(1) DNA Structure and Function

- Nucleotide monomer
  - Nitrogenous Bases (B)
  - 5-C Sugar (S)
  - Phosphate (P)

- Nucleotide Structure
  - 5-C sugar
    - RNA – ribose
    - DNA – deoxyribose

- Nitrogenous Base
  - N – attaches to 1°C of sugar
  - Double or single ring
    - Four Bases – Adenine, Guanine, Thymine, Cytosine

- Phosphate
  - Attached to 5°C of sugar

- Nucleic Acids
  - Polymer of nucleotide monomers:
    - DNA – deoxyribonucleic acid
      - The heredity compound of life
      - Directs cellular activities
      - Sequence of nucleotide bases is unique for each individual
    - RNA – ribonucleic acid
      - Sugar – contains ribose sugar instead of deoxyribose
      - Bases – Uracil replaces Thymine found in DNA
      - Involved in protein synthesis
    - ATP – Adenosine Triphosphate
      - nucleotide consisting of ribose sugar, adenine & 3 phosphates
    - Coenzyme – NAD, FAD, NADP
      - nucleotides that assist enzymes by carrying electrons & hydrogen
DNA
- Information center of the cell
- Particular sequence of nucleotide bases forms a gene
- Gene codes for proteins
- Before a protein is made, genes must be transcribed into RNA

DNA Replication
- DNA must be copied before cell division
- Synthesis of DNA – during S-phase of interphase
- DNA Replication Semi-Conservative Model

Chromosome Structure
- Chromatin – relaxed form of genetic material is necessary during protein synthesis
- Chromosomes – condensed form of genetic material necessary during cell division
  - unduplicated (no chromatids)
  - Replicated (two chromatids)
(2) Gene Expression and Control: Protein Synthesis

- Steps from DNA to Proteins
  - Two steps produce all proteins:

\[
\text{DNA} \xrightarrow{\text{transcription}} \text{RNA} \xrightarrow{\text{translation}} \text{PROTEIN}
\]

- Gene Mutations
  - Base-pair substitutions
  - Insertions
  - Deletions
- Base-Pair Substitution

- Original base triplet

\[
\begin{array}{c}
\text{original base triplet: CAT} \\
\text{a base substitution: CTA}
\end{array}
\]

- Possible outcomes:

\[
\begin{array}{c}
\text{original, unmutated: CAT} \\
\text{a gene mutation: CTA}
\end{array}
\]

- During replication, proofreading enzymes make a substitution.

- Frameshift Mutations
  - Insertion
    - Extra base added into gene region
  - Deletion
    - Base removed from gene region
  - Both shift the reading frame
  - Result in altered amino acid sequence

- Mutations
  - Each gene has a characteristic mutation rate
  - Natural and synthetic chemicals, and radiation, increase mutation rate
Only mutations that arise in germ cells can pass on to next generation
Important evolutionary consequences

(3) How Cells Reproduce: Mitosis and Meiosis

Division Mechanisms
- Eukaryotic organisms
  - Mitosis
  - Meiosis
- Prokaryotic organisms
  - Prokaryotic fission

Roles of Mitosis
- Multicelled organisms
  - Growth
  - Cell replacement
- Some protistans, fungi, plants, animals
  - Asexual reproduction

Chromosome
- A DNA molecule and attached proteins
- Duplicated in preparation for cell division

Mitosis
- Period of nuclear division
- Usually followed by cytoplasmic division
- Four stages:
  ♦ Prophase
  ♦ Metaphase
  ♦ Anaphase
  ♦ Telophase

Chromosome Number
- Total number of chromosomes in a cell
- Somatic cells
  - Chromosome number is diploid (2n)
  - Two of each type of chromosome
- Gametes
  - Chromosome number is haploid (n)
  - One of each chromosome type

Human Chromosome Number
- Diploid chromosome number (n) = 46
- Two sets of 23 chromosomes
  - One set from father
  - One set from mother
- Mitosis produces cells with 46 chromosomes: two of each type
Asexual Reproduction
- Single parent produces offspring
- All offspring are genetically identical to one another and to parent

Sexual Reproduction
- Involves
  - Meiosis
  - Gamete production
  - Fertilization
- Produces genetic variation among offspring

Homologous Chromosomes
- Cell has two of each chromosome
- Chromosome pairs: one from mother, one from father
- Paternal and maternal chromosomes carry different alleles

Sexual Reproduction Shuffles Alleles
- Through sexual reproduction, offspring inherit new combinations of alleles, which lead to variations in traits
- Variation in traits is the basis for evolutionary change

Gamete Formation
- Gametes are sex cells (sperm, eggs)
- Arise from germ cells in reproductive organs

Sexual Reproduction and Genetic Variation
- Two functions of meiosis provide variation in traits:
  - crossing over
  - random alignment

Mitosis: Division of duplicated DNA
- chromosomes (unduplicated) in parent cell at interphase
- same chromosomes (duplicated) in parental cell
- chromosome (unduplicated)
Genetics

- **Genes**
  - Units of information about specific traits
  - Passed from parents to offspring
  - Each has a specific location (locus) on a chromosome

- **Alleles**
  - Different molecular forms of a gene found on homologous chromosomes
  - Arise by mutation
  - Dominant allele masks a recessive allele that is paired with it

