General Pathology

Human Genetics

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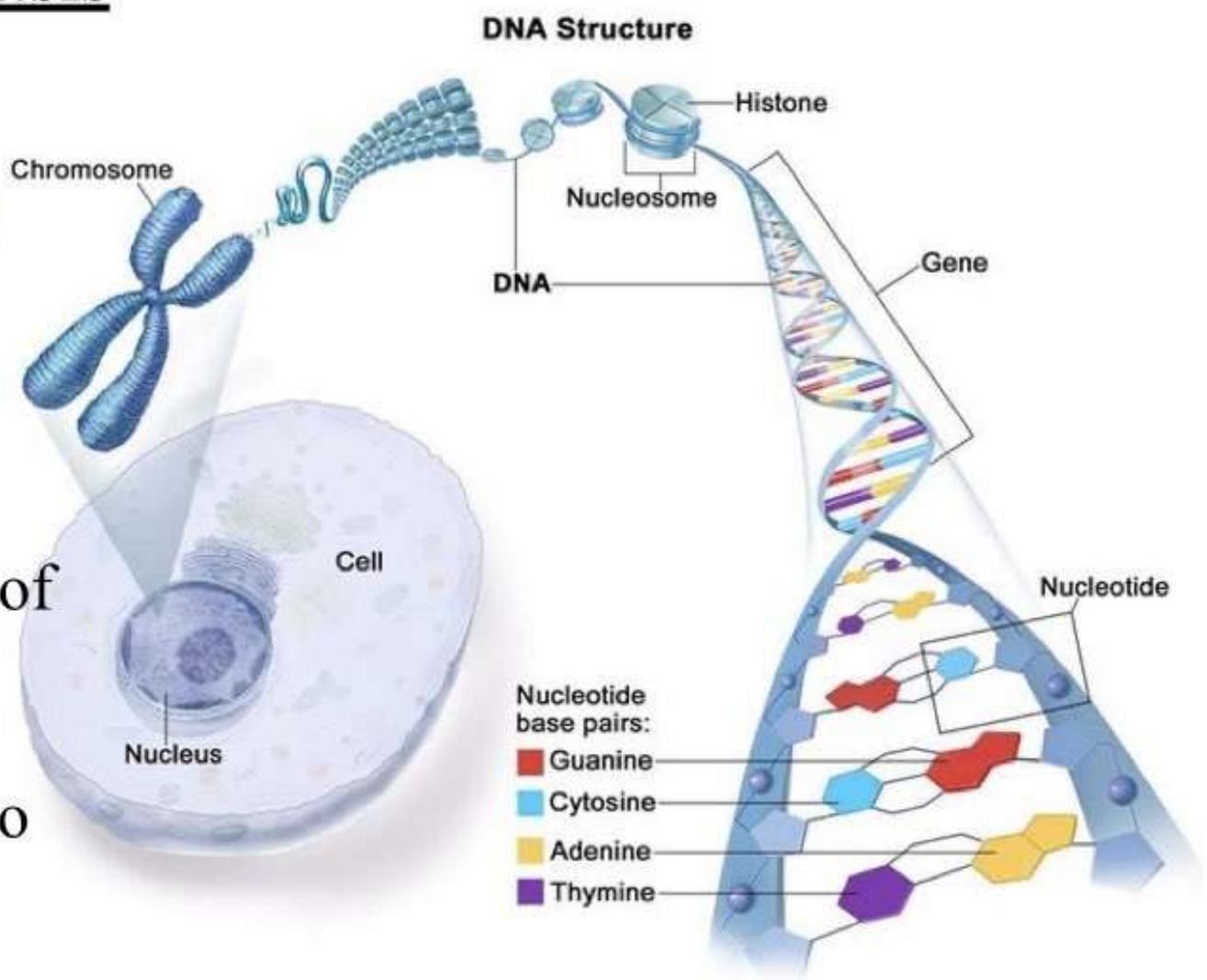
Cellular & Genetic Basis

The cell is the smallest functional unit.

DNA (Deoxyribonucleic acid) stores genetic information in the cell nucleus.

Genes, the basic units of inheritance, are segments of DNA and provide the blueprint for proteins.

Chromatin condenses into chromosomes before cell division. Humans have **20,000–25,000 genes and 23 pairs of chromosomes**.



