Al-Mustaqbal University College
Department of Nursing

Pathophysiology

Lecture \3

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Gene and Chromosome

A gene : is the basic physical and functional unit of heredity .Genes , which are made up of DNA , that provide the coded instructions for synthesis of RNA, which, when translated into protein, leads to the expression of hereditary character.

In humans, genes vary in size from a few hundred DNA bases to more than 2 million bases. Different forms of genes, called alleles, determine how these characters are expressed in a given individual. Humans are thought to have about 35,000 genes, while bacteria have between 500 and 6,000. Humans have 23 pairs of chromosomes or a total of 46 , anything that lives depends on genes. They possess the data to build and maintain cells and pass genetic information to offspring.

Gene component :

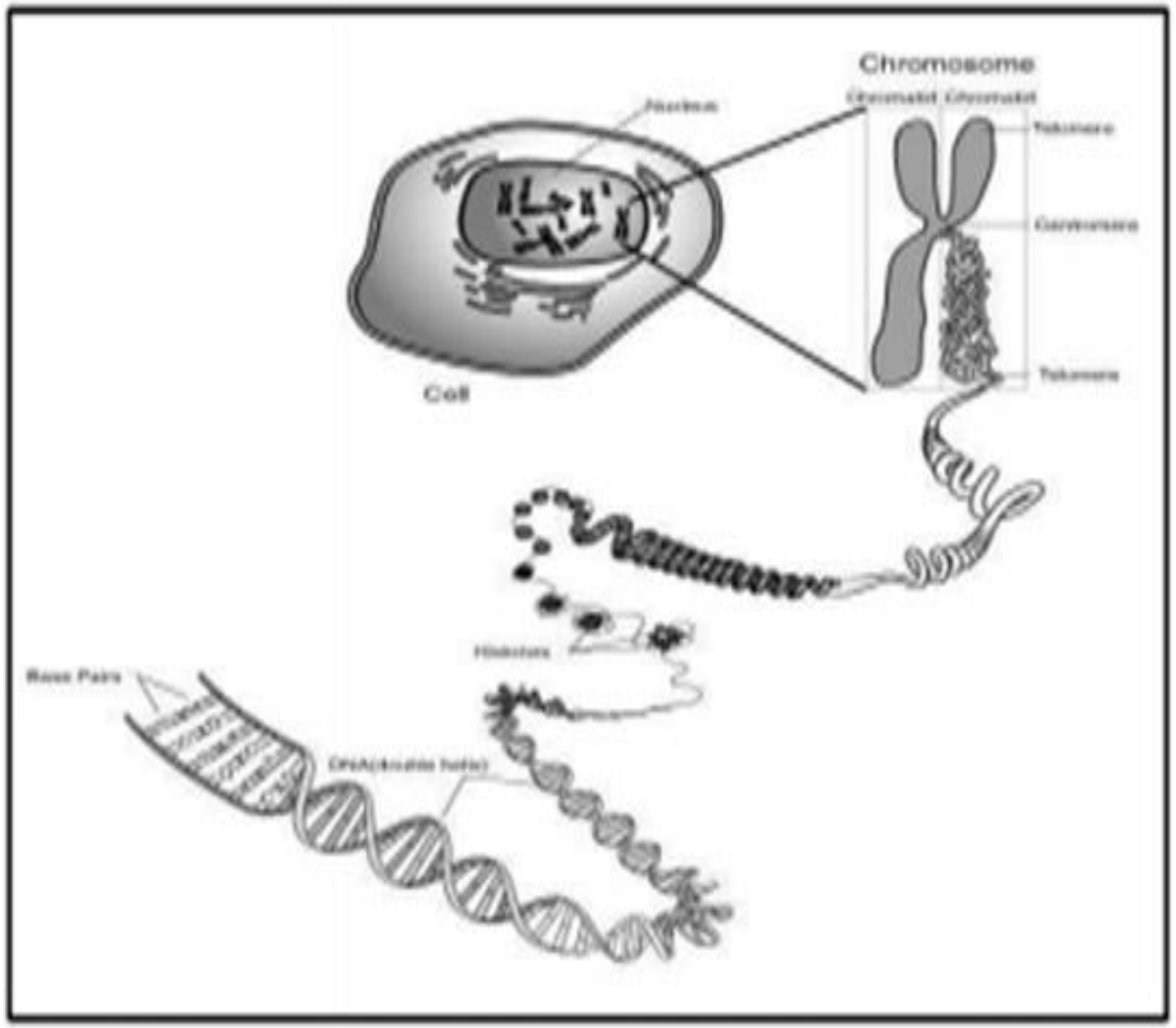
A gene consists of a long combination of four different nucleotide bases (chemicals) , the four nucleotides are: A(adenine) C(cytosine) G(guanine) T(thymine). Different combinations of the letters ACGT give people different characteristics. For example, a person with the following combination -AAACCGGTTTTT - may have green eyes, while somebody whose combination is - AAACCGGTTTAA - may have blue eyes.