- **Allele Combinations**
  - Homozygous
    - having two identical alleles
    - Homozygous dominant, AA
    - Homozygous recessive, aa
  - Heterozygous
    - having two different alleles
    - Aa

- **Genotype & Phenotype**
  - Genotype refers to particular genes an individual carries (RR or Rr or rr)
  - Phenotype refers to an individual’s observable traits (flower color, seed shape, etc)

- **Other Definitions**
  - Dominant allele – in a heterozygous individual, a trait that is fully expressed in the phenotype
  - Recessive allele – in a heterozygous individual, a trait that is completely masked by the expression of the dominant allele
  - Pure (true) breeding – a population with only one type of allele for a given trait
  - Self cross – when individuals of a generation fertilize themselves (e.g., self-fertilized flower)

- **Gregor Mendel (1822-1884)**
  - Father of Genetics
  - Austrian Monk
  - Strong background in mathematics
  - observed evidence of how parents transmit genes to offspring
  - Unaware of cells, chromosomes or genes

- **Mendel studied the Garden Pea**
  - Mendel began by examining varieties of peas suitable for study
    - Character- an observable feature, such as flower color
    - Trait – actual flower color, such as purple or white
    - Heritable trait – is this character passed on to progeny
- Experimentally cross-pollinated

- Mendel’s Methods
  - Mendel crossed round x wrinkle seeded plants
    - P (parental generation) → round x wrinkled
    - F1 (1st filial generation offspring) → round
    - F2 (2nd filial generation offspring) → round & wrinkled

- Dominant / Recessive Traits
  - Mendel observed each parent carried two “units” for a given trait
  - We know these “units” are genes on chromosomes
  - Dominant traits – show up each generation
  - Recessive traits – may be masked by dominant traits

- Monohybrid Cross
  - Experimental cross between two F1 heterozygotes
    - AA x aa → Aa (F1 monohybrids)
    - Aa x Aa → (?) F2
      - Genotype: 1 AA: 2 Aa: 1 aa
      - Phenotype: 3:1 (purple: white)
      - Mendel found 3:1 ratio in F2 for all traits

- Mendel’s Theory of Segregation
  - Individual inherits a unit of information (allele) for a trait from each parent
  - During gamete formation, the alleles segregate from each other

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**A Monohybrid Cross**

![Diagram of Mendel's Monohybrid Cross](image)
Dihybrid Cross
- **AB x ab**
  - Experimental cross between individuals that are homozygous for different versions of two traits
- **Dihybrid Cross: F1 Results**
  - AABB x aabb → AaBb (F1 dihybrids)
  - All have same trait (tall with purple flowers)
- **Dihybrid Cross: F2 Results**

**Dihybrid Cross x AAbb:**

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<th>1/4 Ab</th>
<th>1/4 aB</th>
<th>1/4 ab</th>
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- **Dominance Relations**
  - Complete dominance
  - Incomplete dominance
  - Codominance

- **Codominance (Multiple Alleles): ABO Blood Types**
  - Gene that controls ABO type codes for enzyme that determines structure of a glycolipid on blood cells
  - Two alleles (IA and IB) are codominant when paired
  - Third allele (i) is recessive to others

- **ABO and Transfusions**
  - Type O is universal donor – neither type A nor type B antigens produced
  - Type AB is universal receiver – no immune response to A or B antigens

- **Incomplete Dominance**
  - $F_2$ shows three phenotypes in 1:2:1 ratio
  - Example: crossing white and red flowered snap dragons appears to produce pink flowered hybrids.

- **Pleiotropy**
  - Alleles at a single locus may affect two or more traits
  - Marfan syndrome
  - Cystic fibrosis
  - Color and crossed eyes in Siamese cats
Gene interactions and phenotypic expression
- Genes may interact with each other: one gene influences phenotypic expression of others
- Complex variations: phenotype influenced by gene interactions and/or environmental conditions

Interactions among Gene Pairs
- Common among genes for hair color in mammals
- Genetics of Coat Color in Labrador Retrievers
  - Epistasis: phenotypic expression of one gene governed by another
  - Two genes involved
    - One gene influences melanin production
      - Two alleles - B (black) is dominant over b (brown)
    - Other gene influences melanin deposition
      - Two alleles - E promotes pigment deposition and is dominant over e

Continuous Variation
- A continuous range of small differences in a given trait among individuals
- The greater the number of genes and environmental factors that affect a trait, the more continuous the variation in that trait
  - Examples in humans:
    - Eye color: involves two genes
    - Height: multiple genes, alleles and environmental conditions
    - Skin Color: three genes with multiple alleles

Environmental Effects on Phenotype
- Genotype and environment can interact to affect phenotype
  - Himalayan rabbit ice pack experiment
  - Transplantation of plant cuttings to different elevations
  - Human depression
  - Hydrangeas and Soil
- Phenotypic Plasticity
  - Phenotype change in response to the environment. Examples:
    - Humans tan in response to sun exposure; increased melanin protects cells from harmful solar radiation
    - Mussels exposed to seastar “scents” develop stronger adductor muscles
    - Mussels exposed to dog whelk “scent” develop thicker shells