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HUMAN ANATOMY AND PHYSIOLOGY – II

UNIT 5

TOPIC :

- **Introduction to genetics**

  Chromosomes, genes and DNA, protein synthesis, genetic pattern of inheritance

Introduction to Genetics

- Genetics is the science of heredity and variation. It deals with the study of genes, inheritance of traits, and how characteristics are passed from parents to offspring in all living organisms. Genetics forms a fundamental part of biology and provides the foundation for understanding evolution, development, and disease.
- The idea that offspring resemble their parents and inherit characteristics from them has been known for centuries. However, modern genetics goes further to explain the mechanisms behind inheritance at the molecular level, particularly through the study of DNA and genes.
- The unit of heredity is the gene, which is composed of deoxyribonucleic acid (DNA). DNA is tightly coiled and organized into chromosomes. Each gene contains coded instructions for making specific proteins that control various biological functions and traits. Some disorders in humans arise from mutations in a single gene, while others involve multiple genes or complex interactions with environmental factors.
- In recent years, advances in genetics have revealed that individuals with the same genetic disease often share similar gene patterns. This has led to the development of personalized medicine, where treatment can be tailored to an individual's genetic makeup. This approach helps improve treatment effectiveness and reduces the risk of adverse drug reactions.
- Genetics also helps us understand the structure and function of genes, their locations in the nucleus, and how gene sequences determine the phenotypic traits (observable characteristics) of organisms. It explains which traits are inherited, how they are transmitted across generations, and how gene expression (turning genes on or off) influences cell behaviour. It also includes the study of cell division processes like mitosis and meiosis, which are essential for growth, repair, and reproduction.

Chromosomes

- Chromosomes are thread-like structures composed of DNA and proteins located in the nucleus of eukaryotic cells. They carry genetic information essential for the regulation of growth, development, and reproduction.

  - In prokaryotes: Single circular chromosome (not enclosed in a nucleus)
  - In eukaryotes: Multiple linear chromosomes found inside the nucleus

Structure of a Chromosome

- **Chromatin** – DNA + histone proteins in uncondensed form; becomes chromosome during cell division.
- **Chromatid** – Each chromosome has two identical sister chromatids after replication.
- **Centromere** – The point where two chromatids are joined.
- **Telomeres** – Protective ends that prevent degradation.
- **Arm** – Short arm (p) and long arm (q).
- **Kinetochore** – Protein complex on the centromere; attaches to spindle fibers during cell division.

Types of Chromosomes (Based on Centromere Location):

Type	Centromere Position	Shape
Metacentric	Center	V-shaped
Submetacentric	Slightly off center	L-shaped
Acrocentric	Near one end	Rod-like
Telocentric	At the terminal end	I-shaped

Packaging of DNA in Chromosomes

- **DNA wraps around histone proteins** to form nucleosomes.
- Nucleosomes coil to form **chromatin fiber**.
- Fibers condense into **chromosomes** during cell division.

Human Chromosome Number

- Total: **46 chromosomes** (23 pairs)
  - **22 pairs = Autosomes** (non-sex chromosomes)
  - **1 pair = Sex chromosomes** (XX for females, XY for males)

Types of Chromosomes (Based on Function)

1. **Autosomes**:
   - Control general body traits and functions
   - Numbered 1 to 22
2. **Sex Chromosomes**:
   - Determine the **sex** of an individual
   - XX (female), XY (male)

Functions of Chromosomes

- ✓ **Genetic Information Storage**:
  - Each chromosome contains hundreds to thousands of **genes**.
  - Genes encode proteins that perform essential life functions.
- ✓ **Transmission of Hereditary Traits**:
  - Chromosomes pass genetic information **from one generation to the next** during reproduction.
- ✓ **Gene Regulation**:
  - Chromosomes regulate **which genes are turned on or off**, controlling cell function and differentiation.
- ✓ **Cell Division**:
  - During mitosis and meiosis, chromosomes **ensure proper distribution of DNA** to daughter cells.
- ✓ **Determination of Sex**:
  - **Sex chromosomes (X and Y)** decide the biological sex of the offspring.