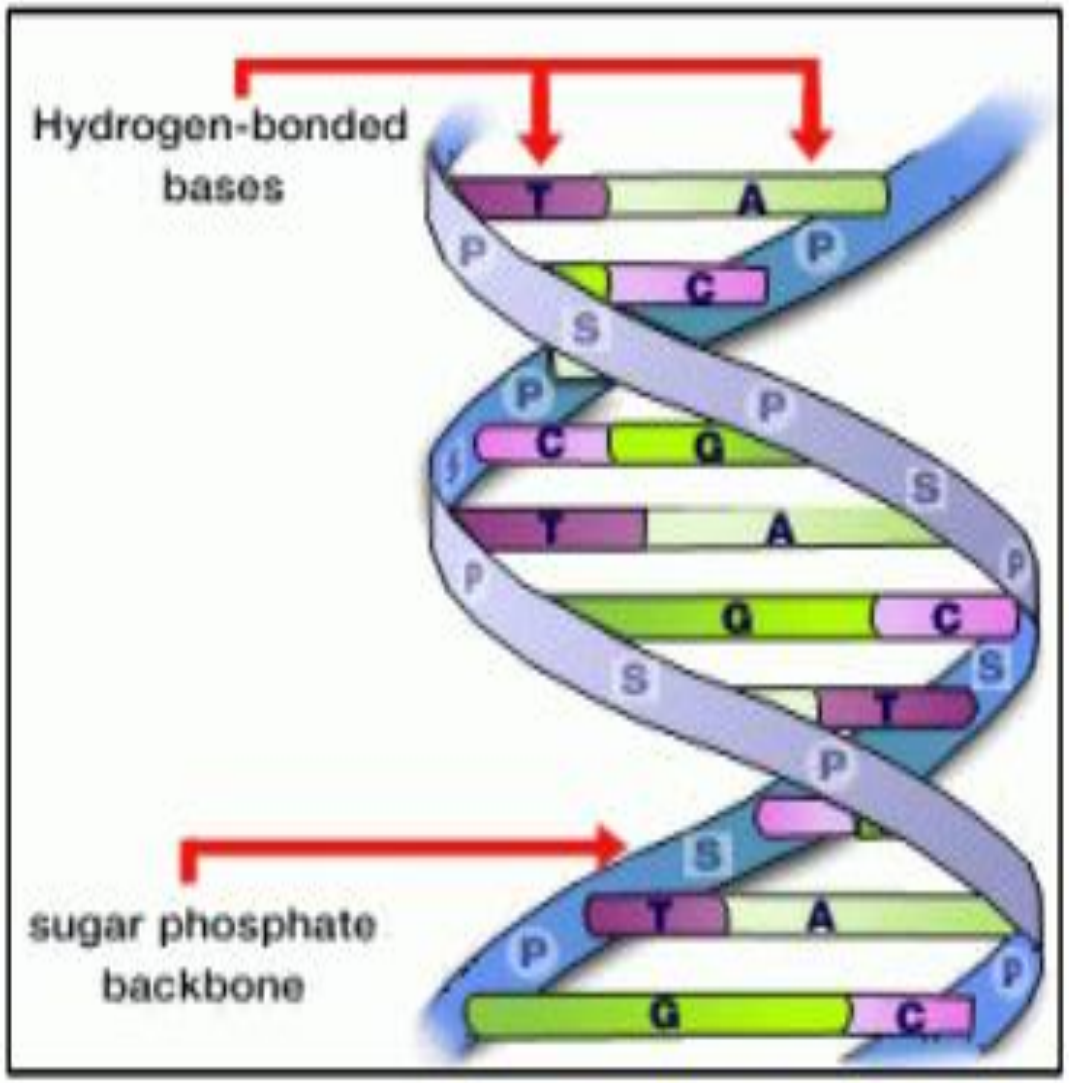
The DNA :

DNA, or deoxyribonucleic acid, is the hereditary material in humans and almost all other organisms. Nearly every cell in a person's body has the same DNA. Most DNA is located in the cell nucleus (where it is called nuclear DNA), but a small amount of DNA can also be found in the mitochondria (where it is called mitochondrial DNA or mtDNA).

