

Introduction to Genetics

Introduction to Genetics – Study Notes

1. What Is Genetics?

- **Definition:** The branch of biology that studies heredity, variation, and the mechanisms by which traits are passed from one generation to the next.
 - **Key Questions:**
 1. How are traits transmitted?
 2. Why do individuals of the same species differ?
 3. What molecular structures carry hereditary information?
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2. Historical Milestones

Year	Scientist	Contribution
1865	Gregor Mendel	Established the laws of inheritance using pea plants (dominant/recessive traits, segregation, independent assortment).
1902	Walter Sutton & Theodor Boveri	Proposed the chromosome theory of inheritance – genes reside on chromosomes.
1953	James Watson & Francis Crick	Described the double-helix structure of DNA.
1961	Marshall Nirenberg & Johann Matthaei	Deciphered the first codon, linking DNA to protein synthesis.
1970s-80s	Recombinant DNA technology	Enabled gene cloning, sequencing, and manipulation.
2003	Human Genome Project	Completed the first draft of the human genome (~3 billion base pairs).

3. Core Concepts

3.1 Genes and Alleles

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- **Gene:** A functional unit of DNA that encodes a product (usually a protein or functional RNA).
- **Allele:** Different versions of the same gene that arise by mutation.

3.2 Chromosomes

- Linear DNA molecules packaged with proteins (histones).
- Humans: 46 chromosomes (23 pairs) – 22 autosomes + 1 pair of sex chromosomes (XX or XY).

3.3 DNA ? RNA ? Protein (Central Dogma)

1. **Replication:** DNA copies itself (semi-conservative).
2. **Transcription:** DNA ? messenger RNA (mRNA).
3. **Translation:** mRNA ? polypeptide chain (protein) via ribosomes and tRNA.

3.4 Mendelian Inheritance

Law	Principle	Typical Ratio (monohybrid cross)
Segregation	Two alleles for a trait separate during gamete formation.	3 (dominant) : 1 (recessive)
Independent Assortment	Genes for different traits segregate independently (if on different chromosomes).	9 : 3 : 3 : 1 (dihybrid cross)
Dominance	Dominant allele masks recessive allele in heterozygote.	—

3.5 Non-Mendelian Patterns

- **Incomplete dominance:** Heterozygote phenotype intermediate (e.g., pink flowers from red × white).
- **Codominance:** Both alleles expressed (e.g., AB blood type).
- **Multiple alleles:** More than two alleles in a population (e.g., ABO blood groups).
- **Polygenic inheritance:** Many genes contribute to a trait (e.g., height, skin color).
- **Sex-linked inheritance:** Genes located on sex chromosomes (e.g., hemophilia, red-green color blindness).
- **Mitochondrial inheritance:** Maternal transmission of mitochondrial DNA.

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4. Molecular Basis of Variation

Source of Variation	Mechanism	Example
Point mutation	Single base substitution, insertion, or deletion.	Sickle