GLOSSARY

aliquot: To measure out, then dispense, an equal portion of a solution or material from a whole.

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. *This term appears in the Student Guide.* See also: unstable angina.

angioplasty: A procedure to remove blockages and/or insert stents in coronary arteries using X-ray-guided flexible catheters.

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. *This term appears in the Student Guide.*

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. *This term appears in the Student Guide.*

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. *This term appears in the Student Guide.*

chelate: To bind chemically.

cofactors: Metallic ions required for the catalytic activity of enzymes such as DNases. *This term appears in the Student Guide.*

complete dominance: A phenotype, resulting from a heterozygous genotype, in which a dominant protein haplotype completely masks the variant function of the recessive haplotype.

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. *This term appears in the Student Guide.*

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

denaturation (in PCR): Heating double-stranded template DNA to break hydrogen bonds so that it separates into two single strands**.**

diplotype: A pair of haplotypes, one from each parent.

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.

DNA sequencing: The process of determining the nucleotide sequence of a particular segment of DNA using molecular biological techniques. See also: Sanger sequencing, capillary electrophoresis sequencing.

DNases: Enzymes that break down DNA.

exon: A segment of DNA that encodes a protein. Exons are spliced together to make mRNA. *This term appears in the Student Guide.*

extension: The stage of PCR during which heating allows new strands of DNA to be made by the enzyme *Taq* polymerase.

gametes: Sperm and eggs. See also: germ cells.

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: The basic unit of heredity, consisting of a specific sequence of nucleotides that can be transcribed to produce a functional RNA or translated into a protein.

gene product: The protein that results from the expression of a gene, such as a human therapeutic protein like insulin. *This term appears in the Student Guide.*

genotype: The genetic makeup of an individual.

germ cells: Precursor reproductive cells that undergo meiosis to produce mature sperm and eggs (gametes).

germline DNA: The haploid genome of gametes (sperm or eggs) generated by meiosis. *This term appears in the Student Guide.*

haplotype: An allele on one chromosome of a homologous pair.

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. *This term appears in the Student Guide.*

incomplete dominance: A phenotype, resulting from a heterozygous genotype, in which two protein haplotypes blend to produce an intermediate form of protein expression.

***in vitro*:** A laboratory technique that occurs outside a living organism. *This term appears in the Student Guide.*

***in vivo*:** A laboratory technique that occurs inside a living organism. *This term appears in the Student Guide.*

lysis: The process of breaking down a cell's plasma and nuclear membranes using heat or chemical means. *This term appears in the Student Guide.*

mutagenesis: The process of permanent changes to DNA due to external agents. *This term appears in the Student Guide.*

nucleotide: The basic building block of DNA. *This term appears in the Student Guide.*

PCR-RFLP: The combination of PCR with diagnostic restriction enzyme digestion and gel electrophoresis to identify single nucleotide polymorphisms. RFLP stands for restriction fragment length polymorphism, the variation in DNA fragment length that results when variations in genomic DNA alter restriction enzyme recognition sites.

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. *This term appears in the Student Guide.*

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!

polymorphism: A difference in sequence between homologous strands of DNA from different individuals.

PCR-RFLP analysis: A molecular biology technique combining PCR amplification of a genetically variable portion of a target gene with restriction enzyme digestion and gel electrophoresis to determine individual genotypes. "RFLP" stands for **R**estriction **F**ragment **L**ength **P**olymorphism.

primers: Short fragments of single-stranded DNA designed specifically to target desired regions of DNA for amplification by polymerase chain reaction.

primer dimers: Artifacts of PCR where primers anneal to each other, creating nonspecific products. These may be visible even in the absence of a DNA template, such as in a reaction control.

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. *This term appears in the Student Guide.*

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 (see the definition for bacteriophages in this glossary). Each bacterial species cuts DNA at a different sequence of nucleotides (see also restriction sites).

restriction enzyme digestion: A laboratory technique using specialized bacterial proteins that 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. *This term appears in the Student Guide.*

Sanger sequencing: An early method of DNA sequencing involving the synthesis and gel electrophoresis of labeled DNA fragments of varying lengths, developed by Frederick Sanger and his colleagues.

sequencing: The process of determining the order of nucleotides in a nucleic acid sample. *This term appears in the Student Guide.*

single nucleotide polymorphism (SNP): A variation at a single position in an individual's DNA sequence. 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): A single-nucleotide variation in an individual's DNA sequence (see also SNP) that occurs with less than 1% frequency in populations.

somatic: Non-reproductive parts of the body, including cells.

***Taq* polymerase:** A DNA-replicating enzyme from the thermophilic bacterium *Thermus aquaticus,* which is capable of copying its DNA at high temperatures. This enzyme is used in PCR, where DNA must be denatured at high temperatures. See also: PCR.

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

terminus: The end of a chain of nucleotides. *This term appears in the Student Guide.*

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

thermophile: A heat-loving microorganism. *This term appears in the Student Guide.*

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

transformation: The process of introducing a plasmid to bacteria. *This term appears in the Student Guide.*

umami: The so-called "5th taste," due to the amino acid glutamate. It is described as a meaty or savory taste.

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