History of Genetics

People have known about inheritance for a long time.

- Children resemble their parents.
- ❖ Domestication of animals and plants, selective breeding for good characteristics.
- ❖ Sumerian horse breeding records.
- **\$** Egyptian data palm breeding.
- Ability to identify a person as a member of a particular family by certain physical traits. (Hair, nose....etc.).

The mechanisms were not readily apparent.

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. Arisotle 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".

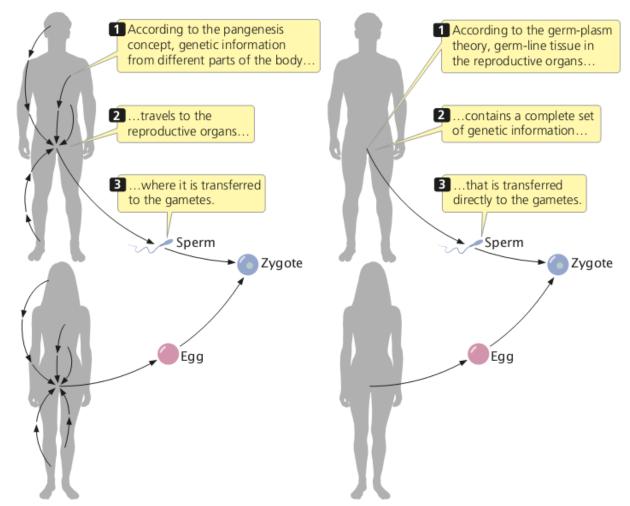
There are different old theories proposed to explain the similarities and dissimilarities between individuals:

- 1. Blending theory, blending theory of inheritance supplanted the spermists and ovists during the 19th century. The mixture of sperm and egg resulted in progeny that were a "blend" of two parents' characteristics. The blending theory means mixing between two colors (for example white and black produce a gray color, then it is very difficult to return to the original colors). the idea that individuals inherit a smooth blend of traits from their parents. Mendel's work disproved this, showing that traits are composed of combinations of distinct genes rather than a continuous blend.
- **2.** Acquired character inheritance: Another theory that had some support at that time was the <u>inheritance of acquired characteristics</u>: the belief that individuals inherit traits strengthened by their parents. This theory (commonly associated with <u>Jean-Baptiste Lamarck</u>) is now known to be wrong—the experiences of individuals do not affect the genes they pass to their children.
- **3.** Pangenesis theory: Other theories included the <u>pangenesis</u> of <u>Charles Darwin</u> (which had both acquired and inherited aspects) and <u>Francis Galton</u>'s reformulation of pangenesis as both particulate and inherited. The debate culminated in the concept of pangenesis, in which specific particles, later called gemmules, carry information from various parts of the body to the reproductive organs, from where they are passed to the embryo at the moment of conception. Although incorrect, the concept of pangenesis was

highly influential and persisted until the late 1800s.

(a) Pangenesis concept

(b) Germ-plasm theory



- **4.** Performation and Epigenesis: Organisms develop by expressing information carried in their hereditary material. As opposed to "preformation", the idea that in each sperm (or egg) is a tiny, fully-formed human that merely grows in size.
- **5.** Cell theory: The observations of Hooke, Leeuwenhoek, Schleiden, Schwann, Virchow ,and others led to the development of the cell theory. The cell theory is a widely accepted explanation of the relationship between cells and living things.

The cell theory states:

- **1.** All living things are composed of one or more cells.
- 2. Cells are the basic units of structure and function in living things.
- **3.** New cells are produced from existing cells .
- **4.** The cell theory holds true for all living things, no matter how big or small, or how simple or complex. Since according to research, cells are common to all living things, they can provide information about all life. And because all cells come from other cells, scientists can study cells to learn about growth, reproduction, and all other functions that living things perform .By learning about cells and how they function, you can learn about all types of living things.

5. Credit for developing cell theory is usually given to three scientists: Theodor Schwann, Matthias Jakob Schleiden, and Rudolf Virchow. In 1839, Schwann and Schleiden suggested that cells were the basic unit of life. Their theory accepted the first two tenets of modern cell theory) see next section, below. (However the cell theory of Schleiden differed from modern cell theory in that it proposed a method of spontaneous crystallization that he called Formation. "In 1858, Rudolf Virchow concluded that all cells come from preexisting cells, thus completing the classical cell theory.

Classical interpretation

- All organisms are made up of one or more cells.
- Cells are the fundamental functional and structural unit of life.
- All cells come from pre-existing cells.
- The cell is the unit of structure, physiology, and organization in living things.
- The cell retains a dual existence as a distinct entity and a building block in the construction of organisms.

