**Cytology**

**Endocytosis**: The uptake of material into cells by inclusion within an invagination of the plasma membrane; the uptake of solid material is phagocytosis, and that of dissolved material is pinocytosis.

**Exocytosis**: A type of bulk transport out of cells in which a vacuole fuses with the plasma membrane, discharging the vacuole’s contents to the outside.

**Pinocytosis**: The process of l uid uptake by endocytosis in a cell.

**Autophagy**: Intracellular materials, such as old organelles, are brought into a lysosome by a process called autophagy

**Microvilli**: (sing., microvillus) Minute projections of the plasma membrane that increase the surface area of the cell; found mainly in cells concerned with absorption or secretion, such as those lining the intestine or the kidney tubules.

**MTOC: microtubule organizing center**: The region of the cell from which microtubules are anchored and possibly assembled. The MTOCs of many organisms (including animals, but not flowering plants or most gymnosperms) contain a pair of

**centrioles**.(sentree-ohl) One of a pair of small, cylindrical organelles lying at right angles to each other near the nucleus in the cytoplasm of animal cells and certain protist and plant cells; each centriole is in the form of a cylinder composed of nine triplets of microtubules (9 3 structure).

**Lamellipodia**: extensive, lamellar cellular projections involved in attachment of eukaryotic cells such as ﬁbroblasts to solid surfaces. Lamellipodia mark the forward edges of moving cells such as macro phages and are also called rufﬂed edges.

**Filopodia**: very thin, ﬁngerlike extensions of the plasma membrane; used by cells that move by amoeboid locomotion.

**Chaperones**: eukaryotic proteins that help some nascent polypeptide chains fold correctly into their tertiary shapes, stabilizing and protecting them in the process, and/or preventing them from making premature or nonproductive intermolecular associations. Note that a chaperone forms a complex with a second protein to facilitate its folding, but chaperones are not part of the mature structure. Some of these molecular chaperones are heat-shock proteins (q.v. ). Some chaperones may bind to nascent polypeptide chains while they are being synthesized on ribosomes, and they may also help the polypeptide move out of the tunnel of 60S ribosomal subunit. Other chaperones may keep the polypeptide in an unfolded conformation as it is being translated. This facilitates subsequent passage across membranes, as when protein enters the endoplasmic reticulum or a mitochondrion. Also called chaperonins or molecular chaperones. See prions.

**Clathrin**: A protein that is the major constituent of the ‘coat’ of the coated pits and coated vesicles formed during endocytosis of materials at the surface of cells (see [endosome](http://www.oxfordreference.com/view/10.1093/acref/9780199204625.001.0001/acref-9780199204625-e-1469)). The clathrin molecules are arranged in a localized polyhedral lattice on the membrane, which subsequently invaginates to form the coated pit and vesicle. When the coated vesicle has delivered its contents within the cell, the clathrin-coated membrane is recycled and returns to the cell surface. A similar process also tranfers materials between membranous organelles within the cell.

**Cell checkpoints**: The dynamics of mitosis are similar to a state machine. In a healthy cell, checkpoints between phases, permit a new phase to begin only when the previous phase is complete and successful. At these checkpoints, gatekeeper molecules block or allow events, depending on their level of phosphorylation.

desmosomes: (dezmoh-sohmz) Buttonlike plaques, present on two opposing cell surfaces, that hold the cells together by means of protein filaments that span the intercellular space.

**Cadherins**: glycoproteins composed of 700–750 amino acids that function as cell–cell adhesion molecules. The N -terminal end of the molecule projects from the membrane surface and contains Ca2+ binding sites. The C -terminal tail binds to the actin of the cytoskeleton. In between is a segment that functions as an integral part of the cell membrane. E-cadherins are the best characterized of the cadherins. They are present in many types of epithelial cells and are usually concentrated in the **adhesion belts** that hold the cells together. See cell–cell adhesion molecules (CAMs).

**Genetics**

**Mosaic**: an individual composed of two or more cell lines of different genetic or chromosomal constitution, both cell lines being derived from the same zygote; in contrast with a chimera (q.v. ). See Appendix C, 1962, Beutler, Yeh, and Fairbanks; dosage compensation, Lyonization, tortoiseshell cat.

**Alleles** (al-leelz): Genes governing variation of the same character that occupy corresponding positions (loci) on homologous chromosomes; alternative forms of a gene.

**Allele**: a shorthand form of allelomorph, one of a series of possible alternative forms of a given gene (cistron, q.v. ), differing in DNA sequence, and affecting the functioning of a single product (RNA and/or protein). If more than two alleles have been identiﬁed in a population, the locus is said to show multiple allelism. See heteroallele, homoallele, isoallele, null allele, silent allele.

