An introduction to Human Genetics

Lecture by
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What is the secret behind the transmission of hereditary characteristics from generation to generation?

Basic Concepts of Genetics/ Mendel`s laws

Mendel's observations led to two laws, regarding the transmission of hereditary characteristics from generation to generation.

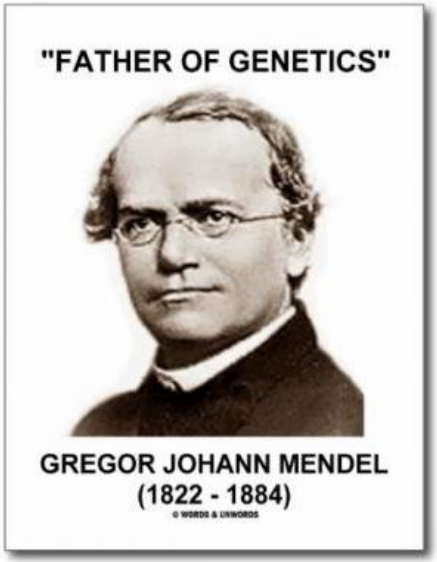
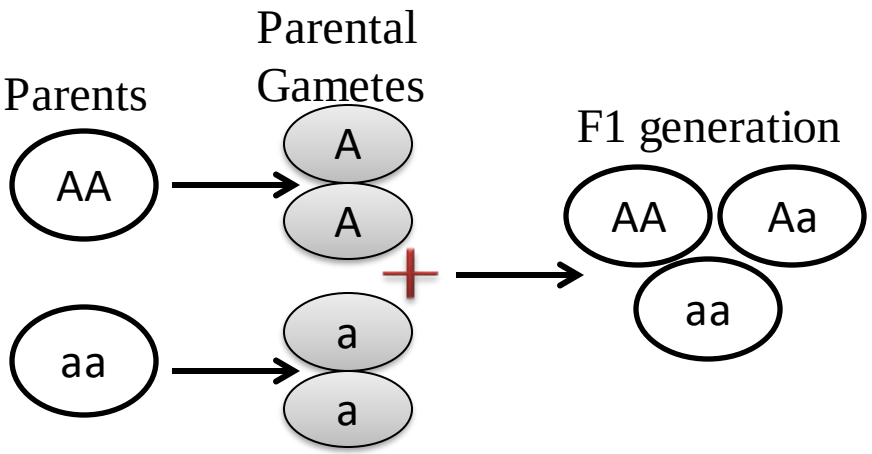


Image obtained from https://www.google.iq/search?q=Gregor+Mendel

First law: Principle of Segregation:

Two alleles segregate randomly from each other during the formation of gametes.

It means, that each gene has two copies (alleles), and each parent will give one copy to a child.



Basic Concepts of Genetics/ Mendel`s laws

Second law: Principle of Independent Assortment:

Two genes will assort independently of one another in gamete production

Important teams to know:

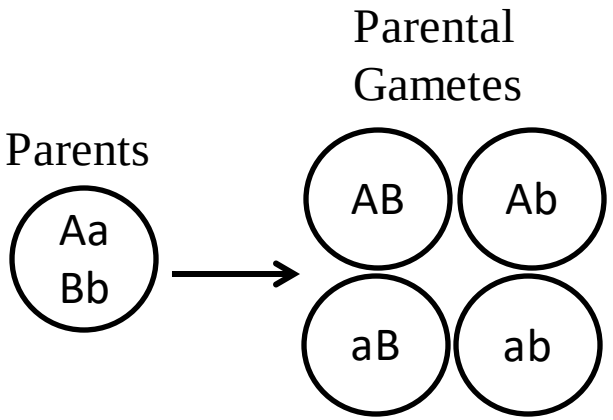
Alleles: are different versions of the same gene.

Homozygous: refers to an individual with two identical alleles.

Heterozygous: refers to an individual with two different alleles.

Genotype: it refers to the specific allelic composition of an individual.

Phenotype refers to the outward appearance of an individual.