Modern interpretation

The generally accepted parts of modern cell theory include:

- The cell is the fundamental unit of structure and function in living things.
- All cells come from pre-existing cells by division.
- Energy flow (<u>metabolism</u> and <u>biochemistry</u>) occurs within cells.
- Cells contain hereditary information (DNA) which is passed from cell to cell during cell division.
- All cells are basically the same in chemical composition.
- All known living things are made up of cells.
- Some organisms are unicellular, i.e., made up of only one cell.
- Others are multicellular, composed of a number of cells.
- The activity of an organism depends on the total activity of independent cells.

Mendelian and classical genetics

The modern science of genetics traces its roots to <u>Gregor Johann Mendel</u>, a German-Czech Augustinian <u>monk</u> and scientist who studied the nature of inheritance in plants. In his paper "Versuche über Pflanzenhybriden" ("<u>Experiments on Plant Hybridization</u>"), presented in 1865 to the *Naturforschender Verein* (Society for Research in Nature) in <u>Brünn</u>, Mendel traced the inheritance patterns of certain traits in pea plants and described them mathematically. Although this pattern of inheritance could only be observed for a few traits, Mendel's work suggested that heredity was particulate, not acquired, and that the inheritance patterns of many traits could be explained through simple rules and ratios.

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. 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. However, the modern science of genetics, which seeks to understand the process of inheritance, only began with the work of Gregor Mendel in the mid-nineteenth century. Although he did not know the physical basis for heredity, Mendel observed that organisms inherit traits via discrete units of inheritance, which are now called genes.

Genetics: The science of heredity that involves the structure and function of genes and the way genes are passed from generation to the next.

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.

Genetics often is divided into four major subdisplines:

- **A.** Transmission Genetics: Which deals with the transmission of genes from generation to generation.
- **B.** Molecular Genetics: Which deals with the structure and function at the molecular level.
- C. Population Genetics: Which deals with heredity in groups of individuals for traits that are determined by one or only a few genes.
- **D.** Quantitative Genetics: Which deals with heredity of traits in groups of individuals where the traits that are determined by many genes simultaneously.

Also the science of genetics has been subdivided into many active and interrelated fields such as biochemical genetics, immunogenetics, medical genetics, human genetics, developmental genetics and behavioral genetics.

Prokaryotes vs. Eukaryotes

Prokaryotes:

- o Eubacteria and Archaea. Usually unicellular.
- o No internal membrane-bound compartments: DNA floats free in the cytoplasm.
- o 1 circular chromosome (plus optional plasmids, which are also circular)
- o reproduction usually asexual
- o sexual processes (mixing DNA from 2 individuals) occur, but with unequal contributions from the 2 partners
- o transcription and translation simultaneous

Eukaryotes:

- o Plants, animals, fungi, protists. Often multicellular.
- o DNA contained within a membrane-bound nucleus.
- o linear chromosomes (usually more than 1)

Histone octamer plus 147 base pairs of DNA

(a) Nucleosomes

Lec. 1

o careful division of chromosomes in cell division: mitosis and meiosis

- o transcription separated from translation
- o sexual reproduction: 2 partners contribute equally to offspring
- o life cycle: alternation of haploid and diploid phases (i.e. 1 vs. 2 copies of each gene and chromosome).
- ❖ (almost) all inheritance is based on DNA: the sequence of ACGT nucleotides encodes all instructions needed to build and maintain an organism.
- ❖ A chromosome is a single DNA molecule together with other molecules (proteins and RNA) needed to support and read the DNA.
- ❖ A gene is a specific region of a chromosome that codes for a single polypeptide (linear chain of amino acids).
- ❖ Proteins_are composed of one or more polypeptides, plus in some cases other small helper molecules (co-factors). Proteins do most of the work of the cell.

Gene Expression

Genes are expressed in a 2 step process:

- First, an RNA copy of a single gene is made (transcription).
- Then, the nucleotide sequence of the RNA copy (messenger RNA) is translated into the amino acid sequence of the polypeptide.

The genetic code is a list of which 3 base DNA or RNA sequence (codon) encodes which amino acid. The same genetic code is used in (almost) all organisms.

((All cells in the body have the same DNA, but different genes are expressed in different cells and under different conditions.))
Genome:

The total amount of genetic material in a chromosome set; in eukaryotes, this is the amount of genetic material in the haploid set of chromosomes of the organism.

Chromatin:

The piece of DNA-protein complex that is studied and analyzed. Each chromatin fragment reflects the general features of chromosomes but not the specifics of any individual chromosome.

