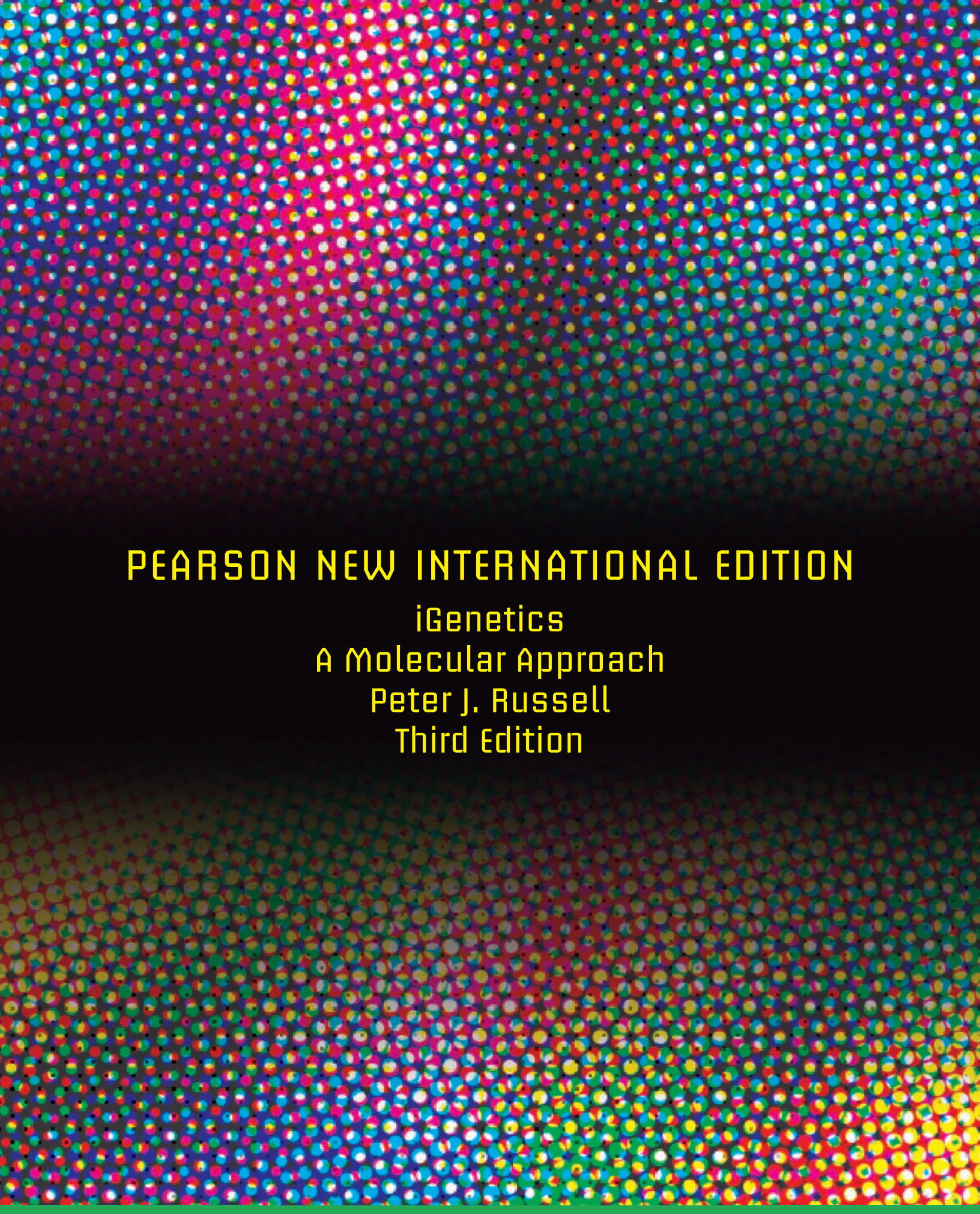

PEARSON NEW INTERNATIONAL EDITION

iGenetics
A Molecular Approach
Peter J. Russell
Third Edition

ALWAYS LEARNING

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Glossary

10-nm chromatin fiber The least compact form of **chromatin.** It is approximately 10 nm in diameter and has a "beads-on-a-string" morphology. It consists of **nucleosomes** which consist of a core of eight histone proteins around which is wrapped the DNA. Linker DNA bridges each nucleosome. *See also* **30-nm chromatin fiber.**

30-nm chromatin fiber The next level of **chromatin** condensation beyond the **10-nm chromatin** fiber brought about by H1 histone binding to the linker DNA and to DNA bound to the histones of the nucleosome. It is about 30 nm in diameter. *See also* **10-nm chromatin fiber.**

acrocentric chromosome A chromosome with the centromere near one end such that it has one long arm plus a stalk and a satellite.

activators The major class of transcription regulatory proteins in eukaryotes. Binding of these proteins to regulatory DNA sequences associated with specific genes determines the efficiency of transcription initiation. Some bacterial genes are controlled by activators. *See also* **repressor(s).**

adenine (A) A **purine** base found in DNA and RNA. In double-stranded DNA, adenine pairs with thymine, a **pyrimidine,** by hydrogen bonding. In double-stranded RNA, adenine pairs with uracil, a **pyrimidine,** by hydrogen bonding.

agarose gel electrophoresis An experimental procedure in which an electric field is used to move DNA or RNA molecules, which are negatively charged, through a gel matrix of agarose from the negative pole to the positive pole.

allele One of two or more alternative forms of a single gene that can exist at the same **locus** in the genome. All the alleles of a gene determine the same hereditary trait (e.g., seed color), but each has a unique nucleotide sequence, which may result in different phenotypes (e.g., yellow or green seeds). *See also* **DNA polymorphism.**

allele frequency Proportion of a particular allele at a locus within a gene pool. The sum of the allele frequencies at a given locus is 1.

allele-specific oligonucleotide (ASO) hybridization A procedure, using PCR primers, to distinguish alleles that differ by one base pair.

allelomorph *See* **allele.**

allopolyploidy Condition in which a cell or organism has two or more genetically distinct sets of chromosomes that originate in different, though usually related, species.

alternation of generations Type of life cycle characteristic of green plants in which haploid cells **(gametophytes)** alternate with diploid cells **(sporophytes).**

alternative polyadenylation Process for generating different functional mRNAs from a single gene by cleavage and polyadenylation of the primary transcript at different **poly(A) sites.**

alternative splicing In eukaryotes, a process for generating different functional mRNAs from a single precursor mRNA (pre-mRNA) by incorporating different exons in the mature mRNA.

Ames test An assay that measures the ability of chemicals to cause mutations in certain bacteria. It can identify potential carcinogens.

amino acid Any of the small molecules, containing a carboxyl group and amino group, that are joined together to form polypeptides and proteins.

aminoacyl–tRNA A tRNA molecule covalently bound to an amino acid; also called *charged tRNA*. This complex brings the amino acid to the ribosome so that it can be used in polypeptide synthesis.

aminoacyl–tRNA synthetase An enzyme that catalyzes the addition of a specific amino acid to the tRNA for that amino acid.

amniocentesis A procedure in which a sample of amniotic sac fluid is withdrawn from the amniotic sac of a developing fetus and cells are cultured and examined for chromosomal abnormalities.

analysis of variance (ANOVA) A series of statistical procedures for determining whether differences in the **means** of a variable in two samples are significant and for partitioning the **variance** into components.

anaphase The stage in mitosis when the **sister chromatids** separate and migrate toward the opposite poles of the cell.

anaphase I The stage in meiosis I when the chromosomes in each **bivalent** separate and begin moving toward opposite poles of the cell.

anaphase II The stage in meiosis II when the **sister chromatids** are pulled to the opposite poles of the cell.

aneuploid Referring to an organism or cell that has a chromosome number that is not an exact multiple of the haploid set of chromosomes.

aneuploidy Any condition in which the number of chromosomes differs from an exact multiple of the normal haploid number in a cell or organism. It commonly results from the gain or loss of individual chromosomes but also can result from the duplication or deletion of part(s) of a chromosome or chromosomes.

antibody A protein molecule that recognizes and binds to a foreign substance introduced into the organism.

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anticodon A group of three adjacent nucleotides in a tRNA molecule that pairs with a **codon** in mRNA by complementary base pairing.

antigen Any large molecule that stimulates the production of specific antibodies or binds specifically to an antibody.

antiparallel In the case of double-stranded DNA, referring to the opposite orientations of the strands, with the 5′ end of one strand paired with the 3′ end of the other strand.

antisense mRNA An mRNA transcribed from a cloned gene that is complementary to the mRNA produced by the normal gene.

apoptosis Controlled process leading to cell death that is triggered by intracellular damage (e.g., DNA lesions) or by external signals from neighboring cells. Also called *programmed cell death*.

aporepressor protein An inactive repressor that is activated when bound to an effector molecule.

applied research Research done with the objective of developing products or processes that can be commercialized or at least made available to humankind for practical benefit.

Archaea Prokaryotes that constitute one of the three main evolutionary domains of organisms. Members of this domain are called *archaeans*.

artificial selection Process for deliberating changing the phenotypic traits of a population by determining which individuals will survive and reproduce.

attenuation A regulatory mechanism in certain bacterial biosynthetic operons that controls gene expression by causing RNA polymerase to terminate transcription.

autonomously replicating sequence (ARS) A specific sequence in yeast chromosomes that, when included as part of an extrachromosomal, circular DNA molecule, confers on that molecule the ability to replicate autonomously; one type of eukaryotic **replicator.**

autopolyploidy Condition in which a cell or organism has two or more genetically distinct sets of chromosomes of the same species.

autosome A chromosome other than a **sex chromosome.**

auxotroph A mutant strain of an organism that cannot synthesize a molecule required for growth and therefore must have the molecule supplied in the growth medium for it to grow. Also called *auxotrophic mutant* or *nutritional mutant*.

auxotrophic mutant *See* **auxotroph.**

auxotrophic mutation A mutation that affects an organism's ability to make a particular molecule essential for growth. Also called *nutritional mutation*.

back mutation *See* **reverse mutation.**

Bacteria Prokaryotes that constitute one of the three main evolutionary domains of organisms. Members of this domain are called *bacteria*.

bacterial artificial chromosome (BAC) A vector for **cloning** DNA fragments up to about 200 kb long in *E. coli*. A BAC contains the origin of replication of the *F* factor, a multiple cloning site, and a selectable marker.

bacteriophages Viruses that attack bacteria. Also called *phages*.

Barr body A highly condensed and transcriptionally inactive X chromosome found in the nuclei of somatic cells of female mammals. *See also* **lyonization.**

base Also called **nitrogenous base.** Purine or pyrimidine component of a **nucleotide.**

base analog A chemical whose molecular structure is very similar to that of one of the bases normally found in DNA. Some chemical **mutagens,** such as 5-bromouracil (5BU), are base analogs.

base excision repair An enzyme-catalyzed process for repairing damaged DNA by removal of the altered base, followed by excision of the baseless nucleotide. The correct nucleotide then is inserted in the gap.

base-modifying agent A chemical **mutagen** that modifies the chemical structure of one or more bases normally found in DNA. Nitrous oxide, hydroxylamine, and methylmethane sulfonate are common base-modifying agents.

base-pair substitution mutation A change in the genetic material such that one base pair is replaced by another base pair; for instance, an `A-T` is replaced by a `G-C` pair.

basic research Research done to further knowledge for knowledge's sake.

bidirectional replication Synthesis of DNA in both directions away from an **origin of replication.**

bioinformatics Application of mathematics and computer science to store, retrieve, and analyze biological data, particularly nucleic acid and protein sequence data.

bivalent A pair of homologous, synapsed chromosomes, consisting of four **chromatids,** during the first meiotic division. *See also* **synapsis.**

bootstrap procedure A method for determining confidence levels attached to the branching patterns of a **phylogenetic tree** chosen by the parsimony approach.

bottleneck effect A form of **genetic drift** that occurs when a population is drastically reduced in size and some genes are lost from the gene pool as a result of chance.

branch-point sequence Specific sequence within introns of precursor mRNAs (pre-mRNAs) of eukaryotes containing an adenylate (A) nucleotide to which the free 5′ end of an intron binds during mRNA splicing.

broad-sense heritability The proportion of the **phenotypic variance** within a population that results from genetic differences among individuals.

cAMP (cyclic AMP) Adenosine 3′,5′ monophosphate; an intracellular regulatory molecule involved in controlling gene expression and some other processes in both prokaryotes and eukaryotes.

cancer Disease characterized by the uncontrolled and abnormal division of cells and by the spread of malignant tumor cells (metastasis) to disparate sites in the organism.

5′ capping The addition of a methylated guanine nucleotide (a "cap") to the 5′ end of a **precursor mRNA (pre-mRNA)** molecule in eukaryotes; the cap is retained on the mature mRNA molecule.

carcinogen Any physical or chemical agent that increases the frequency with which cells become cancerous.

carrier An individual who is heterozygous for a recessive mutation. A carrier usually does not exhibit the mutant phenotype.

catabolite activator protein (CAP) A regulatory protein that binds with cyclic AMP **(cAMP)** at low glucose concentrations, forming a complex that stimulates transcription of the *lac* operon and some other bacterial operons.

catabolite repression The inactivation of some **inducible operons** in the presence of glucose even though the operon's inducer is present. Also called *glucose effect*.

cDNA DNA copies made from RNA templates in a reaction catalyzed by the enzyme **reverse transcriptase.**

cDNA library Collection of cloned **complementary DNAs (cDNAs)** produced from the entire mRNA population of a cell.

cell cycle The cyclical process of growth and cellular reproduction in unicellular and multicellular eukaryotes. The cycle includes nuclear division, or **mitosis,** and cell (cytoplasmic) division, or **cytokinesis.**

cell division A process whereby one cell divides to produce two cells. *See also* **cytokinesis.**

***CEN* sequence** Nucleotide sequence of DNA in the centromere region of yeast chromosomes. Centromeresequences differ among species and between chromosomes in the same species.

centimorgan (cM) The unit of distance on a **genetic map.** Equivalent to *map unit*.

centromere The region of a chromosome containing DNA sequences to which mitotic and meiotic spindle fibers attach. Under the microscope a centromere is seen as a constriction in the chromosome. The centromere region of each chromosome is responsible for the accurate segregation of replicated chromosomes to the daughter cells during mitosis and meiosis. *See also* **kinetochore.**

chain-terminating codon *See* **stop codon.**

character *See* **hereditary trait.**

charged tRNA *See* **aminoacyl–tRNA.**

charging Addition of an amino acid to a tRNA that contains an **anticodon** for that animo acid. Also called *aminoacylation*.

checkpoints, cell-cycle Stages in the cell cycle at which progression of a cell through the cycle is blocked if there is damage to the genome or the mitotic machinery.

chiasma (*plural*, **chiasmata**) A cross-shaped structure formed during **crossing-over** and visible during the diplonema stage of meiosis.

chiasma interference *See* **interference.**

chi-square (χ^2) test A statistical procedure that determines what constitutes a significant difference between observed results and results expected on the basis of a particular hypothesis; a goodness-of-fit test.

chloroplasts Triple-membraned, chlorophyll-containing organelles found in green plants in which photosynthesis occurs.

chorionic villus sampling A procedure in which a sample of chorionic villus tissue of a developing fetus is examined for chromosomal abnormalities.

chromatid One of the two visibly distinct replicated copies of each chromosome that becomes visible between early prophase and metaphase of mitosis and is joined to its sister chromatid at their **centromeres.**

chromatin The DNA–protein complex that constitutes eukaryotic chromosomes and can exist in various degrees of folding or compaction.

chromatin remodeling Alteration of the structure of chromatin in the vicinity of a **core promoter** in a way that stimulates or represses transcription initiation. Remodeling is carried out by enzymes catalyzing histone acetylation or deacetylation and by nucleosome remodeling complexes.

chromosomal aberration *See* **chromosomal mutation.**

chromosomal mutation The variation from the wild-type condition in chromosome number or structure.

chromosome In eukaryotic cells, a linear structure composed of a single DNA molecule complexed with protein. Each eukaryotic species has a characteristic number of chromosomes in the nucleus of its cells. Most prokaryotic cells contain a single, usually circular chromosome.

chromosome library Collection of cloned DNA fragments produced from a particular chromosome (e.g., the human X chromosome).

chromosome theory of inheritance The theory that genes are located on chromosomes and that the transmission of chromosomes from one generation to the next accounts for the inheritance of hereditary traits.

