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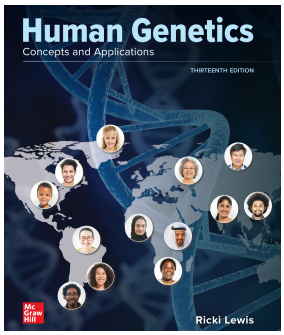

Chapter 1

The Information in a Human Genome

Thirteenth Edition

Ricki Lewis

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Learning Outcomes 1

1. Explain what genetics is and what it is not.
2. Distinguish among gene, exome, and genome.
3. Define *bioethics*.
4. List the levels of genetics.
5. Explain how DNA is maintained and how it provides the information to construct a protein.
6. Explain how a mutation can cause a disease.
7. State the basis of genetic diversity

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Learning Outcomes 2

8. Explain the relationship between DNA and chromosomes.
9. Distinguish between Mendelian and complex traits.
10. Explain how genetics underlies evolution.
11. List some practical uses of DNA information.
12. Distinguish between traditional breeding and genetically modifying organisms.
13. Explain how investigating genomes extends beyond interest in ourselves.

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Introduction

Genetics is the study of inherited traits and their variations and transmission.

It has recently exploded into a powerful source of information about our identities.

- Human genetics touches forensics, bioethics, psychology and even history
- Consumer genetics enables anyone to learn about their DNA

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Consumer Genetics

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What is Genetics?

Genetics is a life science that should not be confused with genealogy

- **Genetic genealogy** examines how people are related.
- **Heredity** concerns the transmission of traits and biological information between generations.

Certain difficult-to-define human characteristics might appear to be inherited if they affect several family members, but may reflect shared genetic and environmental influences.

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The Vocabulary of Genetics

Genes are the instructions to manufacture proteins, which determine inherited traits.

- Are composed of **deoxyribonucleic acid (DNA)**.

A **genome** is a complete set of genetic information.

A **cell**, the basic unit of life, contains two genomes.

The **exome** is the part of the genome that encodes protein.

Genomics compares and analyzes the functions of many genes.

Bioethics addresses issues and controversies that arise in applying medical technology and using genetic information.

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The Levels of Genetics 1

Genetics considers the transmission of information at several levels.

- It begins with the molecular level and broadens through cells, tissues and organs, individuals, families, and finally to populations and the evolution of species.

A DNA molecule resembles a spiral staircase or double helix

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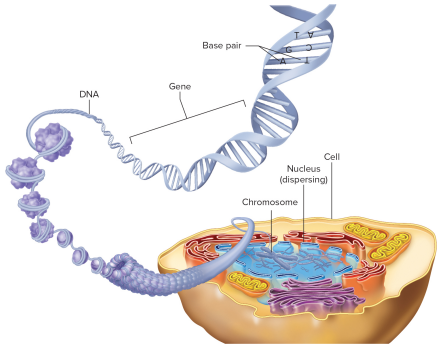
The Levels of Genetics 2



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Deoxyribonucleic Acid

Components:

- **Phosphate**
- **Sugar**
- **Base**: Adenine **(A)** Thymine **(T)**
  Cytosine **(C)** Guanine **(G)**

The information that a DNA molecule imparts is in the sequences of A, T, C, and G.

The chemical structure of DNA gives it two key abilities essential for being the basis of life:

- Can replicate itself
- Is accessible to manufacture proteins

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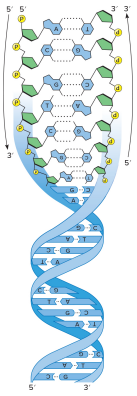
The DNA Double Helix



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From Gene to Protein 1

In **DNA replication**, a new double helix is formed from the old one using free DNA bases.

- Thus, the two "daughter" cells inherit identical copies of the genome during cell division.

Transcription copies the DNA information into a related molecule called messenger **ribonucleic acid (RNA)**

- This process is also called **gene expression**

Translation uses the information in RNA to assemble amino acids into proteins.

- Proteins provide the traits associated with genes.