Role in Mitosis and Meiosis

- In **Mitosis**: Ensures equal chromosome distribution to daughter cells (for growth and repair)
- In **Meiosis**: Ensures halving of chromosome number in gametes, enabling genetic diversity through recombination and independent assortment

Gene

- A gene is a specific segment of DNA that codes for a particular protein or functional RNA. Genes are the basic units of heredity and are passed from parents to offspring.
  - Each gene contains a specific sequence of nucleotides.
  - It determines a trait or function such as eye color, blood type, or enzyme production.

Location of Genes

- Genes are located on chromosomes within the nucleus of a cell.
- Each human cell contains around 20,000 to 25,000 genes.
- Genes also exist in mitochondria (mitochondrial genes), passed maternally.

Structure of a Gene

Gene Region	Function
Promoter	Starting point for transcription; RNA polymerase binds here
Exons	Coding regions that are expressed into proteins
Introns	Non-coding regions removed during RNA splicing
Terminator	Signals the end of transcription

Types of Genes

Type	Function
Structural genes	Code for proteins (enzymes, receptors, etc.)
Regulatory genes	Control the activity of other genes
Housekeeping genes	Always active for basic cellular functions
Non-coding genes	Produce functional RNA (e.g., tRNA, rRNA) but not proteins

Functions of Genes

- ✓ **Protein synthesis** – Each gene codes for a specific protein.
- ✓ **Heredity** – Genes carry traits from parents to offspring.
- ✓ **Cell function** – Genes regulate growth, repair, and metabolism.
- ✓ **Development** – Guide the formation of tissues and organs.
- ✓ **Gene expression control** – Some genes are turned on/off depending on need.

DNA (Deoxyribonucleic Acid)

- DNA is the genetic material found in all living organisms. It carries the hereditary information responsible for the growth, development, reproduction, and functioning of all life forms.
  - DNA stands for Deoxyribonucleic Acid.
  - It is present in the nucleus of eukaryotic cells and in the cytoplasm of prokaryotic cells.

Composition of DNA

- DNA is a polymer made up of repeating units called nucleotides. Each nucleotide is composed of three main components:
  - Nitrogenous base
  - Pentose sugar (deoxyribose)
  - Phosphate group
- These components form the basic building block of DNA.

1. **Nitrogenous Bases :** Organic molecules containing nitrogen; responsible for base pairing

   There are **four bases** in DNA:

   - **Purines** (double-ring structures):
     - **Adenine (A)**
     - **Guanine (G)**
   - **Pyrimidines** (single-ring structures):
     - **Cytosine (C)**
     - **Thymine (T)**

   Base pairing rule:

   - A pairs with T (2 hydrogen bonds)
   - C pairs with G (3 hydrogen bonds)

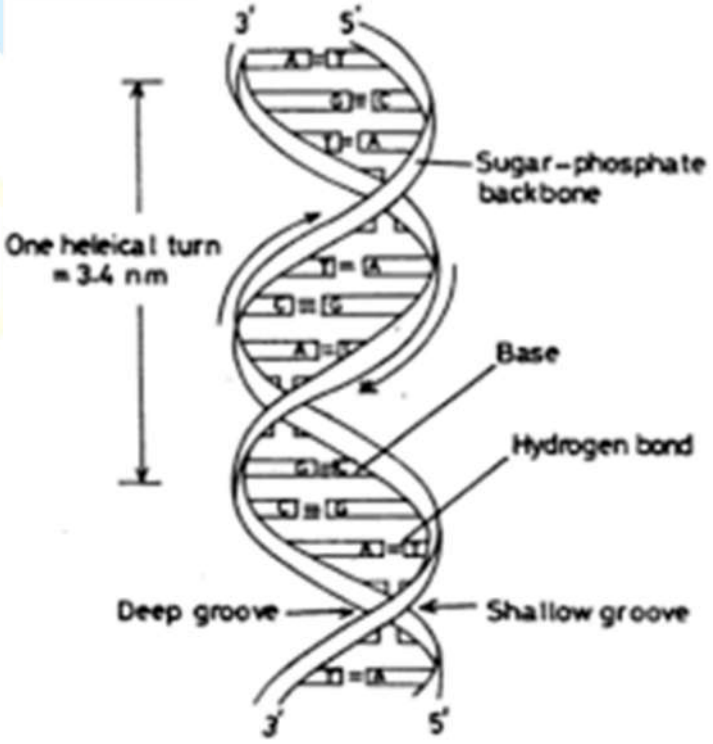
2. **Pentose Sugar :** A 5-carbon sugar (Deoxyribose); forms the backbone of DNA

