Quick Study. Academic

BarCharts

Evolution, cytology, energy & life, cell transport, cell reproduction, organismal reproduction & meiosis, genetics & Mendel, molecular & population genetics

WORLD'S #1 ACADEMIC OUTLINE

BASIC CONCEPTS

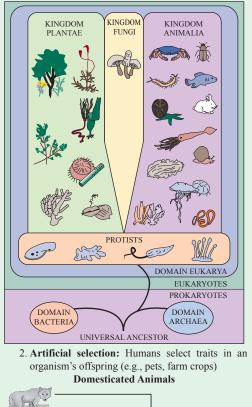
- 1. **Biology** is the study of life
- 2. The characteristics of life are metabolism, reproduction, growth, movement, responsiveness, and complex organization
- 3. **The scientific method:** How scientists study biology A. Based on the observation of phenomena to formulate hypotheses that are both testable and falsifiable (in case they are wrong)
 - B. Used to test hypotheses, collect data, and analyze data statistically (if necessary)

EVOLUTION

Evolution is the concept that all organisms are related to each other by common ancestry; it is the unifying theme in biology

- 1. **Natural selection:** A mechanism, formulated by 19thcentury biologists Charles Darwin and Alfred Wallace, for the occurrence of evolution based on the survival of those offspring best adapted to the conditions in which they live
 - A. Individuals sexually reproduce, creating many more offspring than could possibly survive
 - B. These offspring are not identical (in most situations) but show variations based on genetic differences
 - C. Essentially, those individuals with variations that allow them to survive (i.e., **adaptations**) to the age of reproduction can pass their genes on to the next generation
 - D. Thus, nature is selecting offspring and shaping the evolution of species

Organismal Evolution

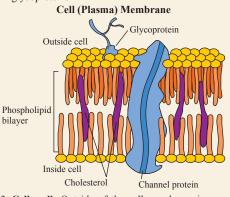


CYTOLOGY: THE STUDY OF CELLS

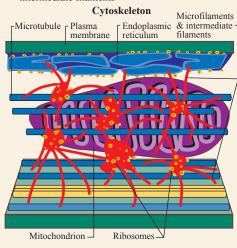
Cell Theory

All living things are composed of cells and come from cells

- 1. Cell size: Small to maximize the ratio of surface area to volume for regulating the internal cell environment
- Cell (plasma) membrane: Composed of a fluidlike phospholipid bilayer, proteins, cholesterol, and glycoproteins

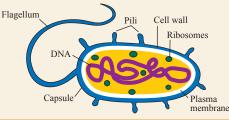


- 3. **Cell wall:** Outside of the cell membrane in some organisms; composed of carbohydrates (e.g., cellulose for plants; chitin for fungi) or carbohydrate derivatives (e.g., peptidoglycan for bacteria)
- 4. **Cytoplasm:** Material outside the nucleus A. Site for metabolic activity
 - B. **Cytosol:** Solution with dissolved substances such as glucose, CO₂, O₂, etc.
 - C. Organelles: Membrane-bound subunits of cells Centrioles with specialized functions
- Cytoskeleton: Supportive and metabolic structure composed of microtubules, microfilaments, and intermediate filaments



Prokaryotic Cells

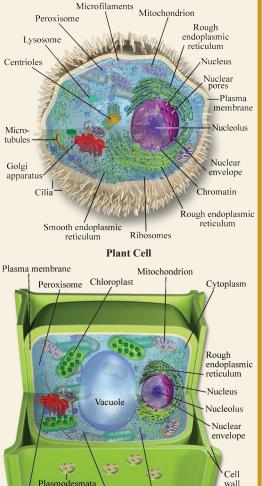
Prokaryotes have a simple cellular organization with no nucleus or other membrane-bound organelles



Eukaryotic Cells

Eukaryotes have a complex cellular organization; membrane-bound organelles, located inside the cell membrane, include the following:

- 1. Nucleus: Contains DNA in the form of chromosomes; controls cellular activities via genes
- 2. **Nucleolus:** Located within the nucleus; site for ribosome synthesis
- 3. Rough endoplasmic reticulum: Has ribosomes, which are necessary for protein synthesis
- 4. **Smooth endoplasmic reticulum:** Involved primarily in lipid synthesis, as it lacks ribosomes
- 5. Golgi apparatus: Packaging center for molecules; synthesizes carbohydrates
- 6. Lysosome: Contains hydrolytic enzymes for intracellular digestion
- 7. **Peroxisome:** Involved in hydrogen peroxide synthesis and degradation
- 8. Chloroplast: Site of photosynthesis
- 9. Chromoplast: Contains nongreen pigments
- 10. Leukoplast: Stores starch
- 11. **Mitochondrion:** Produces adenosine triphosphate (ATP)
- 12. Vacuole: General storage and space-filling structure Animal Cell



Ribosomes

Smooth endoplasmic reticulum

Golgi apparatus

QuickStudy. ENERGY & LIFE

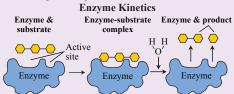
Energy & Cells

- 1. Energy can be generally categorized relative to its passive or active state
 - A. Potential energy is stored for subsequent use to do work
 - B. Kinetic energy is in action or actively doing some kind of work as a result of motion
- 2. Energy can also be categorized based on specific types, including nuclear, magnetic, electrical, radiant, chemical, and thermal (heat)
 - A. Laws of natural physics dictate that when conversions between any specific kinds of energy occur, thermal energy will be involved, even if unintended
 - B. For example, if radiant energy is converted into stored chemical energy (e.g., potential energy as a result of photosynthesis), heat will be produced
- 3. Laws of thermodynamics: The association between heat and all other energy forms is the foundation of the following natural laws:
 - A. First law (conservation of energy): The total energy in any closed system (i.e., energy cannot enter or exit) is always constant, but energy can change from one type to another; thus, energy cannot be created or destroyed
 - B. Second law (increased entropy): The free or potential energy to do work in a closed system decreases, whereas entropy or disorder increases; thus, no energy transfer is 100% efficient in doing work (e.g., ATP production during cell respiration will lose free energy in the form of heat)

4. Earth is an open system

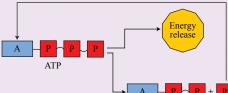
- A. Radiant energy from the Sun supplies organisms with renewable energy to battle entropy and remain alive and highly organized
- B. Photosynthetic organisms (producers) convert radiant energy directly into organic molecules (i.e., chemical energy), which are eaten by herbivores (primary consumers), which in turn are eaten by carnivores (secondary consumers)

- 5. Metabolism: Energy transfers within organisms are based on sequences (metabolic pathways) of chemical reactions involved in releasing (catabolic or exergonic) or absorbing (anabolic or endergonic) free energy
- 6. Enzymes: Biological catalysts; facilitate metabolic chemical reactions by speeding up reaction rates and lowering heat requirements



7. Adenosine triphosphate (ATP): A high-energy molecule; energy stored in ATP is released by breaking phosphate-to-phosphate bonds and creating adenosine diphosphate (ADP) or adenosine monophosphate (AMP); ATP is recycled by adding back phosphate groups using energy from the Sun

Energy & ATP



Photosynthesis

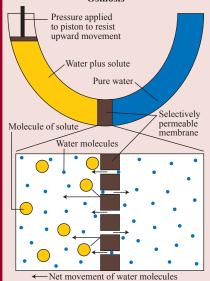
Sunlight or radiant energy is captured by chlorophyll Glucose and carotenoid photopigments (found in cytoplasm in prokaryotes and chloroplasts in eukaryotes) in two main steps

1. Light-dependent reactions (light reactions): The captured light energy is transferred to electrons that come from H₂O; O₂ is a by-product

Passive Transport

of matter; the cell does not do work; there are four categories:

- 1. Diffusion: Movement from an area of high concentration to one of low concentration
- 2. Facilitated diffusion: A permease, or membrane enzyme, carries a substance
- 3. Osmosis: Diffusion across a semipermeable membrane
- 4. Bulk flow: Mass movements of fluids affected by pressure and solutes Osmosis



