### **Nucleic Acids:** Hereditary Material

# All cells <u>store information</u> required to build and maintain the cell (*genetic information*) and <u>constantly use it</u>.





### Laws of Mendelian Inheritance

**Gregor Mendel**, 1856-1863:





		A	а
ea Parent	A	AA	Aa
remale P	а	aA	aa

A = Yellow Seeds a = Green Seeds Because a is recessive, only aa has green seeds. An Example of a Mendelian Genetic Trait pea plant experiments

- Cultivated and tested some 29,000 pea plants in the monastery's 2 hectares (4.9 acres) experimental garden.
- Worked with seven characteristics: plant height, pod shape and color, seed shape and color, and flower position and color.
- Law of Segregation: one random allele (gene variation) from each parent.
- Law of Independent Assortment: alleles for different traits are independent.
- Law of Dominance: some alleles are dominant while others are recessive; an organism with at least one dominant allele will display the effect of the dominant allele.

#### "Father of modern genetics"





# **DNA Discovery**

 Swiss physician Friedrich Miescher discovered DNA ("nuclein") in 1869, athough <u>scientists</u> did not understand what it was until...







...1943: Avery-MacLeod-McCarty experiment showed that DNA is the hereditary material in bacteria.

• In 1953, James Watson and Francis Crick suggested the double-helix model of DNA structure based on a single X-ray diffraction image.

### DNA

DNA is a long polymer made from repeating units called nucleotides, or bases.

- Four types of bases:
  - T Thymine (Uracil in RNA)
  - A Adenine
  - **G** Guanine
  - C Cytosine
- In living organisms DNA does not usually exist as a single molecule, but instead as a pair of molecules that are held tightly together, entwined in the shape of a double helix.



 Within cells, DNA is organized into long structures called *chromosomes*.

## Genome and Genetic Code

#### What is Genome?

- Genetic material of an organism, essentially the instructions on making proteins and RNAs.
- Inscribed in DNA: complete DNA sequence.
- Includes both the genes and the non-coding regions.

#### What is Genetic Code?

- The set of rules by which information encoded within DNA or RNA is translated into proteins.
- In general, the genetic code specifies 20 standard amino acids by means of triple nucleotide codons and is <u>basically the</u> same for all organisms on Earth.

#### What is Gene?

- The portion of the genome that codes for a <u>single</u> protein or an RNA.
- The molecular unit of heredity of a living organism.
- The size of a single gene may vary greatly, ranging from ~1,000 bases to ~1 million bases in humans.



# **Human DNA**

- The Human Genome Project (1990-2003) produced the first complete sequences of individual human genomes.
- <u>Human genome</u> contains ~3 billion bases and ~20,500 genes.
- Over 98% of the human DNA comprises non-coding repetitive sequences (the role, functions and descriptions of these sequences are currently being investigated by scientists).



- By 2012, thousands of human genomes have been completely sequenced.
- All humans have the DNA that is 99.9% similar, however the rest 0.01% is enough to identify different individual DNA sequences (*i.e. tell apart which DNA belongs to whom*).
- Primary (and now standard routine!) applications include paternity testing as well as DNA profiling in criminal investigations.

# **DNA Damage**

#### DNA is damaged up to 1 million times per cell per day.



- The cells have an elaborate type-of-damage-specific system of DNA repair that is constantly active.
- A cell that has accumulated a large amount of DNA damage, or one that no longer effectively repairs damage incurred to its DNA, can enter one of three possible states:
  - 1. an <u>irreversible state of dormancy</u>, known as *senescence*
  - 2. <u>cell suicide</u> (apoptosis) or programmed cell death
  - 3. <u>unregulated cell division</u>, which can lead to cancer

# **DNA Mutations**

#### A <u>mutation</u> is a permanent change in the DNA sequence.

- Mutations can be:
  - spontaneous (by chance)

induced by mutagens (physical, chemical or biological agents)

- Factors that cause mutations:
  - <u>external</u> environmental factors such as sunlight, radiation, and smoking
  - <u>native</u> errors during DNA replication
- Mutations can lead to:
  - an evolutionary advantage of a certain genotype
  - <u>disease</u>, developmental delays, <u>structural abnormalities</u>, or other negative effects.



Example: Sickle cell anemia is a disorder in which the body makes sickle-shaped red blood cells as a result of DNA mutation.

## **DNA Half-Life**

In 2012, researchers have calculated that DNA from bones has a <u>521 year half-life</u>, which means that the oldest clone-able samples of DNA could be no more than 2 million years old.



This result rules out any possibility of ever replicating dinosaurs, as the youngest dinosaurs were around more than 65 million years ago...



### Miller–Urey experiment, 1953: chemical origins of life



Stanley L. Miller Harold C. Urey



- Test for the occurrence of chemical origins of life by <u>simulating the conditions</u> thought at the time to be present on <u>the early Earth</u>.
- The experiment used water (H<sub>2</sub>O), methane (CH<sub>4</sub>), ammonia (NH<sub>3</sub>), and hydrogen (H<sub>2</sub>) all sealed inside a sterile loop array of glass flasks; one flask was half-full of liquid water ("ocean") and another flask contained a pair of electrodes. The liquid water was heated to induce evaporation, sparks were fired between the electrodes to simulate "lightning through the atmosphere" and water vapor; then water could "precipitate" that is condense and trickle back into the first flask in a continuous cycle.
- After two weeks: 10–15% of the carbon was now in the form of organic compounds; >20 amino acids formed; sugars were also formed. However, *nucleic acids were not formed* within the reaction...