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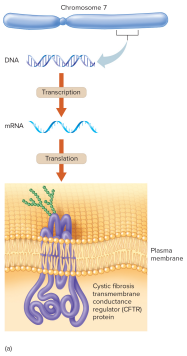
From Gene to Protein 2



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Mutations

A change in a gene is a **mutation**, and it can have an effect at the whole-person level

Alleles are variants of genes

- Are inherited or arise by mutations

Mutations in sperm or egg cells are passed on to the next generation

Mutations may be positive, negative, or neutral

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Cystic Fibrosis 1

The disease cystic fibrosis (CF) illustrates how a missing or abnormal protein causes the symptoms of an inherited disease

In CF, the protein is the cystic fibrosis transmembrane conductance regulator (*CFTR*).

- The functioning protein works like a selective doorway in cells lining the airways and other parts
- Can cause thickening secretions when it doesn't work properly

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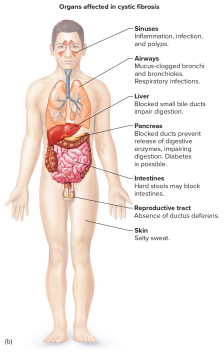
Cystic Fibrosis 2



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Mutation of Cystic Fibrosis 1

The disease cystic fibrosis is caused by a mutation in the *CFTR* gene.

- The mutation causes the replacement of the amino acid glycine with aspartic acid at a specific site.
- This alters the CFTR protein, so that it cannot open at the cell's surface
- Difficulty breathing, impaired digestion, and other symptoms result

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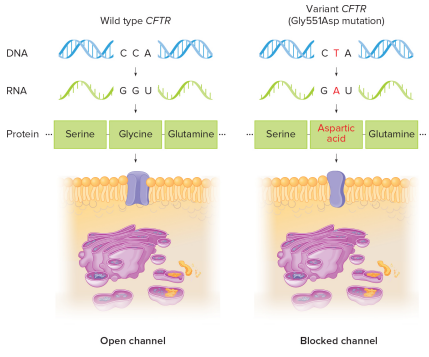
Mutation of Cystic Fibrosis 2



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Chromosomes and More

Chromosomes consist of DNA and protein.

- When a cell is not dividing, the chromosomes are unwound and in a structure called the **nucleus**.

A **somatic cell** in humans has 23 chromosome pairs.

- 22 pairs of **autosomes**
- A pair of **sex chromosomes**
  - Females have two X chromosomes
  - Males have one X and a Y

Karyotypes display the chromosome pairs from largest to smallest

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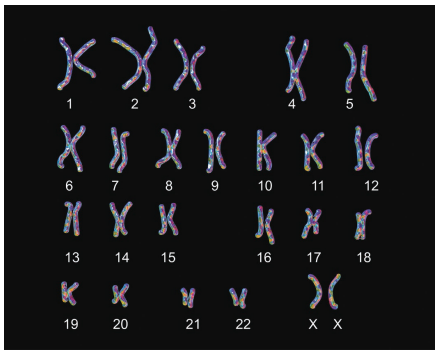
Karyotype



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Traits

A trait caused predominantly by a single gene is termed Mendelian

- Named for Gregor Mendel, who discovered the patterns of trait transmission.

Most characteristics are **complex traits**

- They are determined by one or more genes and environmental factors
- The more factors that contribute to a trait or illness—inherited or environmental—the more difficult it is to predict the risk of occurrence in a particular family member

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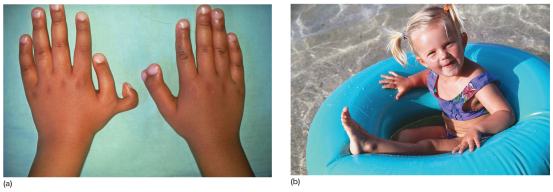
Mendelian versus complex traits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The Body: Cells, Tissues, and Organs

A human body consists of approximately 30 trillion cells.

- Cells differ in appearance and activities because they use only some of their genes.

Cells undergo **differentiation,** or specialization, of distinctive cell types.

Stem cells divide to yield other stem cells and cells that differentiate.

Tissues are groups of cells with a shared function.

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Relationships: From Individuals to Families

- **Genotype** refers to the underlying DNA instructions (alleles present)
- **Phenotype** is the visible trait, biochemical change, or effect on health (alleles expressed).
- Alleles can be **dominant** (exerting an effect in a single copy) or **recessive** (requiring two copies for expression).
- **Pedigrees** are charts that depict the members of a family and indicate which individuals have particular inherited traits.