***cis*-dominant** Referring to a gene or DNA sequence that can control genes on the same DNA molecule but not on other DNA molecules.

***cis-trans* test** *See* **complementation test.**

classical model An early model for genetic variation that was based on the assumption that most natural populations had a wild-type allele with very few mutant alleles present.

cline A systematic change in **allele frequencies** within a continuous population distributed over a geographic region.

clonal selection A process whereby cells that express cell-surface antibodies specific for a particular antigen are stimulated to proliferate and secrete that antibody by exposure to that antigen.

cloning (a) The production of many identical copies of a DNA molecule by replication in a suitable host; also called *DNA cloning*, *gene cloning*, and *molecular cloning*. (b) The generation of cells (or individuals) genetically identical to themselves and to their parent.

cloning vector A double-stranded DNA molecule that is able to replicate autonomously in a host cell and into which a DNA fragment (or fragments) can be inserted to form a recombinant DNA molecule for cloning.

coactivator In eukaryotes, a large multiprotein complex that interacts with activators bound at enhancers, general transcription factors bound near the promoter, and RNA polymerase II. These interactions help stimulate transcription of regulated genes.

coding sequence The part of an mRNA molecule that specifies the amino acid sequence of a polypeptide during translation.

codominance The condition in which an individual heterozygous for a gene exhibits the phenotypes of both homozygotes.

codon A group of three adjacent nucleotides in an mRNA molecule that specifies either one amino acid in a polypeptide chain or the termination of polypeptide synthesis.

codon usage bias A disproportionate use of one or a few synonymous codons within a codon family for a particular gene or across a genome.

coefficient of coincidence A measure of the extent of chiasma interference throughout a genetic map; ratio of the observed to the expected frequency of double crossovers. *See also* **interference.**

combinatorial gene regulation In eukaryotes, control of transcription by the combined action of several activators and repressors, which bind to particular gene regulatory sequences.

comparative genomics Comparison of the nucleotide sequences of entire genomes of different species, with the

goal of understanding the functions and **evolution** of genes. Such comparisons can identify which genome regions are evolutionarily conserved and likely to represent functional genes.

complementary base pairs The specific A-T and G-C base pairs in double-stranded DNA. The bases are held together by hydrogen bonds between the purine and pyrimidine base in each pair.

complementary DNA *See* **cDNA.**

complementation test A test used to determine whether two independently isolated mutations that confer the same phenotype are located within the same gene or in two different genes. Also called *cis-trans test*.

complete dominance The condition in which an allele is phenotypically expressed when one or both copies are present, so that the phenotype of the heterozygote is essentially indistinguishable from that of the homozygote.

complete medium For a microorganism, a medium that supplies all the ingredients required for growth and reproduction, including those normally produced by the wild-type organism.

complete recessiveness The condition in which an allele is phenotypically expressed only when two copies are present.

conditional mutation A mutation that results in a wild-type phenotype under one set of conditions but a mutant phenotype under other conditions. Temperature-sensitive mutations are a common type of conditional mutation.

conjugation In bacteria, process of unidirectional transfer of genetic material through direct cellular contact between a donor ("male") cell and a recipient ("female") cell.

consensus sequence The series of nucleotides found most frequently at each position in a particular DNA sequence among different species.

conservative model A model for DNA replication in which the two parental strands of DNA remain together and serve as a template for the synthesis of a new daughter double helix. The results of the Meselson–Stahl experiment did not support this model.

constitutive gene A gene whose expression is unregulated. The products of constitutive genes are essential to the normal functioning of the cell and are always produced in growing cells regardless of the environmental conditions.

constitutive heterochromatin Condensed chromatin that is always transcriptionally inactive and is found at homologous sites on chromosome pairs.

continuous trait *See* **quantitative trait.**

contributing allele An allele that affects the phenotype of a **quantitative trait.**

coordinate induction The simultaneous transcription and translation of two or more genes brought about by the action of an inducer.

core enzyme The portion of *E. coli* RNA polymerase that is the active enzyme; it is bound to the sigma factor, which directs the enzyme to the **promoter** region of genes.

corepressor In eukaryotes, a large multiprotein complex that interacts with repressors bound at enhancers, general transcription factors bound near the promoter, and RNA polymerase II. These interactions help inhibit transcription of regulated genes.

core promoter In eukaryotic genomes, the gene regulatory elements closest to the transcription start site that are required for RNA synthesis to begin at the correct nucleotide.

correlation coefficient A statistical measure of the strength of the association between two variables. *See also* **regression.**

cotransduction The simultaneous transduction of two or more bacterial genes, a good indication that the bacterial genes are closely linked.

coupling In individuals heterozygous at two genetic loci, the arrangement in which the wild-type alleles of both genes are on one homologous chromosome and the recessive mutant alleles are on the other; also called *cis configuration*. *See also* **repulsion.**

covariance A statistical measure of the tendency for two variables to vary together; used to calculate the **correlation coefficient** between the two variables.

CpG island DNA region containing many copies of the dinucleotide CpG. Many genes in eukaryotic DNA have CpG islands in or near the promoter. Methylation of the cytosines (C) in these islands represses transcription.

crisscross inheritance Transmission of a gene from a male parent to a female child to a male grandchild.

cross The fusion of male gametes from one individual and female gametes from another.

cross-fertilization *See* **cross.**

crossing-over The process of reciprocal chromosomal interchange that occurs frequently during meiosis and gives rise to **recombinant chromosomes.**

C-value The amount of DNA found in the haploid set of chromosomes.

cyclin Any of a group of proteins whose concentrations increase and decrease in a regular pattern through the cell cycle. The cyclins act in conjunction with **cyclin-dependent kinases** to regulate cell-cycle progression.

cyclin-dependent kinase (Cdk) Any of a group of protein kinases, activated by binding of specific cyclins, that regulate cell-cycle progression.

cytokinesis Division of the cytoplasm following mitosis or meiosis I and II during which the two new nuclei compartmentalize into separate daughter cells.

cytosine (C) A **pyrimidine** found in RNA and DNA. In double-stranded DNA, cytosine pairs with guanine, a **purine,** by hydrogen bonding.

dark repair *See* **excision repair.**

Darwinian fitness (*w*) The relative reproductive ability of individuals with a particular genotype.

daughter chromosomes Detached sister chromatids after they separate at the beginning of mitotic anaphase or meiotic anaphase II.

deamination Removal of an amino group from a nucleotide in DNA.

degeneracy In the **genetic code,** the existence of more than one codon corresponding to each amino acid.

degradation control The regulation of the rate of breakdown (turnover) of RNA molecules in the cell.

deletion A chromosomal mutation resulting in the loss of a segment of a chromosome and the gene sequences it contains.

deoxyribonuclease (DNase) An enzyme that catalyzes the degradation of DNA to nucleotides.

deoxyribonucleic acid (DNA) A polymeric molecule consisting of deoxyribonucleotide building blocks that in a double-stranded, double-helical form is the genetic material of all living organisms.

deoxyribonucleotide Any of the nucleotides that make up

DNA, consisting of a sugar (deoxyribose), a base, and a phosphate group.

deoxyribose The pentose (five-carbon) sugar found in DNA.

depurination Loss of a purine base (adenine or guanine) from a nucleotide in DNA.

determination Process early in development that establishes the fate of a cell, that is, the differentiated cell type it will become.

development Overall process of growth, differentiation, and morphogenesis by which a zygote gives rise to an adult organism. It involves a programmed sequence of phenotypic events that are typically irreversible.

diakinesis The final stage in prophase I of meiosis, during which the replicated chromosomes (bivalents) are most condensed, the nuclear envelope breaks down, and the spindle begins to form.

dicentric bridge *See* **dicentric chromosome.**

dicentric chromosome A homologous chromosome pair in meiosis I in which one chromatid has two centromeres as the result of crossing-over within a paracentric inversion. As the two centromeres begin to migrate to opposite poles, a dicentric bridge stretching across the cell forms and eventually breaks.

dideoxynucleotide (ddNTP) A modified nucleotide that has a 3′-H on the deoxyribose sugar rather than a 3′-OH. A ddNTP can be incorporated into a growing DNA chain, but no further DNA synthesis can occur because no phosphodiester bond can be formed with an incoming nucleotide. *See also* **dideoxy sequencing.**

dideoxy sequencing A method for rapidly sequencing DNA molecules in which the DNA to be sequenced is used as the template for *in vitro* DNA synthesis in the presence of **dideoxynucleotides (ddNTPs).** When a dideoxynucleotide is incorporated into a growing DNA chain, no further DNA synthesis occurs, generating a truncated chain in which the terminal dideoxynucleotide corresponds to the normal nucleotide that occurs at that point in the sequence.

differentiation Series of cell-specific changes in gene expression by which determined cells give rise to cell types with characteristic structures and functions.

dihybrid cross A cross between two individuals of the same genotype that are heterozygous for two pairs of alleles at two different loci (e.g., *Ss Yy* × *Ss Yy*).

dioecious Referring to plant species in which individual plants possess either male *or* female sex organs. *See also* **monoecious.**

diploid (2N) A cell or an individual with two copies of each chromosome.

diplonema The stage in prophase I of meiosis during which the **synaptonemal complex** disassembles and homologous chromosomes begin to move apart.

discontinuous trait A heritable characteristic that exhibits a small number of distinct phenotypes, which commonly are determined by variant alleles at a single locus. *See also* **quantitative trait.**

dispersed repeated DNA Repetitive DNA sequences that are distributed at irregular intervals in the genome.

dispersive model A model for DNA replication in which the parental double helix is cleaved into double-stranded DNA segments that act as templates for the synthesis of new double-stranded DNA segments, which are reassembled into complete DNA double helices, with parental and progeny DNA segments interspersed. The results of the Meselson–Stahl experiment did not support this model.

DNA A polymeric molecule consisting of deoxyribonucleotide building blocks that in a double-stranded, double-helical form is the genetic material of all living organisms.

DNA chip *See* **DNA microarray.**

DNA-dependent RNA polymerase The more complete name for **RNA polymerase**, the enzyme responsible for **transcription,** the process of RNA synthesis using a DNA template. *See* **RNA polymerase.**

DNA fingerprinting *See* **DNA typing.**

DNA helicase An enzyme that catalyzes unwinding of the DNA double helix at a **replication fork** during DNA replication.

DNA ladder Also known as DNA size markers, a set of DNA molecules of known size used in agarose gel electrophoresis experiments.

DNA ligase An enzyme that catalyzes the formation of a phosphodiester bond between the 5′ end of one DNA chain and 3′ end of another DNA chain during DNA replication and DNA repair.

DNA markers Sequence variations among individuals in a specific region of DNA that are detected by molecular analysis of the DNA and can be used in genetic analysis. *See also* **gene markers.**

DNA microarray An ordered grid of DNA molecules of known sequence—*probes*—fixed at known positions on a solid substrate, either a silicon chip, glass, or less commonly, a nylon membrane. Labeled free DNA molecules—*targets*—are added to the unlabeled fixed probes to analyze identities or quantities of target molecules. DNA microarrays allow for the simultaneous analysis of thousands of DNA target molecules.

DNA molecular testing A type of genetic testing that focuses on the molecular nature of mutations associated with a particular disease.

DNA polymerase Any enzyme that catalyzes the polymerization of deoxyribonucleotides into a DNA chain. All DNA polymerases synthesize DNA in the 5′ to 3′ direction.

DNA polymerase I (DNA Pol I) One of several *E. coli* enzymes that catalyze DNA synthesis; originally called the Kornberg enzyme.

DNA polymorphism Variation in the nucleotide sequence or number of tandem repeat units at a particular locus in the genome. Most commonly, this term is used for DNA markers, variations that are located outside of genes and that are detected by molecular analysis.

DNA primase An enzyme that catalyzes formation of a short RNA primer in DNA replication.

DNA profiling *See* **DNA typing.**

DNA typing Molecular analysis of **DNA polymorphisms** to identify individuals based on the unique characteristics of their DNA. Also called *DNA fingerprinting*.

domain shuffling Proposed mechanism for **evolution** of genes with new functions by the duplication and rearrangement of exons encoding protein domains in different combinations. Also called *exon shuffling*.

dominant Describing an allele or phenotype that is expressed in either the homozygous or the heterozygous state.

dominant lethal allele An allele that results in the death of an organism that is homozygous or heterozygous for the allele.

dosage compensation Any mechanism in organisms with **genotypic sex determination** for equalizing expression of genes on the sex chromosomes in males and females. *See also* **Barr body.**

Down syndrome *See* **trisomy-21.**

duplication A chromosomal mutation that results in the doubling of a segment of a chromosome and the gene sequences it contains.

EF *See* **elongation factor.**

effective population size The effective number of adults contributing gametes to the next generation.

effector A small molecule involved in controlling expression of a regulated gene or the activity of a protein.

elongation factor (EF) Accessory proteins required for the elongation phase of translation in prokaryotes and eukaryotes.

embryonic stem (ES) cell A cell derived from a very early embryo that retains the ability to differentiate into a cell type characteristic of any part of the organism.

enhancer A set of gene regulatory elements in eukaryotic genomes that can act over distances up to thousands of base pairs upstream or downstream from a gene. Most enhancers bind activators and act to stimulate transcription. *See also* **silencer element.**

environmental genomics *See* **metagenomics.**

environmental variance (V_E) Component of the **phenotypic variance** for a trait that is due to any nongenetic source of variation among individuals in a population. V_E includes variation arising from general environmental effects, which permanently influence phenotype; special environmental effects, which temporarily influence phenotype; and family environmental effects, which are shared by family members.

epigenetic Referring to a heritable change in gene expression that does not result from a change in the nucleotide sequence of the genome.

episome In bacteria, a **plasmid** that is capable of integrating into the host cell's chromosome.

epistasis Interaction between two or more genes that controls a single phenotype. For instance, the expression of a gene at one locus can mask or suppress the expression of a second gene at another locus.

epitope The specific short region of a protein (or other molecule recognized by an **antibody**) that is bound specifically by the antibody.

essential gene A gene that when mutated can result in the death of the organism.

euchromatin Chromatin that is condensed during mitosis but becomes uncoiled during interphase, when it can be transcribed. *See also* **heterochromatin.**

Eukarya One of the three major evolutionary domains. Organisms in this domain have genetic material in a membrane-bound nucleus as well as a number of membrane-bounded organelles such as mitochondria. *See also* **eukaryote.**

eukaryote Any organism whose cells have a membrane-bound nucleus in which the genetic material is located and membrane-bound organelles (e.g., mitochondria). Eukaryotes can be unicellular or multicellular and constitute one of the three major evolutionary domains of organisms. *See also* **Eukarya** and **prokaryote.**

euploid Referring to an organism or cell that has one complete set of chromosomes or an exact multiple of complete sets.

evolution Genetic change that takes place over time within a group of organisms.

evolutionary domains The three major lineages of organisms—**Bacteria, Archaea,** and **Eukarya**—thought to have evolved from a common, single-celled ancestor.

excision repair An enzyme-catalyzed process for removal of thymine dimers from DNA and synthesis of a new DNA segment complementary to the undamaged strand.

exon A segment of a protein-coding gene and its precursor (pre-mRNA) that specifies an amino acid sequence and is retained in the functional mRNA. *See also* **intron.**

exon shuffling *See* **domain shuffling.**

expected heterozygosity (H_e) The number of heterozygotes expected if the population is in **Hardy–Weinberg** equilibrium.

expression vector A cloning vector carrying a **promoter** and other sequences required for expression of a cloned gene in a host cell.

expressivity The degree to which a particular gene is expressed in the phenotype. A gene with variable expressivity can cause a range of phenotypes.

extranuclear inheritance The inheritance of characters determined by genes located on mitochondrial or chloroplast chromosomes. Such extranuclear genes show inheritance patterns distinctly different from those of genes on chromosomes in the nucleus. Also called *non-Mendelian inheritance*.

facultative heterochromatin Chromatin that may become condensed and therefore transcriptionally inactive in certain cell types, at different developmental stages, or in one member of a homologous chromosome pair.

familial trait A characteristic shared by members of a family as the result of shared genes and/or environmental factors.

fate map A diagram of an early embryo showing the cell types and tissues that different embryonic cells subsequently develop into.

