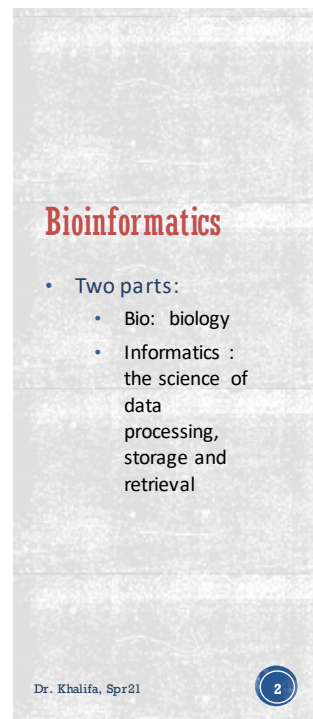
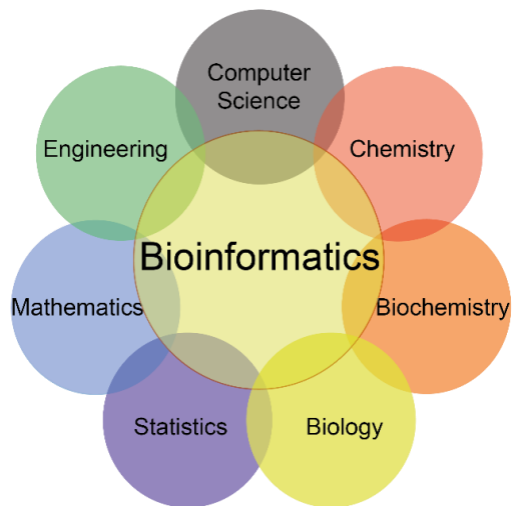


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## 3 Goal

Understand the properties of biological molecules in order to be represented and stored in the computer.

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- The Genome
- What is a cell?
- What is a chromosome?
- What is DNA?
- What is a gene?
- How genes work?
- What is a gene mutation?
- What is genome editing?



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## The genome

- Living organisms carry with them their own miniaturized part list, the genome.
- The complete set of genes or genetic material present in a cell or organism
- Since 1995, a growing number of genomes have been sequenced.
  - Discovering the succession of base-pairs



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## Discoveries

- Three main domains of life:
  - archebacteria (A)
  - bacteria (B)
  - Eukaryotes (E)
- A heavy bias toward bacteria because of:
  - its medical importance
  - small in size

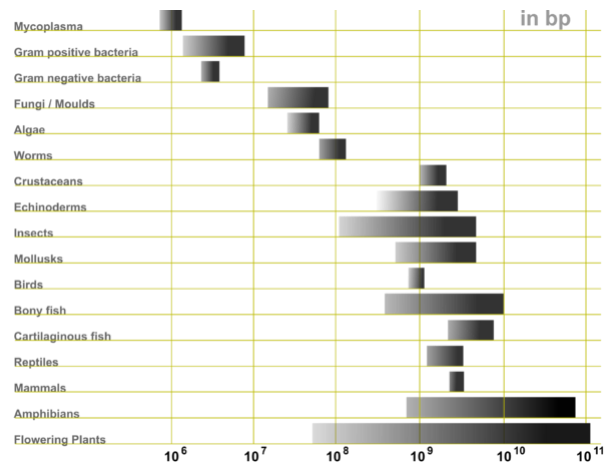


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## Genome size



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## What is a cell?

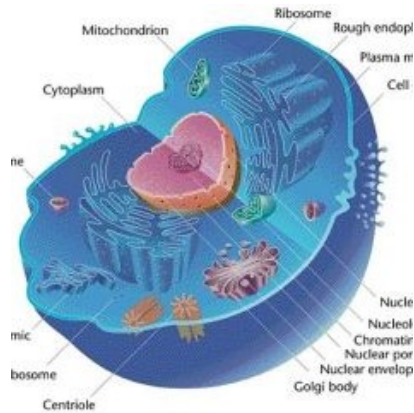
- Cells are the basic building blocks of all living things.
- The human body is composed of trillions of cells :
  - provide structure for the body,
  - convert nutrients in food into energy,
  - carry out specialized functions.
- Cells also contain the body's hereditary material and can make copies of themselves.