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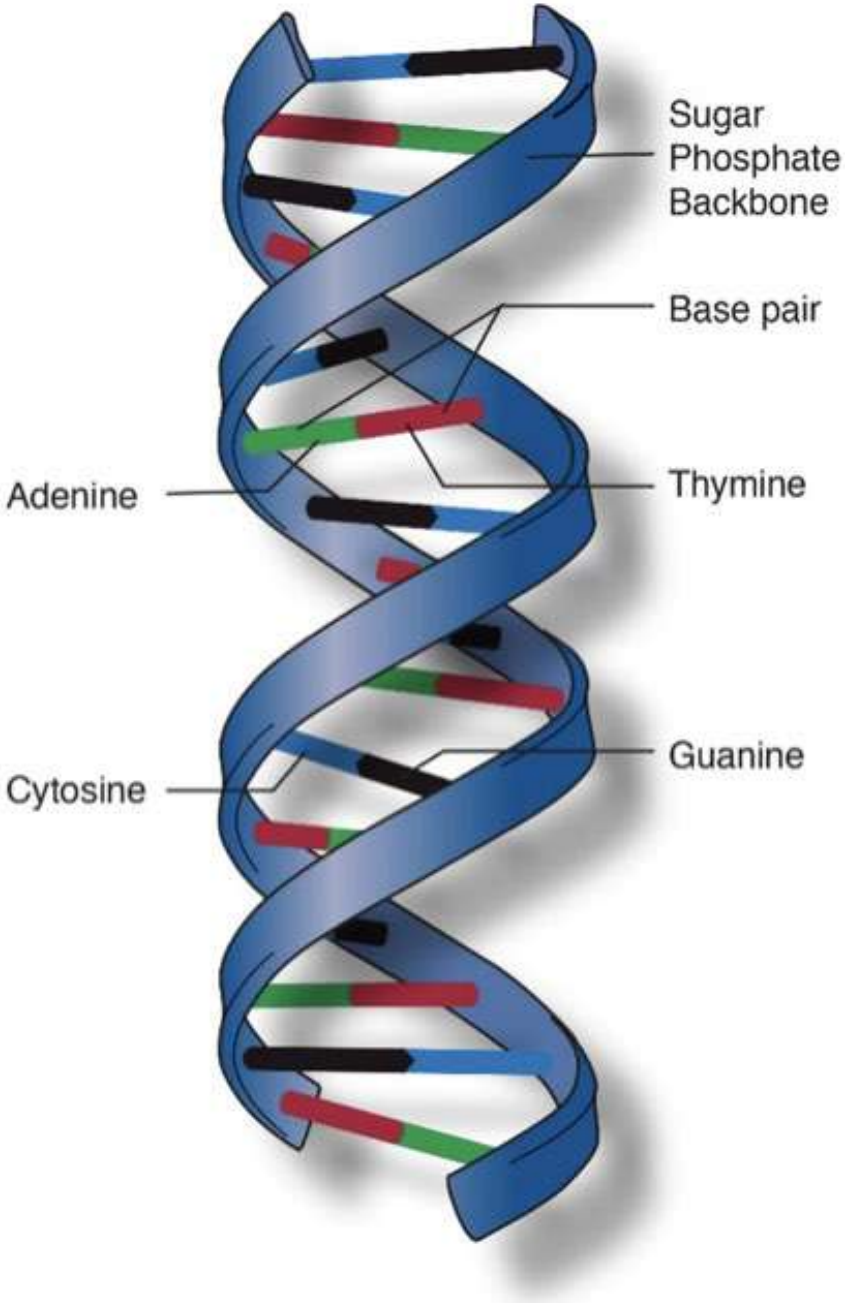
DNA, RNA, and Protein Synthesis

DNA Structure: A double-stranded helix made of nucleotides (sugar, phosphate, and a nitrogenous base: A, T, C, G). A pairs with T; C pairs with G.

DNA Replication: The semiconservative process of copying DNA prior to cell division.

RNA (Ribonucleic acid) is involved in protein synthesis.