***F*-duction** Transfer of host genes carried on an F' factor in **conjugation** between an F' and an F^- cell. If the genes are different between the two cell types, the recipient becomes partially diploid for the genes on the F'.

***F* factor** In *E. coli*, a **plasmid**—a self-replicating circular DNA molecule—that confers the ability to act as a donor cell in **conjugation.** Excision of an *F* factor from the bacterial chromosome may generate an F' factor, which may carry host cell genes. *See also* ***F*-duction.**

F_1 generation The offspring that result from the first experimental crossing of two parental strains of animals or plants; the first filial generation.

F_2 generation The offspring that result from crossing F_1 individuals; the second filial generation.

fine-structure mapping Procedures for generating a high-resolution map of allele sites within a gene.

first filial generation *See* **F_1 generation.**

first law *See* **principle of segregation.**

fitness *See* **Darwinian fitness.**

formylmethionine (fMet) A modified form of the amino acid methionine that has a formyl group attached to the amino group. It is the first amino acid incorporated into a polypeptide chain in prokaryotes and in eukaryotic organelles.

forward mutation A point mutation in a wild-type allele that changes it to a mutant allele.

founder effect A form of **genetic drift** that occurs when a

population is formed by migration of a small number of individuals from a large population.

***F*-pili** (*singular,* ***F*-pilus**) Hairlike cell surface components produced by cells containing the ***F* factor,** which allow the physical union of F^+ and F^- cells or *Hfr* and F^- cells to take place. Also called *sex pili*.

frameshift mutation A mutational addition or deletion of a base pair in a gene that disrupts the normal **reading frame** of the corresponding mRNA.

frequency distribution In genetics, a graphical representation of the numbers of individuals within a population who fall within the same range of phenotypic values for a continuous **quantitative trait.** Typically, the phenotypic classes are plotted on the horizontal axis and the number of individuals in each class are plotted on the vertical axis.

functional genomics The comprehensive analysis of the functions of genes and of nongene sequences in entire genomes, including patterns of gene expression and its control.

gain-of-function mutation A mutation that confers a new property on a protein, causing a new phenotype.

gamete Mature reproductive cell that is specialized for sexual fusion. Each gamete is haploid and fuses with a cell of similar origin but of opposite sex to produce a diploid zygote.

gametic disequilibrium Deviations from what is expected of loci that assort independently as a result of hybridization, genetic drift, and migration.

gametogenesis The formation of male and female gametes.

gametophyte The haploid sexual generation in the life cycle of plants that produces the gametes by mitotic division of spores.

GC box A promoter-proximal element upstream of the promoter of a eukaryotic gene at about 90 bp away from the transcription start site. The GC box has the consensus sequence 5′-GGGCGG-3′.

gene The physical and functional unit that helps determine the traits passed on from parents to offspring; also called *Mendelian factor*. In molecular terms, a gene is a nucleotide sequence in DNA that specifies a polypeptide or RNA. Alterations in a gene's sequence can give rise to species and individual variation.

gene conversion A nonreciprocal recombination process in which one allele in a heterozygote is changed to the other allele, thus converting a heterozygous genotype to a homozygous genotype.

gene expression The overall process by which a gene produces its product and the product carries out its function.

gene flow The movement of genes that takes place when organisms migrate and then reproduce, contributing their genes to the gene pool of the recipient population.

gene locus *See* **locus.**

gene markers Alleles that produce detectable phenotypic differences useful in genetic analysis. *See also* **DNA markers.**

gene mutation A heritable alteration in the sequence of a gene, usually from one allele form to another, or in the sequences regulating the gene.

gene pool All of the alleles in a breeding population existing at a given time.

generalized transduction A type of transduction in which any gene may be transferred from one bacterium to another.

general transcription factor (GTF) One of several proteins required for the initiation of transcription by a eukaryotic RNA polymerase.

gene segregation *See* **principle of segregation.**

gene silencing Inactivation of a gene due to its location in the genome, DNA methylation, or **RNA interference (RNAi).** This type of gene control often represses transcription of multiple genes in a region of DNA.

genetic code The set of three-nucleotide sequences **(codons)** within mRNA that carries the information for specifying the amino acid sequence of a polypeptide.

genetic correlation Phenotypic correlation due to genetic causes such as **pleiotropy** or genetic **linkage.**

genetic counseling Evaluation of the probabilities that prospective parents will have a child who expresses a particular genetic trait (deleterious or not) and discussion with the couple of their options for avoiding or minimizing the possible risk.

genetic drift Random change in allele frequencies within a population over time; observed most often in small populations due to **sampling error.**

genetic engineering Alteration of the genetic constitution of cells or individuals by directed and selective modification, insertion, or deletion of an individual gene or genes.

genetic hitchhiking During the process in which an allele that is advantageous or detrimental and thus is a target of natural selection may sweep to fixation or be lost very rapidly in the population, variants that are selectively neutral, or nearly so, and lie in positions on the chromosome nearby a new mutation may hitchhike along with the mutation to fixation or loss.

genetic map A representation of the relative distances separating genes on a chromosome based on the frequencies of recombination between nonallelic gene loci; also called *linkage map*. *See also* **physical map.**

genetic marker Any gene or DNA region whose sequence varies among individuals and is useful in genetic analysis, for example, in the detection of genetic recombination events.

genetic recombination A process by which parents with different alleles give rise to progeny with genotypes that differ from either parent. For example, parents with *A B* and *a b* genotypes can produce recombinant progeny with *A b* and *a B* genotypes.

genetics The science that deals with the structure and function of genes and their transmission from one generation to the next (heredity).

genetic structure of populations The patterns of genetic variation found among individuals within groups.

genetic testing Analysis to determine whether an individual who has symptoms of a particular genetic disease or is at high risk of developing it actually has a gene mutation associated with that disease.

genetic variance (V_G) Component of the **phenotypic variance** for a trait that is due to genetic differences among individuals in a population. V_G includes variation arising from the dominance effects of alleles, the additive effects of genes, and epistatic interactions among genes.

gene tree A **phylogenetic tree** based on the divergence observed within a single homologous gene. Gene trees are not always a good representation of the relationships among species because polymorphisms in any given gene may have arisen before speciation events. *See also* **species tree.**

genic sex determination System of sex determination, found primarily in eukaryotic microorganisms, in which sex is determined by different alleles at a small number of gene loci. *See also* **genotypic sex determination.**

genome The total amount of genetic material in a chromosome set; in eukaryotes, this is the amount of genetic material in the haploid set of chromosomes of the organism.

genomic imprinting Phenomenon in which the phenotypic expression of certain genes is determined by whether a particular allele is inherited from the female or male parent.

genomic library Collection of cloned DNA fragments in which every DNA sequence in the genome of an organism is represented at least once.

genomics The development and application of new mapping, sequencing, and computational procedures to analyze the entire genome of organisms.

genotype The complete genetic makeup (allele composition) of an organism. The term is commonly used in reference to the specific alleles present at just one or a limited number of genetic loci.

genotype frequency Percentage of individuals within a population that have a particular genotype. The sum of the genotype frequencies at a given locus is 1.

genotypic sex determination Any system in which sex chromosomes play a decisive role in the inheritance and determination of sex. *See also* **genic sex determination.**

germ-line mutation In sexually reproducing organisms, a change in the genetic material in germ-line cells (those that give rise to gametes), which may be transmitted by the gametes to the next generation, giving rise to an individual with the mutant genotype in both its somatic and germ-line cells. *See also* **somatic mutation.**

glucose effect *See* **catabolite repression.**

Goldberg–Hogness box *See* TATA **box.**

GTF *See* **general transcription factor.**

guanine (G) A **purine** found in RNA and DNA. In double-stranded DNA, guanine pairs with cytosine, a **pyrimidine,** by hydrogen bonding.

Haldane's rule Common observation that among the offspring of crosses between two species, one sex is sterile, absent, or rare. Often, male hybrids are sterile and female hybrids are fertile.

haploid (N) A cell or an individual with one copy of each nuclear chromosome.

haplosufficient Describing a gene that can support the normal wild-type phenotype when present in only one copy (heterozygous condition) in a diploid cell. A haplosufficient gene exhibits **complete dominance** in genetic crosses.

haplotype A set of specific SNP alleles at particular SNP loci that are close together in one small region of a chromosome.

haplotype map (hapmap) A complete description of all of the haplotypes known in all human populations tested, as well as the chromosomal location of each of these haplotypes.

Hardy–Weinberg law An extension of Mendel's laws of inheritance that describes the expected relationship between gene frequencies in natural populations and the frequencies of individuals of various genotypes in the same populations.

hemizygous Possessing only one copy (allele) of a gene in a diploid cell. Usually applied to genes on the X chromosome in males with the XY genotype.

hereditary trait A characteristic that results from gene action and is transmitted from one generation to another. Also called *character*.

heritability The proportion of phenotypic variation in a population attributable to genetic factors.

hermaphroditic Referring to animal species in which each individual has both testes and ovaries and to plant species in which both stamens and pistils are on the same flower.

heterochromatin Chromatin that remains condensed throughout the cell cycle and is usually not transcribed. *See also* **euchromatin.**

heterodimer A dimer containing one copy each of two different polypeptides.

heteroduplex DNA A region of double-stranded DNA with different sequence information on the two strands.

heterogametic sex In a species, the sex that has two types of sex chromosomes (e.g., X and Y) and therefore produces two kinds of gametes with respect to the sex chromosomes. In mammals, the male is the heterogametic sex.

heterogeneous nuclear RNA (hnRNA) A group of RNA molecules of various sizes that exist in a large population in the nucleus and include **precursor mRNAs (pre-mRNAs).**

heteroplasmon Cell of individuals with diseases caused by mtDNA defects in which there is a mixture of normal and mutant mitochondria. Also called *cytohet*.

heterosis The superiority of heterozygous genotypes regarding one or more characters compared with the corresponding homozygous genotypes based on growth, survival, phenotypic expression, and fertility. Also called *heterozygote superiority* or *overdominance*.

heterozygosity (*H*) A measure of genetic variation; with respect to a particular locus, the proportion of individuals within a population that are heterozygous at that locus.

heterozygote superiority *See* **heterosis.**

heterozygous Describing a diploid organism having different alleles of one or more genes and therefore producing gametes of different genotypes.

***Hfr* (high-frequency recombination)** Designation for an *E. coli* cell that has an ***F* factor** integrated into the bacterial chromosome. When an *Hfr* cell conjugates with a recipient (F^-) cell, bacterial genes are transferred to the recipient with high frequency.

highly repetitive DNA A class of DNA sequences, each of which is present in 10^5 to 10^7 copies in the haploid chromosome set.

histone One of a class of basic proteins that are complexed with DNA in **chromatin** and play a major role in determining the structure of eukaryotic nuclear chromosomes.

holandric trait *See* **Y-linked trait.**

homeobox A 180-bp consensus sequence found in many genes that regulate development.

homeodomain The 60-amino acid part of proteins that corresponds to the homeobox sequence in genes. All homeodomain-containing proteins can bind to DNA and function in regulating transcription.

homeotic genes Group of genes in *Drosophila* that specify the body parts (appendages) that will develop in each segment, thus determining the identity of the segments.

homeotic mutation Any mutation that alters the identity of a particular body segment, transforming it into a copy of a different segment.

homodimer A dimer containing two copies of the same polypeptide monomer.

homogametic sex In a species, the sex that has one type of sex chromosome (e.g., X) and therefore produces only one kind of gamete with respect to the sex chromosomes. In mammals, the female is the homogametic sex.

homolog Each individual member of a pair of homologous chromosomes.

homologous Referring to genes that have arisen from a common ancestral gene over evolutionary time; also used in reference to proteins encoded by homologous genes.

homologous chromosomes Chromosomes that have the same arrangement of genetic loci, are identical in their visible structure, and pair during meiosis.

homologous recombination Recombination between identical or highly similar DNA sequences; it is most common during meiosis.

homozygous Describing a diploid organism having the same alleles at one or more genetic loci and therefore producing gametes of identical genotypes.

homozygous dominant A diploid organism that has the same dominant allele for a given gene locus on both members of a homologous pair of chromosomes.

homozygous recessive A diploid organism that has the same recessive allele for a given gene locus on both members of a homologous pair of chromosomes.

Human Genome Project (HGP) A project to determine the sequence of the complete 3 billion (3×10^9) nucleotide pairs of the human genome and to map all of the genes along each chromosome.

hybridization In experiments, the complementary base-pairing a single-stranded DNA or RNA probe to a single-stranded DNA or RNA target molecule. One of the probe and target molecules is labeled, which one depending on the experiment.

hypersensitive sites Regions of DNA around transcriptionally active genes that are highly sensitive to digestion by DNase I. Also called *hypersensitive regions*.

hypothetico-deductive method of investigation Research method involving making observations, forming hypotheses to explain the observations, making experimental predictions based on the hypotheses, and, finally, testing the predictions. The last step produces new observations, so a cycle is set up leading to a refinement of the hypotheses and perhaps eventually to the establishment of a law or an accepted principle.

IF *See* **initiation factor.**

imaginal disc In the *Drosophila* blastoderm, a group of undifferentiated cells that will develop into particular adult tissues and organs.

immunoglobulins (Igs) Specialized proteins (antibodies) secreted by B cells that circulate in the blood and lymph and are responsible for humoral immune responses.

immunoprecipitation An experimental technique in which an **antibody** is allowed to bind to a specific target molecule in a solution, and then the antibody molecules, and all of the molecules bound to them, are collected (precipitated) from the solution.

inborn error of metabolism A biochemical disorder caused by mutation in a gene encoding an enzyme in a particular metabolic pathway.

inbreeding Preferential mating between close relatives.

incomplete dominance The condition in which neither of two alleles is completely dominant to the other, so that the heterozygote has a phenotype between the phenotypes of individuals homozygous for either allele involved. Also called *partial dominance*.

indels Gaps in a sequence alignment where it is not possible to determine whether an insertion occurred in one sequence or a deletion occurred in another.

independent assortment *See* **principle of independent assortment.**

induced mutation Any mutation that results from treating a cell or organism with a chemical or physical **mutagen.**

inducer A chemical or environmental agent that stimulates transcription of specific genes.

inducible operon An **operon** whose transcription is turned on in the presence of a particular substance (inducer). The lactose (*lac*) operon is an example of an inducible operon. *See also* **repressible operon.**

induction (1) Stimulation of the synthesis of a gene product in response to the action of an inducer, that is, a chemical or environmental agent. (2) In development, the ability of one cell or group of cells to influence the developmental fate of other cells.

inferred tree A phylogenetic tree generated with molecular data from real organisms.

initiation factor (IF) Any of various proteins involved in the initiation of translation.

initiator protein A protein that binds to the **replicator,** stimulates local unwinding of the DNA, and helps recruit other proteins required for the initiation of replication.

insertion sequence (IS element) The simplest **transposable element** found in prokaryotes. An IS element contains a single gene, which encodes transposase, an enzyme that catalyzes movement of the element within the genome.

insulator A DNA regulatory element, located between a promoter and associated enhancer, that blocks the ability of activators bound at the enhancer to stimulate transcription from the promoter.

interaction variance (V_I) Genetic variation among individuals resulting from **epistasis.**

intercalating agent A chemical mutagen that can insert between adjacent nucleotides in a DNA strand.

interference Phenomenon in which the presence of one crossover interferes with the formation of another crossover nearby. Mathematically defined as 1 minus the **coefficient of coincidence.** Also called *chiasma interference*.

intergenic suppressor A mutation whose effect is to suppress the phenotypic consequences of another (primary) mutation in a different gene.

interspersed repeated DNA *See* **dispersed repeated DNA.**

intragenic suppressor A mutation whose effect is to suppress the phenotypic consequences of another (primary) mutation within the same gene.

introgression Transfer of genes across species barriers.

intron A segment of a protein-coding gene and its precursor mRNA (pre-mRNA) that does not specify an amino acid sequence. Introns in pre-mRNA are removed by **mRNA splicing.** *See also* **exon.**

inversion A chromosomal mutation in which a segment of a chromosome is excised and then reintegrated in an orientation 180° from the original orientation.

karyotype A complete set of all the metaphase chromatid pairs in a cell.

kinetochore Specialized multiprotein complex that assembles at the centromere of a chromatid and is the site of attachment of spindle microtubules during mitosis.