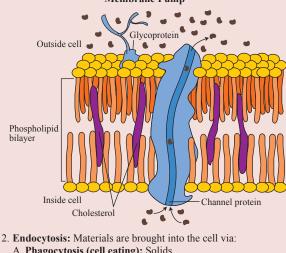
Active Transport

CELL TRANSPORT

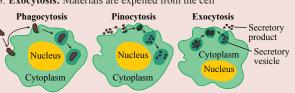
Passive transport relies on the thermal energy Active transport relies on the cell to provide the energy supply to move materials; there are three categories:

1. Membrane pumps: ATP is required; a permease is used to move a substance, usually in the opposite direction of diffusion

Membrane Pump



- A. Phagocytosis (cell eating): Solids
- B. Pinocytosis (cell drinking): Liquids
- 3. Exocytosis: Materials are expelled from the cell



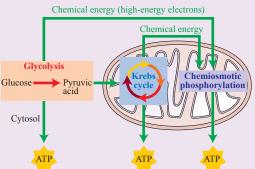
2. Light-independent reactions (dark reactions): Energized electrons are transferred to CO₂ (reduction reactions) to form glucose (in the Calvin cycle)

Cell Respiration

Highly energized electrons stored temporarily in glucose are removed (oxidation reactions) in a stepwise fashion to maximize the energy captured at each step:

- 1. Glycolysis: Anaerobic process in cytoplasm in which glucose, a six-carbon compound, is oxidized to two pyruvate molecules, which are both three-carbon chains
- 2. Krebs cycle: Aerobic process that oxidizes pyruvate molecules to CO.
- 3. Chemiosmotic phosphorylation: The energized electrons released during the previous steps are used to concentrate hydrogen ions in one area (of the cell membrane in prokaryotes; of the mitochondrion in eukaryotes) to create a chemical gradient between positively and negatively charged ions (like a battery); the potential energy resulting from this osmotic gradient is used to resynthesize ATP from ADP and AMP; after electrons have been used, they must be transferred to O₂

Cell Respiration



CELL REPRODUCTION

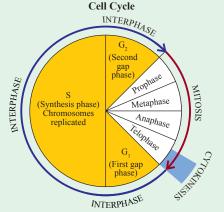
The Two Steps to Cell Reproduction

- 1. Mitosis: Division of nuclear material 2. Cytokinesis: Division of remaining cellular
- contents of the cytoplasm

The Cell Cycle

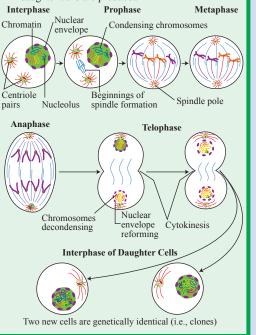
Cells go through four stages during cell reproduction:

- 1. G1: Active growth and metabolism
- 2. S: DNA synthesis and duplication
- 3. G.: Synthesis of molecules in preparation for cell division
 - A. Stages G₁, S, and G₂ are collectively referred to as interphase
 - B. Interphase chromosomes are referred to as chromatin, a diffuse, loosely scattered arrangement of chromosomes
- 4. Mitosis and cytokinesis: Mitotic chromosomes in the mitosis-cytokinesis stage are highly condensed and coiled and thus distinct



The Four Stages of Mitosis

- Prophase: Chromosomes condense and organize; nuclear membrane and nucleoli disappear; spindle apparatus is assembled and attaches to centromeres of duplicated chromosomes
- 2. **Metaphase:** Spindles line up duplicated chromosomes along equator of cell, one spindle to each half, or chromatid, of duplicated chromosome
- 3. **Anaphase:** The centromere of each duplicated chromosome is separated, and paired chromatids are pulled apart
- Telophase: Chromosomes uncoil, nucleoli reappear, cytokinesis occurs, and two genetically identical daughter cells are produced



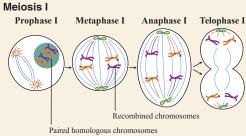
ORGANISMAL REPRODUCTION & MEIOSIS

Sexual Processes

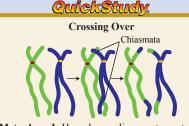
- 1. **Sexual reproduction** involves the fusion of genetic material (gametes) from two parental organisms
- 2. To ensure the proper chromosomal numbers in the **zygote** (fertilized egg), each gamete must have **haploid**, or half (N, or one set of chromosomes), of the original **diploid** (2N, or two sets of chromosomes) amount of DNA
- 3. **Meiosis:** The process by which the chromosome number is reduced by half, resulting in new genetic combinations in the gametes

The Two Stages of Meiosis

Meiosis is preceded by interphase; many meiotic events are similar to those of mitosis; the following points note the differences

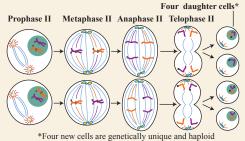


 Prophase I: Chromosomes condense and organize; matched, or homologous, chromosomes (one maternal and one paternal in each pair) are physically paired; segments of chromatids can cross over at breakage points called chiasmata within each chromosome pair



- 2. Metaphase I: Homologues line up at equator
- 3. Anaphase I: Homologues are separated into two groups, with each group having a mixture of maternal and paternal chromosomes
- 4. **Telophase I:** New haploid nuclei form for two new daughter cells
- Interkinesis: No replication of DNA occurs because each chromosome is still duplicated and consists of two chromatids (although crossing over results in some chromatids with maternal and paternal segments)

Meiosis II

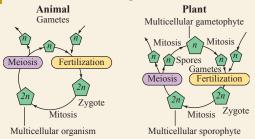


- 1. Prophase II: Chromosomes condense
- 2. Metaphase II: Chromosomes line up at equator
- Anaphase II: Chromatids of each chromosome are separated
- 4. **Telophase II:** Each daughter cell from meiosis I will form two more cells for a total of four cells

Faunal & Floral Gametogenesis

- 1. In animals, meiosis occurs in germinal tissues and is called **spermatogenesis** in males and **oogenesis** in females; each results in gametes
- 2. In plants, the process is similar, except that more mitotic divisions may follow meiosis to produce gametes

Gametogenesis

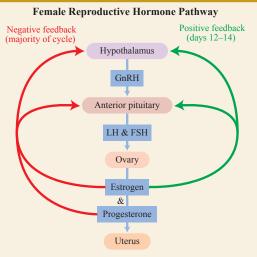


Reproductive Cycles & Hormones in Animals

Many animals, including humans, regulate the production of gametes and sexual behavior based on the presence of and complex interplay between circulating hormones that cycle based on monthly and seasonal rhythms

Reproductive Control in the Human Female 1. Hormones

- A. At puberty, the hypothalamus releases gonadotropin-releasing hormone (GnRH); this stimulates the anterior pituitary to release folliclestimulating hormone (FSH) and luteinizing hormone (LH)
- B. FSH and LH (but primarily FSH) trigger ovarian structures called follicles to grow and produce an **oocyte** (potential egg), as well as **estrogen** and **progesterone**
- C. Estrogen and progesterone have major effects on the morphology and physiology of the uterus in anticipation that ovulation and subsequent sexual intercourse will result in a successful fertilization



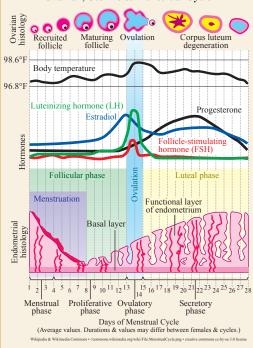
2. Ovarian cycle

- A. Under the stimulation of FSH and LH, some (but not all) follicles grow at different rates (during the **follicular phase**), with the **Graafian follicle** maturing the fastest; as the follicles grow, they also produce estrogen, with the Graafian follicle producing the most
- B. Estrogen has a negative feedback role on the brain (both the hypothalamus and anterior pituitary) for most of the monthly cycle, thereby keeping FSH and LH levels relatively low; however, continually rising levels of estrogen from growing follicles trigger a positive feedback role that causes a temporary surge in FSH and LH, with the latter hormone spiking the most
- C. This surge in LH triggers ovulation, in which the Graafian follicle bursts at the surface of the ovary and releases a **secondary oocyte** covered in associated follicular tissues (which will later become a barrier through which sperm must pass to contact the oocyte)
- D. The remnants of the Graafian follicle now function as a **corpus luteum** (during the **luteal phase**), partly to prevent any additional follicles from developing and releasing potential eggs by combining estrogen production with the additional hormone progesterone (many birth control pills use this same hormonal strategy to prevent unwanted pregnancies)
- E. The corpus luteum will eventually disintegrate, unless fertilization and implantation subsequently occur