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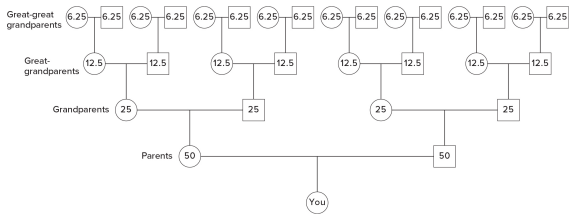
Pedigrees R Us



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The Bigger Picture: From Populations to Evolution

Above the family level of genetic organization is the population.

- In a strict biological sense, a population is a group of individuals that can have healthy offspring together.
- In a genetic sense, a population is a large collection of alleles, distinguished by their frequencies

Genetic populations are defined by their collections of alleles, termed the **gene pool**.

Genome comparisons among species reveal evolutionary relationships.

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Applications of Genetics and Genomics

Genetics is impacting several areas of our lives, from health care choices, to what we eat, to unraveling our pasts and guiding our futures.

"Citizen scientists" are discovering genetic information about themselves while helping researchers compile genomic databases

Thinking about genetics evokes fear, hope, anger, wonder, and despair, depending on context and circumstance.

- This has to do with how DNA information is used

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Establishing Identity

Comparing DNA sequences among individuals can rule out identity, relationships, or ancestry, or indicate the probability that two individuals are related.

DNA profiling refers to the techniques, statistical analyses, and machine learning approaches used to compare DNA sequences between & among individuals.

- Used most often in the context of forensic science
- Also useful in identifying victims of natural disasters
- Another use is to analyse foods to determine authenticity of gourmet items

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Illuminating History

DNA analysis is a time machine of sorts.

- DNA evidence may confirm findings from anthropology and history or contradict it.

DNA testing can provide views into past epidemics.

- Analysis of DNA in the mummy of the Egyptian king Tutankhamun revealed that the boy king likely died from complications of malaria and nothing else

DNA analysis confirmed that president Thomas Jefferson had children with his slave Sally Hemings.

- Today the extended family holds reunions

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DNA reveals and clarifies history

 Leslie Close/AP Photo 30

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Conservation Genetics

- Combining analysis of genetic diversity with reproductive technologies creates a way to rebuild populations that are headed toward extinction.
- This is the case for the northern white rhinoceros of Africa.
- Researchers are working on ways to bring back the species by borrowing from the genomes of the subspecies to the south

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Precision Medicine

In several nations, people are volunteering to have their genomes sequenced so that researchers can learn more about health and disease.

- Evaluating genetic data is a large part of precision medicine, which is tailoring of treatments to individuals.

The **microbiome** is the symbiotic relationship between an individual's genome, diet, lifestyle factors, and the many microbes in the body.

Pharmacogenetics considers gene variants to predict whether a specific drug will be effective or cause side effects in an individual.

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Genetic Modification

Genetic modification means altering a gene or genome in a way that does not occur in nature

Genetically modified organisms (GMOs) and drugs have been available for many years.

- They arise from **recombinant DNA technology**, which adds a gene from a different species.

A newer technology, **genome editing**, can replace, remove, or add specific genes into the cells of any organism.

- The most talked-about tool is CRISPR-Cas9

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Exome Sequencing

Exome sequencing determines the order of the DNA bases of all parts of the genome that encode proteins.

- The information is compared to databases that list many gene variants (alleles) and their associations with specific phenotypes, such as diseases.

Exome sequencing is valuable in identifying extremely rare diseases—swiftly.

- Used to be years – not a matter of hours!

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Global Perspective on Genomes 1

We share the planet with many thousands of other species.

- Many of these we cannot grow in the lab

Metagenomics is a field that involves sequencing all of the DNA in a habitat.

- Shows how species interact, and may yield new drugs and reveal novel energy sources
- The first metagenomics project described life in the Sargasso Sea.

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Global Perspective on Genomes 2

Efforts are underway to limit "genetic prospecting"

Genetics is a special branch of life science because it affects us intimately.

Social issues that parallel scientific progress

- Equal access to genetic tests and treatments
- Misuse of genetic information
- Abuse of genetics to cause harm

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Global Perspective on Genomes 3

Genetics and genomics are spawning technologies that may vastly improve the quality of life

Human genome information has tremendous potential for the entire globe.

- World organizations are discussing how nations can share new diagnostic tests and therapeutics.
- Individual nations are adopting guidelines to use genetic information to suit their strengths.

Bioethics discusses instances when genetic testing can affect privacy.

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