Klinefelter syndrome A human clinical syndrome that results from disomy for the X chromosome in a male, which results in a 47,XXY male. Many of the affected males are mentally deficient, have underdeveloped testes, and are taller than average.

knockout mouse A mouse in which a nonfunctional allele of a particular gene has replaced the normal alleles, thereby knocking out the gene's function in an otherwise normal individual.

lagging strand In DNA replication, the DNA strand that is synthesized discontinuously from multiple RNA primers in the direction opposite to movement of the replication fork. *See also* **leading strand** and **Okazaki fragments.**

leader sequence *See* **5′ untranslated region (5′ UTR).**

leading strand In DNA replication, the DNA strand that is synthesized continuously from a single RNA primer in the same direction as movement of the replication fork. *See also* **lagging strand.**

leptonema The first stage in prophase I of meiosis during which the chromosomes begin to coil and become visible.

lethal allele An allele whose expression results in the death of an organism.

light repair *See* **photoreactivation.**

LINEs (long interspersed elements) One class of **dispersed repeated DNA** consisting of repetitive sequences that are several thousand base pairs in length. Some LINEs can move in the genome by **retrotransposition.**

linkage The association of genes located on the same chromosome such that they tend to be inherited together.

linkage disequilibrium Deviations from the expectations of **independent assortment** and Hardy–Weinberg equilibrium caused either by physical linkage or population demography.

linkage map *See* **genetic map.**

linked genes Genes that are located on the same chromosome and tend to be inherited together. A collection of such genes constitutes a *linkage group.*

linker *See* **restriction site linker.**

locus (*plural,* **loci**) The position of a gene on a genetic map; the specific place on a chromosome where a gene is located. More broadly, a locus is any chromosomal location that exhibits variation detectable by genetic or molecular analysis.

lod score method The lod (logarithm of odds) score method is a statistical analysis, usually performed by computer programs, based on data from pedigrees. It is used to test for linkage between two loci in humans.

long interspersed elements *See* **LINEs.**

looped domains Loops of supercoiled DNA that serve to compact the chromosomes.

loss-of-function mutation A mutation that leads to the absence or decreased biological activity of a particular protein.

Lyon hypothesis *See* **lyonization.**

lyonization A mechanism of dosage compensation, discovered by Mary Lyon, in which one of the X chromosomes in the cells of female mammals becomes highly condensed and genetically inactive.

lysogenic Referring to a bacterium that contains the genome of a temperate phage in the **prophage** state. On induction, the prophage leaves the bacterial chromosome, progeny phages are produced, and the bacterial cell lyses.

lysogenic pathway One of two pathways in the life cycle of temperate phages in which the phage genome is integrated into the host cell's chromosome and progeny phages are not formed.

lysogeny The phenomenon in which the genome of a temperate phage is inserted into a bacterial chromosome, where it replicates when the bacterial chromosome replicates. In this state, the phage genes are repressed and progeny phages are not formed.

lytic cycle Bacteriophage life cycle in which the phage takes over the bacterium and directs its growth and reproductive activities to express the phage genes and to produce progeny phages.

mapping function Mathematical formula used to correct the observed recombination frequencies for the incidence of multiple crossovers.

map unit (mu) A unit of measurement for the distance between two genes on a **genetic map.** A recombination (crossover) frequency of 1% between two genes equals 1 map unit. *See also* **centimorgan.**

maternal effect (a) The phenotype established by expression of **maternal effect genes** in the oocyte before fertilization. (b) An influence derived from the maternal environment (e.g., uterus size, quantity and quality of milk) that affects the phenotype of offspring, expressed as V_{Em}; one of the family environmental effects that influence the variation of **quantitative traits.**

maternal effect gene A nuclear gene, expressed by the mother during oogenesis, whose product helps direct early development in the embryo.

maternal inheritance A type of uniparental inheritance in which the mother's phenotype is expressed exclusively.

mating types In lower eukaryotes, two forms that are morphologically indistinguishable but carry different alleles and will mate; equivalent to the sexes in higher organisms. *See also* **genic sex determination.**

maximum parsimony Property of the **phylogenetic tree** (or trees) that invokes the fewest number of mutations and therefore is most likely to represent the true evolutionary relationship between species or their genes.

MCS *See* **multiple cloning site.**

mean The average of a set of numbers, calculated by adding all the values represented and dividing by the number of values.

meiosis Two successive nuclear divisions of a diploid nucleus, following one DNA replication, that result in the formation of haploid gametes or of spores having one-half the genetic material of the original cell.

meiosis I The first meiotic division, resulting in the reduction of the number of chromosomes from diploid to haploid.

meiosis II The second meiotic division, resulting in the

separation of the chromatids and formation of four haploid cells.

Mendelian factor *See* **gene.**

Mendelian population A group of interbreeding individuals who share a common **gene pool;** the basic unit of study in population genetics.

messenger RNA (mRNA) Class of RNA molecules that contain coded information specifying the amino acid sequences of proteins.

metabolomics The study of all of the small chemicals that are intermediates or products of metabolic pathways.

metacentric chromosome A chromosome with the centromere near the center such that the chromosome arms are of about equal lengths.

metagenomics A branch of comparative genomics involving the analysis of genomes in entire communities of microbes isolated from the environment. Also called *environmental genomics*.

metaphase The stage in mitosis or meiosis during which chromosomes become aligned along the equatorial plane of the spindle.

metaphase I The stage in meiosis I when each homologous chromosome pair (bivalent) becomes aligned on the equatorial plate.

metaphase II The stage of meiosis II during which the chromosomes (each a sister chromatid pair) line up on the equatorial plate in each of the two daughter cells formed in meiosis I.

metaphase plate The plane in the cell where the chromosomes become aligned during metaphase.

metastasis The spread of malignant tumor cells throughout the body so that tumors develop at new sites.

methyl-directed mismatch repair An enzyme-catalyzed process for repairing mismatched base pairs in DNA *after* replication is completed; contrast to **proofreading,** a process for correcting mismatched base pairs *during* replication.

methylome The complete set of DNA methylation modifications in the cell.

microbiome The community of microorganisms in a particular environment.

microRNA (miRNA) Noncoding, single-stranded regulatory RNA molecule about 21–23 nt long derived from an RNA transcript. An miRNA regulates the expression of a target mRNA by binding to the 3′ UTR causing either inhibition of translation of the mRNA or degradation of that molecule, depending on the extent of complementary base-pairing between the two molecules.

microsatellite *See* **short tandem repeat.**

minimal medium For a microorganism, a medium that contains the simplest set of ingredients (e.g., a sugar, some salts, and trace elements) required for the growth and reproduction of wild-type cells.

minisatellite *See* **variable number tandem repeat.**

missense mutation A **point mutation** in a gene that changes one codon in the corresponding mRNA so that it specifies a different amino acid than the one specified by the wild-type codon.

mitochondria Organelles found in the cytoplasm of all aerobic animal and plant cells in which most of the cell's ATP is produced.

mitosis The process of nuclear division in haploid or diploid cells producing daughter nuclei that contain identical chromosome complements and are genetically identical to one another and to the parent nucleus from which they arose.

moderately repetitive DNA A class of DNA sequences, each of which is present from a few to about 10^5 copies in the haploid chromosome set.

modifier gene A gene that interacts with another nonallelic gene causing a change in the phenotypic expression of the alleles of that gene.

molecular clock hypothesis The hypothesis that for any given gene, mutations accumulate at an essentially constant rate in all evolutionary lineages as long as the gene retains its original function.

molecular cloning *See* **cloning (a).**

molecular evolution Study of how genomes and macromolecules evolve at the molecular level and how genes and organisms are evolutionarily related.

molecular genetics Study of how genetic information is encoded within DNA and how biochemical processes of the cell translate the genetic information into the phenotype.

monoecious Referring to plant species in which individual plants possess *both* male and female sex organs and thus produce male and female gametes. Monoecious plants are capable of self-fertilization. *See also* **dioecious.**

monohybrid cross A cross between two individuals that are both heterozygous for the same pair of alleles (e.g., $Aa \times Aa$). By extension, the term also refers to crosses involving the pure-breeding parents that differ with respect to the alleles of one locus (e.g., $AA \times aa$).

monoploidy Condition in which a normally diploid cell or organism lacks one complete set of chromosomes.

monosomy A type of **aneuploidy** in which one chromosome of a homologous pair is missing from a normally diploid cell or organism. A monosomic cell is $2N - 1$.

morphogen A substance that helps determine the fate of cells in early development in proportion to its concentration.

morphogenesis Overall developmental process that generates the size, shape, and organization of cells, tissues, and organs.

mRNA splicing Process whereby an intron (intervening sequence) between two exons (coding sequences) in a **precursor mRNA (pre-mRNA)** molecule is excised and the exons ligated (spliced) together.

multifactorial trait A characteristic determined by multiple genes and environmental factors.

multigene family A set of genes encoding products with related functions that have evolved from a common ancestral gene through gene duplication.

multiple alleles Many alternative forms of a single gene. Although a population may carry multiple alleles of a particular gene, a single diploid individual can have a maximum of only two alleles at a locus.

multiple cloning site (MCS) A region within a cloning vector that contains many different restriction sites. Also called *polylinker.*

multiple-gene hypothesis for quantitative inheritance *See* **polygene hypothesis for quantitative inheritance.**

mutagen Any physical or chemical agent that significantly increases the frequency of mutational events above a spontaneous mutation rate.

mutagenesis The creation of mutations.

mutant allele Any form of a gene that differs from the wild-type allele. Mutant alleles may be **dominant** or **recessive** to wild-type alleles.

mutation Any detectable and heritable change in the genetic material not caused by genetic recombination; mutations may occur within or between genes and are the ultimate source of all new genetic variation.

mutation frequency The number of occurrences of a particular kind of mutation in a population of cells or individuals.

mutation rate The probability of a particular kind of mutation as a function of time.

mutator gene A gene that, when mutant, increases the spontaneous mutation frequencies of other genes.

narrow-sense heritability The proportion of the **phenotypic variance** that results from the additive effects of different alleles on the phenotype.

natural selection Differential reproduction of individuals in a population resulting from differences in their **genotypes.**

negative assortative mating Preferential mating between phenotypically dissimilar individuals that occurs more frequently than expected for **random mating.**

neutral mutation A **point mutation** in a gene that changes a codon in the corresponding mRNA to that for a different amino acid but results in no change in the function of the encoded protein.

neutral theory The hypothesis that much of the pattern of evolutionary changes in protein molecules can be explained by the opposing forces of mutation and random genetic drift.

nitrogenous base A nitrogen-containing **purine** or **pyrimidine** that, along with a pentose sugar and a phosphate, is one of the three parts of a **nucleotide.**

noncontributing allele An allele that has no effect on the phenotype of a **quantitative trait.**

nondisjunction A failure of homologous chromosomes or sister chromatids to separate at anaphase. *See also* **primary nondisjunction** and **secondary nondisjunction.**

nonhistone An acidic or neutral protein found in **chromatin.**

nonhomologous chromosomes Chromosomes that contain dissimilar genetic loci and that do not pair during meiosis.

nonhomologous recombination Recombination between DNA sequences that are not identical or highly similar. *See* **homologous recombination.**

non-Mendelian inheritance *See* **extranuclear inheritance.**

nonsense codon *See* **stop codon.**

nonsense mutation A **point mutation** in a gene that changes an amino-acid-coding codon in the corresponding mRNA to a stop codon.

nonsynonymous Referring to nucleotides in a gene that when mutated cause a change in the amino acid sequence of the encoded wild-type protein.

normal distribution Common probability distribution that exhibits a bell-shaped curve when plotted graphically.

norm of reaction Range of phenotypes produced by a particular genotype in different environments.

northern blot analysis A technique for detecting specific RNA molecules in which the RNAs are separated by gel electrophoresis, transferred to a nitrocellulose filter, and then hybridized with labeled complementary probes; also called *northern blotting. See also* **Southern blot analysis.**

nuclease An enzyme that catalyzes the degradation of a nucleic acid by breaking phosphodiester bonds.

nucleic acid High-molecular-weight polynucleotide. The main nucleic acids in cells are DNA and RNA.

nucleoid Central region in a bacterial cell in which the chromosome is compacted.

nucleoside A **purine** or **pyrimidine** covalently linked to a sugar.

nucleoside phosphate A nucleoside with an attached phosphate group. Also called *nucleotide.*

nucleosome The basic structural unit of eukaryotic **chromatin,** consisting of two molecules each of the four core histones (H2A, H2B, H3, and H4, the histone octamer), a single molecule of the linker histone H1, and about 180 bp of DNA.

nucleosome remodeling complex Large, multiprotein complex that uses the energy released by ATP hydrolysis to alter the position or structure of nucleosomes, thereby remodeling chromatin structure.

nucleotide The type of monomeric molecule found in RNA and DNA. Nucleotides consist of three distinct parts: a pentose (ribose in RNA, deoxyribose in DNA), a nitrogenous base (a purine or pyrimidine), and a phosphate group.

nucleotide excision repair (NER) *See* **excision repair.**

nucleus A discrete structure within eukaryotic cells that is bounded by a double membrane (the nuclear envelope) and contains most of the DNA of the cell.

null hypothesis A hypothesis that states there is no real difference between the observed data and the predicted data.

nullisomy A type of **aneuploidy** in which one pair of homologous chromosomes is missing from a normally diploid cell or organism. A nullisomic cell is $2N - 2$.

null mutation A mutation that results in a protein with no function.

nutritional mutant *See* **auxotroph.**

observed heterozygosity (H_o) The number of individuals in the population that are heterozygous at that locus.

Okazaki fragments The short, single-stranded DNA fragments that are synthesized on the lagging-strand template during DNA replication and are subsequently covalently joined to make a continuous strand, the **lagging strand.**

oligonucleotide A short DNA molecule.

oncogene A gene whose protein product promotes cell proliferation. Oncogenes are altered forms of **proto-oncogenes.**

oncogenesis Formation of a tumor (cancer) in an organism.

one-gene–one-enzyme hypothesis The hypothesis that each gene controls the synthesis of one enzyme.

one-gene–one-polypeptide hypothesis The hypothesis that each gene controls the synthesis of a polypeptide chain.

oogenesis Development of female gametes (egg cells) in animals.

open reading frame (ORF) In a segment of DNA, a potential protein-coding sequence identified by a start codon in frame with a stop codon.

operator A short DNA region, adjacent to the promoter of a bacterial operon, that binds repressor proteins responsible for controlling the rate of transcription of the operon.

operon In bacteria, a cluster of adjacent genes that share a common operator and promoter and are transcribed into a single mRNA. All the genes in an operon are regulated

coordinately; that is, all are transcribed or none are transcribed.

optimal alignment In the comparison of nucleotide or amino acid sequences from two or more organisms, an approximation of the true alignment of sequences where gaps are inserted to maximize the similarity among the sequences being aligned. *See also* **indels.**

ORF *See* **open reading frame.**

origin A specific site on a DNA molecule at which the double helix denatures into single strands and replication is initiated.

origin recognition complex (ORC) A multisubunit complex that functions as an **initiator protein** in eukaryotes.

origin of replication A specific region in DNA where the double helix unwinds and synthesis of new DNA strands begins.

overdominance *See* **heterosis.**

ovum (*plural,* **ova**) A mature female **gamete** (egg cell); the larger of the two cells that arise from a secondary oocyte by meiosis II in the ovary of female animals.

pachynema The stage in prophase I of meiosis during which the homologous pairs of chromosomes undergo **crossing-over.**

paracentric inversion A chromosomal mutation in which a segment on one chromosome arm that does not include the centromere is inverted.

parental *See* **parental genotype.**

parental class *See* **parental genotype.**

parental genotype The genetic makeup (allele composition) of individuals in the parental generation of genetic crosses. Progeny in succeeding generations may have combinations of linked alleles like one or the other of the parental genotypes or new (nonparental) combinations as the result of **crossing-over.**

partial reversion A **point mutation** in a mutant allele that restores all or part of the function of the encoded protein but not the wild-type amino acid sequence.

particulate factors The term Mendel used for the entities that carry hereditary information and are transmitted from parents to progeny through the gametes. These factors are now called *genes*.