    - A five-carbon sugar ($C_5H_{10}O_4$)
    - Lacks one oxygen atom on the 2′ carbon (hence, "deoxy")
    - Bonds to the nitrogenous base at carbon 1′ and to the phosphate group at carbon 5′

3. **Phosphate Group :** Links the sugar of one nucleotide to the next via phosphodiester bond
    - ♦ Contains one phosphorus atom bonded to four oxygen atoms (PO_4^{3-})
    - ♦ Forms phosphodiester bonds between adjacent nucleotides
    - ♦ Connects the 3′ carbon of one sugar to the 5′ carbon of the next sugar, forming the sugar-phosphate backbone

Structure of DNA (Double Helix Model by Watson & Crick)

- DNA as an acidic substance present in the nucleus was first identified by Frederich Meischer in 1869. He named it as 'nucleon'. Due to technical limitations in isolating such a long polymer intact the elucidation of structure of DNA remained elusive for a long period of time.
- It was only in 1953 that James Watson and Francis Crick proposed the very simple but famous double helix model for the structure of DNA.
- The main opposition was base pairing between the two strands of polynucleotide chains



Watson and Crick Model of DNA Molecule

The salient features of double helix structure of DNA are as follows :

- It is made up of two polynucleotide chains.
- The two chains have antiparallel polarity if one has polarities and the second chain must have polarity.

- The base into strands is paired through hydrogen bond forming base pairs. Adenine forms to hydrogen bonds with thymine from opposite strands and vice versa.
- Similarly guanine forms three H bonds with cytosine. As a result, purine comes opposite to pyrimidine.
- Because of this approximate a uniform distance between the two strengths of The Helix occurs.
- The two chains are called in a right-handed fashion. Pitch of the helix is and there are roughly 10bp in each turn.
- The plane of one base pair is stacked over the other in a double helix. This confirms stability of the helical structure.

Function of DNA

- ✓ **Genetic Information :** It transfer genetic information from mother to the child.
- ✓ **Replication :** It makes its copy during cell division.
- ✓ **Transcription :** When RNA is formed from DNA it is called transcription.
- ✓ **DNA finger print :** It help to determine the identity of suspected person on the basis of nucleotide sequences

Protein Synthesis

→ Protein synthesis is the biological process by which cells produce proteins, which are essential for the structure, function, and regulation of the body's tissues and organs. The process involves two main stages: Transcription and Translation.

Transcription (in the Nucleus)

What Happens

- A **gene on the DNA** is **copied into mRNA (messenger RNA)**.
- This process is **regulated by enzymes** and **RNA polymerase**.

Steps of Transcription

1. **Initiation**:
   - RNA polymerase binds to a **promoter region** on DNA.
2. **Elongation**:
   - RNA polymerase reads the DNA template and synthesizes **mRNA** by matching complementary RNA bases (A-U, C-G).
   - Note: **Uracil (U)** replaces **Thymine (T)** in RNA.
3. **Termination**:
   - Transcription ends when RNA polymerase reaches a **stop signal.**
   - The newly formed **pre-mRNA** detaches from DNA.

Translation (in the Cytoplasm at the Ribosome)

What Happens

- **mRNA is read** to assemble a specific **sequence of amino acids** (protein).
- Involves **tRNA, ribosomes**, and **amino acids**.