The genetic material: Genome

- Genome as a term was suggested in 1920 by Hans Winkler, professor of biology (botany) at the University of Hamburg. Genome word is a blend of the words ***gene*** and ***chromosome*.**

- The **genome** can be defined as the **genetic material** of an organism. It consists of DNA (or RNA in RNA viruses). The genome includes both the genes, (the coding regions), the non-coding DNA and the genomes of the mitochondria and chloroplasts.

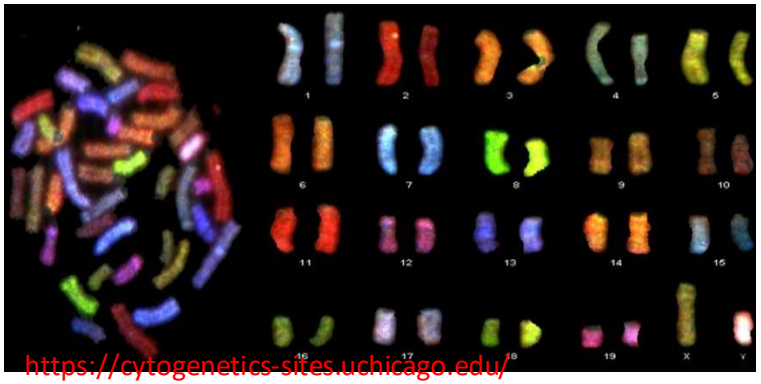
Cytogenetics: the study of chromosomes

- **Cytogenetics** is the study of chromosomes and their role in heredity. Cytogenetics focuses on studying structure, composition of chromosomes as well as, diagnosis of chromosomal abnormalities associated with diseases.

- A **karyotype** refers to the number and appearance of chromosomes in the nucleus of a eukaryotic cell. It is also refers to the complete set of chromosomes in a species.

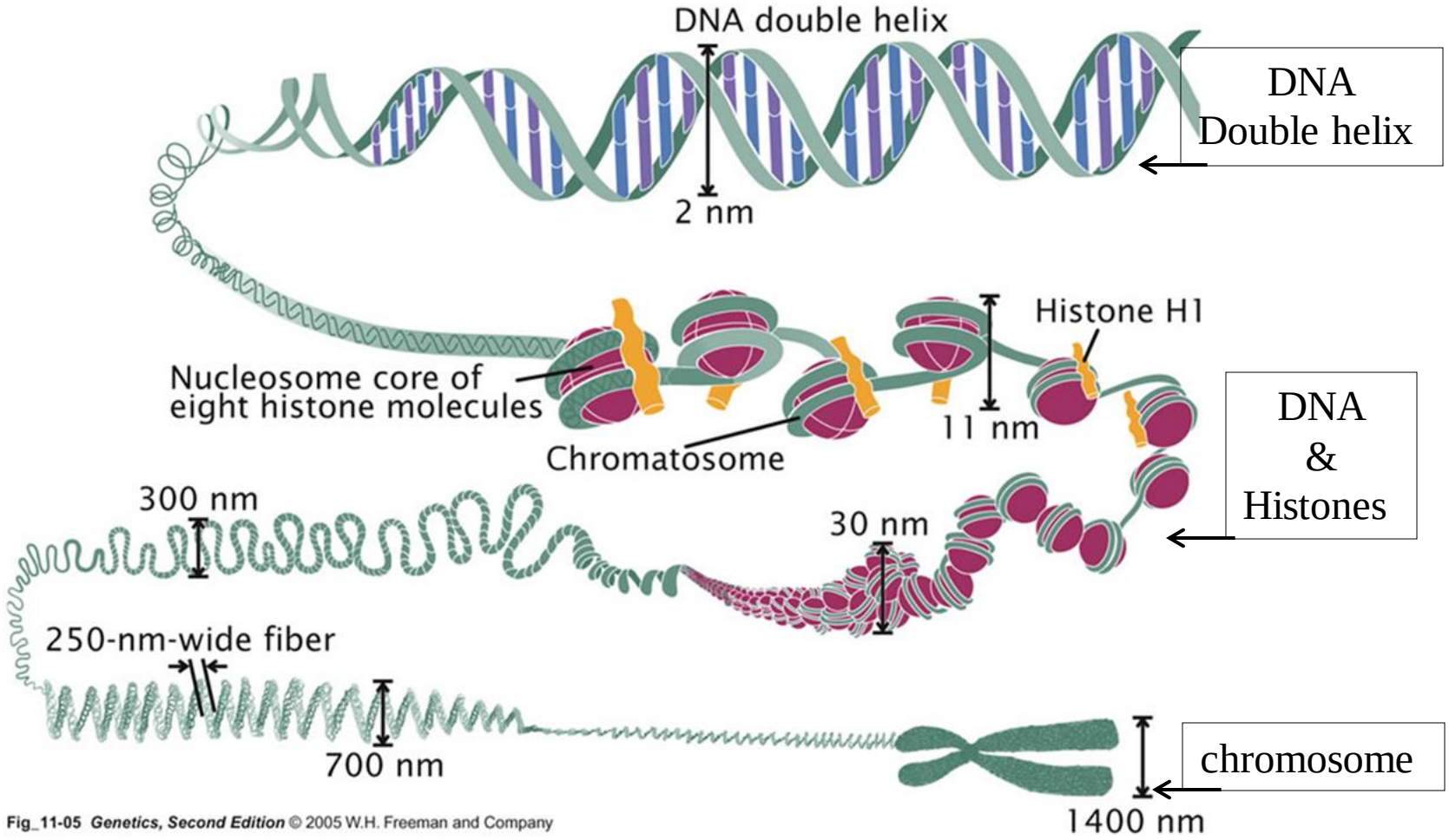
- Among the members of a species, the number of chromosomes is uniform.