The information in DNA is stored as a code made up of four chemical bases : adenine (A), guanine (G), cytosine (C), and thymine (T).

Human DNA consists of about 3 billion bases, and more than 99 % of those bases are


Chromosome
Cell
Base Pairs


Hydrogen-bonded bases
sugar phosphate backbone

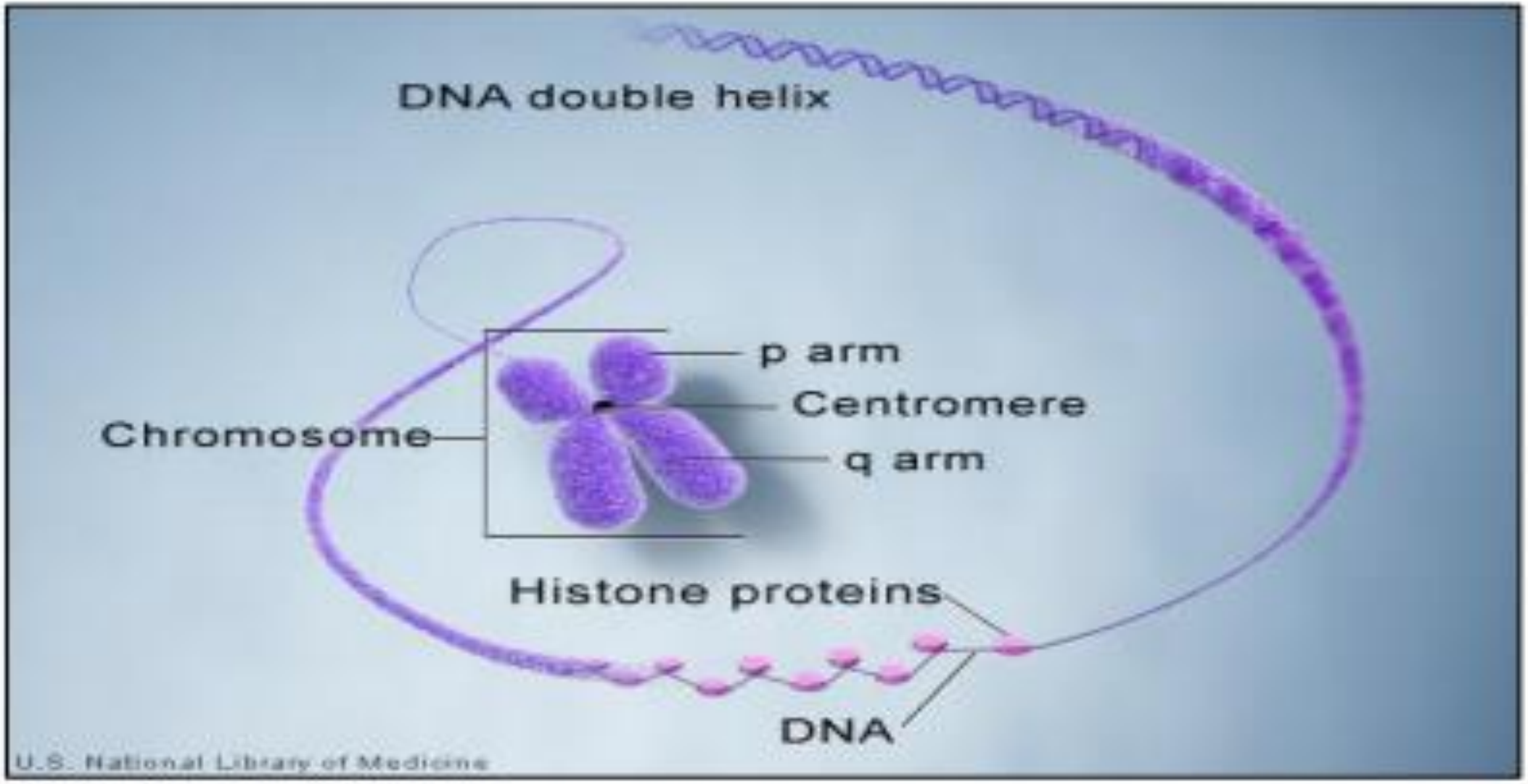
Chromosomes :

Chromosomes comes from the Greek Chroma, which means color, and Soma, which means body (chromosomes are stained very strongly by some dyes), humans have 23 pairs of chromosomes.