Steps of Translation

1. **Initiation**:
   - Ribosome binds to mRNA at the **start codon (AUG)**.
   - First tRNA (carrying **methionine**) binds to start codon.
2. **Elongation**:
   - tRNA molecules bring **specific amino acids** based on codon-anticodon pairing.
   - Ribosome links amino acids with **peptide bonds**, forming a **polypeptide chain.**
3. **Termination**:
   - When a **stop codon (UAA, UAG, UGA)** is reached, translation ends.
   - The completed protein is released and undergoes **folding and modification.**

Genetic Pattern of Inheritance

→ The genetic pattern of inheritance refers to the manner in which genes and traits are passed from parents to offspring.

→ These patterns follow the principles of Mendelian genetics and include both dominant and recessive traits that may be autosomal or sex-linked.

Basic Terminology

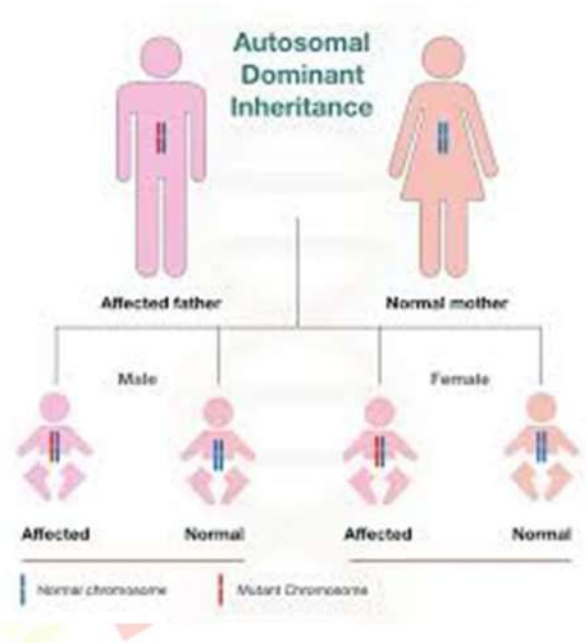
Term	Meaning
Gene	A segment of DNA that codes for a specific trait
Allele	Alternative forms of a gene (e.g., A and a)
Dominant allele	Expressed even if only one copy is present (e.g., Aa = trait shown)
Recessive allele	Expressed only if both copies are present (e.g., aa = trait shown)
Homozygous	Two identical alleles (AA or aa)
Heterozygous	Two different alleles (Aa)
Genotype	Genetic makeup (e.g., AA, Aa, aa)
Phenotype	Physical expression of the genotype (e.g., brown eyes, blue eyes)

Types of Inheritance Patterns

Type	Inheritance Description
1. Autosomal Dominant	Trait appears in every generation; only one dominant allele needed
2. Autosomal Recessive	Trait appears when both alleles are recessive; may skip generations
3. X-linked Dominant	Dominant gene on X chromosome; affects males and females
4. X-linked Recessive	Recessive gene on X; males more affected (only one X chromosome)
5. Y-linked	Trait passed only from father to son via Y chromosome
6. Mitochondrial Inheritance	Passed from mother to all children via mitochondrial DNA
7. Codominance	Both alleles expressed equally in phenotype
8. Incomplete Dominance	Blending of traits in heterozygous condition
9. Polygenic Inheritance	Multiple genes contribute to a single trait (e.g., height, skin color)
10. Multifactorial Inheritance	Trait influenced by genes and environment (e.g., diabetes, heart disease)

1. Autosomal Dominant Inheritance:

- Only one copy of the dominant gene needed for trait to be expressed
- Affects males and females equally
- Appears in every generation



Examples:

- Huntington's disease
- Marfan syndrome
- Familial hypercholesterolemia

Genotypes:

- AA = Affected
- Aa = Affected
- aa = Normal

2. Autosomal Recessive Inheritance:

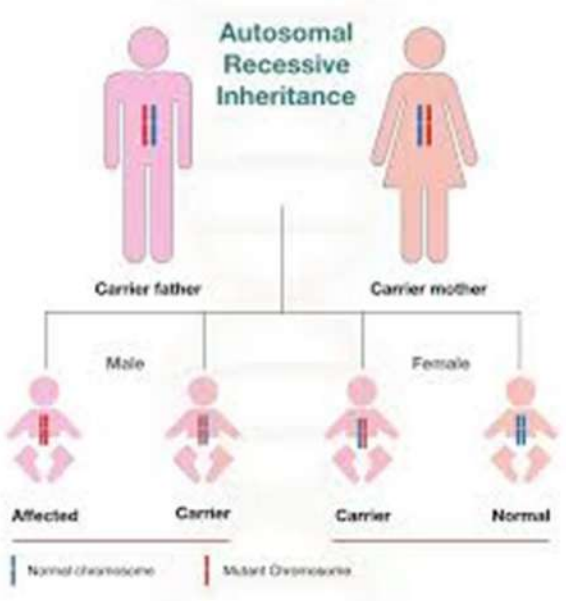
- Two copies of the recessive allele required
- Often skips generations
- Carriers (heterozygous) do not show symptoms

Examples:

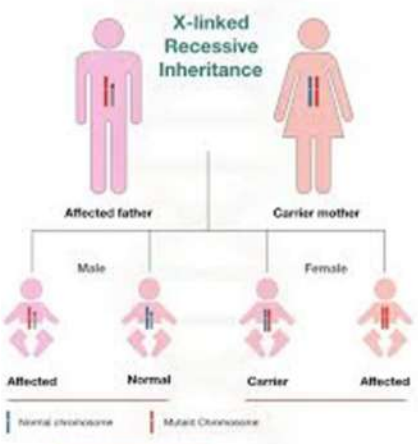
- Cystic fibrosis
- Sickle cell anemia
- Phenylketonuria (PKU)

Genotypes:

- AA = Normal
- Aa = Carrier
- aa = Affected



3. X-linked Dominant Inheritance:

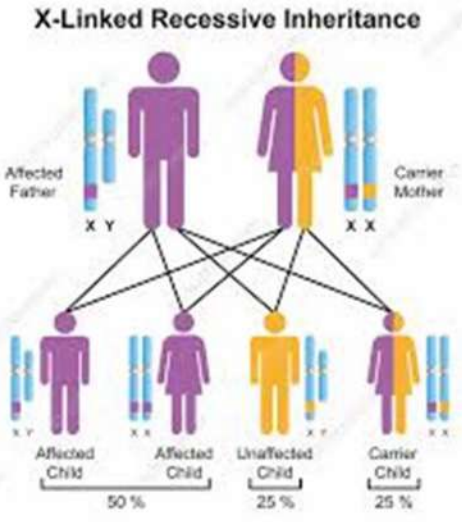


- Dominant gene on the X chromosome
- Affects both males and females, but females may be less severe
- An affected father passes to all daughters, not sons

Examples:

- Rett syndrome
- Fragile X syndrome

4. X-linked Recessive Inheritance:

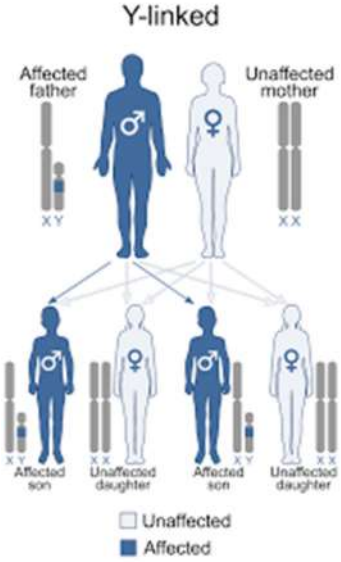


- Gene located on X chromosome
- Males more affected (only one X)
- Females are usually carriers

Examples:

- Hemophilia
- Duchenne muscular dystrophy
- Red-green color blindness

5. Y-linked Inheritance:

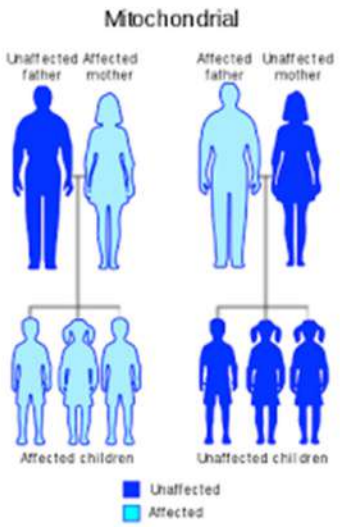


- Gene located on Y chromosome
- Only males are affected
- Passed from father to son only