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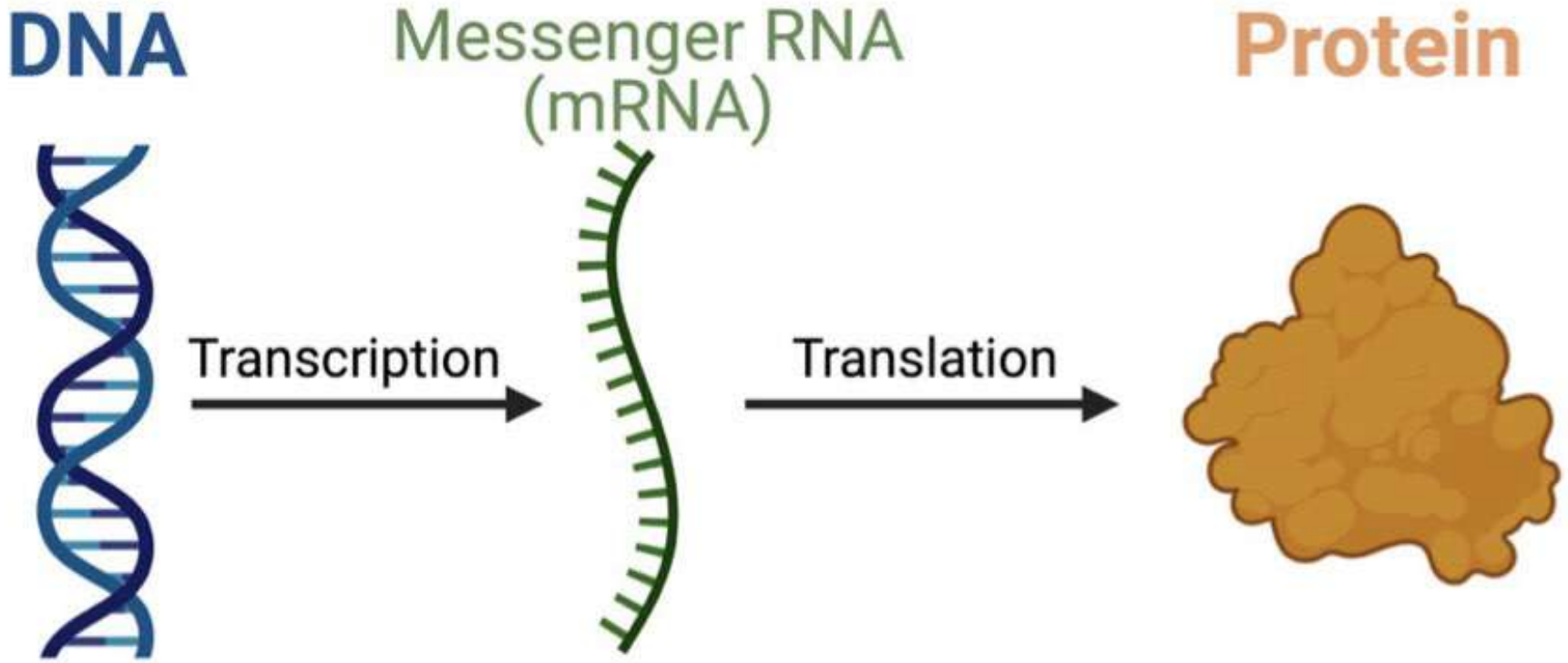

DNA, RNA, and Protein Synthesis

Transcription: DNA is copied into mRNA (messenger RNA). (Uracil 'U' replaces Thymine 'T' in RNA, so A pairs with U).

Translation: mRNA instructions are read by ribosomes (rRNA) and used by tRNA (transfer RNA) to deliver amino acids to synthesize a protein. A group of three mRNA nucleotides is a codon.