3. Uterine and menstrual cycle

- A. Estrogen and progesterone from the ovaries stimulate the uterus to prepare for a potential embryo; specifically, the internal lining of the uterus (endometrium) is partially shed at the beginning of each cycle (menstrual flow phase or "period") if there is no successful fertilization and subsequent implantation; increasing estrogen (from growing follicles) stimulates a thickening of the endometrium (proliferative phase)
- B. After ovulation, increasing progesterone levels additionally cause the endometrium to become highly vascularized and to secrete nutrients (secretory phase) for embryo development; the disintegrating corpus luteum, and the concomitant drop in estrogen and progesterone, causes vasoconstriction of endometrial blood vessels, eventually leading to a shedding of tissue and blood
- C. If fertilization and implantation do occur during the secretory phase and right after ovulation, the subsequent implanted embryo releases a hormone called **human chorionic gonadotropin (HCG)** into the maternal circulation that helps maintain both the corpus luteum and its secretion of the two primary steroid hormones, estrogen and progesterone
- D. The maintenance of the corpus luteum allows the endometrium to continue to develop and work with the **placenta** of the embryo, while also preventing further follicle development and egg release

Ovarian, Uterine & Menstrual Cycle

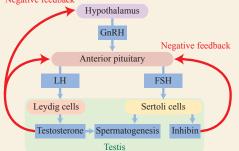


- E. After several months, the placenta takes over directly for the production of estrogen and progesterone for the duration of the pregnancy
 - i. **Menopause:** Human females typically ovulate for a finite period, after which they also cease having a menstrual cycle
 - ii. Estrous cycle: The endometrial lining thickens in all mammals; however, in most nonhuman and nonprimate animals, the endometrium is not shed but rather reabsorbed if fertilization and implantation do not occur; behaviorally, females are only receptive to a male for reproduction during estrous (a.k.a. "heat"), which is the time associated with their uterus being ready for implanting an embryo; the estrous cycle may occur seasonally or yearly in some animals (e.g., bears) or much more frequently in others (e.g., rats)

Reproductive Control in the Human Male

- 1. At puberty the hypothalamus releases GnRH, which stimulates the anterior pituitary to release FSH and LH (called **interstitial cell-stimulating hormone [ICSH]** in males)
- FSH stimulates spermatogenesis through the activity of Sertoli cells inside seminiferous tubules located in the testes
- 3. LH (ICSH) stimulates testosterone production by **Leydig cells**, which are positioned between neighboring seminiferous tubules
 - A. Human males can normally produce functional sperm throughout their entire lives, although men generally produce less testosterone and sperm as they age, likely because of a variety of healthrelated issues
- 4. Negative feedback involving testosterone and **inhibin** (produced by Sertoli cells) regulates overall hormone levels

Male Reproductive Hormone Pathway Negative feedback

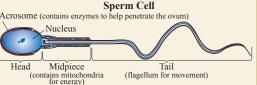


Fertilization & Development in Animals

The gametes produced during spermatogenesis and oogenesis are then used during fertilization to produce offspring

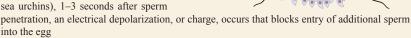
QuickStudy

 A sperm's function is to find and penetrate an egg; sperm are much smaller than eggs and include the following structures: A. Head, which contains:



Egg Fertilization

- i. Acrosome: Contains enzymes necessary for fertilization; a high sperm count is required for the minimum, collective amount of enzyme to break through physical barriers surrounding the potential egg so that a single sperm can be involved in actual fertilization
- ii. Nucleus: Contains one complete set of DNA (i.e., haploid) from the male parent
- B. **Midpiece:** Contains tightly packed mitochondria to make ATP for propelling sperm toward the egg C. **Tail:** The flagellum that propels sperm
- An egg's function is to attract and receive sperm; eggs are much larger than sperm and include the following external structures through which sperm must penetrate for successful fertilization to occur:
 A. Corona radiata: Outer layer of attached follicular cells
 - B. Zona pellucida: Middle layer essential for sperm bonding and acrosomal activity
- C. Vitelline membrane: Present in many nonmammalian animals; similar function to zona pellucida
- 3. Fertilization: Sperm and egg combine
 - A. Initial stages before contact by sperm i. In many animals, including humans, chemical signals attract sperm to the egg
 - ii. Acrosome reactions prepare for a sperm's entry into the oocyte
 - B. Final stages after contact by sperm
 - i. **Polyspermy prevention** (i.e., allowing only one sperm to enter the oocyte)
 - (a) **Fast block:** In some animals (e.g.,



Corona radiata

Zona pellucida

Secondary oocyte

First polar bod

- (b) Slow block: In many animals, entry of the first sperm triggers the formation of a barrier layer called the fertilization membrane directly outside the plasma membrane of the oocyte; the timing of completion of the slow block can vary from less than a minute to up to an hour (e.g., mammals are typically slow)
- ii. The penetrating sperm's head breaks off inside the oocyte, delivering one full set of chromosomes (i.e., haploid) from the male; the sperm nucleus is now called the **male pronucleus**
- iii. The penetrating sperm triggers the oocyte to complete the second meiotic division and produce the actual ovum (and a nonfunctional polar body); at this point, the male pronucleus resides in the cytoplasm of the ovum with the **female pronucleus**
- iv. Both pronuclei replicate their DNA and then fuse to complete fertilization to form the diploid zygote
- 4. Cleavage divisions: With the replicated DNA, the first cell divisions can now occur; in almost all animals there is an urgency to produce many cells rapidly; thus, the relatively large zygote cell is "cleaved" into numerous small cells
- 5. Embryonic stages: Most animals go through the following sequentially named stages:
 - A. Morula: Solid ball of cells
 - B. Blastula: Hollow ball of cells
 - C. Gastrula: A tube grows into the blastula (a process called gastrulation) that will become the first digestive tract (archenteron) lined with endoderm; the outside is now the ectoderm; a middle layer called the mesoderm will develop in most animals; at this point, the tissues and organs of the adult form and begin to take shape (morphogenesis)

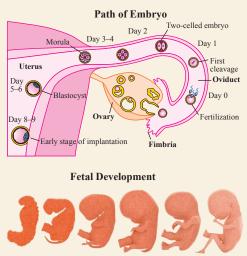


- 6. Embryo-parental relationships: Fertilized eggs vary in their physiological connection to and development within the female parent
 - A. **Oviparity:** Female lays fertilized eggs outside the body; the eggs continue to develop inside protective coverings (e.g., the hard shells of birds) using available nutrients directly available inside; embryos then hatch while outside the female's body
 - B. Ovoviviparity: Female produces eggs that are fertilized and surrounded with protective coverings using available nutrients directly inside; embryos hatch from egg coverings while still inside the oviducts of the female's body; embryo eventually exits the female during birth
 - C. **Viviparity:** Female produces eggs that are fertilized and subsequently nourished inside the uterus of the female's body; embryo eventually exits the female during birth
 - i. **Histotrophic viviparity:** Embryos consume eggs or other embryos (e.g., intrauterine cannibalism in some shark species) for nutrients
 - ii. Hemotrophic viviparity: Embryos consume nutrients typically provided via a placental connection with the female parent's blood supply
- 7. **Human embryology:** The blastula stage in humans is called the **blastocyst**, which is responsible for implanting (i.e., attaching) to the endometrium lining of the uterus; subsequently, the placenta will form to gain nutritional support from the mother's blood supply

ORGANISMAL REPRODUCTION & MEIOSIS (continued)