PCR *See* **polymerase chain reaction.**

pedigree analysis Study of the inheritance of human traits by compilation of phenotypic records of a family over several generations.

penetrance The frequency with which a dominant or homozygous recessive gene is phenotypically expressed within a population.

pentose sugar A five-carbon sugar that, along with a nitrogenous base and a phosphate group, is one of the three parts of a **nucleotide.**

peptide bond A covalent bond in a polypeptide chain that joins the α-carboxyl group of one amino acid to the α-amino group of the adjacent amino acid.

peptidyl transferase Catalytic activity of an RNA component of the ribosome that forms the peptide bond between amino acids during translation.

pericentric inversion A chromosomal mutation in which a segment including the centromere and parts of both chromosome arms is inverted.

P generation The parental generation; the immediate parents of F_1 offspring.

phage Shortened form of **bacteriophage.**

phage lysate The progeny phages released after lysis of phage-infected bacteria.

phage vector A phage that carries pieces of bacterial DNA between bacterial strains in the process of transduction.

pharmacogenomics Study of how a person's unique genome affects the body's response to medicines.

phenotype The observable characteristics of an organism that are produced by the genotype and its interaction with the environment.

phenotypic correlation An association between two or more **quantitative traits** in the same individual.

phenotypic variance (V_P) A measure of all the variability for a **quantitative trait** in a population; mathematically is identical to the **variance.**

phosphate group An acidic chemical component that, along with a pentose sugar and a nitrogenous base, is one of the three parts of a **nucleotide.**

phosphodiester bond A covalent bond in RNA and DNA between a sugar of one nucleotide and a phosphate group of an adjacent nucleotide. Phosphodiester bonds form the repeating sugar–phosphate array of the backbone of DNA and RNA.

photoreactivation Repair of **thymine dimers** in DNA by exposure to visible light in the wavelength range 320–370 nm. Also called *light repair.*

phylogenetic relationship A reconstruction of the evolutionary history of groups of organisms (taxa) or genes.

phylogenetic tree A graphic representation of the evolutionary relationships among a group of species or genes. It consists of *branches* (lines) connecting *nodes*, which represent ancestral or extant organisms. *See also* **maximum parsimony.**

physical map A representation of the physical distances, measured in base pairs, between identifiable regions or markers on genomic DNA. A physical map is generated by analysis of DNA sequences rather than by genetic recombination analysis, which is used in constructing a **genetic map.**

physical marker Cytologically detectable visible (under the microscrope) changes in the chromosomes that make it possible to distinguish the chromosomes and, hence, the results of crossing-over.

pistil The female reproductive organ in flowering plants. It usually consists of a pollen-receiving stigma, stalklike style, and ovary.

plaque A round, clear area in a lawn of bacteria on solid medium that results from the lysis of cells by repeated cycles of phage lytic growth.

plasmid An extrachromosomal, double-stranded DNA molecule that replicates autonomously from the host chromosome. Plasmids occur naturally in many bacteria and can be engineered for use as **cloning vectors.**

pleiotropic Referring to genes or mutations that result in multiple phenotypic effects.

point mutant An organism whose mutant phenotype results from an alteration of a single nucleotide pair.

point mutation A heritable alteration of the genetic material in which one base pair is changed to another.

poly(A)+ mRNA An mRNA molecule in eukaryotes with a 3′ **poly(A) tail.**

poly(A) polymerase (PAP) The enzyme that catalyzes formation of the poly(A) tail at the 3′ end of eukaryotic mRNA molecules.

poly(A) site In eukaryotic precursor mRNAs (pre-mRNAs), the sequence that directs cleavage at the 3′ end and subsequent addition of adenine nucleotides to form the poly-A tail, during RNA processing.

poly(A) tail A sequence of 50 to 250 adenine nucleotides at the 3′ end of most eukaryotic mRNAs. The tail is added during processing of pre-mRNA.

polycistronic mRNA An mRNA molecule, transcribed from a bacterial or bacteriophage **operon,** that is translated into all the polypeptide encoded by the structural genes in the operon.

polygene hypothesis for quantitative inheritance The hypothesis that **quantitative traits** are controlled by many genes.

polygenes Two or more genes whose additive effects determine a particular **quantitative trait.**

polylinker *See* **multiple cloning site.**

polymerase chain reaction (PCR) A method for producing many copies of a specific DNA sequence from a DNA mixture without having to clone the sequence in a host organism.

polynucleotide A linear polymeric molecule composed of **nucleotides** joined by phosphodiester bonds. DNA and RNA are polynucleotides.

polypeptide A linear polymeric molecule consisting of **amino acids** joined by peptide bonds. *See also* **protein.**

polyploidy Condition in which a cell or organism has more than two sets of chromosomes.

polyribosome (polysome) The complex between an mRNA molecule and all the ribosomes that are translating it simultaneously.

polytene chromosome A special type of chromosome representing a bundle of numerous chromatids that have arisen by repeated cycles of replication of single chromatids without nuclear division. This type of chromosome is characteristic of various tissues of Diptera.

population A specific group of individuals of the same species.

population genetics Study of the consequences of Mendelian inheritance on the population level, including the mathematical description of a population's genetic composition and how it changes over time.

population viability analysis Analysis of the survival probabilities of different genotypes in the population.

position effect A change in the phenotypic effect of one or more genes as a result of a change in their position in the genome.

positive assortative mating Preferential mating between phenotypically similar individuals that occurs more frequently than expected for **random mating.**

postzygotic isolation Reduction in mating between closely related species by various mechanisms that act after fertilization, resulting in nonviable or sterile hybrids or hybrids of lowered fitness. *See also* **prezygotic isolation.**

precursor mRNA (pre-mRNA) The initial (primary) transcript of a protein-coding gene that is modified or processed to produce the mature, functional mRNA molecule.

precursor rRNA (pre-rRNA) The initial (primary) transcript produced from ribosomal DNA that is processed into three different rRNA molecules in prokaryotes and eukaryotes.

precursor tRNA (pre-tRNA) The initial (primary) transcript of a tRNA gene that is extensively modified and processed to produce the mature, functional tRNA molecule.

prezygotic isolation Reduction in mating between closely related species by various mechanisms that prevent courtship, mating, or fertilization. *See also* **postzygotic isolation.**

Pribnow box A part of the **promoter** sequence in bacterial genomes that is located at about 10 base pairs upstream from the transcription start site. Also called the *–10 box*.

primary nondisjunction A rare event in cells with a normal chromosome complement in which sister chromatids (in mitosis or meiosis II) or homologous chromosomes (in meiosis I) fail to separate and move to opposite poles. *See also* **nondisjunction** and **secondary nondisjunction.**

primary oocytes Diploid cells that arise by mitotic division of primordial germ cells (oogonia) and undergo meiosis in the ovaries of female animals.

primase *See* **DNA primase.**

primer *See* **RNA primer.**

primosome A complex of *E. coli* primase, helicase, and other proteins that functions in initiating DNA synthesis.

principle of independent assortment Mendel's second law stating that the factors (genes) for different traits assort independently of one another. In other words, genes on different chromosomes behave independently in the production of gametes.

principle of segregation Mendel's first law stating that two members of a gene pair (alleles) segregate (separate) from each other during the formation of gametes. As a result, one-half the gametes carry one allele and the other half carry the other allele.

probability The ratio of the number of times a particular event occurs to the number of trials during which the event could have happened.

proband In human genetics, an affected person with whom the study of a trait in a family begins. *See also* **proposita; propositus.**

product rule The rule that the probability of two independent events occurring simultaneously is the product of each of their probabilities.

programmed cell death *See* **apoptosis.**

prokaryote Any organism whose genetic material is not located within a membrane-bound nucleus. The prokaryotes are divided into two evolutionarily distinct groups, the **Bacteria** and the **Archaea.** *See also* **eukaryote.**

prometaphase Stage in **mitosis** in which the mitotic spindle that has been forming between the separating centriole pairs enters the former nuclear area, a kinetochore binds to each centromere, and kinetochore microtubules originating at one or other of the poles attach to each kinetochore.

prometaphase I Stage in **meiosis I** in which the nucleoli disappear, the nuclear envelope breaks down, the meiotic spindle that has been forming between the separating centriole pairs enters the former nuclear area, a kinetochore binds to each centromere, and kinetochore microtubules originating at one or other of the poles attach to each kinetochore.

prometaphase II Stage in **meiosis II** in which the nuclear envelopes (if formed in **telophase I**) break down, the spindle organizes across the cell, and kinetochore microtubules from the opposite poles attach to the kinetochores of each chromosome.

promoter A DNA region containing specific **gene regulatory elements** to which RNA polymerase binds for the initiation of transcription. *See also* **core promoter.**

promoter-proximal elements Gene regulatory elements in eukaryotic genomes that are located 50–200 base pairs

from the transcription start site (upstream of the TATA **box**) and help determine the efficiency of transcription.

proofreading In DNA synthesis, the process of recognizing a base-pair error during the polymerization events and correcting it. Proofreading is carried out by some DNA polymerases in prokaryotic and eukaryotic cells.

prophage The genome of a temperate bacteriophage that has been integrated into the chromosome of a host bacterium in the **lysogenic pathway.** A prophage is replicated during replication of the host cell's chromosome.

prophase The first stage in mitosis or meiosis during which the replicated chromosomes condense and become visible under the microscope.

prophase I The first stage of meiosis, divided into several substages, during which the replicated chromosomes condense, homologues undergo **synapsis,** and **crossing-over** occurs.

prophase II The first stage of meiosis II during which the chromosomes condense.

proportion of polymorphic loci (*P*) A ratio calculated by determining the number of loci with more than one allele present and dividing by the total number of loci examined.

proposita In human genetics, an affected female person with whom the study of a trait in a family begins. *See also* **proband.**

propositus In human genetics, an affected male person with whom the study of a trait in a family begins. *See also* **proband.**

protein A macromolecule composed of one or more **polypeptides.** The functional activity of a protein depends on its complex folded shape and composition.

protein array A collection of different proteins, immobilized on a solid substrate, that serve as probes for detecting labeled target proteins that bind to those affixed to the substrate. Also called *protein microarray* and *protein chip*.

proteome The complete set of proteins in a cell.

proteomics The cataloging and analysis of the proteins in a cell to determine when they are expressed, how much is made, and which proteins interact.

proto-oncogene A gene that in normal cells functions to control the proliferation of cells and that when mutated can become an **oncogene.** *See also* **tumor suppressor gene.**

prototroph A strain of an organism that is wild type for all nutritional requirements and can grow on minimal medium. *See also* **auxotroph.**

prototrophic strain *See* **prototroph.**

pseudodominance The phenotypic expression of a single recessive allele resulting from deletion of a dominant allele on the homologous chromosome.

pseudogene A nonfunctional gene that has sequence homology to one or more functional genes elsewhere in the genome.

Punnett square A matrix that describes all the possible genotypes of progeny resulting from a genetic cross.

pure-breeding strain *See* **true-breeding strain.**

purine One of the two types of cyclic nitrogenous bases found in DNA and RNA. Adenine and guanine are purines.

pyrimidine One of the two types of cyclic nitrogenous bases found in DNA and RNA. Cytosine (in DNA and RNA), thymine (in DNA), and uracil (in RNA) are pyrimidines.

pyrosequencing A DNA sequencing technique using a single-stranded template DNA molecule attached to a bead in which the release of the pyrophosphate in DNA chain growth is detected enzymatically. Pyrosequencing does not involve chain termination.

QTL *See* **quantitative trait loci.**

quantitative genetics Study of the inheritance of complex characteristics that are determined by multiple genes.

quantitative trait A heritable characteristic that shows a continuous variation in phenotype over a range. Also called *continuous trait*.

quantitative trait loci (QTL) The individual loci that contribute to a **quantitative trait.**

random mating Matings between individuals of the same or different genotypes that occur in proportion to the frequencies of the genotypes in the population.

rDNA repeat unit Set of ribosomal RNA (rRNA) genes—encoding 18S, 5.8S, and 28S rRNAs—that are located adjacent to each other and repeated many times in tandem arrays in eukaryotic genomes.

reading frame Linear sequence of codons (groups of three nucleotides) in mRNA that specify amino acids during translation beginning at a particular start codon.

real-time PCR A PCR method for measuring the increase in the amount of DNA as it is amplified (which gives the technique its "real-time" name). Also called *real-time quantitative PCR*.

recessive Describing an allele or phenotype that is expressed only in the homozygous state.

recessive lethal allele An allele that results in the death of organisms homozygous for the allele.

reciprocal cross A pair of crosses in which the genotypes of the males and females for a particular trait is reversed. In the garden pea, for example, a reciprocal cross for smooth and wrinkled seeds is smooth female × wrinkled male and wrinkled female × smooth male.

recombinant A chromosome, cell, or individual that has nonparental combinations of **genetic markers** as a result of genetic recombination.

recombinant chromosome A daughter chromosome that emerges from meiosis with an allele composition that differs from that of either parental chromosome.

recombinant DNA molecule Any DNA molecule that has been constructed in the test tube and contains sequences from two or more distinct DNA molecules, often from different organisms.

recombinant DNA technology A collection of experimental procedures for inserting a DNA fragment from one organism into DNA from another organism and for cloning the new recombinant DNA.

recombination *See* **genetic recombination.**

regression A statistical analysis assessing how changes in one variable are quantitatively related to changes in another variable.

regression coefficient The slope of the **regression line** drawn to show the relationship between two variables.

regression line A mathematically computed line that represents the best fit of a line to the data values for two variables plotted against each other. The slope of the regression line indicates the change in one variable (y) associated with a unit increase in another variable (x).

regulated gene A gene whose expression is controlled in response to the needs of a cell or organism.

reinforcement A model which states that, if populations

harbor genetic variation for mate recognition, then the alleles that allow the adults to discriminate successfully will increase in frequency.

release factor (RF) One of several proteins that recognize stop codons in mRNA and then initiate a series of specific events to terminate translation.

replica plating Procedure for transferring the pattern of colonies from a master plate to a new plate. In this procedure, a velveteen pad on a cylinder is pressed lightly onto the surface of the master plate, thereby picking up a few cells from each colony to inoculate onto the new plate.

replication bubble A locally unwound (denatured) region of DNA bounded by replication forks at which DNA synthesis proceeds in opposite directions.

replication fork A Y-shaped structure formed when a double-stranded DNA molecule unwinds to expose the two single-stranded **template strands** for DNA replication.

replicator The entire set of DNA sequences, including the **origin of replication,** required to direct the initiation of DNA replication.

replicon A stretch of DNA in eukaryotic chromosomes extending from an origin of replication to the two termini of replication on each side of that origin. Also called *replication unit*.

replisome The complex of closely associated proteins that forms at the replication fork during DNA synthesis in bacteria.