- A normal human karyotype contains 22 pairs of autosomes and one pair of sex chromosomes.



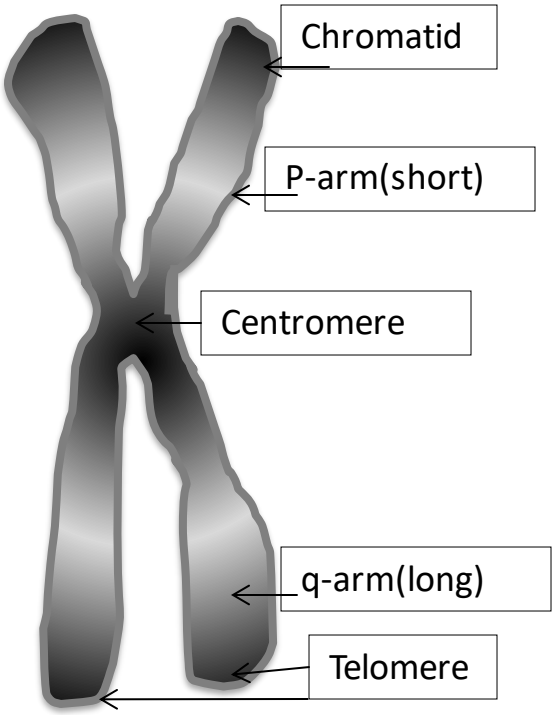
What is chromosome?

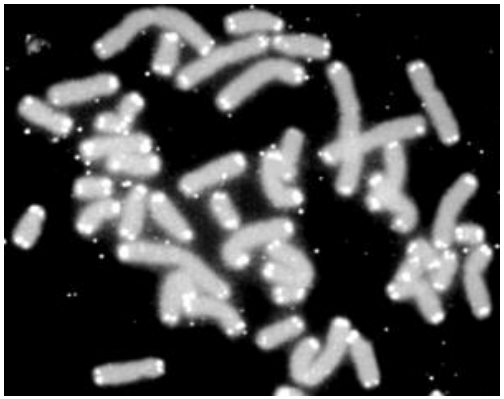
Chromosome is a thread-like structures, it is consisted of DNA molecule packed tightly and coiled around a specific proteins called histones. Chromosomes are located in the nucleus of eukaryotic cells.