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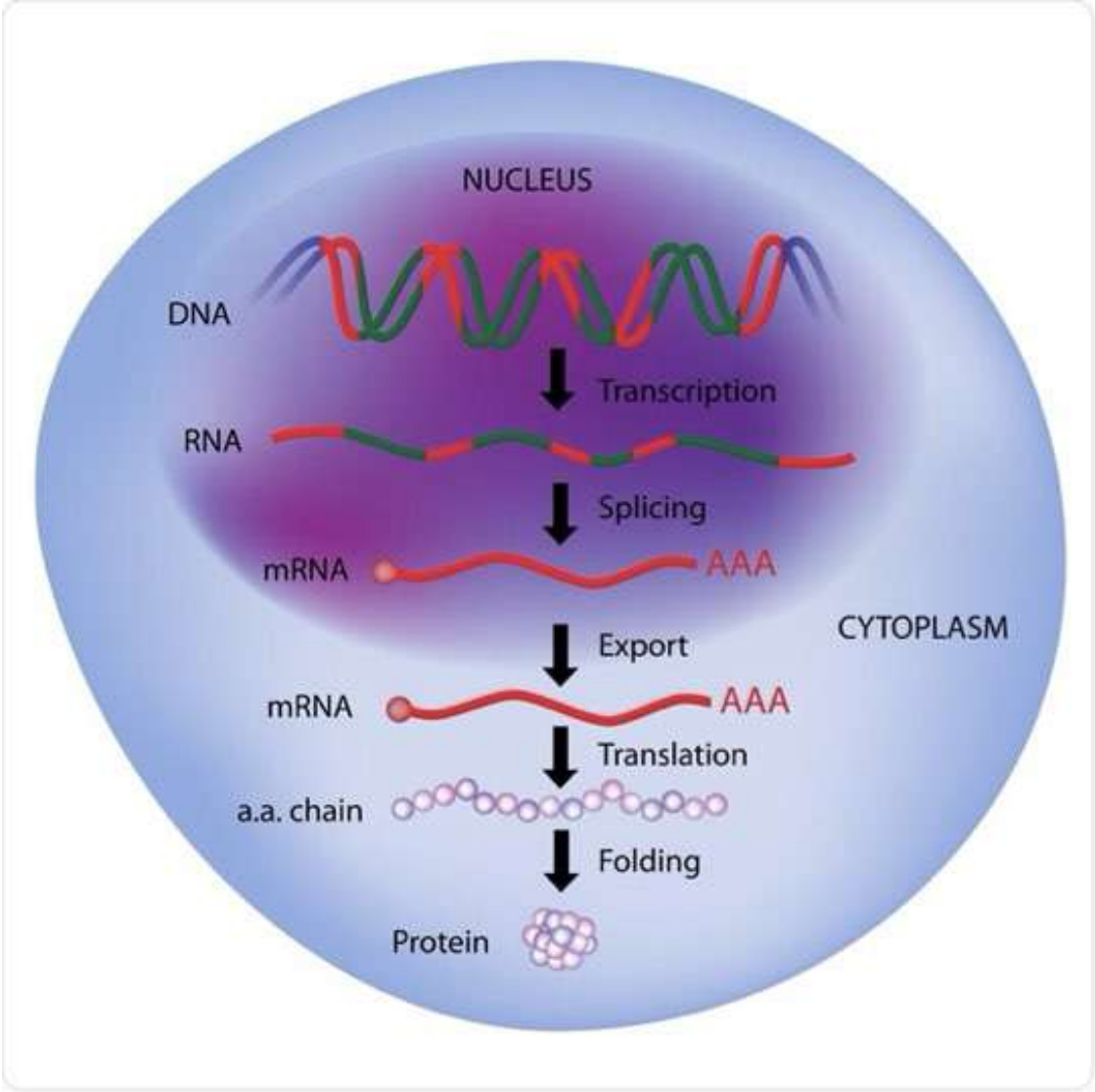

Gene expression is the fundamental process by which the information encoded in a gene (a segment of DNA) is used to create a functional gene product, typically a **protein** or a functional RNA molecule. This process determines the structure and function of a cell and is central to life.