repressible operon An **operon** whose transcription is reduced in the presence of a particular substance, often the end product of a biosynthetic pathway. The tryptophan (*trp*) operon is an example of a repressible operon. *See also* **inducible operon.**

repressor The major class of transcription regulatory proteins in prokaryotes. Bacterial repressors usually bind to the **operator** and prevent transcription by blocking binding of RNA polymerase. In eukaryotes, repressors act in various ways to control transcription of some genes. *See also* **activators.**

repressor gene A regulatory gene whose product is a protein that controls the transcriptional activity of a particular operon or gene.

repulsion In individuals heterozygous for two genetic loci, the arrangement in which each homologous chromosome carries the wild-type allele of one gene and the mutant allele of the other gene; also called *trans configuration*. *See also* **coupling.**

restriction endonuclease *See* **restriction enzyme.**

restriction enzyme Enzyme that cleaves double-stranded DNA molecules within or near a specific nucleotide sequence (restriction site), which often is present in multiple copies with a genome. These enzymes are used in analyzing DNA and constructing recombinant DNA. Also called *restriction endonuclease*.

restriction fragment length polymorphism (RFLP) Variation in the lengths of fragments generated by treatment of DNA with a particular restriction enzyme. RFLPs result from point mutations that create or destroy restriction enzyme cleavage sites.

restriction mapping Procedure for locating the relative positions of restriction enzyme cleavage sites in a cloned DNA fragment, yielding a restriction map of the fragment.

restriction site Sequence in DNA recognized by a **restriction enzyme**. Many restriction enzymes cut both strands of DNA within the restriction site. Some restriction enzymes cut both strands of DNA near the restriction site.

restriction site linker A double-stranded oligodeoxyribonucleotide about 8 to 12 base pairs long that contains the cleavage site for a specific restriction enzyme and is used in cloning cDNAs. Also called *linker.*

retrotransposition The movement of certain mobile genetic elements (retrotransposons) in the genome by a mechanism involving an RNA intermediate.

retrotransposon A type of mobile genetic element, found only in eukaryotes, that encodes **reverse transcriptase** and moves in the genome via an RNA intermediate.

retrovirus A virus with a single-stranded RNA genome that replicates via a double-stranded DNA intermediate produced by **reverse transcriptase,** an enzyme encoded in the viral genome. The DNA integrates into the host's chromosome where it can be transcribed.

reverse genetics An experimental approach in which investigators attempt to find what phenotype, if any, is associated with a cloned gene.

reverse mutation A point mutation in a mutant allele that changes it back to a wild-type allele. Also called *reversion*.

reverse transcriptase An enzyme (an RNA-dependent DNA polymerase) that makes a double-stranded DNA copy of an RNA strand.

reverse transcriptase PCR (RT-PCR) A two-step method for detecting and quantitating a particular RNA in an RNA mixture by first converting the RNAs to cDNAs and then performing the **polymerase chain reaction (PCR)** using primers specific for the RNA of interest.

reversion *See* **reverse mutation.**

ribonuclease (RNase) An enzyme that catalyzes degradation of RNA to nucleotides.

ribonucleic acid (RNA) A usually single-stranded polymeric molecule consisting of ribonucleotide building blocks. The major types of RNA in cells are **ribosomal RNA (rRNA), transfer RNA (tRNA), messenger RNA (mRNA), small nuclear RNA (snRNA),** and **microRNA (miRNA),** each of which performs an essential role in protein synthesis (translation). In some viruses, RNA is the genetic material.

ribonucleotide Any of the nucleotides that make up RNA, consisting of a sugar (ribose), a base, and a phosphate group.

ribose The pentose (five-carbon) sugar found in RNA.

ribosomal DNA (rDNA) The regions of the genome that contain the genes for rRNAs in prokaryotes and eukaryotes.

ribosomal proteins A group of proteins that along with rRNA molecules make up the ribosomes of prokaryotes and eukaryotes.

ribosomal RNA (rRNA) Class of RNA molecules of several different sizes that, along with ribosomal proteins, make up ribosomes of prokaryotes and eukaryotes.

ribosome A large, complex cellular particle composed of ribosomal protein and rRNA molecules that is the site of amino acid polymerization during protein synthesis (translation).

ribosome-binding site (RBS) The nucleotide sequence in an mRNA molecule on which the ribosome becomes oriented in the correct reading frame for the initiation of translation. More commonly called the **Shine–Dalgarno sequence.**

ribosome recycling factor (RRF) A protein shaped like a tRNA molecule that, after translation termination, participates with

EF-G in steps to release the uncharged tRNA and to cause the two ribosomal subunits to dissociate from the mRNA.

ribozyme An RNA molecule that has catalytic activity.

RNA *See* **ribonucleic acid.**

RNA editing Unusual type of RNA processing in which the nucleotide sequence of a pre-mRNA is changed by the posttranscriptional insertion or deletion of nucleotides or by conversion of one nucleotide to another.

RNA enzyme *See* **ribozyme.**

RNA interference (RNAi) Silencing of the expression of a specific gene by double-stranded RNA whose sequence matches a portion of the mature mRNA encoded by the gene. Also called *RNA silencing*.

RNA polymerase Any enzyme that catalyzes the synthesis of RNA molecules from a DNA template in a process called **transcription.**

RNA polymerase I An enzyme in eukaryotes that catalyzes transcription of 18S, 5.8S, and 28S rRNA genes.

RNA polymerase II An enzyme in eukaryotes that catalyzes transcription of mRNA-coding genes and some snRNA genes.

RNA polymerase III An enzyme in eukaryotes that catalyzes transcription of tRNA and 5S rRNA genes and of some snRNA genes.

RNA primer A short RNA chain, produced by DNA primase during DNA replication, to which DNA polymerase adds nucleotides, thereby extending the new DNA strand.

RNA silencing *See* **RNA interference (RNAi).**

RNA splicing *See* **mRNA splicing.**

RNA synthesis *See* **transcription.**

RNA world hypothesis Theory proposing that RNA-based life predates the present-day DNA-based life, with the RNA carrying out the necessary catalytic reactions required for life in the presumably primitive cells of the time.

Robertsonian translocation A type of nonreciprocal translocation in which the long arms of two nonhomologous **acrocentric chromosomes** become attached to a single centromere.

rolling circle replication Process that occurs when a circular, double-stranded DNA replicates to produce linear DNA.

rooted tree A **phylogenetic tree** in which one internal node is represented as a common ancestor to all the other nodes on the tree.

rRNA transcription unit *See* **ribosomal DNA.**

RRF *See* **ribosome recycling factor.**

RT-PCR *See* **reverse transcriptase PCR.**

sample Subset of individuals belonging to a population. Study of a sample can provide accurate information about the population if the sample is large enough and randomly selected.

sampling error Chance deviations from expected results that arise when the observed sample is small.

secondary nondisjunction Abnormal segregation of the X chromosomes during meiosis in the progeny of females with the XXY genotype produced by a primary nondisjunction. *See also* **nondisjunction,** and **primary nondisjunction.**

secondary oocyte The larger of the two daughter cells produced by unequal cytokinesis during meiosis I of a primary oocyte in the ovaries of female animals.

second law *See* **principle of independent assortment.**

segmentation genes Group of genes in *Drosophila* that determine the number and organization of segments in the embryo and adult.

selection The favoring of particular combinations of genes in a given environment.

selection coefficient (*s*) A measure of the relative intensity of selection against a genotype; equals $1 - w$ **(Darwinian fitness).**

selection differential (*s*) In natural and artificial selection, the difference between the mean phenotype of the selected parents and the mean phenotype of the unselected population.

selection response (*R*) The amount by which a phenotype changes in one generation when natural or artificial selection is applied to a group of individuals.

self-fertilization (selfing) The union of male and female gametes from the same individual.

selfing *See* **self-fertilization.**

self-splicing The excision of introns from some pre-RNA molecules that occurs by a protein-independent reaction in certain organisms.

semiconservative model A model for DNA replication in which each daughter molecule retains one of the parental strands. The results of the Meselson–Stahl experiment supported this model.

semidiscontinuous Concerning DNA replication, when one new strand (the **leading strand**) is synthesized continuously and the other strand (the **lagging strand**) is synthesized discontinuously.

sex chromosome A chromosome in eukaryotic organisms that differs morphologically or in number in the two sexes. In many organisms, one sex possesses a pair of visibly different chromosomes. One is an X chromosome, and the other is a Y chromosome. Commonly, the XX sex is female and the XY sex is male.

sex-influenced trait A characteristic controlled by autosomal genes that appears in both sexes, but either the frequency of its occurrence or the relationship between genotype and phenotype is different in males and females.

sex-limited trait A characteristic controlled by autosomal genes that is phenotypically exhibited in only one of the two sexes.

sex-linked *See* **X-linked.**

sexual reproduction Mode of reproduction involving the fusion of haploid gametes produced directly or indirectly by meiosis.

Shine–Dalgarno sequence A sequence in prokaryotic mRNAs upstream of the start codon that base-pairs with an RNA in the small ribosomal subunit, allowing the ribosome to locate the start codon for correct initiation of translation. Also called the **ribosome-binding site (RBS).**

short interfering RNA (siRNA) Short double-stranded RNAs that function in gene silencing by **RNA interference (RNAi).**

short interspersed elements *See* **SINEs.**

short tandem repeat (STR) A type of **DNA polymorphism** involving variation in the number of short identical sequences (2 to 6 bp in length) that are tandemly repeated at a particular locus in the genome. Also called *microsatellite* and *simple sequence repeat*.

shuttle vector A **cloning vector** that can be introduced into and replicate in two or more host organisms (e.g., *E. coli* and yeast).

signal hypothesis The hypothesis that secreted proteins are synthesized on ribosomes that are directed to the endoplasmic

reticulum (ER) by an amino terminal **signal sequence** in the growing polypeptide chain.

signal peptidase An enzyme in the cisternal space of the endoplasmic reticulum that catalyzes removal of the **signal sequence** from growing polypeptide chains.

signal recognition particle (SRP) A cytoplasmic ribonucleoprotein complex that binds to the ER **signal sequence** of a growing polypeptide, blocking further translation of the mRNA in the cytosol.

signal recognition particle (SRP) receptor *See* **SRP receptor.**

signal sequence Hydrophobic sequence of 15–30 amino acids at the amino end of a growing polypeptide chain that directs the chain–mRNA–ribosome complex to the endoplasmic reticulum (ER) where translation is completed. The signal sequence is removed and degraded in the cisternal space of the endoplasmic reticulum.

signal transduction Process by which an external signal, such as a growth factor, leads to a particular cell response.

silencer element In eukaryotes, an **enhancer** that binds a repressor and acts to decrease RNA transcription rather than stimulating it, as most enhancers do.

silent mutation A **point mutation** in a gene that changes a codon in the mRNA to another codon for the same amino acid, resulting in no change in the amino acid sequence or function of the encoded protein.

simple telomeric sequences Short, tandemly repeated nucleotide sequences at or very close to the extreme ends of chromosomal DNA molecules. The same species-specific sequence is present at the ends of all chromosomes in an organism.

SINEs (short interspersed elements) One class of **dispersed repeated DNA** consisting of sequences that are 100 to 400 bp in length. SINEs can move in the genome by **retrotransposition.**

single nucleotide polymorphism (SNP) A difference in one base pair at a particular site (SNP locus) within coding or noncoding regions of the genome. SNPs that affect restriction sites cause **restriction fragment length polymorphisms (RFLPs).**

single-strand DNA-binding (SSB) protein A protein that binds to the unwound DNA strands at a **replication bubble** and prevents them from reannealing.

sister chromatids Two identical copies of a chromosome derived from replication of the chromosome during interphase of the cell cycle. Sister chromatids are held together by the replicated but unseparated centromeres.

site-specific mutagenesis Introduction of a mutation at a specific site in a particular gene by one of several in vitro techniques.

slope of the line *See* **regression coefficient.**

small nuclear ribonucleoprotein particle (snRNP) Large complex formed by small nuclear RNAs (snRNAs) and proteins in which the processing of pre-mRNA molecules occurs.

small nuclear RNA (snRNA) Class of RNA molecules, found only in eukaryotes, that associate with certain proteins to form small nuclear ribonucleoprotein particles (snRNPs).

SNP (single nucleotide polymorphism) locus Site of a simple, single base-pair alteration found between individuals that can be used as a **DNA marker.**

somatic mutation In multicellular organisms, a change in the genetic material of somatic (body) cells. It may affect the phenotype of the individual in which the mutation occurs but is not passed on to the succeeding generation.

sonicate The use of very high-frequency sound (well beyond what we can hear) to disrupt cells or molecules.

Southern blot analysis A technique for detecting specific DNA fragments in which the fragments are separated by gel electrophoresis, transferred from the gel to a nitrocellulose filter, and then hybridized with labeled complementary probes; also called *Southern blotting*. *See also* **northern blot analysis.**

specialized transducing phage A temperate bacteriophage that can transfer only a certain section of the bacterial chromosome from one bacterium to another.

specialized transduction A type of transduction in which only specific genes are transferred from one bacterium to another.

species tree A **phylogenetic tree** based on the divergence observed within multiple genes. A species tree is better than a **gene tree** for depicting the evolutionary history of a group of species.

spermatogenesis Development of male gametes (sperm cells) in animals.

sperm cell A mature male **gamete,** produced by the testes in male animals. Also called *spermatozoon* (plural: *spermatozoa*).

spliceosome Large complex in the nucleus of eukaryotic cells that carries out **mRNA splicing.** It consists of several small nuclear ribonucleoprotein particles (snRNPs) bound to a pre-mRNA molecule.

spontaneous mutation Any mutation that occurs without the use of a chemical or physical mutagenic agent.

sporophyte The haploid asexual generation in the life cycle of plants that produces haploid spores by meiosis.

SRP receptor The signal recognition particle (SRP) receptor is an integral protein in the membrane of the **endoplasmic reticulum (ER)** to which binds the complex of a growing **polypeptide, signal recognition particle (SRP),** and **ribosome.** This interaction facilitates binding of the ribosome to the outside surface of the ER and the insertion of the polypeptide into the lumen of the ER.

stamen The male reproductive organ in flowering plants. It usually consists of a stalklike filament bearing a pollen-producing anther.

standard deviation The square root of the **variance;** a common measure of the extent of variability in a population for **quantitative traits.**

standard error of allele frequency A statistical measure of the amount of variation in allele frequency among populations.

steroid hormone response element (HRE) DNA sequence to which a complex of a specific steroid hormone and its receptor binds, resulting in activation of genes regulated by that hormone.

stop codon One of three codons in mRNA for which no normal tRNA molecule exists and that signals the termination of polypeptide synthesis.

STR *See* **short tandem repeat.**

submetacentric chromosome A chromosome with the centromere nearer one end than the other such that one arm is longer than the other.

substitution A mutation that has passed through the filter of selection on at least some level.

sum rule The rule that the probability of either of two mutually exclusive events occurring is the sum of their individual probabilities.

supercoiled Referring to a double-stranded DNA molecule that is twisted in space about its own axis.

suppressor gene A gene that when mutated causes suppression of mutations in other genes.

suppressor mutation A mutation at a second site that totally or partially restores a function lost because of a primary mutation at another site.

synapsis The intimate association of replicated homologous chromosomes brought about by the formation of a zipper-like structure (the synaptonemal complex) between the homologues during prophase I of meiosis.

synaptonemal complex A complex structure that spans the region between meiotically paired (synapsed) chromosomes and facilitates **crossing-over.**

synonymous Referring to nucleotides in a gene that when mutated do not result in a change in the amino acid sequence of the encoded wild-type protein.

tag SNP One (or more) SNP locus used to test for and represent an entire haplotype.

tandemly repeated DNA Repetitive DNA sequences that are clustered together in the genome, so that each such sequence is repeated many times in a row within a particular chromosomal region.