Spacer DNA plus H1 histone

Chromosome:

The genetic material of the cell, complexes with protein and organized into a number of linear structures. It literally means "colored body" because the threadlike structures are visible under the microscope only after they are attained with dyes.

Homologous chromosomes:

Paired chromosomes similar in shape, size, gene arrangement, and gene information.

1st Semester

Gene (Mendelian factor):

A segment of DNA that performs a specific function, such as coding for a particular protein.

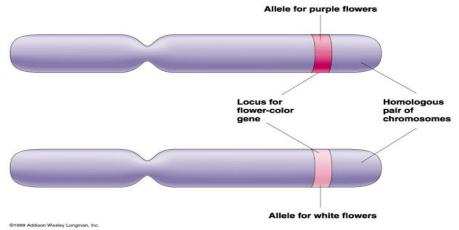
Allele: One of two or more alternative forms of a single gene sequence, and their activities are all concerned with the same biochemical and developmental process, although their individual phenotypes may differ.

Homozygous: Having identical alleles for the same gene.

Heterozygous: Having different alleles for the same gene.

Genotype: The complete genetic make up of an organism. (The genetic complement of an organism).

Phenotype: The observable properties of an organism (that are produced by the genotype and its interaction with the environment).



The genotype is the genetic constitution of an organism. The phenotype is the observable manifestation of the genetic traits. The genes give the potential for development of characteristic; this potential often is affected by interactions with other genes and with the environment. Thus individuals with the same genotype can have different phenotypes, and individuals with the same phenotypes may different genotypes.

The genotype is essentially a fixed character of an individual organisms; the genotype remains constant throughout life and is essentially unchanged by environmental effects. Most phenotypes change continually throughout the life of an organism as its genes interact with a sequence of environments. Fixity of genotype does not imply fixity of phenotype.

The Future of Genetics

The information content of genetics is increasing at a rapid pace. The genome sequences of many organisms are added to DNA databases every year, and new details about gene structure and function are continually expanding our knowledge of heredity. All of this information provides us with a better understanding of numerous biological processes and evolutionary relationships. The flood of new genetic information requires the continuous development of sophisticated computer programs to store, retrieve, compare, and analyze genetic data and has given rise to the field of bioinformatics, a merging of molecular biology and computer science.

1st Semester

Questions and problems:

- 1. Outline some of the ways in which genetics is important to each of us.
- 2. Briefly explain why genetics is crucial to modern biology.
- 3. List the three traditional subdisciplines of genetics and summarize what each covers.
- 4. Outline the notion of pangenesis and explain how it differs from the germ-plasm theory.
- 5. What does the concept of the inheritance of acquired characteristics propose and how is it related to the notion of pangenesis?
- 6. What is preformationism? What did it have to say about how traits are inherited?
- 7. Define blending inheritance and contrast it with preformationism.
- 8. Briefly define the following terms: (a) gene; (b) allele; (c) chromosome; (d) DNA; (e) RNA; (f) genetics; (g) genotype; (h) phenotype; (i) mutation; (j) evolution.
- 9. Outline the relations between genes, DNA, and chromosomes.
- 10. Genetics is said to be both a very old science and a very young science. Explain what is meant by this statement.
- 11. Match the theory or concept on the left with the correct description on the right.
- Preformationism (a) Each reproductive cell contains a complete set of genetic information.

Pangenesis (b) All traits are inherited from one parent.

Germ-plasm theory (c) Genetic information may be altered by the use of a feature.

Inheritance of acquired (d) Cells of different tissues characteristics contain different genetic information.

- 12. For each of the following genetic topics, indicate whether it focuses on transmission genetics, molecular genetics, or population genetics.
- **a.** Analysis of pedigrees to determine the probability of someone inheriting a trait.
- **b.** Study of the genetic history of people on a small island to determine why a genetic form of asthma is so prevalent on the island.
- **c.** The influence of nonrandom mating on the distribution of genotypes among a group of animals.
- **d.** Examination of the nucleotide sequences found at the ends of chromosomes.
- e. Mechanisms that ensure a high degree of accuracy during DNA replication.
- **f.** Study of how the inheritance of traits encoded by genes on sex chromosomes (sex-linked traits) differs from the inheritance of traits encoded by genes on nonsex chromosomes (autosomal traits).
- 13. The following concepts were widely believed at one time but are no longer accepted as valid genetic theories. What experimental evidence suggests that these concepts are incorrect and what theories have taken their place? (a) pangenesis; (b) the inheritance of acquired characteristics; (c) preformationism;
- (d) blending inheritance.