Example:

- Y-chromosome infertility
- Hypertrichosis (hairy ears)

6. Mitochondrial Inheritance:



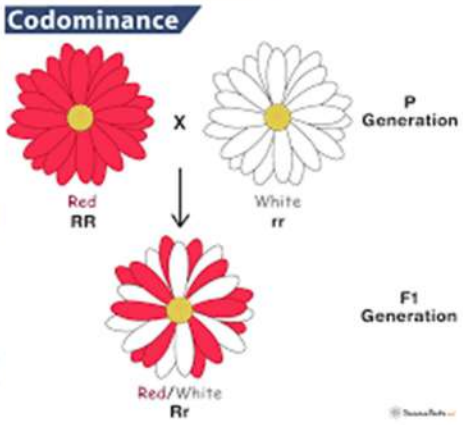
- Gene inherited only from mother
- All children of an affected mother may show the trait
- Only females pass it to the next generation

Examples:

- Leber's hereditary optic neuropathy
- Mitochondrial myopathy

7. Codominance:

- Both alleles in a heterozygous genotype are fully expressed
- No blending

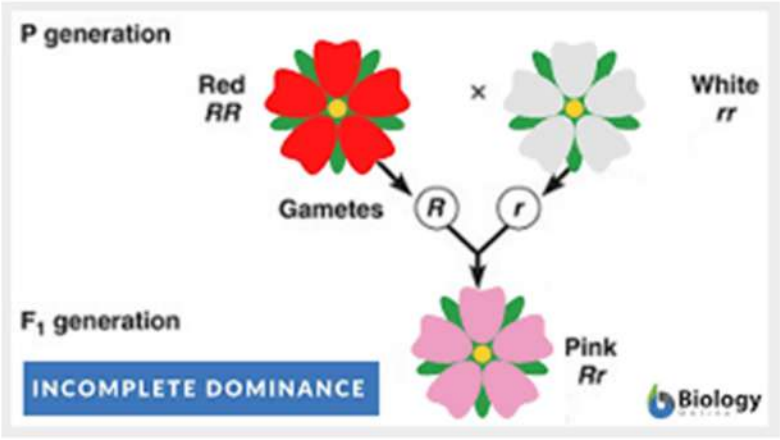


Example:

- AB blood group (A and B alleles both expressed)
-

8. Incomplete Dominance:

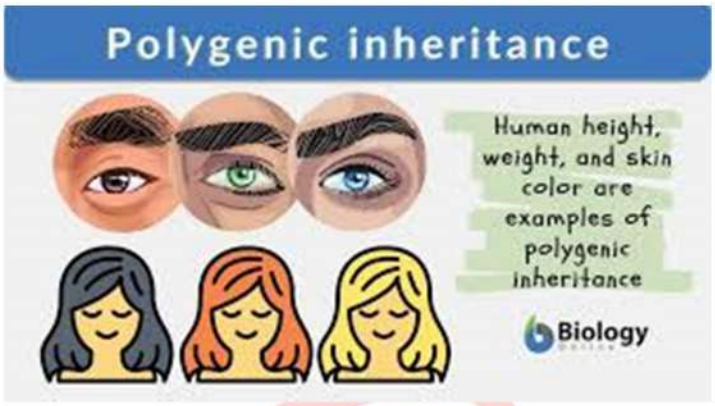
- Heterozygous condition shows a blend of traits



Example:

- Red (RR) × White (rr) = Pink (Rr) flower in snapdragon

9. Polygenic Inheritance:



- Multiple genes influence a single trait
- Trait shows continuous variation

Examples:

- Height
- Skin color
- Intelligence

10. Multifactorial Inheritance:

- Trait influenced by genetic and environmental factors

Examples:

- Diabetes
- Hypertension
- Cleft lip
- Congenital heart disease