- A. Early human embryos closely resemble other vertebrates at similar developmental stages
- B. After about 8–10 weeks, the embryo will more closely resemble the form of the adult and is called a **fetus**; **gestation** (i.e., development inside the mother) is about 280 days or 9 months (from the time of the beginning of the cycle in which ovulation occurred) and is typically divided into three trimesters
- 8. **Birth (parturition):** Can be divided into three major events in humans
 - A. **Dilation of cervix:** The inferior opening to the uterus must dilate to accommodate the circumference of the baby's head
 - B. **Expulsion of fetus:** Normally, the head will pass through the cervix and vagina first; breech births involving the opposite end of the fetus are often problematic and may require surgical delivery via **Caesarean section**

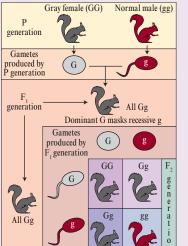
- C. **Expulsion of placenta (afterbirth):** The membranous support system maintaining the fetus is eventually (typically within 30 minutes of fetal expulsion) released from the endometrium and passes through the cervix and vagina
- 9. Mammalian embryology: Classification of mammals is based on developmental differences in embryos
 - A. **Monotremes** (e.g., platypus and echidnas) lay eggs (i.e., oviparity)
 - B. **Marsupials** (e.g., kangaroos, opossums, koalas) have live births with embryonic nutrition provided through a placenta (i.e., hemotrophic viviparity); however, the embryo is born in an extremely underdeveloped state; typically, the embryo subsequently crawls into a special pouch, or **marsupium**, on the mother where further embryonic development occurs while the embryo nurses from mammary glands
 - C. **Placentals** (most mammals, including humans) have live births after embryos develop using nutrients from a more complex placenta than marsupials; additionally, placental mammal gestation periods are typically much longer than those of marsupials



GENETICS & MENDEL

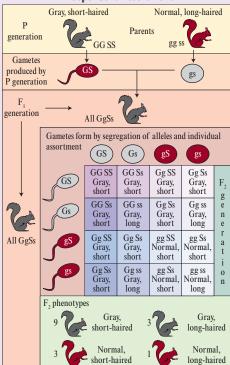
- 1. Genetics: The study of traits and their inheritance
- 2. 19th-century biologists believed that traits blended; if blending occurred, things would become more similar, not different; Darwin and Wallace stated that variations or differences in offspring were necessary for natural selection to occur
- 3. Gregor Mendel provided the most plausible hypothesis for genetics, known as **Mendelian genetics**, which is based on two laws developed by using statistics to analyze results of crosses involving distinguishing traits of garden peas
 - A. Mendel's first law, the **law of segregation of alternate factors**, was developed using single-trait crosses
 - Two true-breeding (i.e., those that consistently yield the same form when crossed with each other) parents (P₁) with different strains were crossed (e.g., roundversus wrinkled-seed-producing plants)
 - ii. The offspring (F₁) from this cross all showed only one trait (e.g., a round seed), and this was called the dominant trait; thus, the traits from the parents did *not* blend
 - iii. The F₁ individuals were crossed with each other to produce F₂ individuals
 - iv. $\frac{3}{4}$ of the F₂ individuals expressed the dominant trait, whereas $\frac{1}{4}$ expressed the trait of the other P₁ parent (e.g., a wrinkled seed) that had not been expressed in the F₁ generation and was thus recessive (*see* figure)

Segregation of Alternate Factors



- v. Mendel's first conclusions: Discrete factors (now known as genes) were responsible for the traits, and these factors were paired, separated (during meiosis), and recombined (during fertilization); alternate forms of factors or genes exist called alleles; the F₁ individuals had two alleles, as their genotype (i.e., specific allele types present for each gene) consisted of a dominant and recessive allele (e.g., Rr, with R for round and r for wrinkled seed); thus, the F₁ individuals were hybrids, as their phenotype (i.e., physical characteristics or traits) was similar to only one of the original parents (e.g., round seed)
- B. Mendel's second law, the **law of independent** assortment, was developed using multiple-trait crosses
 - i. Two true-breeding parents of different strains for two traits were crossed; the F₁ individuals were then crossed, producing F₂ individuals (*see* figure)

Independent Assortment



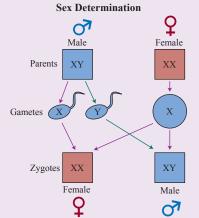
Mendel concluded statistically that these results occurred because alleles for one trait or gene did not affect the inheritance of alleles for another trait

Mendel Updated

- Genes are found on chromosomes, and thus multiple traits assort independently as long as they are located on different chromosomes; Mendel studied traits in peas that were each on separate chromosomes; genes on the same chromosome are linked and thus will not normally assort independently
- 2. Interactions between alleles
 - A. Complete dominance: One allele dominates another allele
 - B. **Incomplete dominance:** Neither allele is expressed fully
 - C. Codominance: Both alleles are expressed fully
 - D. Multiple alleles: More than two alleles for a
 - gene are found within a population E. **Epistasis:** One gene alters the affect of another gene
 - F. **Polygenic inheritance:** Many genes contribute to a phenotype
 - G. **Pleiotropy:** One gene can effect several phenotypes
 - H. Environmental influences: The genotype and environment interact to form a phenotype

Chromosomes & Sex Determination

- In many animals, special chromosomes determine sex and are called **allosomes**; the remaining chromosomes are called **autosomes**
- 2. In humans, there are 44 autosomes and 2 sex chromosomes: X and Y in males, X and X in females



Sex-Linked Traits

In humans, the Y chromosome contains the determinant for maleness; the X contains many genes; if a male gets a recessive (or dominant) allele on the X chromosome from his mother, he will express the trait; therefore, males are frequently afflicted with X-linked disorders

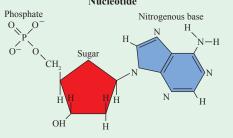
<u>QuickStudy</u>

MOLECULAR GENETICS

Genes, DNA & Nucleic Acids

1. Gene functions

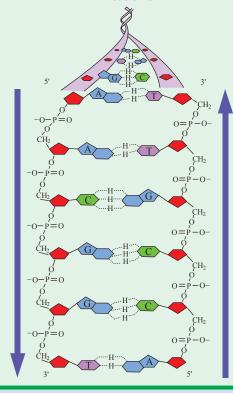
- A. To be preserved and transmitted
- B. To control various biological functions through the production of proteins (i.e., large, complex sequences of amino acids) and RNA
- 2. Gene structure is based on two nucleic acids:
 - A. Deoxyribonucleic acid (DNA)
 - B. Ribonucleic acid (RNA)
- 3. Nucleotides: The components of nucleic acids; made of three subunits: Nucleotide



A. Sugar (deoxyribose in DNA; ribose in RNA)

B. Phosphate

- C. Nitrogenous base (one of five possible bases)
 - i. In DNA, the nucleic acid of chromosomes, four nitrogenous bases are found: adenine (A), guanine (G), cytosine (C), and thymine (T)
 - ii. RNA consists of similar bases, except uracil (U) replaces thymine (T)
 - iii. DNA is a double helix molecule; that is, it is similar to a spiral staircase or twisted ladder, with the sides formed by repeating sugar-phosphate groups from each nucleotide, and the horizontal portions (i.e., steps) formed by hydrogen bonds involving A with T or C with G
 - iv. Hereditary information: Genes found along the linear sequence of nucleotides in the DNA molecule **DNA Double Helix**

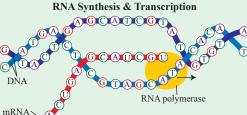


The Central Dogma

1. Replication: DNA is copied from other DNA by unzipping the helix and pairing new nucleotides with the proper bases (i.e., A with T and C with G) on each separated side of the original DNA

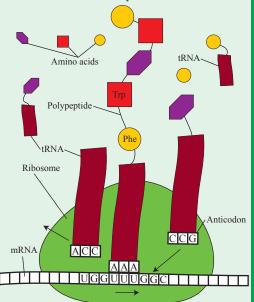
2. Transcription

- A. Messenger RNA (mRNA) is copied from DNA by unzipping a portion of the DNA helix that corresponds to a gene
- B. Only one side of the DNA will be transcribed, and nucleotides with the proper bases (A with U and C with G) will be sequenced to build pre-mRNA
- C. Sequences of nucleotides called introns are removed, and the remaining segments called exons are spliced together
- D. The mature mRNA leaves the nucleus to be translated by the ribosomes