A chromosome consists of DNA and has proteins attached to it called histones that support its structure.

These chromosomes are located in the body's cells, which then contain this important genetic information held in the long strands of DNA.

Each chromosome has a tightening point called the centromere, which divides the chromosome into two sections, or "arms." The short arm of the chromosome is labeled the "p arm" , the long arm of the chromosome is labeled the "q arm" .



(DNA and **histone** proteins are **packaged** into structures called **chromosomes**) .

Each of the cells contains two sets of chromosomes - one set came from the mother while the other came from the father , male sperm and the female ovum carry a single set of 23 chromosomes each - 22 autosomes and an X or Y sex chromosome. If you are female you inherited an X chromosome from each parent - if you are male you inherited an X chromosome from your mother and a Y from your father.

Alteration of gene : The alteration (mutation) : means that the information contained in the particular gene is either changed or absent.

The four main ways of inheriting an altered gene are:

❖ **Autosomal dominant -** the alteration is present in every generation and may cause the condition in every person who has the alteration. This is because the altered copy of the gene is dominant over the healthy copy. Examples include : Huntington's disease and familial hypercholesterolemia (genetically linked high cholesterol levels).

❖ **Autosomal recessive -** the affected person has two copies of the altered gene (they have inherited an altered copy of the gene from both parents). They develop the disorder because they do not have a functioning copy of the gene. Examples of autosomal recessive genetic disorders include cystic fibrosis, phenylketonuria (PKU) and sickle cell anaemia .

❖ **X-linked dominant -** this type of disorder generally occurs in females. The "X" refers to one of the sex chromosomes that decide gender. The mother always provides an X, while the father provides either X (female child) or Y (male child). Women with an X-linked dominant disorder have one altered copy and one normal copy of a gene that is on the X chromosome. An example of an X-linked dominant genetic disorder is a rare form of rickets known as hypophosphatasemia or vitamin D resistant rickets.

❖ **X-linked recessive** - this type of disorder is more common in males. It is caused by an alteration in a gene on the X chromosome. Since a male has one X and one Y (XY), he does not have a second „healthy" copy of the gene. Examples of X-linked recessive genetic disorders include: Duchenne muscular dystrophy and hemophilia.

Genetic Disorder : A genetic disorder is caused by an altered or faulty gene or set of genes. **The three broad groups of genetic disorders are include :**

❖ **Single gene disorder** : single gene disorder is the result of a single mutated gene. Over 4000 human diseases are caused by single gene defects. Single gene disorders can be passed on to subsequent generations in several ways for example sickle cell disease, Fragile X syndrome, muscular dystrophy, or Huntington disease.

❖ **Chromosome abnormalities** A chromosome disorder means there is a change in either the structure or the number of chromosomes. This can happen in three main ways:

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• The altered chromosome is passed from the parent to the child

• The abnormality happens when either the sperm or egg (germ cells) is created

• Soon after conception.

❖ **Chromosomal aberrations** : are disruptions in the normal chromosomal content of a cell, and are a major cause of genetic conditions in humans, such as Down syndrome, although most aberrations have little to no effect. The gain or loss of DNA from chromosomes can lead to a variety of genetic disorders. **Human examples include :**

• **Down syndrome** : the most common trisomy, usually caused by an extra copy of chromosome 21 (trisomy 21). Characteristics include **decreased**

muscle tone, **stockier build, asymmetrical skull, slanting eyes and mild to moderate developmental disability.**

• **Edwards syndrome** : or trisomy-18, the second most common trisomy. Symptoms include **motor retardation, developmental disability** and **numerous congenital anomalies** causing serious health problems. Ninety percent of those affected die in infancy. They have characteristic : **clenched hands** and **overlapping fingers .**

Any Question?