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## Organelles, cell parts



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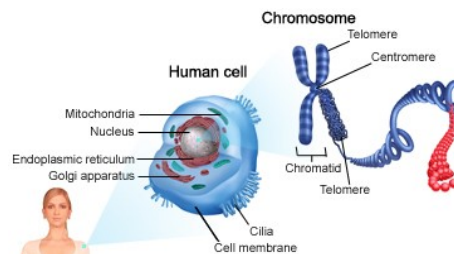
- **nucleus** :
  - houses cell's hereditary material
  - The cell's command center
  - sends directions to the cell to grow, mature, divide, or die.
- **Mitochondria** :
  - convert energy from food into a form that the cell can use.
  - They have their own genetic material, separate from the one in the nucleus, and can make copies of themselves.
- **Ribosomes**
  - process the cell's genetic instructions to create proteins.

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## Genetic information

- contains all necessary information required to produce/create/direct activities any living organism.
- Packed in the nucleus
  - Human : 46 (23-pairs) Chromosome



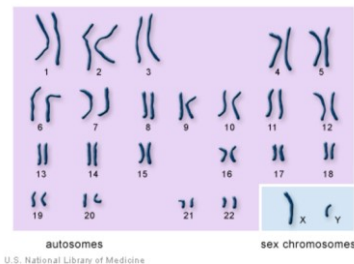
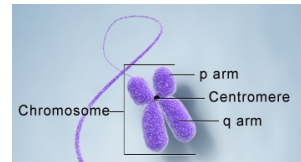
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## What is a chromosome?

- A thread-like structure packing the DNA molecule
- Chromosomes can be visible -under a microscope - only during cell division.
  - The short arm → "p arm."
  - The long arm → "q arm."
- The location of the centromere on each chromosome gives the chromosome its characteristic shape
- In humans:
  - 23 pairs of chromosomes (total of 46)
    - 22 pairs : look the same in both males and females.
    - The 23rd pair, the sex chromosomes:
      - Females : 2 copies of the X chromosome
      - Males: one X and one Y chromosome.



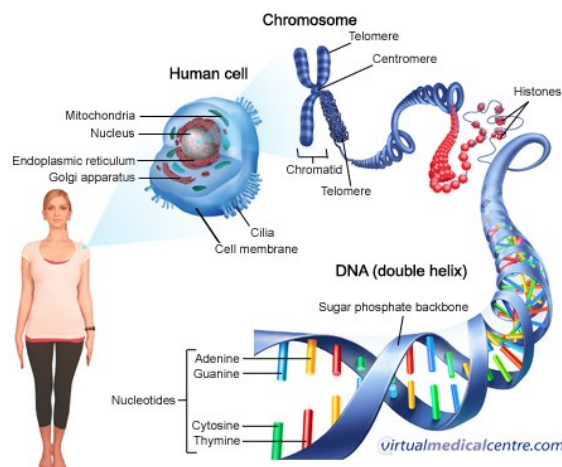
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## What is DNA?

- The life's most important molecule!
  - the hereditary material in humans and almost all other organisms.
- Every cell in a person's body has the same DNA (deoxyribonucleic acid)
  - located in the cell nucleus
    - called nuclear DNA



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## DNA Structure

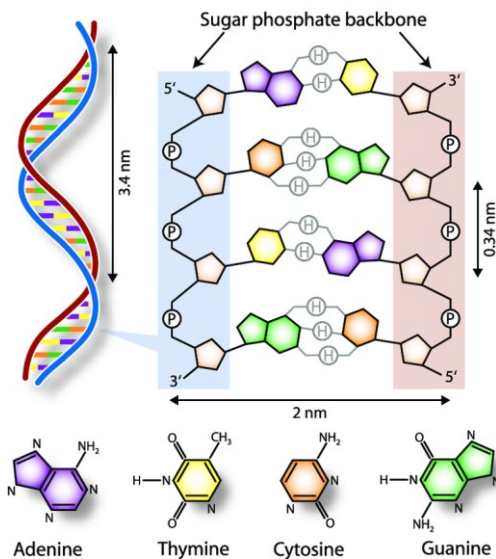
- Life encodes itself into the DNA code bases:
  - A (adenine),
  - C (cytosine),
  - G (guanine) and
  - T (thymine)
- composed of two polynucleotide chains (strands) that coil around each other to form a double helix