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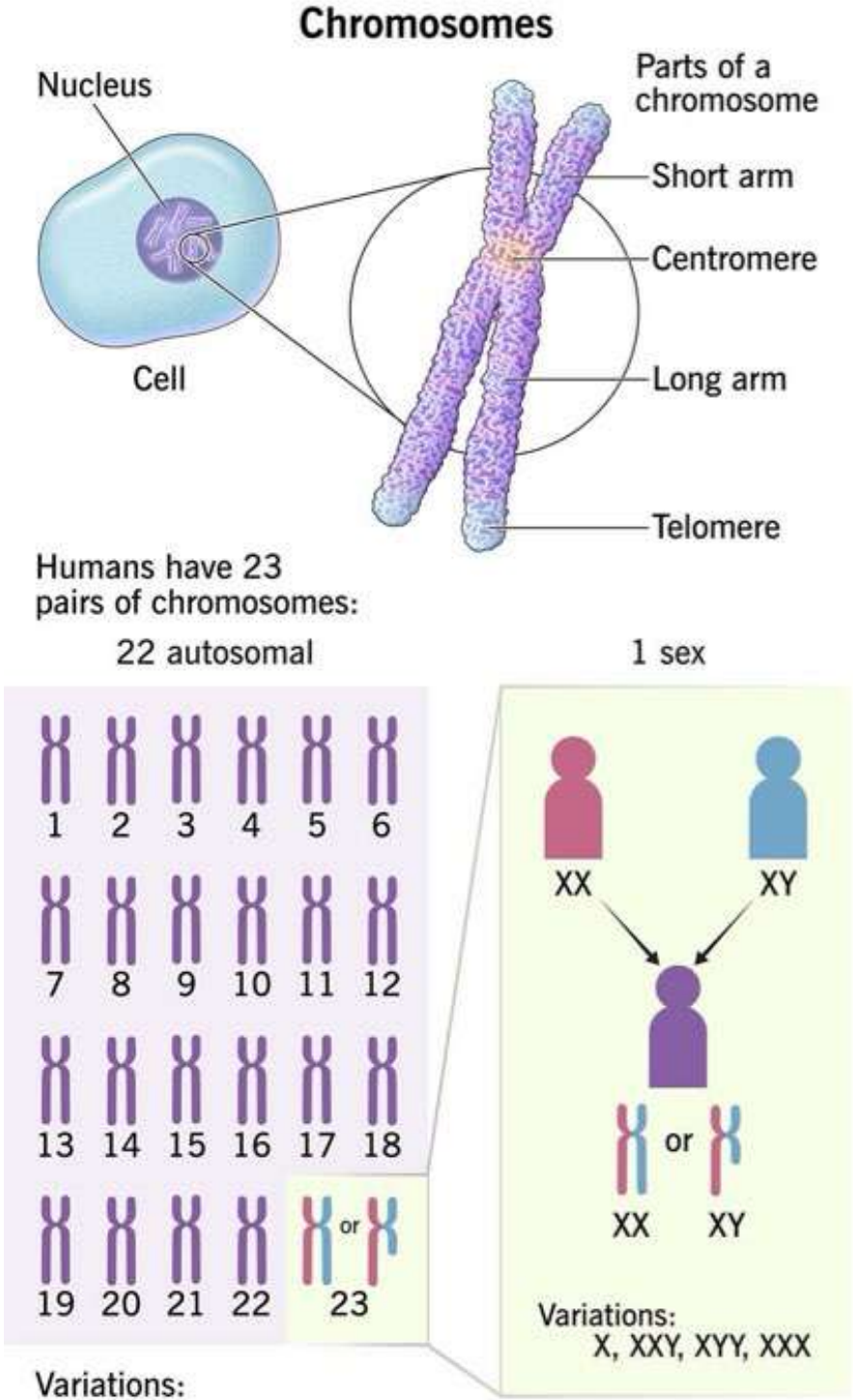

Chromosomes & Inheritance

Chromosomes: Coiled DNA and protein. Structurally divided by a centromere into a short arm (p) and a long arm (q).

Chromosomes Pairs: 22 pairs of autosomes and 1 pair of sex chromosomes (XX-female, XY-male).