The structure of the chromosome

- **Chromatids** are two identical parts of each chromosomes.
- These chromatids are held together at a point named the **centromere**, which can be located at any point along the length of the chromosome.
- **Telomere** is a region of repetitive nucleotide sequences. Telomere is located at each end of a chromosome, it protects the end of the chromosome from deterioration or from fusion with neighbouring chromosomes





Chromosomal abnormalities

- Almost every cell in our body contains **23 pairs** of chromosomes, for a total of 46 chromosomes.
- **22** pairs are called **autosomes** and **1 pair of sex** chromosomes, XX in female and XY in male in each cell.

- **Chromosomal abnormalities** can be categorised as:

1- **Numerical abnormalities** refer to a missing or existing of a whole chromosome to the normal pair.

2- **Structural abnormalities** occur when part of an individual chromosome is missing, extra, switched to another chromosome, or turned upside down.

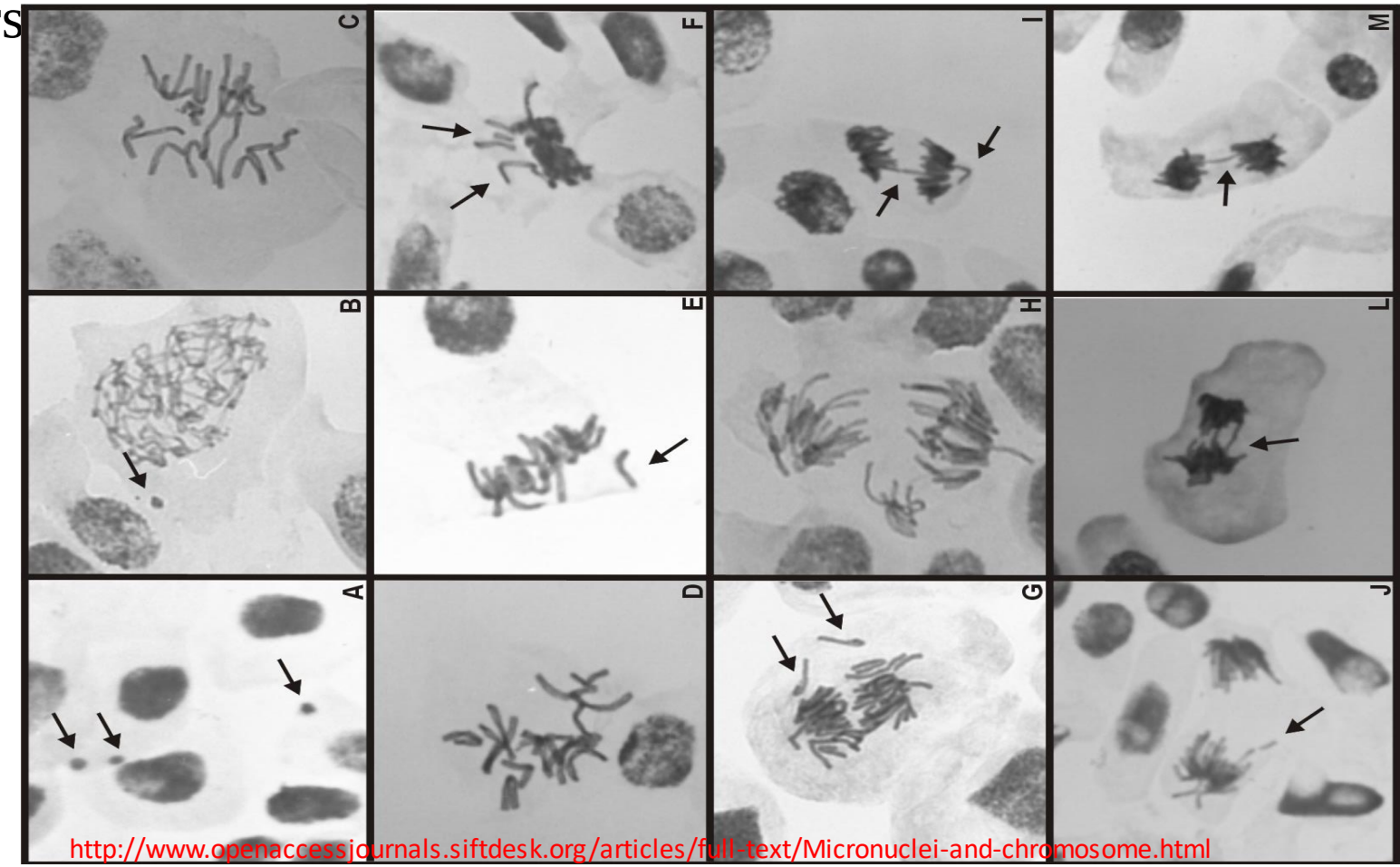
How chromosomal abnormalities may occur?

