N201
Heredity - 2025

Overview

- **What is genetics?**
  The study of heredity and how traits are passed from parents to offspring.
- **What are genes?**
  Segments of DNA that carry instructions for traits and functions.

📚II. Basic Genetics

A. Chromosomes

- 44 autosomes (22 pairs)
- 2 sex chromosomes (1 pair)
- = **46 total chromosomes in humans**

Sex determination:

- XX → Female
- XY → Male

B. Phenotype – outward appearance or expression of genes.
C. Genotype – actual genetic makeup of an organism.
D. Genome – complete set of genes/DNA in an organism.

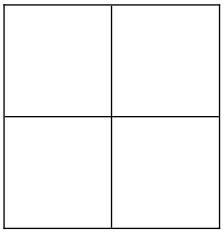
E. Mendelian Inheritance – ability to predict inheritance.

- **Homozygous** – two like genes for a trait
- **Heterozygous** – two different genes for a trait
- **Dominant** – expressed trait even if only one allele present
- **Recessive** – masked unless both alleles are recessive

III. Inheritance Patterns

Example:

- Father: **BB** (brown eyes, homozygous dominant)
- Mother: **bb** (blue eyes, homozygous recessive)
- Offspring prediction →



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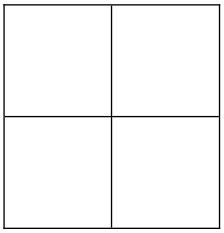
IV. Autosomal Dominance

- Dominant gene causes disease expression.
- One parent usually has the disease.
- Both sexes equally affected.
- Examples: Huntington's chorea, Osteogenesis Imperfecta, Dwarfism.

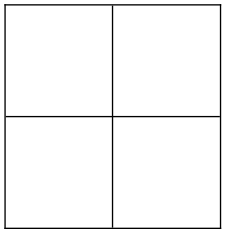
Example 1: D = Disease trait, h = healthy trait

- Father: **Dh** (disease)
- Mother: **hh** (healthy)
- Offspring Prediction:



Example 2: D = Disease trait, h = healthy trait

- Both parents: **Dh**
- Prediction:

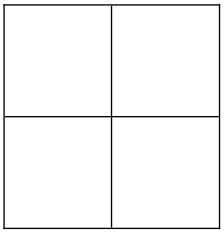


V. Autosomal Recessive

- Disorder occurs only if **two recessive genes** are present.
- Carriers (heterozygous) are healthy but carry the gene.
- Examples: Cystic fibrosis, Tay Sachs, Galactosemia, PKU.

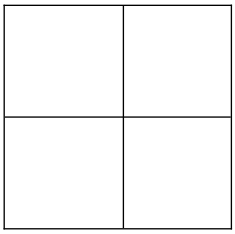
Example 1:

- Parents: **Hd × Hd** (both carriers, disease free)
- Prediction:



Example 2:

- Mother: **Hd** (carrier)
- Father: **HH** (healthy)
- Prediction:



♀ VI. X-Linked Dominant (sex linked)

- Passed on the **X chromosome (female)**.
- One gene = symptoms.
- Example: Fragile X Syndrome.

♂ VII. X-Linked Recessive

- Most X-linked disorders are recessive.
- Females less often affected (normal second X blocks disorder).
- More common in males.
- Example: Hemophilia.

VIII. Chromosomal Abnormalities

- Abnormal formation of chromosomes
- Can be **numerical or structural**.
- Lead to profound abnormalities or disabilities.

🔍 I X. Diagnostic Testing

A. Karyotyping – visual map of chromosomes.
   a. Blood sample or scraping of cells from buccal membrane, stain, magnify, and photograph

B. Alpha-Fetoprotein (AFP) – blood test, 15–20 weeks, screens for neural tube defects (NTD).
   a. glycoprotein produced by the fetal liver
   b. Present in the amniotic fluid or maternal serum
   c. If increased- possible NDT. False positives if date of conception is not accurate

C. Amniocentesis – after 15 weeks, detects NTDs, risk of complications.

D. Chorionic Villi Sampling (CVS) – 10–13 weeks, Chromosomal analysis that tests placental tissue. Cannot dx NTD.

E. Cell-Free Fetal DNA Screen – maternal blood test (11–13 weeks) for chromosome

abnormalities, sex, Rh, and some single-gene disorders. Positive results needs f/u with CVS or amniocentesis.

⚕X. Genetic Counseling

- Purpose: Estimate risk of genetic disease in a child.
- Timing: Before conception.
- Nursing implications:
  - Stay updated on genetic testing advances.
  - Provide support and understanding.
  - Patient teaching.