide is actually the nucleotide in the DNA sequence.

cytochrome P450: A member of a class of enzymes active in the endoplasmic reticulum, where they metabolize substances such as medications.

DNA chromatogram: A computer-generated chart of a DNA sequence that shows the nucleotides in each position and the level of fluorescence emitted by each nucleotide in the sequence.

DNA polymerase: The enzyme that builds new DNA molecules by connecting nucleotides.

DNA replication: The process by which organisms assemble new strands of DNA from existing templates using free nucleotides and the enzyme DNA polymerase.

DNases: Enzymes that break down DNA.

enzyme: A protein that catalyzes a chemical reaction without being altered itself.

exon: A segment of DNA that encodes a protein. Exons are spliced together to make mRNA.

gel electrophoresis: A laboratory technique for separating nucleic acids or proteins based on their relative size by applying an electric current that pulls materials through a gel in a buffer solution.

gene cloning: Using reagents derived from living organisms, such as restriction enzymes and DNA polymerase, to isolate and make copies of specific genes.

gene product: The protein that results from the expression of a gene, such as a human therapeutic protein like insulin.

genotype: The genetic makeup of an individual.

germline DNA: The haploid genome of gametes (sperm or eggs) generated by meiosis.

Human Genome Project: A large, collaborative, international project that generated the first full sequence of the human genome. This used Sanger sequencing; see also Sanger sequencing.

in vitro: A laboratory technique which occurs outside a living organism.

in vivo: A laboratory technique which occurs inside a living organism.

lysis: Breaking down a cell's plasma and nuclear membranes using heat or chemical means.

mutagenesis: The process of permanent changes to DNA due to external agents.

nucleotide: The basic building block of DNA.

percutaneous coronary intervention: (PCI): Also known as "angioplasty," a procedure to remove blockages in coronary arteries using X-ray-guided flexible catheters. May involve the placement of stents.

pharmacogenomics: The science of understanding how individuals' genotypes influence their response to medications.

phenotype: The set of observable characteristics of an individual based on how their genotype is expressed; see genotype.

phenylthiocarbamide (PTC): A chemical compound which some individuals perceive as bitter, while others do not taste it at all, depending on their genetics. It is commonly used to study inherited taste traits.

plasmids: Circles of DNA found in bacteria and other microorganisms that are separate from chromosomal DNA and can replicate independently.

polymerase chain reaction (PCR): A technique to make many copies of a portion of DNA ranging from as small as 200 base pairs to as large as 40 kilobases!

precision medicine: An approach to the prevention and treatment of disease that considers individual variability in genes, environment, and lifestyle.

promoter: A region of DNA upstream from a gene where proteins bind to initiate transcription of the gene.

restriction enzyme: A species-specific bacterial protein that restricts the growth of the harmful viruses known as bacteriophages by recognizing and destroying the phage DNA without damaging the host (bacterial) DNA. Each bacterial species cuts DNA at a different sequence of nucleotides (see also restriction sites).

restriction enzyme digest: A laboratory technique using specialized bacterial proteins which cut double-stranded DNA at particular sites for diagnostic and gene cloning purposes (see also restriction enzymes and restriction sites).

restriction sites: Short sequences of DNA that bind specific bacterial restriction enzymes, allowing the enzymes to cut the DNA.

running buffer: A solution of electrolytes capable of forming ions and maintaining a stable pH; used to facilitate gel electrophoresis.

Sanger sequencing: An early method of DNA sequencing developed by Frederick Sanger and his colleagues.

sequencing: The process of determining the order of nucleotides in a nucleic acid sample.

single nucleotide polymorphism (SNP): A variation at a single position in individuals' DNA sequences. This can occur either in coding or non-coding DNA. If it occurs within coding DNA in a gene, each SNP represents a different allele. SNPs occur with greater than 1% frequency in populations.

single nucleotide variant (SNV): Single nucleotide variation in individuals' DNA sequences (see also SNP), but that occurs with less than 1% frequency in populations.

somatic cells: Body cells, excluding reproductive cells.

stent: A miniature, expandable flexible wire "cage" used to hold open a major blood vessel.

***Taq* polymerase:** A DNA-replicating enzyme from the thermophilic bacterium *Thermus aquaticus* that can copy its DNA at high temperatures. This enzyme is used in PCR, where DNA must be denatured at high temperatures.

template DNA: The sample DNA that contains the target sequence.

terminus: The end of a chain of nucleotides.

thermocycler: A laboratory instrument that controls the temperature and length of time for different phases of a reaction.

thermophile: A heat-loving microorganism.

trait: A characteristic of living organisms that can be described, quantified, or measured, and that is due to the influence of genes, the environment, or both.

transformation: The process of introducing a plasmid to bacteria.

unstable angina: Sudden, severe chest pain brought on by a temporary blockage of a coronary artery which limits blood flow to the heart.

wild-type: The most prevalent form of a gene in a species, as found in nature.