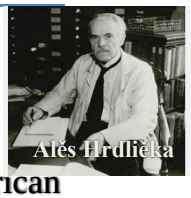
Basic Concepts in Genetics



History and Background:

- In 1900, Aleš Hrdlička, an anthropologist and physician working for the American Museum of Natural History, visited the Hopi villages of Black Mesa (Figure 1) and reported a startling discovery. Among the Hopis were 11 white people—not Caucasians, but white Hopi Native Americans. These Hopis had a genetic condition known as albinism (Figure 2).
- **Albinism** is caused by a defect in one of the enzymes required to produce **melanin,** the pigment that darkens our skin, hair, and eyes. People with albinism either don't produce **melanin** or produce only small amounts of it and, consequently, have white hair, light skin, and no pigment in the irises of their eyes.
- Melanin normally protects the DNA of skin cells from the damaging effects of ultraviolet radiation in sunlight, and melanin's presence in the developing eye is essential for proper eyesight.
- The **genetic basis of albinism** was first described by the English physician Archibald Edward Garrod (**Figure 3**), who recognized in 1908 that the condition was inherited as **an autosomal recessive trait**, meaning that a person must receive two copies of an albino mutation—one from each parent—to have albinism.
- He was the first to connect a **human genetic disorder** with **Mendel's laws of inheritance**. He also proposed the idea that **diseases** came about through a **metabolic route** leading to the molecular basis of inheritance.

Figure3: Sir Archibald Garrod, around 1910.

Figure1: A Hopi pueblo on Black Mesa.

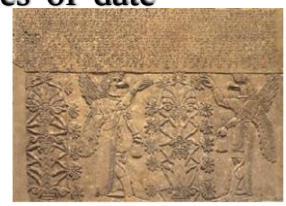
Figure2: Albinism among the Hopi Native Americans.

History: The Early Use and Understanding of Heredity

- The first evidence that people understood and applied the **principles of heredity** in earlier times is found in the **domestication of plants and animals**, which began between approximately 10,000 and 12,000 years ago.

- The first **domesticated organisms** included wheat, peas, lentils, barley, goats, and sheep.
- By 4000 years ago, genetic techniques of **selective breeding** were already in use in the Middle East., for example **Egyptian** data palm breeding.

- The **Assyrians** and **Babylonians** developed several hundred varieties of date palms that differed in fruit size, color, taste, and time of ripening.



- The fact that **living things** ***inherit traits*** from their parents has been used since prehistoric times to improve crop plants and animals through selective breeding.
- Other crops and domesticated animals were developed by cultures in Asia, Africa, and the Americas in the same period.
- Also **ancient writings** demonstrate that early humans were also aware of their own **heredity**.
- So people have known about **inheritance** for a long time that children resemble their parents.

History: Early Concepts of Heredity:

- Some early **concepts of heredity** were **incorrect**, but **reflect** human interest in heredity and our attempts to explain the inheritance of traits. The **ancient Greeks** gave careful consideration to **human reproduction** and **heredity**.
- The Greek philosophers had a variety of ideas:
  1) **Theophrastus** proposed that *male flowers* caused *female flowers* to ripen.
  2) **Hippocrates** speculated that *"seeds"* were produced by various body parts and transmitted to offspring at the time of conception.
  3) **Aristotle** thought that male and female semen mixed at conception.
  4) **Aeschylus** proposed the male as the parent, with the female as a "nurse for the young life sown within her".

Introduce to Genetics:

- **Genetics,** (from Ancient Greek genetikos, "genitive" and that from ***genesis*, "origin"),** a discipline of biology, is the science of ***heredity*** and ***variation*** in living organisms.
- **William Bateson**, a proponent of **Mendel's work**, coined the word ***genetics*** in 1905.
- Prior to Mendel, **Imre Festetics**, a Hungarian noble, was the first *who used the word "genetics."* He described several **rules of genetic inheritance** in his work (**The genetic law of the Nature**) (Die genetische Gesätze der Natur, 1819). Also his **second law is the same as what Mendel published**. In his third law, **he developed the basic principles of mutation.**

Description of Genetics:

- **Genetics** is a discipline of **biology**, is the science of ***Heredity*** and ***Variation*** in living organisms.
- **Medical genetics** is the branch of medicine that involves the **diagnosis** and **management of hereditary disorders.**
- **Heredity: a** *study of the* ***transmission of genetic characteristics*** *from* ***parents*** *to* ***offspring***.
- **Variation:** ***Differences that are seen among the members of same species*.**
- **Inheritance:** ***Process*** *by which* ***genetic characteristics*** *are transferred from parent to offspring.*
- **Genetics** is the science of **heredity** that involves the **structure** and **function of genes** and **the way genes** are **passed from one generation to the next**.
- **So Genetics** is concerned primarily with understanding biological properties that are **transmitted from parent to offspring**.

The subject matter of genetics includes:

- **Heredity.**
- The **molecular nature of the genetic material**.
- The **ways in which genes**, which determine the characteristics of organisms, control life functions.
- The **distribution and behavior of genes** in populations.