- Chromosomal abnormality may occur accidentally during:

1- The formation of the egg or the sperm.

2- The early developmental stages of the foetus.

3- Certain environmental factors may play a role in the occurrence of genetic errors



http://www.openaccessjournals.siftdesk.org/articles/full-text/Micronuclei-and-chromosome.html

Causes of chromosomal abnormalities

1) Some medicines
2) Street drugs
3) Alcohol
4) Tobacco
5) Toxic chemicals
6) Some viruses and bacteria
7) Some kinds of radiation
8) Certain health conditions, such as uncontrolled diabetes

The deoxyribonucleic acid (DNA)

- DNA is the hereditary material in humans and almost all other organisms.
- Nearly every cell in the 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 structure of DNA

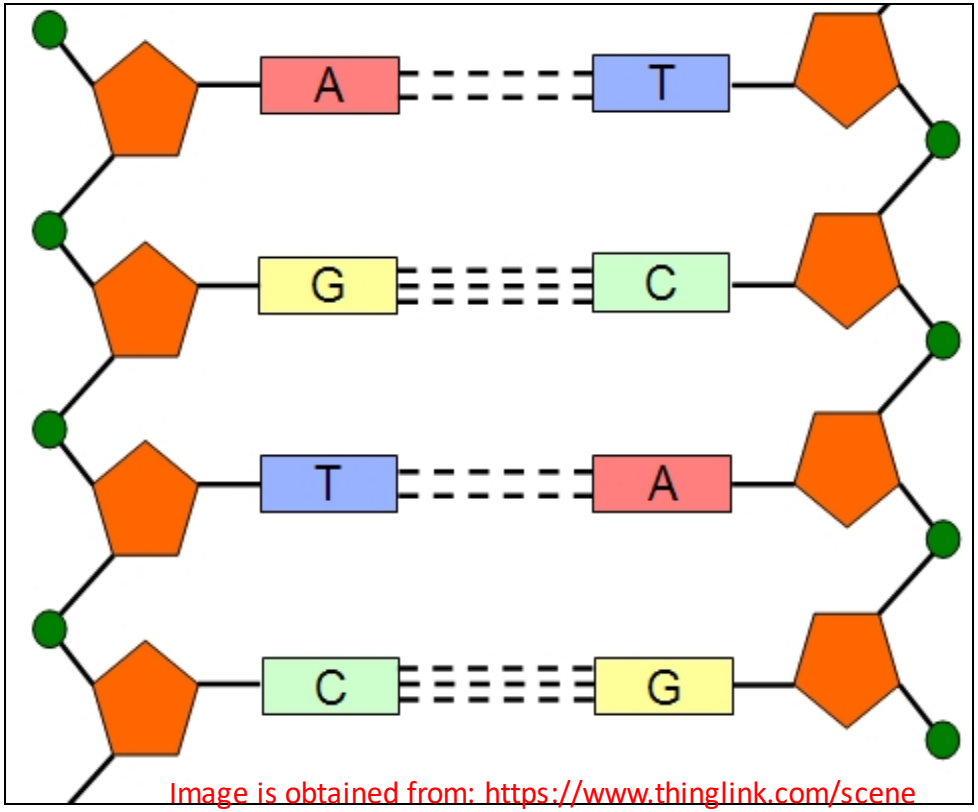
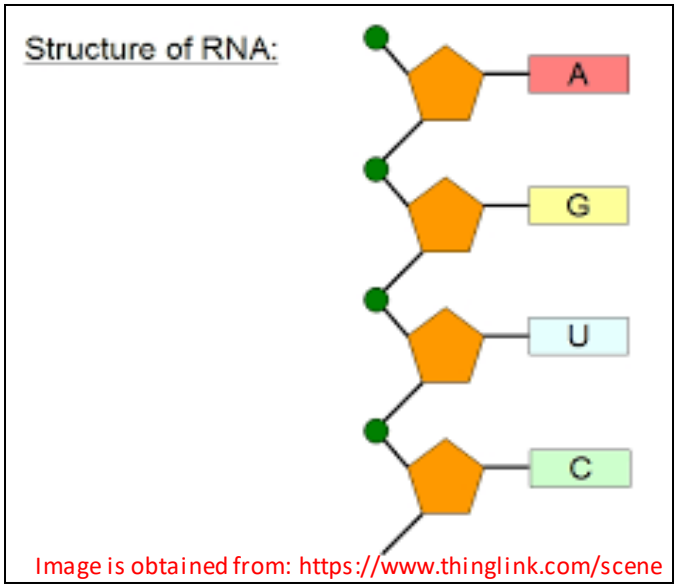