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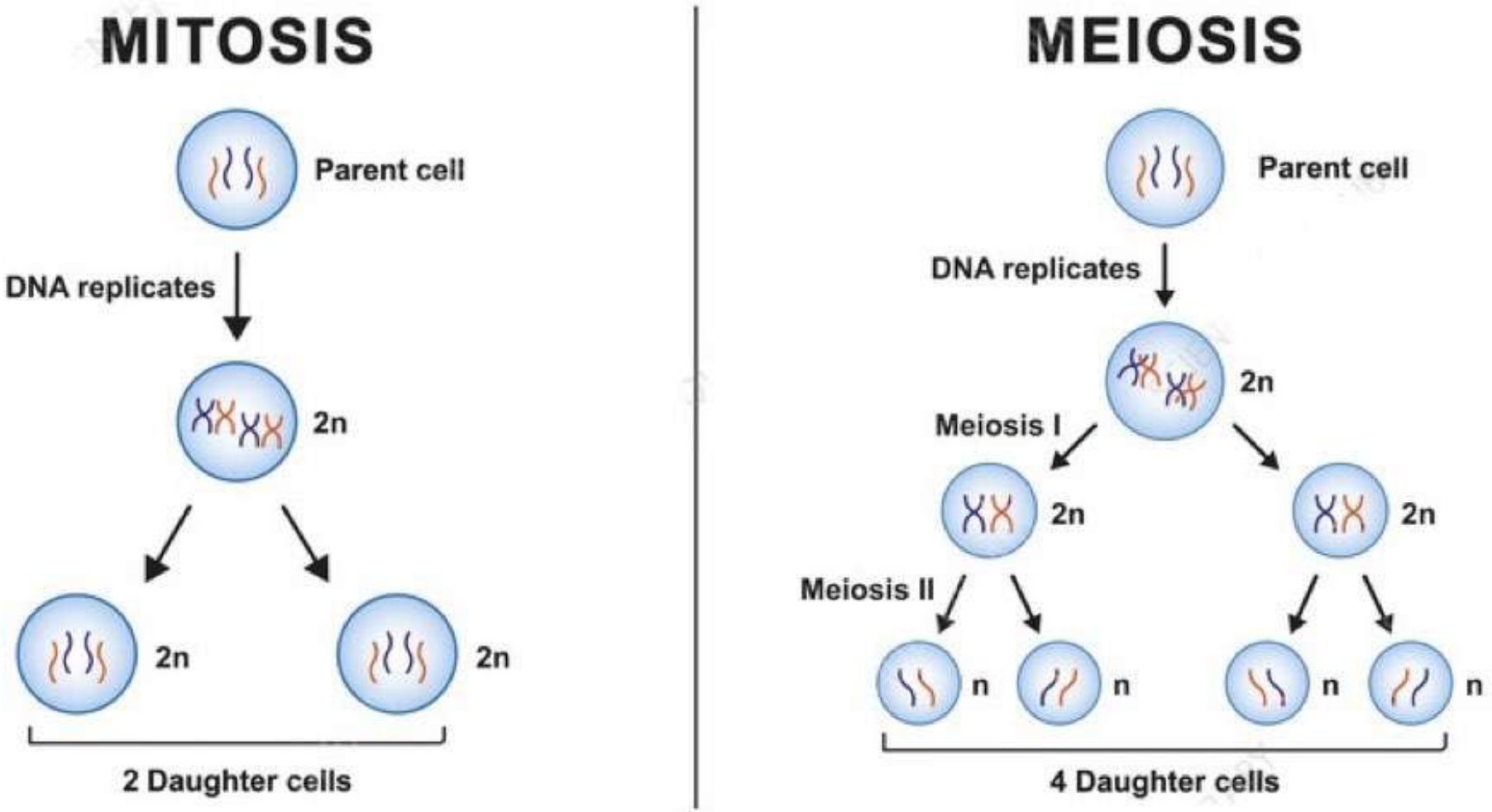

Cell Division:

Mitosis: Somatic cell division, results in diploid (2n) daughter cells.

Meiosis: Germ cell division, results in haploid (n) gametes (sperm/ovum).

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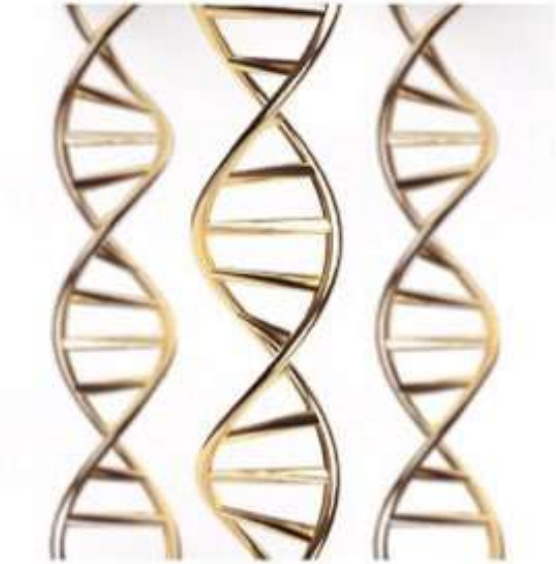
Genotype

Genotype is the genetic makeup of an organism, including all the DNA sequences that determine inherited traits.