**Pseudo gene**: a gene bearing close resemblance to a known gene at a different locus, but rendered non-functional by additions or deletions in its structure that prevent normal transcription and/or translation. Pseudo genes are usually ﬂanked by direct repeats of 10 to 20 nucleotides; such direct repeats are considered to be a hallmark of DNA insertion. Two classes of pseudo genes exist: (1) Traditional pseudo genes (as exempliﬁed in the globin gene families) appear to have originated by gene duplication and been subsequently silenced by point mutations, small insertions, and deletions; they are usually adjacent to functional copies and show evidence of being under some form of selective constraint for several millions of years after their formation. (2) Processed pseudo genes lack introns, possess a remnant of a poly-A tail, are often ﬂanked by short direct repeats, and are usually unassociated with functional copies; all of which suggests their formation by the integration into germ line DNA of a reverse-transcribed processed RNA. Processed pseudo genes are rare in yeast and Drosophila, but common in mammals. For example, in humans there are 20 pseudo genes that are believed to have arisen from actin and beta tubulin mRNAs. See Appendix C, 1977, Jacq et al.; hemoglobin genes, leprosy bacterium, orphons, processed gene.

**Dosage compensation**: a mechanism that regulates the expression of sex-linked genes that differ in dose between females and males in species with an XXXY method of sex determination. In *Drosophila melanogaster*, dosage compensation is accomplished by raising the rate of transcription of genes on the single X chromosome of males to double that of genes on either X chromosome in females. In mammals, the compensation is made by inactivating at random one of the two X chromosomes in all somatic cells of the female. The inactivated X forms the Barr body or sex chromatin. In cases where multiple X chromosomes are present all but one are in activated. See Appendix C, 1948, Muller; 1961, Lyon, Russell; 1962, Beutler, et al.; Fabry disease, glucose-6-phosphate dehydrogenase deﬁciency, LeschNyhan syndrome, Lyon hypothesis, Lyonization, mosaic, MSL proteins, ocular albinism, Ohno hypothesis.

**Supergene**: a chromosomal segment protected from crossing over and so transmitted from generation to generation as if it were a single recon.

**Apoptosis**: (pronounced “apo-tosis”) the programmed death of cells in various tissues at speciﬁc times during embryogenesis and metamorphosis or during cell turnover in adult tissues. For example, 12% of the cells formed during the development of an adult hermaphroditic *Caenorhabditis elegans* are destined to die because of a genetically controlled suicide program. If genes functioning in this system are inactivated by mutation, cells that normally die will survive. Recently the term apoptosis has been broadened to include all forms of cell death controlled by caspases (q.v.). Apoptosis as a part of normal development (programmed cell death), is now called physiologic apoptosis; whereas that occurring in diseased tissues is called aberrant apoptosis. See Appendix C, 1986, Ellis and Horvitz.

**Allopolyploid:** allopolyploid (also alloploid) a polyploid organism arising from the combination of genetically distinct chromosome sets. See isosyndetic alloploid, segmental alloploid.

**Autopolyploid**: a polyploid that originates by the multiplication of one basic set of chromosomes. See autotetraploid.

**Phenocopy**: the alteration of the phenotype, by nutritional factors or the exposure to environmental stress during development, to a form imitating that characteristically produced by a speciﬁc gene. Thus, rickets due to a lack of vitamin D would be a phenocopy of vitamin D-resistant rickets.

**Genocopy**: A genetically determined CHARACTER that mimics the appearance of another genetically determined CHARACTER, though caused by an ALLELE at a different LOCUS.

**Gynandromorph**: an individual made up of a mosaic of tissues of male and female genotypes. The fruit ﬂy illustrated on page 191 is a bilateral gynandromorph, with the right side female and the left side male. The zygote was ++/wm . Loss of the X chromosome containing the dominant (+) genes occurred at the ﬁrst nuclear division. The cell with the single X chromosome containing the recessive marker genes gave rise to the male tissues. Therefore, the left eye is white and the left wing is miniature. Note the male abdominal pigmentation and the sex comb.

**Centimorgan (Morgan unit)**: a unit for expressing the relative distance between genes on a chromosome. One morgan (M) equals a crossover value of 100%. A crossover value of 10% is a decimorgan (dM); 1% is a centimorgan (cM); named in honor of Thomas Hunt Morgan.

**Barr body**: the condensed single X chromosome seen in the nuclei of somatic cells of female mammals. See Appendix C, 1949, Barr and Bertram; dosage compensation, drumstick, late replicating X chromosome, Lyon hypothesis, sex chromatin.