Image is obtained from: https://www.thinglink.com/scene

The ribonucleic acid (RNA)

RNA is a polymeric molecule essential in various biological roles in:

- coding,
- decoding,
- regulation,
- and expression of genes.



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The types of RNA

1. messenger RNA (***mRNA***) to convey genetic information

2. transfer RNA (***tRNA***) molecules to deliver amino acids to the ribosome

3. ribosomal RNA (***rRNA***) links amino acids together to form proteins

The main differences between DNA and RNA

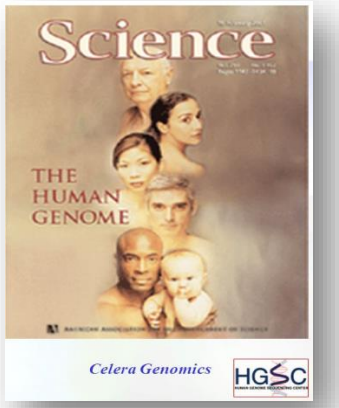
Difference	DNA	RNA
Definition	It contains the genetic instructions used in the development and functioning of all modern living organisms.	It contains the genetic information that transcripted from DNA to RNA
Function	Medium of long-term, stable storage and transmission of genetic information	Transfers genetic code needed for the creation of proteins from the nucleus to the ribosome.
Structure	Double-stranded	Single-stranded.
Base Pairing	Adenine links to thymine (A-T) and cytosine links to guanine (C-G).	Adenine links to uracil (A-U) and cytosine links to guanine (C-G).

The main differences between DNA and RNA

Difference	DNA	RNA
Location	DNA is found in the nucleus of a cell and in mitochondria	Is is found in the nucleus, cytoplasm, ribosome.
Stability	Stable in alkaline conditions.	Less Stable in alkaline conditions.
Propagation	DNA is self-replicating	RNA is synthesized from DNA when needed
Unique Features	DNA is protected in the nucleus, as it is tightly packed. DNA can be damaged by exposure to ultra-violet rays	RNA strands are continually made, broken down and reused. RNA is more resistant to damage by Ultra-violet rays.

The Human Genome Project (HGP)

- The **Human Genome Project** (**HGP**) is an international scientific research project aims to determine the sequence of DNA, to identify and to map all of the genes of the human genome from both a physical and a functional standpoint.



- The **International Human Genome Sequencing Consortium** published the first draft of the human genome in 2001 with the sequence of the entire genome's three billion base pairs some 90 percent complete.
- The full sequence was completed and published in April 2003, the number of human genes appeared to be ranged between **<u>50,000</u>** genes and **<u>140,000</u>** genes.