3. Translation

- A. Proteins are synthesized from mRNA by ribosomes (which are composed of ribosomal RNA, or rRNA, and proteins), which read from a universal triplet code (i.e., codons)
- B. The ribosomes instruct transfer RNAs (tRNAs) to bring specific amino acids in the sequence dictated by the mRNA, which in turn was built based on the sequence of nucleotides in the original gene portion of the DNA



Mutations

A mutation is any random, permanent change in the DNA molecule; many mutations are harmful, some have no effect, and a few actually benefit the organism; nature selects those mutations that are beneficial or adaptive in organisms to help shape the course of evolution

6

POPULATION GENETICS

Genes in Populations vs. Individuals

- 1. Populations evolve just as species do
- 2. Genotype: Genetic composition of an individual 3. Gene pool: Genetic composition of a
- population of individuals (i.e., all alleles for all genes in a population)
- 4. Evolution involves changes in gene pools over time; to understand changes in gene pools as populations evolve, an understanding of nonevolving populations is necessary

The Hardy-Weinberg Law

- 1. Both allelic frequencies and genotypic ratios (i.e., gene pools) remain constant from generation to generation in sexually reproducing populations if the following conditions of equilibrium exist:
 - A. Mutations do not occur
 - B. There is no net movement of individuals out of or into a population
 - C. All offspring produced have the same chances for survival, and mating is random (i.e., no natural selection occurs)
 - D. The population is so large that chance would not alter frequencies of alleles

2. Algebraic equivalent of the Hardy-Weinberg law:

- $p^2 + 2pq + q^2 = 1$, where:
- p = frequency of dominant allele
- q = frequency of recessive allele
- $p^2 = AA$ genotype
- 2pq = Aa genotype
- q^2 = aa genotype

Example

- 1. If a group of six individuals has nine dominant (A) alleles and three recessive (a) alleles, then p = 9/12 or 0.75 and q =3/12 or 0.25; a total of 12 gametes will be produced, 9 of which will have the dominant allele and 3 of which will have the recessive allele
- 2. Use the equation to predict the ratios of the three possible genotypes as a result of fertilizations
 - A. Frequency of AA genotypes is p^2 , or $(0.75)^2 = 0.56$
 - B. Frequency of Aa genotypes is 2pq, or 2(0.75)(0.25) = 0.38
 - C. Frequency of aa genotypes is q^2 , or $(0.25)^2$ = 0.06
- 3. The frequencies of dominant and recessive alleles are still the same, but the specific alleles have been redistributed

Hardy-Weinberg & **Natural Populations**

- 1. Few (if any) populations are in equilibrium; therefore, changes in allele frequencies and thus gene pools do occur in natural populations
- 2. The Hardy-Weinberg law helps identify the mechanisms of these evolutionary changes by predicting that one or more of the four conditions required are not met; that is, in natural populations:
 - A. Mutations occur
- B. Individuals leave and enter populations C. Nonrandom mating and natural selection
- occur

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D. Small populations exist



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Author: Randy Brooks, PhD

TSBN

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Protein Synthesis

A. Definitions

Ruick Study. CADEMIC

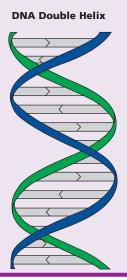
Featuring: Evolution/origins, molecular biology, cancer biology, human aging and immunology

Evolution

- 1. Concept that all organisms are related by common ancestry 2. Fundamental paradigm of biology
- B. Natural selection: The mechanism for how evolution occurs
 - 1. Species have high potential for rapid reproduction
 - 2. Population sizes eventually level off and remain fairly constant over time
 - 3. There is **competition** for reproduction and survival of offspring
 - Variations (from random mutations and shuffling of genes via meiosis) exist in behavior, physiology, structure, etc.
 - 5. Nature selects individuals (i.e., the **fittest** or just fortunate) for survival and reproduction to pass these favorable characteristics (**adaptations**) via their genes to their offspring
 - 6. Over time, natural selection "can" lead to genetic changes in populations i.e., evolution
 - 7. Microevolution: Small-scale changes
 - 8. **Macroevolution:** Larger-scale changes; can lead to evolution of new species and groups

Cellular/Molecular Evidence for Evolution

- A.**Cell Theory** 1. The cell is the basic unit of life
 - 2. Every life form, from bacteria to humans, is made
- of/comes from this basic structure
- **B. Organic Molecules**
- 1.99% of all life consists of carbon, hydrogen, oxygen, nitrogen, phosphorus and sulfur
- Evolutionary relatedness explains organisms' common usage of a small subset of over 90 available elements
- $C. \boldsymbol{DNA}$
 - 1. Genetic, informational molecule in every organism, including viruses (which appear to be molecular fragments of DNA/RNA capable of "living" in host cells)
 - 2. DNA "language" (genetic code) is essentially universal (slightly different dialects exist in some single-celled organisms and in some mitochondrial/chloroplast genomes)
- 3. A common genetic language allows for such phenomena as the insertion of human genes into bacteria, which can then produce "human" proteins (see Molecular Biology)
 D. ATP (Adenosine triphosphate): The primary energy currency



Evidence for Evolution via Natural Selection

molecule used by every organism

A.Artificial selection

- 1. Human-controlled breeding of species strongly supports the idea that, over time, nature could also influence changes in populations
- 2. Humans have selected for traits to increase the attractiveness (to us) of the offspring (e.g., "cute" dogs, chickens that produce many eggs, wheat that yields numerous, plump grains)
- 3. Domesticated species often do poorly in the wild, as traits (i.e., variations) selected by humans would not necessarily be advantageous in nature
- **Artificial Selection For Crop Production**



- B. **Biogeography:** Geographic distribution of species can show organisms are related
- 1. Flightless birds, such as African ostriches, Australian emus, and South American rheas are found (naturally) only in the southern hemisphere; on separate continents

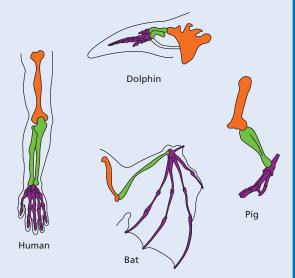
- 2. Either flightlessness in these birds evolved independently three times (possible, but improbable) or they arose from a common, flightless ancestor
- 3. If the latter explanation is correct, and they could not fly, how then could they get to these disparate southern continents while being excluded from the northern hemisphere?
- 4. Geological evidence indicates the continents were once one large land mass that subsequently broke up into pieces (**plate tectonics**) that moved (**continental drift**) first into northern and southern portions, and later into the present-day continents
- 5. This geological concept also explains why **marsupial mammals** (e.g., kangaroos) developed only on Australia, as this continent was geographically isolated from areas where **placental mammals** evolved
- C. Fossils
- 1. Preserved remnants of dead organisms
- 2. Darwin termed evolution "descent with modification"
- 3. Although the fossil record has gaps (some structures/organisms do not fossilize well), fossils provide valuable information about evolutionary changes or modifications in organisms (including transitional forms, e.g., horses with toes, whales with hind limbs, ferns with seeds) that have taken place over many generations
- 4. Estimating the age of fossils involves looking at their physical positions in sedimentary rocks (**relative dating**) and radiometric isotope techniques (**absolute dating**)

5. **Molecular clocks** look at changes in portions of genomes of organisms; also used to help determine the age of evolutionary events

D. Homologies

- 1. Anatomical similarities of related life forms
- 2. Provide strong evolutionary evidence of relatedness
- 3. Example: Forelimbs of vertebrates are composed of the same basic bones in disparate groups, but differ based on adaptations necessary for the specific environmental needs (i.e., walking, swimming, flying)
- 4. Vestigial structures
 - a. Those present are usually in a rudimentary, nonfunctional form
 - b. Show anatomically-related structures that are likely to disappear completely in future generations
 - c. Example: The vestiges of pelvic bones within the body in some modern-day baleen whales

Homologous Forelimb Bones: Evidence for Vertebrate Evolution

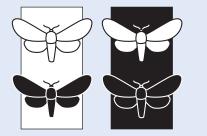


Evidence for Evolution via Natural Selection cont.