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## DNA Chemical Structure

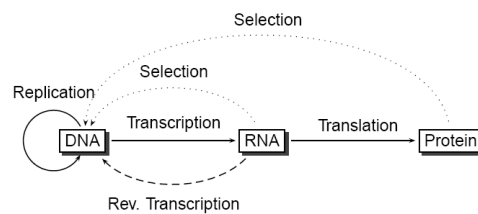
- Each base is attached to:
  - a sugar molecule
  - a phosphate molecule
- Together, a base, sugar, and phosphate are called a nucleotide.
- DNA bases pair up with each other:
  - G always pairing a C
  - A always pairing a T.
- DNA strands run in opposite directions
  - one strand → reverse complement
- Always written left to right:
  - 5' to 3' direction

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## The book of instructions

- An important property of DNA is that it can replicate
  - Each strand of DNA serves as a pattern of bases.
  - This is critical when cells divide
- Constant reading of the genome :
  - Called central dogma
  - DNA → RNA → protein

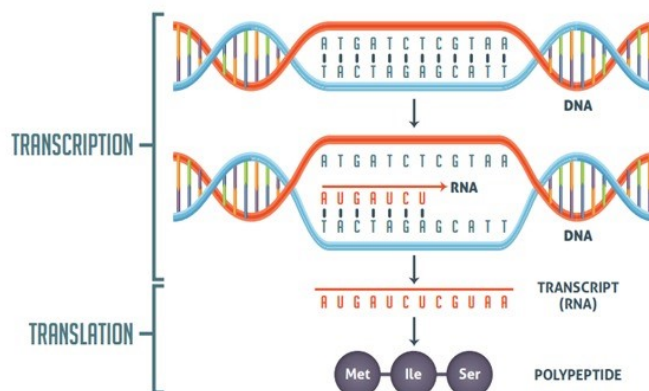


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## Central dogma



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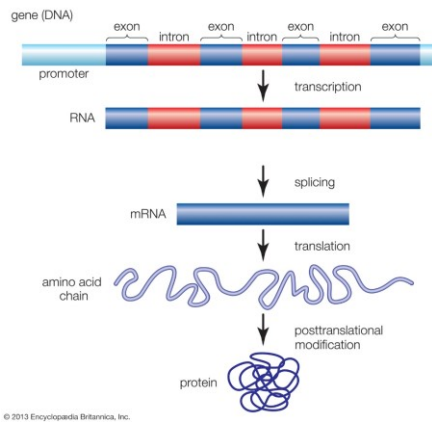
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## What is a gene?

- A gene is the basic physical and functional unit of heredity.
- Genes can vary in size from a few hundred DNA bases to more than 2 million bases
- The Human Genome Project estimated that humans have between 20,000 and 25,000 genes.



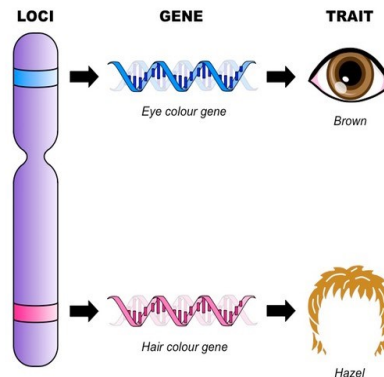
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## From gene to protein!!

- Some genes act as instructions to make molecules called proteins:
  - Some proteins are structural and make up our tissues like bones and muscles.
  - Proteins called enzymes are involved in chemical reactions like breaking down the food we eat.
  - others are like little messengers that send signals around our body, these proteins are known as hormones.
- Scientists giving genes unique names/symbols ( CETR )



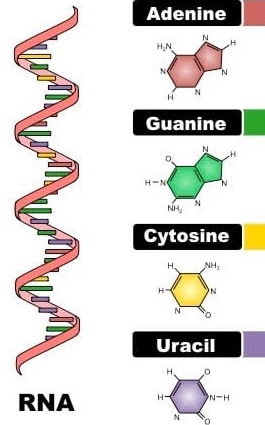
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## What is RNA?

- RNA only has one strand made up of nucleotides.
- RNA shares Adenine ('A'), Guanine ('G') and Cytosine ('C') with DNA, but contains Uracil ('U') rather than Thymine.
- Pairs: (A-U) & (C-G)
- RNA forms in the nucleolus, and then moves to specialized regions of the cytoplasm depending on the type of RNA formed.
- RNA strands are shorter than DNA strands & are more resistant to damage from UV light.