TATA **box** A part of the **core promoter** in eukaryotic genomes; it is located about 30 base pairs upstream from the transcription start point. Also called the TATA element, or the **Goldberg–Hogness box.**

tautomers Alternate chemical forms in which DNA (or RNA) bases are able to exist.

telocentric chromosome A chromosome with the centromere more or less at one end such that only one arm is visible.

telomerase An enzyme that adds short, tandemly repeated DNA sequences **(simple telomeric sequences)** to the ends of eukaryotic chromosomes. It contains an RNA component complementary to the telomeric sequence and has **reverse transcriptase** activity.

telomere A specific set of sequences at the end of a linear chromosome that stabilizes the chromosome and is required for replication. *See also* **simple telomeric sequences** and **telomere-associated sequences.**

telomere-associated sequence Repeated, complex DNA sequence extending inward from the simple telomeric sequence at each end of a chromosomal DNA molecule.

telophase The stage in mitosis or meiosis during which the migration of the daughter chromosomes to the two poles is completed.

telophase I The stage in meiosis I, when chromosomes (each a sister chromatid pair) complete migration to the poles and new nuclear envelopes form around each set of replicated chromosomes.

telophase II The last stage of meiosis II, during which a nuclear membrane forms around each set of daughter chromosomes and **cytokinesis** takes place.

temperate phage A bacteriophage that is capable of following either the **lytic cycle** or **lysogenic pathway.** *See also* **virulent phage.**

temperature-sensitive mutant A strain that exhibits a wild-type phenotype in one temperature range but a defective (mutant) phenotype in another, usually higher, temperature range.

template strand DNA strand on which is synthesized a complementary DNA strand during replication or an RNA strand during transcription.

terminator A DNA sequence located at the distal (downstream) end of a gene that signals the termination of transcription.

testcross A cross of an individual of unknown genotype, usually expressing the dominant phenotype, with a homozygous recessive individual to determine the unknown genotype.

testis-determining factor Gene product in placental mammals that causes embryonic gonadal tissue to develop into testes; in the absence of this factor, the gonadal tissue develops as ovaries.

tetrasomy A type of **aneuploidy** in which a normally diploid cell or organism possesses four copies of a particular chromosome instead of two copies. A tetrasomic cell is 2N + 2.

three-point testcross A cross between an individual heterozygous at three loci with an individual homozygous for recessive alleles at the same three loci. Commonly used in mapping linked genes to determine their order in the chromosome and the distances between them.

thymine (T) A **pyrimidine** found in DNA but not in RNA. In double-stranded DNA, thymine pairs with adenine, a **purine,** by hydrogen bonding.

thymine dimer A common lesion in DNA, caused by ultraviolet radiation, in which adjacent thymines in the same strand are linked in an abnormal way that distorts the double helix at that site.

topoisomerase Any enzyme that catalyzes the supercoiling of DNA.

totipotent Describing a cell that has the potential to develop into any cell type of the organism.

trailer sequence *See* **3′ untranslated region (3′ UTR).**

trait *See* **hereditary trait.**

transconjugant A bacterial cell that incorporates donor DNA received during conjugation into its genome.

transcription The process for making a single-stranded RNA molecule complementary to one strand (the template strand) of a double-stranded DNA molecule, thereby transferring information from DNA to RNA. Also called *RNA synthesis*.

transcriptome The set of mRNA transcripts in a cell.

transcriptomics The study of gene expression at the level of the entire genome.

***trans*-dominant** Referring to a gene or DNA sequence that can control genes on different DNA molecules.

transducing phage Any bacteriophage that can mediate transfer of genetic material between bacteria by **transduction.**

transducing retrovirus Retrovirus that has picked up an **oncogene** from the genome of a host cell.

transductant In bacteria, a recombinant recipient cell generated by **transduction.**

transduction A process by which bacteriophages mediate the transfer of pieces of bacterial DNA from one bacterium (the donor) to another (the recipient).

transfer RNA (tRNA) Class of RNA molecules that bring amino acids to ribosomes, where they are transferred to growing polypeptide chains during translation.

transformant In bacteria, a recombinant recipient cell generated by **transformation.**

transformation (a) In bacteria, a process in which genetic information is transferred by means of extracellular pieces of DNA. (b) In eukaryotes, the conversion of a normal cell with regulated growth properties to a cancer-like cell that can give rise to tumors.

transforming principle Term coined by Frederick Griffith for the unknown agent responsible for the change in genotype via transformation in bacteria. DNA is now known to constitute the transforming principle.

transgene A gene introduced into the genome of an organism by genetic manipulation to alter its genotype.

transgenic Referring to a cell or organism whose genotype has been altered by the artificial introduction of a different allele or gene from the same or a different species.

transition *See* **transition mutation.**

transition mutation A type of **base-pair substitution mutation** that involves a change of one purine-pyrimidine base pair to the other purine–pyrimidine base pair (e.g., A-T to G-C) at a particular site in the DNA.

translation The process that converts the nucleotide sequence of an mRNA into the amino acid sequence of a polypeptide. Also called *protein synthesis*.

translesion DNA synthesis An inducible DNA repair process that allows the replication of DNA beyond a lesion that normally would interrupt DNA synthesis. In *E. coli*, this process is called the *SOS response*.

translocation (a) A chromosomal mutation involving a change in the position of a chromosome segment (or segments) and the gene sequences it contains. (b) In polypeptide synthesis, translocation is the movement of the ribosome, one codon at a time, along the mRNA toward the 3′ end.

transmission genetics Study of how genes are passed from one individual to another. Also called *classical genetics*.

transposable element A DNA segment that can move from one position in the genome to another (nonhomologous) position; also called *mobile genetic element*. Transposable elements are found in both prokaryotes and eukaryotes.

transposase An enzyme encoded by many types of mobile genetic elements that catalyzes the movement **(transposition)** of these elements in the genome.

transposition The movement of a transposable element within the genome. *See also* **retrotransposition.**

transposon (Tn) A mobile genetic element that contains a gene for transposase, which catalyzes transposition, and genes with other functions such as antibiotic resistance.

transversion *See* **transversion mutation.**

transversion mutation A type of **base-pair substitution mutation** that involves a change of a purine–pyrimidine base pair to a pyrimidine–purine base pair (e.g., A-T to T-A or G-C to T-A) at a particular site in the DNA.

trihybrid cross A cross between individuals of the same genotype that are heterozygous for three pairs of alleles at three different loci (e.g., *Ss Yy Cc* × *Ss Yy Cc*).

trisomy A type of **aneuploidy** in which a normally diploid cell or organism possesses three copies of a particular chromosome instead of two copies. A trisomic cell is 2N + 1.

trisomy-13 The presence of an extra copy of chromosome 13, which causes Patau syndrome in humans.

trisomy-18 The presence of an extra copy of chromosome 18, which causes Edwards syndrome in humans.

trisomy-21 The presence of an extra copy of chromosome 21, which causes Down syndrome in humans.

true-breeding strain A strain in which mating of individuals yields progeny with the same genotype as the parents.

true reversion A **point mutation** in a mutant allele that restores it to the wild-type allele; as a result, the wild-type amino acid sequence and function of the encoded protein is restored.

tumor A tissue mass composed of transformed cells, which multiply in an uncontrolled fashion and differ from normal cells in other ways as well; also called *neoplasm*. Benign tumors do not invade the surrounding tissues, whereas malignant tumors invade tissue and often spread to other sites in the body.

tumor suppressor gene A gene in normal cells whose protein product suppresses uncontrolled cell proliferation. *See also* **proto-oncogene.**

tumor virus A virus that induces cells to dedifferentiate and to divide to produce a tumor.

Turner syndrome A human clinical syndrome that results from monosomy for the X chromosome in the female, which gives a 45,X female. Affected females fail to develop secondary sexual characteristics, tend to be short, have weblike necks, have poorly developed breasts, are usually infertile, and exhibit mental deficiencies.

unequal crossing-over The process of chromosomal interchange between misaligned chromosomes that may occur during meiosis.

uniparental inheritance A phenomenon, usually exhibited by mitochondrial and chloroplast genes, in which all progeny have the phenotype of only one parent.

unique-sequence DNA A class of DNA sequences, each of which is present in one to a few copies in the haploid chromosome set; includes most protein-coding genes. Also called *single-copy DNA*.

3′ untranslated region (3′ UTR) The untranslated part of an mRNA molecule beginning at the end of the amino acid-coding sequence and extending to the 3′ end of the mRNA.

5′ untranslated region (5′ UTR) In eukaryotes, the untranslated part of an mRNA molecule extending from the **5′** end to the first (start) codon. It contains coded information for directing initiation of protein synthesis at the translation start site.

unweighted pair group method with arithmetic averages (UPGMA) A statistically based approach used in constructing **phylogenetic trees** that groups taxa based on their overall pairwise similarities to each other. Also called *cluster analysis*.

uracil (U) A **pyrimidine** found in RNA but not in DNA.

variable number tandem repeat (VNTR) A type of **DNA polymorphism** involving variation in the number of identical sequences (7 bp to a few tens of base pairs in length) that are tandemly repeated at a particular locus in the genome. Also called a *minisatellite*.

variance A statistical measure of the extent to which values in a data set differ from the **mean.**

virulent phage A bacteriophage, such as T4, that always follows the **lytic cycle** when it infects bacteria. *See also* **temperate phage.**

visible mutation A mutation that affects the morphology or physical appearance of an organism.

VNTR *See* **variable number tandem repeat.**

whole-genome shotgun approach for genome sequencing An approach for sequencing an entire genome in which the whole genome is broken into partially overlapping fragments, each fragment is cloned and sequenced, and the genome sequence is assembled from the overlapping sequences by computer.

wild type Term describing an allele or phenotype that is designated as the standard ("normal") for an organism and is usually, but not always, the most prevalent in a "wild" population of the organism; also used in reference to a strain or individual.

wild-type allele *See* **wild type.**

wobble hypothesis A proposed mechanism that explains how one **anticodon** can pair with more than one **codon.**

X chromosome A sex chromosome present in two copies in the homogametic sex (the female in mammals) and in one copy in the heterogametic sex (the male in mammals).

X chromosome–autosome balance system A genotypic sex determination system in which the ratio between the numbers of X chromosomes and number of sets of autosomes is the primary determinant of sex.

X chromosome nondisjunction Failure of the two X chromosomes to separate in meiosis so that eggs are produced with two X chromosomes or with no X chromosomes instead of the usual one X chromosome.

X-linked Referring to genes located on the X chromosome.

X-linked dominant trait A characteristic caused by a dominant mutant allele carried on the X chromosome.

X-linked recessive trait A characteristic caused by a recessive mutant allele carried on the X chromosome.

Y chromosome A sex chromosome that when present is found in one copy in the heterogametic sex, along with an X chromosome, and is not present in the homogametic sex. Not all organisms with sex chromosomes have a Y chromosome.

Y chromosome mechanism of sex determination A genotypic system of sex determination in which the Y chromosome determines the sex of an individual. Individuals with a Y chromosome are genetically male, and individuals without a Y chromosome are genetically female.

yeast artificial chromosome (YAC) A vector for cloning large DNA fragments, several hundred kilobase pairs long, in yeast. A YAC is a linear molecular with a telomere at each end, a centromere, an autonomously replicating sequence (ARS), a selectable marker, and a **polylinker.**

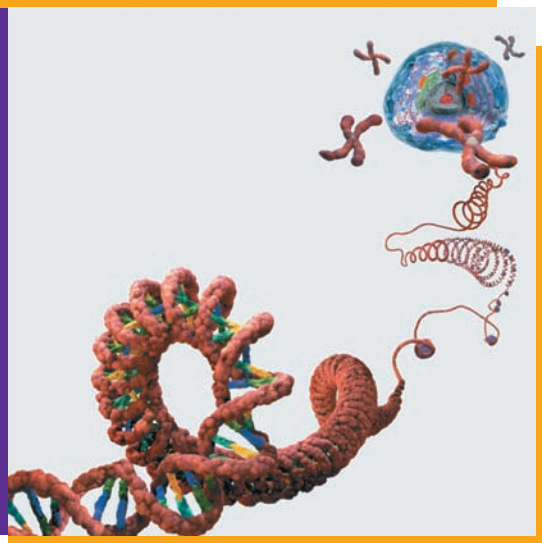
yeast two-hybrid system Experimental procedure to find genes encoding proteins that interact with a known protein. Also called *interaction trap assay*.

Y-linked trait A characteristic controlled by a gene carried on the Y chromosome for which there is no corresponding gene locus on the X chromosome. Also called *holandric* or *"wholly male" trait*.

zygonema The stage in prophase I of meiosis during which homologous chromosomes begin to pair in a highly specific way along their lengths.

zygote The cell produced by the fusion of a male gamete (sperm cell) and a female gamete (egg cell).

Genetics: An Introduction



Sylized diagram of the relationship between DNA, chromosomes, and the cell.

Key Questions

- What are the major subdivisions of genetics?
- What are geneticists, and what is genetics research?

Welcome to the study of **genetics,** the science of heredity. Genetics is concerned primarily with understanding biological properties that are transmitted from parent to offspring. The subject matter of genetics includes heredity, the molecular nature of the genetic material, the ways in which genes (which determine the characteristics of organisms) control life functions, and the distribution and behavior of genes in populations.

Genetics is central to biology because gene activity underlies all life processes, from cell structure and function to reproduction. Learning what genes are, how genes are transmitted from generation to generation, how genes are expressed, and how gene expression is regulated is the focus of this text. Genetics is expanding so rapidly that it is not possible to describe everything we know about it between these covers. The important principles and concepts are presented carefully and thoroughly; readers who want to go further are advised to look for information on the Internet, including searching for research papers using Google Scholar or the PubMed database supported by the National Library of Medicine, National Institutes of Health, at http://www.pubmed.gov.

It is assumed that your experience in your introductory biology course has given you a general understanding of genetics. This chapter provides a contextual framework for your study of genes.

Classical and Modern Genetics

Humans recognized long ago that offspring tend to resemble their parents. Humans have also performed breeding experiments with animals and plants for centuries. However, the principles of heredity were not understood until the mid-nineteenth century, when Gregor Mendel analyzed quantitatively the results of crossing pea plants that varied in easily observable characteristics. He published his results, but their significance was not realized in his lifetime. Several years after his death, however, researchers realized that Mendel had discovered fundamental principles of heredity. We now consider Mendel's work to be the foundation of modern genetics.

Since the turn of the twentieth century, genetics has been an increasingly powerful tool for studying biological processes. An important approach used by many geneticists is to work with mutants of a cell or an organism affecting a particular biological process: by characterizing the differences between the mutants with normal cells or organisms, they develop an understanding of the process. Such research has gone in many directions, such as analyzing heredity in populations, analyzing evolutionary processes, identifying the genes that control the steps in a process, mapping the genes involved, determining the products of the genes, and analyzing the molecular features of the genes, including the regulation of the genes' expression.

Research in genetics underwent a revolution in 1972, when Paul Berg constructed the first recombinant DNA

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molecule *in vitro*, and in 1973, when Herbert Boyer and Stanley Cohen cloned a recombinant DNA molecule for the first time. The development by Kary Mullis in 1986 of the polymerase chain reaction (PCR) to amplify specific segments of DNA spawned another revolution. Recombinant DNA technology, PCR, and other molecular technologies are leading to an ever-increasing number of exciting discoveries that are furthering our knowledge of basic biological functions and will lead to improvements in the quality of human life.

Now the genomics revolution is occurring. That is, the complete genomic DNA sequences have been determined for many viruses and organisms, including humans. As scientists analyze the genomic data, we are seeing major contributions to our knowledge in many areas of biology. Of course, it is natural for us to focus on the expected outcomes from studying the human genome. For example, eventually we will understand the structure and function of every gene in the human genome. Such knowledge undoubtedly will lead to a better understanding of human genetic diseases and contribute significantly to their cures. The science-fiction scenario of each of us carrying our DNA genome sequence on a chip will become reality in the near future. However, knowledge about our genomes will raise social and ethical concerns that must be resolved carefully.