E. Variations in Life

- 1. In England, the peppered moth shifted from predominantly light coloring to dark when air pollution darkened the trees on which it lives
- 2. Predators can easily spot moths that contrast with their background, limiting the abundance of these types of moths in the population
- 3. Subsequent air quality measures have lightened trees and light-colored moths are again the predominant form
- 4. Additional examples of selection observed in living organisms involve increasing drug resistance: e.g., bacteria-antibiotics, insectinsecticides and HIV-drug therapies

Generations of Peppered Moths Changed Color to Match Habitat



- QuickStudy.
- A. Where do humans fit in the evolutionary scheme?
- B. Some of the greatest evidence for evolution is seen when comparing vertebrate chordates, which include humans (see **Homologies**, Evolution & Natural Selection)

C. Comparative anatomy of adults

1. Obvious visual similarities in adult vertebrates (i.e., eyes, ears, mouth, nose, appendages) link humans to other vertebrates, especially the great apes

D. Comparative embryology

- 1. **Earnst Haeckel** coined the phrase "ontogeny recapitulates phylogeny," suggesting the false claim humans start as fish, then progress through a series of developmental stages that retrace the lower vertebrate groups before becoming human
- 2. Early developmental stages of humans share remarkably similar vertebrate characteristics that either disappear or become vestigial in adult humans

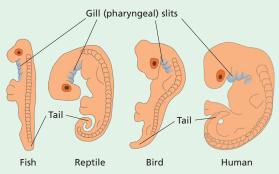
Origins of Life

Stromatolites Form Aquatic Reefs

Human Origins

a. Gill (pharyngeal) slits (they occasionally do not close in infants – cervical (branchial) fistulae – may require surgery)

Embryonic Similarities Among Vertebrates



E. Vestigial structures

- 1. Show clear links to vertebrate ancestry and include the following non-functional structures:
- a. Tail bones (coccyx)
- b. **Ear muscles** (function in other mammals)
- c. Nictitating membrane (3rd eyelid in some vertebrates)
- d. Pointed canine teeth
- (continued pg.3)

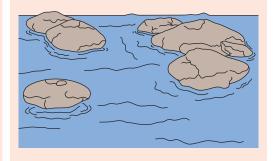
A. The ultimate spark of life may never be known but science provides a controversial scenario of how life "might" have arisen

B. Universe/Earth origins

- 1. First, the universe had to be formed, theoretically via the **Big Bang** about 16-18 billion years ago
- 2. Geologic and other physical evidence date the earth's origin to about 4.6 billion years ago
- 3. The crust and **biosphere** (thin portion of earth where life exists) would not be habitable (too hot) for nearly a billion years

C. **First cells**: How did they form?

- Early hypotheses suggest life arose spontaneously from simple molecules (e.g., CO, CO₂, N₂, H₂O) that combined into larger, complex macromolecules such as proteins, carbohydrates, lipids and nucleic acids
- 2. Some rocks from outer space (meteorites) have pre-formed complex organic molecules, including the five nitrogenous bases that make up DNA/RNA
- 3. Whether life was seeded from outer space (**panspermia**), or macromolecules were synthesized entirely on earth, the next step was to incorporate these organics into cells the basic functional units of life
- 4. These first life forms were likely **heterotrophs**, which consumed the abundant food molecules present in the "**primordial soup**"
- 5. Later, photosynthesis (by **autotrophs**) developed and oxygen levels began increasing in the atmosphere
- 6. The oldest fossils discovered (aged 3.8 billion years) consist of photosynthesizing bacteria called **stromatolites**, which still have representatives in colonies that form large, calcareous structures in some shallow, tropical oceans



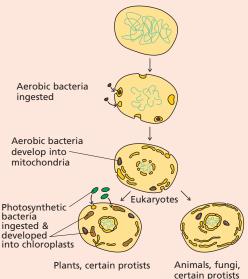
D. Oxygen crisis and the endosymbiotic hypothesis

- 1. Geologic evidence supports increasing oxygen levels via photosynthesis-created "rust" zones at similar ages in ancient sea beds worldwide
- 2. Chemically, oxygen is a corrosive element to organic molecules as well, and likely created a crisis for many of the earliest life forms
- 3. Some bacteria evolved a metabolic pathway that could neutralize as well as produce ATP energy from this highly-reactive oxygen
- Symbioses formed between these oxygenconsuming, energy-producing bacteria and other larger, soft-bodied bacteria that lacked protection against the effects of oxygen
- 5. This was the birth of the **eukaryotic** cell, from **prokaryotic** ancestors; one of the major evolutionary events in life
- 6. This **endosymbiotic hypothesis** is supported by the following facts:
 - i. **Mitochondria** (use oxygen for metabolism) have their own set of DNA, separate from that of the cell nucleus

- ii. **Mitochondrial DNA** is more like present-day bacterial DNA than the **nuclear DNA** of the cell in which it resides
- iii. Chloroplasts have their own genomes
- iv. Today, living organisms provide numerous examples of symbiotic relationships between single-celled organisms; sometimes including bacteria that perform the role of mitochondria in cells lacking ATPproducing organelles
- 7. Eukaryotic cells subsequently evolved into protists, fungi, plants and animals
- 8. Prokaryotes continued to thrive and, though microscopic, are among the most successful groups of organisms on earth

Evolution of Eukaryotic Cells

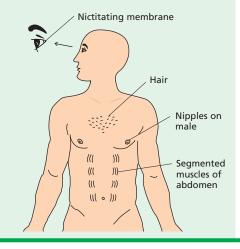
Host Prokaryotic Cell



e. 3rd molar teeth

- f. **Hair** (plays major thermoregulation role in most mammals)
- g. Nipples in males
- h. **Appendix** (functions as digestive caecum in many mammals)
- i. Segmented muscles of abdomen
- j. **Pyramidalis muscle** (absent in 20% of humans; arguably unnecessary; present in other mammals)

Some Vestigial Structures in Humans



Human Origins F. Molecular Comparisons

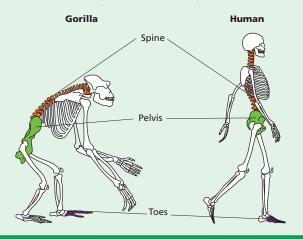
- 1. Comparison of DNA sequences in humans and chimpanzees show average similarity of 98.5%
- 2. Comparison of hemoglobin amino acid sequences (the main carrier of oxygen in the blood of thousands of different animals [by itself evidence for evolution]) between humans and other vertebrates show the same evolutionary patterns as those with skeletal/physical anatomy that is comparative, with the great apes showing the greatest similarity

G.Fossil Record

- 1. Fossils show a transition from apelike forms to the first primitive human forms that were truly **bipedal** (walking on the pelvic appendages or legs)
- Modern apes are not bipedal, but one of the oldest fossil forms (3.2 million years) resembling an ape to walk bipedally was named *Australopithecus afarenesis* or Lucy (named after a famous Beatles song)

- 3. From this origin in Africa, modern humans, *Homo sapiens*, eventually arose
- 4. Debate exists among paleoanthropologists about how to arrange the phylogenetic tree of humans based on the available fossils
- 5. Most agree that Neanderthals were the most recent group of humans to become extinct, and were probably a subspecies called *Homo sapiens neanderthalensis*
- 6. From these origins, humans have spread to most land areas on Earth

Anthropoid Skeletal Comparison



Molecular Biology

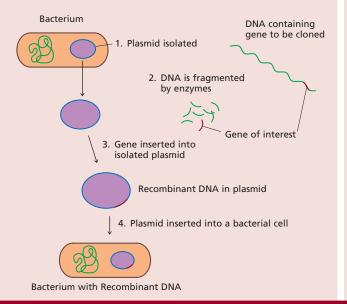
A.The discovery that DNA is the informational molecule housing genes started a revolution in biology

B. **Molecular biotechnology** is now a pervasive component in modern societies

Cloning

- A.Gene Cloning
- 1. Making exact copies of genes
- 2. Involves two major processes:
 - a. Recombinant DNA
 - i. **Restriction enzymes** create DNA fragments with the gene of interest
 - ii. DNA fragments are fused with DNA from a bacterium (**plasmid**)
 - iii. Newly-created **recombinant DNA** is placed into bacteria
 - iv. Bacteria produce protein for which the "cloned" gene coded
 - v. Large quantities of the gene, and thus protein, are produced as the bacterial cell reproduces