**cDNA (copy DNA):** single-stranded, complementary DNA produced from an RNA template by the action of RNA-dependent, DNA polymerase (re verse transcriptase) in vitro. If the RNA template has been processed to remove the introns, the cDNA will be much shorter than the gene from which the RNA was transcribed. The single-stranded, cDNA molecule may subsequently serve as a template for a DNA polymerase. The symbol cDNA is sometimes also applied to the double-stranded DNA molecule that results. See posttranscriptional processing. gene imprinting

**Isochromosome**: a metacentric chromosome produced during mitosis or meiosis when the centromere splits transversely instead of longitudinally. The arms of such a chromosome are equal in length and genetically identical. However, the loci are positioned in reverse sequence in the two arms.

**Bioremediation**: bioremediation the use of living organisms (e.g., bacteria, fungi, and other microorganisms) to reclaim contaminated environmental sites, such as soil, groundwater, or lakes. This is achieved by enhancing the natural ability of microbes already present at the site to break down contaminating molecules. Alternatively, such microbes are added to the affected site. See Pseudomonas, Streptomyces.

**Phytoremediation**: a process of decontaminating soil or water by using plants and trees to absorb or break down pollutants.

**Epistasis**: epistasis the nonreciprocal interaction of non-allelic genes. The situation in which one gene masks the expression of another. The recessive gene apterous (ap )in Drosophila produces wingless homozygotes. In such individuals, any other recessive gene affecting wing morphology will have its action masked. The apterous gene is said to be epistatic to a gene like curled wing, which is hypostatic to ap. See Appendix C, 1900, Bateson; Bombay blood group.

**Eugenics**: the improvement of humanity by altering its genetic composition by encouraging breeding of those presumed to have desirable genes (positive eugenics), and discouraging breeding of those presumed to have undesirable genes (negative eugenics). The term was coined by Francis Galton.

**Phenotype:** the observable characteristics of a cell or an organism, such as its size and shape, its metabolic functions, and its behavior. The genotype (q.v. ) is the underlying basis of the phenotype, and the term is commonly used to describe the effect a particular gene produces, in comparison with its mutant alleles. Some genes control the behavior of the organism, which in turn generates an artefact outside the body. R. Dawkins uses the term extended phenotype to refer to the production of such an artefact (spider webs, bird nests, and beaver dams are examples). See Appendix C, 1909, Johannsen.

**Genotype**: genotype the genetic constitution of a cell or an organism, as distinguished from its physical and behavioral characteristics, i.e., its phenotype (q.v. ). See Appendix C, 1909, Johannsen.

**Junk** **DNA** (also called selﬁsh DNA or parasitic DNA) 1. Functionless segments of DNA that are replicated along with the rest of the chromosomal regions that serve vital functions. Examples would be pseudogenes (q.v.) and tandemly repeated and dispersed repetitive DNA segments that appear to serve no function, yet accumulate by unequal crossing over (q.v. ). 2. The term is also used to refer to a parasitic DNA that has the ability to engineer its host genetically so that the host cell is better able to survive in nature. Examples would be R (resistance) plasmids and Ti plasmids (both of which see). See Appendix C, 1980, Doolittle and Sapienza, Orgel and Crick; 1997, Yoder,Walsh,andBestor;C value paradox, DNA methylation, repetitious DNA.

**Pleiotropy**: the phenomenon in which a single gene is responsible for a number of distinct and seemingly unrelated phenotypic effects.

**Holandric**: Occurring only in males, i.e. a GENE that is on the Y CHROMOSOME.

**IsoAllele**: an allele whose effect can only be distinguished from that of the normal allele by special tests. For example, two + alleles +1 and +2 may be indistinguishable (i.e., +1 /+1 , +2 /+2 , and +2 /+1 individuals are phenotypically wild type). However, when compounded with a mutant allele a , +1 and +2 prove to be distinguishable (i.e., a/+1 and a/+2 individuals are observably different).

**Replicon**: replicon a genetic element that behaves as an autonomous unit during DNA replication. In bacteria, the chromosome functions as a single replicon, whereas eukaryotic chromosomes contain hundreds of replicons in series. Each replicon contains a segment to which a speciﬁc RNA polymerase binds and a replicator locus at which DNA replication commences. The polymerase makes an RNA primer called an initiator. See Appendix C, 1963, Jacob and Brenner; 1968, Huberman and Riggs; autonomously replicating sequences, DNA ﬁber autoradiography, *ori* site, primase.