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Phenotype

Phenotype is the set of observable characteristics or traits of an organism, resulting from the interaction of its genotype with the environment.

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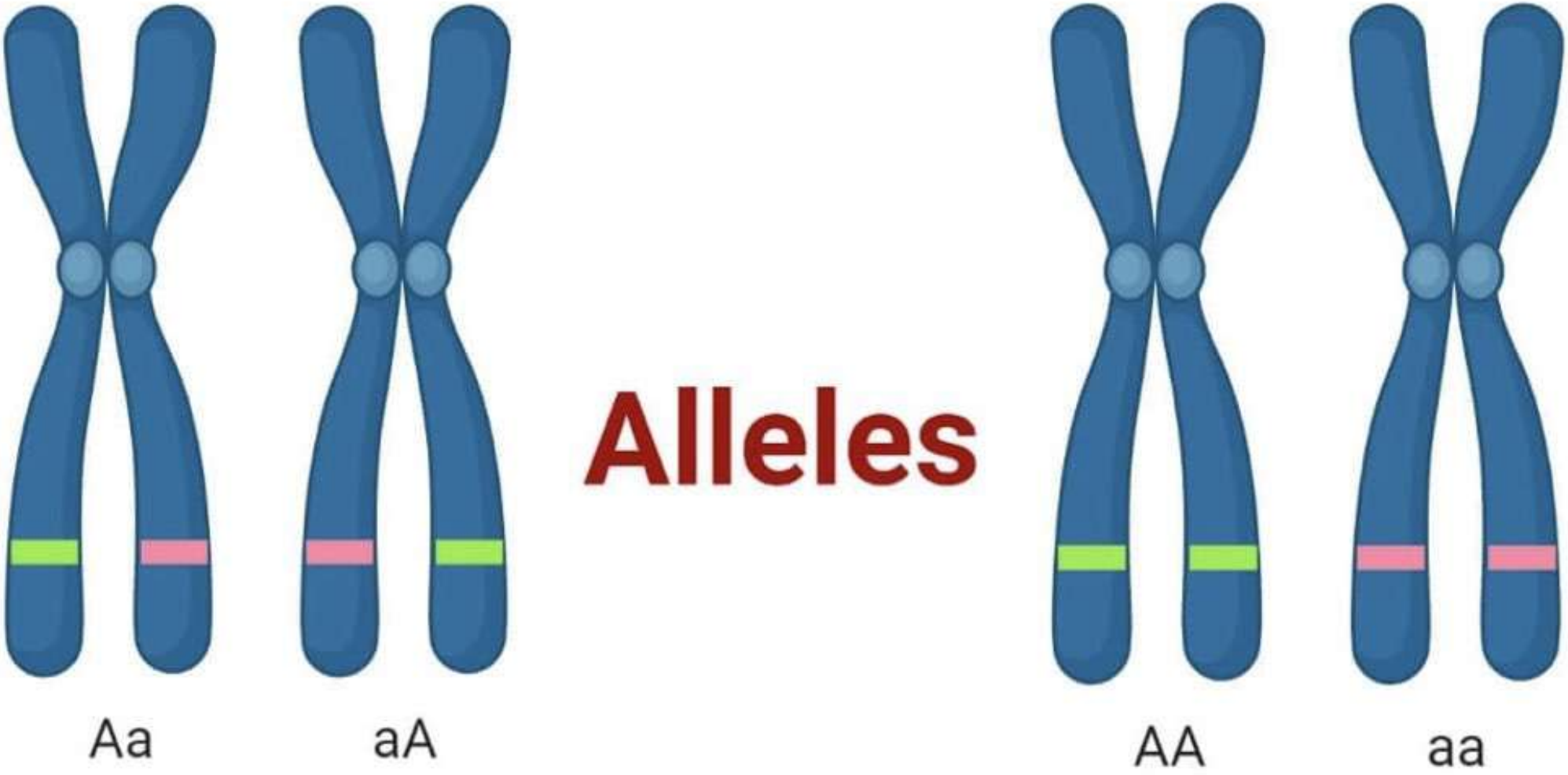

Alleles: Alternate forms of a gene at a locus (position).