Gene Cloning using Recombinant DNA

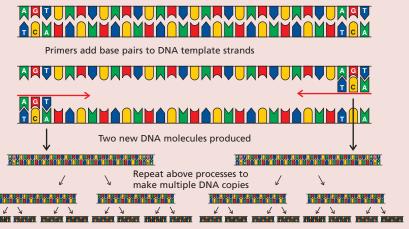


b. Polymerase Chain Reaction (PCR)

- i. Amplifies (copies) a segment of DNA without using a bacterial (or other) host organism
- ii. DNA sample is heated until the double helix denatures (hydrogen bonds are broken), separating the DNA into two single strands
- iii. Heat-resistant, single-stranded DNA primers allow DNA polymerase to add the appropriate nucleotides to each side of the separated DNA strands
- iv. This process results in multiple copies of the original DNA
- v. Repeating the process on the copies, via automation, can amplify a small DNA fraction a billion fold in a short period of time

Using PCR to Amplify DNA

Separate DNA strands by heating



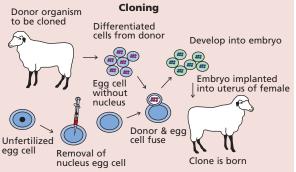
B. Reproductive cloning

- 1. Produces living cells/organisms with exactly the same DNA in the nuclei as that from a donor cell/organism
- 2. Specifically, DNA from the nucleus of a **somatic cell** of the donor is inserted into an **egg cell** from which the original nucleus has been removed
- 3. The new egg cell is electrically or chemically stimulated to begin cell division and embryonic development
- 4. The growing embryo is implanted into a female where development continues until birth

QuickStudy.

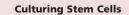
Molecular Biology cont.

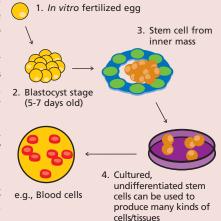
- 5. The new individual is not a true clone of the donor organism, as the mitochondrial DNA is from the organism that donated the egg
- 6. Survival rates have been low as multiple factors (mostly unknown) influence successful development, such as source of donor cells



C. Therapeutic cloning

- 1. Use of reproductive cloning to create human embryos to procure **stem cells**, which have potential to develop into adult tissues
- 2. These special cells may hold the key to treatments for many diseases (heart, cancers, Alzheimer's, Parkinson's) and afflictions (injury to spinal cord, including paralysis)
- Stem cells can also be retrieved from human embryos produced by regular fertilization processes (*in vivo* or *in vitro*) or adults (e.g., bone marrow)
- 4. Stem cell procurement via cloning and embryos is a growing ethical and political issue





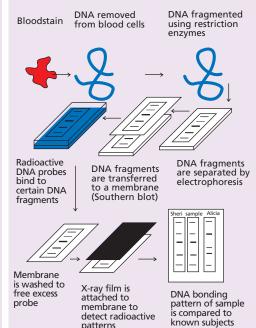
- A.Study of the structural and functional aspects of the entire set of genes in a species (i.e., genome)
- B. Encompasses many different aspects of approach
 - 1. **Bioinformatics** uses computer/statistical applications to access large databases concerning DNA/gene/protein information
 - 2. **Proteomics** studies the functioning of the proteins coded by the genes
- C. Several specific applications of genomics will be discussed further below:
 - 1. Restriction Fragment Length Polymorphisms (RFLP)
 - a. Technique relies on enzymes discovered that protect bacteria from "foreign" DNA of bacteriophages (viruses specific for bacteria) and other invading bacteria
 - b. These bacterial restriction enzymes cut foreign DNA at specific points or **restriction sites**, while protecting their own DNA by adding special "buffering" functional groups to potentially susceptible areas
 - c. Exact positions of restriction points are highly individual, reproducible and measurable
 - d. DNA samples from the same individual will produce the same fragments, but these fragments will be different from others (polymorphic)
 - e. Fragment patterns can be represented visually as a **DNA fingerprint**, by use of special electrophoretic processes
 - f. RFLP is used frequently in forensic, criminal and paternity applications
 - g. Because DNA samples may be minute in some of these applications, PCR amplification may be used to create quantities necessary for RFLP analysis
 - h. A modified DNA fingerprint approach has been developed using polymorphisms of satellite (repetitive)
 DNA regions called Simple Tandem Repeats (STR)



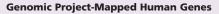
a. Monumental, historical effort to determine the actual sequence of the entire set of chromosomes in humans - gene mapping

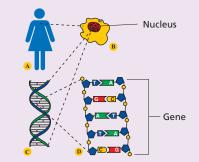
Genomics

DNA fingerprinting using RFLP



- b. Involved over 3 billion base pairs, which if written, would create a book with a half-billion pages and take nearly a lifetime to read
- c. Several molecular techniques were employed, with automated computerassisted analysis paving the way for a rapid conclusion to the project
- d. Although the precise number of genes is still unknown, *a priori* estimates suggested there would be nearly 100,000
- e. Actual number probably does not exceed 40,000, which when compared to simpler organisms suggests human genomics is extremely concise, but complex
- f. Future studies will undoubtedly reveal much about how genes function, which should lead to numerous future benefits

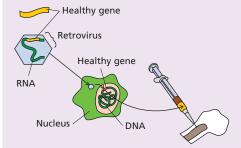




3. Gene therapy

- a. Treating diseases and injury in humans involves the use of harmless retrovirus vectors (or other entry mechanisms) that possess the enzyme **reverse transcriptase**, allowing them to insert genetic information "into" DNA
- b. Normal information flow occurs "from" the DNA
- c. These treatments raise ethical questions, but certainly have tremendous potential
- d. Limited success and legal restrictions using human subjects have made progress in this area challenging

Using Retroviruses to Insert Healthy Genes



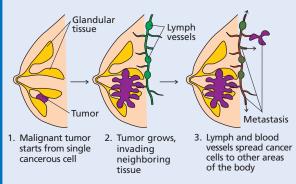
4. Genetic engineering

- a. Research involving gene transfer in non-human organisms has been much more extensive
- b. **Transgenic** and **genetically-modified** plants and animals are becoming more common
- c. Great potential to artificially select desirable traits in crops, farm animals, etc.
- d. Safety concerns are still high as this new technology is incorporated into modern society

Biology of Cancer

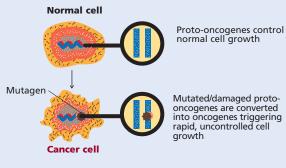
- **Optional review**: "Cell Reproduction" section, p.2 of Quickstudy[®] Biology guide
- A.Cells reproduce by dividing primarily through two processes: 1. Mitosis: Nuclear division
- 2. Cytokinesis: Cytoplasm division
- B. Cell division is part of the cell cycle, which is under a control system involving internal and external factors
- C. Cancer cells have escaped this regulatory process through
- transformation and divide uncontrollably D. Tumors form, which may progress from a benign to a malignant state and interfere with normal tissue functioning

Tumor Formation & Spreading



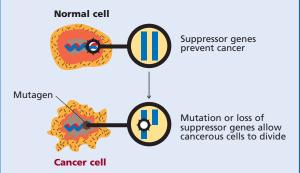
- E. Metastasize: Initial tumor cells can spread and form more malignant tumors in other tissues in the body
- F. Oncogenes stimulate abnormal cell growth and division, which can lead to malignant tumors
 - 1. These abnormal genes are converted from normal genes (proto-oncogenes) that regulate the cell cycle. Viruses can also deliver oncogenes to cells

Oncogene Activation Leading to Cancer



G.Tumor-suppressor genes normally prevent the uncontrolled growth and division of cells and tissues

Tumor-Suppressor Gene Deactivation Leading to Cancer



- H.**Mutations** are primary factors contributing to cancers 1. Mutagens are any factors that can trigger mutations those that cause cancer are called **carcinogens**
- I. All tissues in the human body are susceptible to tumors, because mutations (either induced by carcinogens or inherited) can occur in any cell
- Cancers are prevalent and difficult to cure (in most cases) because of our limited knowledge about: 1. Factors controlling the cell cycle
 - 2. The genomics of humans