Geneticists and Genetic Research

The material presented in this text is the result of an incredible amount of research done by geneticists working in many areas of biology. Geneticists use the standard methods of science in their studies. As researchers, geneticists typically use the **hypothetico-deductive method of investigation.** This consists of making *observations*, forming *hypotheses* to explain the observations, making experimental *predictions* based on the hypotheses, and finally *testing* the predictions. The last step provides new observations, producing a cycle that leads to a refinement of the hypotheses and perhaps, eventually, to the establishment of a theory that attempts to explain the original observations.

As in all other areas of scientific research, the exact path a research project will follow cannot be predicted precisely. In part, the unpredictability of research makes it exciting and motivates the scientists engaged in it. The discoveries that have revolutionized genetics typically were not planned; they developed out of research in which basic genetic principles were being examined. The work of Barbara McClintock on the inheritance of patches of color on corn kernels is an excellent example. After accumulating a large amount of data from genetic crosses, she hypothesized that the appearance of colored patches was the result of the movement (transposition) of a DNA segment from one place to another in the genome. Only many years later were these DNA segments—called *transposons* or *transposable elements*—isolated and characterized in detail. We know now that transposons are ubiquitous, playing a role not only in the evolution of species but also in some human diseases.

The Subdisciplines of Genetics

Geneticists often divide genetics into four major subdisciplines:

1. **Transmission genetics** (sometimes called classical genetics) is the subdiscipline dealing with how genes and genetic traits are transmitted from generation to generation and how genes recombine (exchange between chromosomes). Analyzing the pattern of trait transmission in a human pedigree or in crosses of experimental organisms is an example of a transmission genetics study.
2. **Molecular genetics** is the subdiscipline dealing with the molecular structure and function of genes. Analyzing the molecular events involved in the gene control of cell division, or the regulation of expression of all the genes in a genome, are examples of molecular genetics studies. Genomic analysis is part of molecular genetics.
3. **Population genetics** is the subdiscipline that studies heredity in groups of individuals for traits that are determined by one or only a few genes. Analyzing the frequency of a disease-causing gene in the human population is an example of a population genetics study.
4. **Quantitative genetics** also considers the heredity of traits in groups of individuals, but the traits of concern are determined by many genes simultaneously. Analyzing the fruit weight and crop yield in agricultural plants are examples of quantitative genetics studies.

Although these subdisciplines help us think about genes from different perspectives, there are no sharp boundaries between them. Increasingly, for example, population and quantitative geneticists analyze molecular data to determine gene frequencies in large groups. Historically, transmission genetics developed first, followed by population genetics and quantitative genetics, and then molecular genetics.

Genes influence all aspects of an organism's life. Understanding transmission genetics, population genetics, and quantitative genetics will help you understand population biology, ecology, evolution, and animal behavior. Similarly, understanding molecular genetics is useful when you study such topics as neurobiology, cell biology, developmental biology, animal physiology, plant physiology, immunology, and, of course, the structure and function of genomes.

Basic and Applied Research

Genetics research, and scientific research in general, may be either basic or applied. In **basic research,** experiments are done to gain an understanding of fundamental

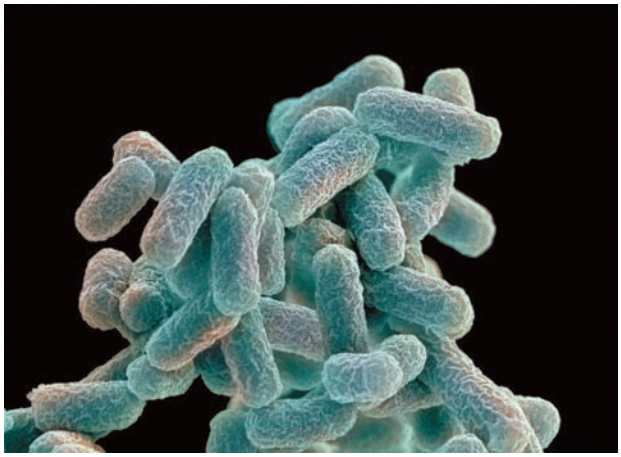
phenomena, whether or not the knowledge gained leads to any immediate applications. Basic research was responsible for most of the facts we discuss in this text. For example, we know how the expression of many prokaryotic and eukaryotic genes is regulated as a result of basic research on model organisms such as the bacterium *Escherichia coli* (*E. coli*) ("esh-uh-REEK-e-uh CO-lie," shown in Figure 1), the yeast *Saccharomyces cerevisiae* ("sack-a-row-MY-seas serry-VEE-see-eye," shown in Figure 4a), and the fruit fly *Drosophila melanogaster* ("dra-SOFF-ee-la muh-LANO-gas-ter," shown in Figure 4b). The knowledge obtained from basic research is used largely to fuel more basic research.

In **applied research,** experiments are done with different goals in mind; namely, with an eye toward overcoming specific problems in society or exploiting discoveries. In agriculture, applied genetics has contributed significantly to improvements in animals bred for food (such as reducing the amount of fat in beef and pork) and in crop plants (such as increasing the amount of protein in soybeans). A number of diseases are caused by genetic defects, and great strides are being made in diagnosis and understanding the molecular bases of some of those diseases. For example, drawing on knowledge gained from basic research, applied genetic research involves developing rapid diagnostic tests for genetic diseases and producing new pharmaceuticals for treating diseases.

There is no sharp dividing line between basic and applied research. Indeed, in both areas, researchers use similar techniques and depend on the accumulated body of information when building hypotheses. For example, **recombinant DNA technology**—procedures that allow molecular biologists to splice a DNA fragment from one organism into DNA from another organism and to clone (make many identical copies of) the new recombinant DNA molecule—has profoundly affected both basic and applied research. Many biotechnology companies owe their existence to recombinant DNA technology as they seek to clone and manipulate genes in developing their products. In the area of plant breeding, recombinant DNA technology has made it easier to introduce traits such as disease resistance from noncultivated species into cultivated species. Such crop improvement traditionally was achieved by using conventional breeding experiments. In animal breeding, recombinant DNA technology is being used in the beef, dairy, and poultry industries, for example, to increase the amount of lean meat, the amount of milk, and the number of eggs. In medicine, the results are equally impressive. Recombinant DNA technology is being used to produce a number of antibiotics, hormones, and other medically important agents such as clotting factor and human insulin (marketed under the name Humulin; Figure 2) and to diagnose and treat a number of human genetic diseases. In forensics, *DNA typing* (also called *DNA fingerprinting* or *DNA profiling*) is being used in paternity cases, criminal cases, and anthropological studies. In short, the science of genetics is currently in an exciting and dramatic growth phase, and there is still much to discover.

Figure 1

Colorized scanning electron micrograph of *Escherichia coli*, a rod-shaped bacterium common in the intestines of humans and other animals.



Keynote

Genetics can be divided into four major subdisciplines: transmission genetics, molecular genetics, population genetics, and quantitative genetics. Depending on whether the goal is to obtain a fundamental understanding of genetic phenomena or to exploit discoveries, genetic research is considered to be basic or applied, respectively.

Genetic Databases and Maps

In this section, we talk about two important resources for genetic research: genetic databases and genetic maps. Genetic databases have become much more sophisticated and expansive as computer analysis tools have been developed and Internet access to databases has become routine. Constructing genetic maps has been part of genetic analysis for about 100 years.

Figure 2

Example of a product developed as a result of recombinant DNA technology. Humulin—human insulin for insulin-dependent diabetics.

Genetic Databases. The amount of information about genetics has increased dramatically. No longer can we learn everything about genetics by going to a college or university library; the computer now plays a major role. For example, a useful way to look for genetic information through the Internet is by entering key terms into search engines such as Google (http://www.google.com). Typically, a vast number of hits are listed, some useful and some not.

There are many specific genetic databases on the Internet, too many to summarize all that are useful in this section. You must search for yourself and be critical about what you find. However, we can consider a set of important and extremely useful genetic databases at the National Center for Biotechnology Information (NCBI) website (http://www.ncbi.nlm.nih.gov). NCBI was created in 1988 as a national resource for molecular biology information. Its role is to "create public databases, conduct research in computational biology, develop software tools for analyzing genome data, and disseminate biomedical information—all for the better understanding of molecular processes affecting human health and disease."

Some of the search tools available at the NCBI site are as follows:

- PubMed is used to access literature citations and abstracts and provides links to sites with electronic versions of research journal articles. These articles can sometimes be viewed, or you must pay a one-time fee or obtain a free subscription. You search PubMed by entering terms, author names, or journal titles. It is highly recommended that you use PubMed to find research articles on genetic topics that interest you.
- OMIM (Online Mendelian Inheritance in Man) is a database of human genes and genetic disorders authored and edited by Dr. Victor A. McKusick and his colleagues. You search OMIM by entering terms in a textbox search window; the result is a list of linked pages, each with a specific OMIM entry number. The pages have detailed information about the gene or genetic disorder specified in the original search, including genetic, biochemical, and molecular data, along with an up-to-date list of references. Throughout the book, each time we discuss a human gene or genetic disease, we refer to OMIM entries and give the OMIM entry number.
- GenBank is the National Institutes of Health (NIH) genetic-sequence database. This database is an annotated collection of all the tens of billions of publicly available DNA sequences. You search GenBank by entering terms in the search window. For example, if you are interested in the human disease cystic fibrosis, enter the term *cystic fibrosis* into the search window, and you will find all sequences that have been entered into GenBank that include those two words in the annotations.
- BLAST (Basic Local Alignment Search Tool) is a tool used to compare a nucleotide sequence or protein sequence with all sequences in the database to find possible matches. This is useful, for example, if you have sequenced a new gene and want to find out whether anything similar has been sequenced previously. Moreover, genes with related functions may be listed in the databases, allowing you to focus your research on the function of the gene you are studying.
- Entrez is a system for searching several linked databases. The particular database is chosen from a pull-down menu. The databases include PubMed; Nucleotide, for the GenBank DNA and RNA sequences database; Protein, for amino acid sequences; Structure, for three-dimensional macromolecular structures; Genome, for complete genome assemblies; RefSeq, an annotated collection of genes, transcripts, and the proteins derived from the transcripts; OMIM, the Online Mendelian Inheritance in Man human gene database; and PopSet, population study datasets. The database can be selected from the hot links, or a pull-down menu choice on the main Entrez page will guide your search terms appropriately. For example, if you are interested in nucleotide sequences related to the human disease cystic fibrosis, you would select "Nucleotide" in the pull-down menu and enter *cystic fibrosis* in the search window. A list of relevant sequence entries will be returned.
- Books is a collection of biomedical books that can be searched directly. Included are some genetics, molecular biology, and developmental biology textbooks.

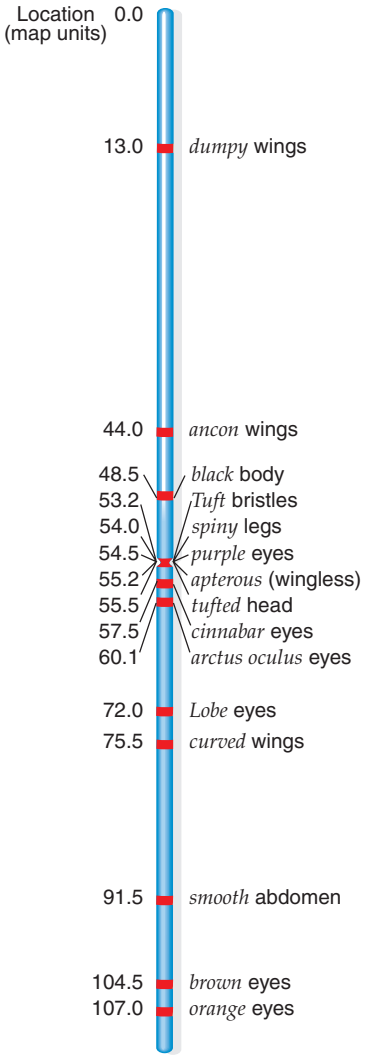
A powerful feature of the NCBI databases is that they are linked, enabling users to move smoothly between them and hence integrate the knowledge obtained in each of them. For example, a literature citation found in PubMed will have links to sequences in nucleotide and protein databases.

Genetic Maps. Since 1902, much effort has been made to construct **genetic maps** (Figure 3) for the commonly used experimental organisms in genetics. Like road maps that show the relative locations of towns along a road, genetic maps show the arrangements of genes along the chromosomes and the genetic distances between the genes. The position of a gene on the map is called a **locus** or **gene locus.** The genetic distances between genes on the same chromosome are calculated from the results of genetic crosses by counting the frequency of recombination—that is, the percentage of the time among the progeny that the genes in the two original parents exchange (i.e., recombine;). The unit of genetic distance is the **map unit** (mu)**.**

The goal of constructing genetic maps has been to obtain an understanding of the organization of genes along the chromosomes (e.g., to inform us whether genes with related functions are on the same chromosome; and if they are, whether they are close to each other). Genetic maps have also proved very useful in efforts to clone and

Figure 3

Example of a genetic map, illustrating some of the genes on chromosome 2 of the fruit fly, *Drosophila melanogaster*. The numerical values represent the positions of the genes from the chromosome end (top) measured in map units.



sequence particular genes of interest—and more recently, as part of genome projects, in efforts to obtain the complete sequences of genomes.

Keynote

Two important resources for genetic research are genetic databases and genetic maps. Databases provide the means to search for specific information about a gene, including its sequence, its function, its position in the genome, research papers written about it, and details about its product. Genetic maps show the positions of genes along a chromosome. They have proved useful in efforts to clone genes, as well as in the efforts to sequence genomes.

Organisms for Genetics Research

The principles of heredity were first established in the nineteenth century by Gregor Mendel's experiments with the garden pea. Since Mendel's time, many organisms have been used in genetic experiments. In general, the goal of the research has been to understand gene structure and function. Because of the remarkable conservation of gene function throughout evolution, scientists have realized that results obtained from studies with a particular organism typically would apply more generally. Among the qualities that historically have made an organism a particularly good model for genetic experimentation are the following:

- The organism has a short life cycle, so that a large number of generations occur within a short time. In this way, researchers can obtain data readily over many generations. Fruit flies, for example, produce offspring in 10 to 14 days.
- A mating produces a large number of offspring.
- The organism should be easy to handle. For example, hundreds of fruit flies can be kept easily in small bottles.
- Most importantly, genetic variation must exist between the individuals in the population or be created in the population by inducing mutations so that the inheritance of traits can be studied.

Both eukaryotes and prokaryotes are used in genetics research. **Eukaryotes** (meaning "true nucleus") are organisms with cells within which the genetic material (DNA) is located in the **nucleus** (a discrete structure bounded by a nuclear envelope). Eukaryotes can be unicellular or multicellular. In genetics today, a great deal of research is done with six eukaryotes (Figure 4a–f): *Saccharomyces cerevisiae* (budding yeast), *Drosophila melanogaster* (fruit fly), *Caenorhabditis elegans* ("see-no-rab-DYT-us ELL-e-gans," a nematode worm), *Arabidopsis thaliana* ("a-rab-ee-DOP-sis thal-ee-AH-na," a small weed of the mustard family), *Mus musculus* ("muss MUSS-cue-lus," a mouse), and *Homo sapiens* ("homo SAY-pee-ens," human). Humans are included although they do not meet the criteria for an organism well suited for genetic experimentation, but because ultimately we want to understand as much as we can about human genes and their function. With this understanding, we will be able to combat genetic diseases and gain fundamental knowledge about our species' development and evolution.

Over the years, research with the following seven eukaryotes has also contributed significantly to our understanding of genetics (Figure 4g–m): *Neurospora crassa* ("new-ROSS-pore-a crass-a," orange bread mold), *Tetrahymena* ("tetra-HI-me-na," a protozoan), *Paramecium* ("para-ME-see-um," a protozoan), *Chlamydomonas reinhardtii* ("clammy-da-MOAN-as rhine-HEART-ee-eye," a green alga), *Pisum sativum* ("PEA-zum sa-TIE-vum," garden pea), *Zea mays* (corn), and *Danio rerio* (zebrafish). Of these, *Tetrahymena, Paramecium, Chlamydomonas,* and *Saccharomyces* are unicellular organisms, and the rest are multicellular.