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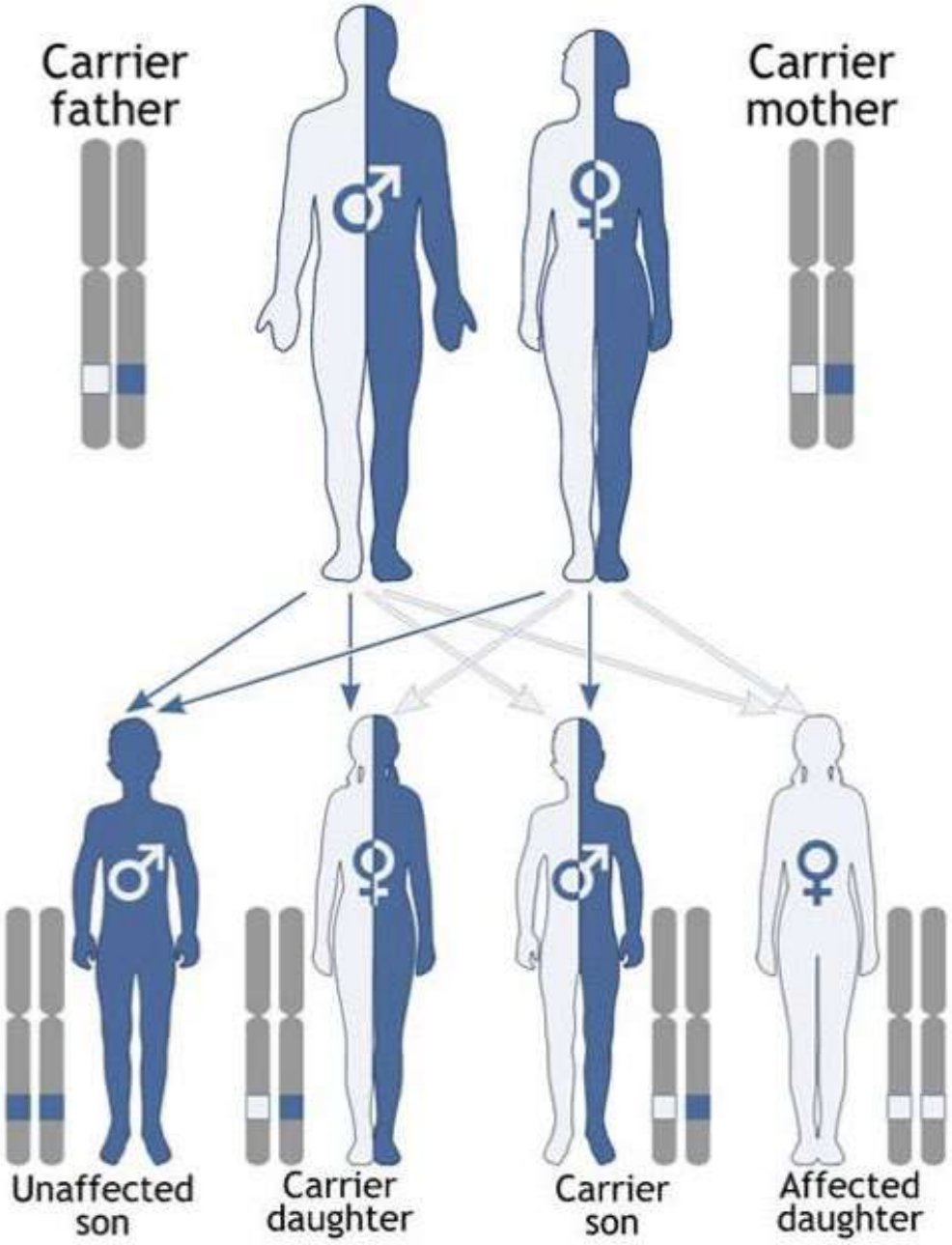

Inheritance: Can be single-gene or polygenic (multiple genes), often including multifactorial (environmental) effects.

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