QuickStudy

Biology of Aging

Human Lifespan Increase

YEAR

All races, male

All races, female

- A.Most animals in nature die shortly after their reproductive years, and in some cases, die immediately after reproduction
- B. Humans and most animals kept under controlled conditions can survive many years after fertility has waned, allowing the phenomenon of aging to be studied
- C. For humans, the potential to live longer has been realized over our history; in the last 50 years, average lifespan in well-developed countries has risen from the 60-70s to nearly 80 years
- D. Considering the longevity of some rare individuals, human lifespan could be up to 120-130 years in the near future

Theories of Aging

A.What prevents all but a few of us from living to our physiological maximum? B. What are the specific causes for the physical transformations that occur as we age? C. Random events may accumulate and contribute to early senescence; some specific

- hypotheses follow: 1. Free radical formation typically involves the production of oxidative metabolic
- by-products such as molecular variants of oxygen, which may damage the DNA, RNA, proteins and mitochondria
 - a. Anti-oxidants produced naturally may eventually lose the battle in cells, causing cell death
 - b. Proponents of this hypothesis suggest supplemental intake of anti-oxidants (e.g., found in vitamins) may slow this form of damage
- 2. Cross-linking suggests as cells age, structural molecules such as DNA and proteins form unsuitable attachments within or between other molecules
 - a. Skin wrinkling, cataracts of the eye, atherosclerosis in blood vessels, kidney function and brain function **Physical Changes During Aging**
 - decline are all possibly related to cross-linking
 - b. Some drugs that prevent or slow cross-linking may be important future therapies





- 3. Wear and tear suggests that 30 years 40 years the mere use of cells and concomitant damage result in aging
- a. This type of damage occurs at the DNA level, which has its own set of repair proteins
- b. Years of exposure to mutagens such as toxins and various forms of radiation are not always repaired
- c. At the ends of DNA molecules are protective caps called telomeres, which are degraded with each cell division event
 - i. Telomere loss eventually can lead to DNA damage
 - ii. Telomerase, an enzyme that repairs these end caps, has been shown to keep cells in a more "youthful" state
- 4. Somatic mutations, those occurring in tissues outside of the egg or sperm, could lead to diminished function; skin and connective tissues lose resiliency, muscles become weaker, brain cells become less efficient, etc.
- 5. Rate of living hypothesis: Suggests those that "live the fastest, die the youngest" a. Theorizes those organisms with the most active metabolisms have the shortest lifespan
 - b. With mammals, this is usually the case (e.g., an elephant lives longer than a mouse)
 - c. Hypothesis may be broadly linked to those under the pre-programmed events (see below)

D. Pre-programmed events may be a cause of senescence in humans; following is a discussion of specific hypotheses:

- 1. Genetic theory suggests our lifespan is determined by the inherited genes
 - a. When food and health issues are maintained at least minimally, humans have roughly the same lifespan
 - b. Females in most instances (including other animals) typically live longer than males
 - c. Offspring of long-lived parents typically live longer than offspring of shorter-lived parents
 - d. The above observations strongly suggest at least part of lifespan determination is related to longevity-assurance genes
- 2. Pacemaker theory suggests there are "biological clocks" or pacemakers that commence at birth and simply slow and stop, ending in death
 - a. Specifically, the immune and neuroendocrine systems are thought to be controlled by pacemakers
 - b. Cessation of these systems could account for body-wide failures, susceptibility to attack by foreign agents, and increase incidence of cancers



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Immunoloav

threatening

healthy tissues

3. Allergies are hypersensitive tissue reactions

a. Specifically, antibodies against specific

symptoms (e.g., hay fever, asthma)

4. Autoimmunity is a condition in which cells

of the specific immune response attack

a. Normally, those antibodies and cells of

deleted to prevent such self attacks

b. The following diseases/afflictions are

triggered or related to autoimmunity:

i. Rheumatoid arthritis

Autoimmune Disease Leading

to Rheumatoid Arthritis

Bone

5. Immunodeficiency diseases are those in

which some aspect of the immune system

Cartilage and bone worn

down narrowing joint cavity

Inflammatory cells

attacking joint surfaces

Swollen/inflammed joint

Synovial membrane

HIV Virus

Attacking Helper

T-cell Lymphocyte

hypertrophy into

nearby bone

ii. Diabetes mellitus

iii. Grave's disease iv. Multiple sclerosis

v. Lupus

2

(usually specific)

is defective, thus

compromising the

ability of the body

to protect itself

a. One of the best

known of these

is Acquired Im-

munodeficiency

Syndrome

disease which is

triggered by the

Human Immun-

odeficiency

Virus (HIV)

- a

(AIDS)

the immune response that could harm

"self" tissues are either suppressed or

b. Severe allergic reactions can lead to

anaphylactic shock, which may be life-

antigens called allergens trigger tissue

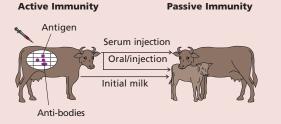
response resulting in typical allergic

to part of the specific immune response

Optional review: "Immunology" section, p.5 of Quickstudy[®] Physiology guide

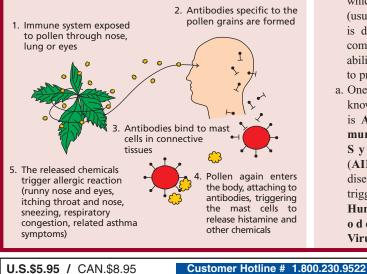
- A. The body has two main lines of defense against injury and infection:
 - 1. Nonspecific immunity involves a generalized, similar response to a wide variety of potentially harmful conditions; a typical component of this response is inflammation, which results in swelling, redness, heat and pain in the affected area
 - 2. Specific immunity is an extremely specific response typically involving the production of **antibodies**, which are designed with the exact purpose of combining with specific cell surface markers, or antigens, of foreign agents (microbes, toxins)
- B. Selected subjects related to immunity are discussed below:
 - 1. Passive immunity involves receiving antibodies or antiserum from another source
 - a. This could involve maternal antibody delivery to the fetus/child via breast milk from the mother or injections (also for treatment of venomous bites/stings)

Antibodies Injected or Passed to Others



- 2. Vaccinations contain weakened versions of pathogens injected into the body to stimulate, among other aspects of specific immunity, B cells to produce two products:
 - a. Plasma cells, which begin synthesizing antibodies within 10-17 days
 - b. Memory cells, which retain the potential (for up to many years) to develop quickly (within 2-5 days) into antibodyproducing plasma cells upon subsequent exposure
 - c. This quicker response could mean the difference between successfully destroying the foreign antigen versus possible death of the individual

Allergic Reaction Events

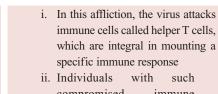








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- compromised immune systems are susceptible to and infections secondary cancers, which untreated usually leads to death
- iii. AIDS is still a worldwide health issue and the leading cause of premature death in some countries
- b. Severe Combined Immunodeficiency Syndrome (SCIDS) is a rare congenital condition in which T and B cells are defective
 - i. In the most severe cases, a person is born essentially with no specific immune response and stands little chance of warding off infection
 - ii. Death can occur within the first year without a bone marrow or stem-cell transplant
- 6. Bacterial resistance to antibiotics can occur when medical drugs are used to supplement the specific immune response, the latter of which may be too slow to prevent serious and possibly fatal symptoms
 - i. When antibiotics are taken, highly resistant forms of bacteria may survive and reproduce
 - ii. These new "resistant" strains may be extremely difficult, if not impossible, to treat
 - iii. Over-prescribing of antibiotics may be a leading cause of resistance
 - iv. As much as half of the roughly 100 million prescriptions for antibiotics written each year may be unnecessary (e.g., colds and flu symptoms are caused by viral infections; therefore, antibiotics are of limited use)
 - v. When prescriptions are given, medication should be taken to completion - only taking a portion of the pills may allow the hardiest bacteria to survive and evolve

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