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RNA codon table

1st position	2nd position				3rd position
U	U	C	A	G	U C A G
U	Phe Phe Leu Leu	Ser Ser Ser Ser	Tyr Tyr stop stop	Cys Cys stop Trp	U C A G
C	Leu Leu Leu Leu	Pro Pro Pro Pro	His His Gln Gln	Arg Arg Arg Arg	U C A G
A	Ile Ile Ile Met	Thr Thr Thr Thr	Asn Asn Lys Lys	Ser Ser Arg Arg	U C A G
G	Val Val Val Val	Ala Ala Ala Ala	Asp Asp Glu Glu	Gly Gly Gly Gly	U C A G

Amino Acids

Ala: Alanine  
 Arg: Arginine  
 Asn: Asparagine  
 Asp: Aspartic acid  
 Cys: Cysteine  
 Gln: Glutamine  
 Glu: Glutamic acid  
 Gly: Glycine  
 His: Histidine  
 Ile: Isoleucine  
 Leu: Leucine  
 Lys: Lysine  
 Met: Methionine  
 Phe: Phenylalanine  
 Pro: Proline  
 Ser: Serine  
 Thr: Threonine  
 Trp: Tryptophane  
 Tyr: Tyrosine  
 Val: Valine

## The genetic code

- Each group of three bases in mRNA constitutes a codon
- Each codon specifies a particular amino acid
- Proteins are made up of hundreds or thousands of smaller units called amino acids, which are attached to one another in long chains.
  - The sequence of amino acids determines each protein's unique 3-dimensional structure and its specific function.
- There are 20 different types of amino acids that can be combined to make a protein.

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## Watch it in real-time!

- Animation video from the DNA learning center

<https://dnlc.cshl.edu/resources/3d/12-transcription-basic.html>

<https://dnlc.cshl.edu/resources/3d/15-translation-basic.html>

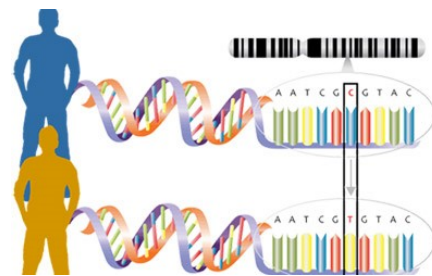
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## What is a gene mutation?

- A gene mutation is a permanent alteration in the DNA sequence that makes up a gene, such that the sequence differs from what is found in most people.
- Gene mutations can be classified in two major ways:
  - **Hereditary**: inherited from a parent, present in virtually every cell
  - **Acquired** (or somatic): occur at some time during a person's life, present only in certain cells, not in every cell in the body, may not be passed to the next generation



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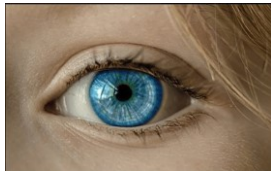
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## Examples

### Blue Eyes

- blue eyes aren't technically a color—[they're a lack of color.](#)



### Lung Cancer

- Besides smoking, other possible causes of lung cancer are secondhand smoke, air pollution, and gene mutations.



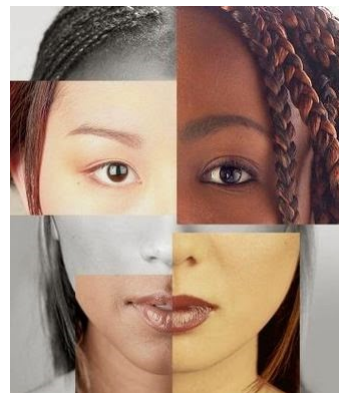
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## Polymorphism

- Polymorphisms:
  - occur in more than 1 percent of the population
  - occur frequently
  - Normal Genetic changes
  - Responsible for many of the normal differences between people such as eye color, hair color, and blood type.
- Most disease-causing gene mutations are uncommon in the general population.



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## What is genome editing?

- Genome editing is a group of technologies that give scientists the ability to change an organism's DNA.
  - Becoming fast, cheap, accurate, and efficient
- It is of great interest in the prevention and treatment of human diseases
- Based on concerns about ethics and safety, germline cell and embryo genome editing are currently illegal in many countries.

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### Questions?

Ref:

Chap 00

<https://ghr.nlm.nih.gov/primer>

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