Chapter 18 – Heredity

Objectives

Given the synopsis in this chapter, competence in each objective will be demonstrated by responding to multiple choices or matching questions, completing fill-in questions, or writing short answers, at the level of 75% or greater proficiency for each student.

A. To explain what is heredity.
B. To explain the basic principles of Mendelian genetics.
C. To explain Mendel's law of segregation and law of recombination.

Heredity and Genetics

Heredity can be the defined as the sum of all biological processes and characteristics that are transmitted from parents to their offspring. These processes and characteristics include the internal workings of the organism, as well as more visible superficial traits.

Every member of a species has a set of genes specific to that species. This set of genes provides the constancy of the species. Among individuals within a species, however, variations occur in the form each gene takes, providing the genetic basis for different traits. The variations that a gene can take are called **alleles**.

The set of genes that an offspring inherits from both parents is called the organism's **genotype**. An organism's bodily structures, physiological processes, and behaviors is called the organism's **phenotype**.

The genotype remains constant throughout an organism's lifetime. However, the phenotype, depend on complex interactions between genes and their environment. Accordingly, as the organism's internal and external environments changes, so does its phenotype.

Because genes are integral to the explanation of hereditary observations, and because the study of genes is called genetics, the terms **heredity** and **genetics** are often used interchangeably.

Recent discoveries show that genes are important determinants of all aspects of an organism's makeup. For this reason, most areas of biological research now have a genetic component, and the study of genetics has a position of central importance in biology. Genetic research also has demonstrated that virtually all known organisms have similar genetic systems, with genes that are built on the same chemical principle and that function according to similar mechanisms. Although species differ in the sets of genes they contain, many similar genes are found across a wide range of species. This genetic unity has radically reshaped the understanding of the relationship between humans and all other organisms. Throughout history humans have modified organisms by using the techniques of selective breeding, and recently are using techniques involving recombinant DNA. Current biomedical research is uncovering critical roles for genes in disease.

This chapter (18) focuses on classic Mendelian patterns of inheritance and the organization of genes into chromosomes. Chapter 19 describes the functioning of genes at the molecular level, particularly the transcription of DNA, into RNA and the translation of RNA into proteins. Finally, the role of heredity in the evolution of species is discussed in chapter 20.

Mendelian genetics

Discovery and rediscovery of Mendel’s laws

Gregor Mendel published his work in the proceedings of the local society of naturalists in Brünn in 1866, but none of his contemporaries appreciated its significance. It was not until 1900, 16 years after Mendel’s death, that his work was rediscovered independently.

Mendel’s experiments looked at hybrids of different varieties of a common pea plant. His methods were unique for the time and focused on following the inheritance of single, easily visible and distinguishable traits, such as round versus wrinkled seed, yellow versus green seed, purple versus white flowers, and so on. In addition, he made exact counts of the numbers of plants bearing each trait; it was from such quantitative data that he deduced the rules governing inheritance.

Mendel obtained pea plants that were self-pollinated and descended for many generations from plants with similar traits (“pure” plants). Mendel crossed them by deliberately transferring the pollen of one variety to the pistils of another. The resulting first-generation hybrids, denoted by the symbol F_1, usually showed the traits of only one parent. For example, the crossing of pure yellow-seeded plants with pure green-seeded ones gave yellow seeds, and the crossing of pure purple-flowered plants with pure white-flowered ones gave purple-flowered plants. Results are shown in Table 1.

Table 1a. Crossing of pure pea plants with yellow seeds with pure pea plants with green seeds to produce first-generation hybrids (F_1).

Color of Seed	Yellow (AA)	Green (aa)
Yellow (AA)		**Yellow** (Aa)
Green (aa)	**Yellow** (Aa)	

Table 1b. Crossing of pure pea plants with Purple flowers with pure pea plants with white flowers to produce first-generation hybrids (F_1).

Color of Flower	Purple (RR)	White (rr)
Purple (RR)		**Purple** (Rr)
White (rr)	**Purple** (Rr)	

It looked to Mendel as if the yellow and purple “bloods” overcame or consumed the green and white “bloods.” Mendel called traits such as the yellow-seed color and the purple-flower color **dominant**; and traits such as the green-seed color and the white-flower color **recessive**.

Mendel then isolated and allowed the F_1 hybrid plants to self-pollinate, resulting in second-generation hybrids (F_2). Here, both the dominant and the recessive traits reappeared, as pure and uncontaminated as they were in the original parents (generation P). Moreover, these traits now appeared in constant proportions: about $^3/_4$ of the plants in the second generation showed the dominant trait and $^1/_4$ showed the recessive, a 3 to 1 ratio. Results are shown in Table 2

Table 2a. Self-pollination of F_1 hybrid pea plants with yellow seeds and F_1 hybrid pea plants with green seeds to produce second-generation hybrids (F_2).

Color of Seed	Yellow (AA)		Yellow (Aa)		Yellow (Aa)		Green (aa)	
Yellow (AA)								
Yellow (Aa)			Y(AA)	Y(Aa)				
			Y(aA)	G(aa)				
Yellow (Aa)					Y(AA)	Y(Aa)		
					Y(aA)	G(aa)		
Green (aa)								

Table 2b. Self-pollination of F_1 hybrid pea plants with purple flowers and F_1 hybrid pea plants with white flowers to produce second-generation hybrids (F_2).

Color of Flower	Purple (RR)		Purple (Rr)		Purple (Rr)		White (rr)	
Purple (RR)								
Purple (Rr)			P(RR)	P(Rr)				
			P(rR)	W(rr)				
Purple (Rr)					P(RR)	P(Rr)		
					P(rR)	W(rr)		
White (rr)								

Mendel concluded that the sex cells (the gametes) of the purple-flowered plants carried some factor that caused the progeny to develop purple flowers, and that the gametes of the white-flowered variety had a variant factor that induced the development of white flowers. In 1909 the Danish biologist Wilhelm Ludvig Johannsen proposed to call these factors genes.

Table 1 and Table 2 also illustrate how the genes are transmitted and in what particular ratios. Dominant genes are conventionally symbolized by capital letters and recessive genes by lowercase letters.

- **Dominant** genes are symbolized by capital letters.
- **Recessive** genes are symbolized by lowercase letters.

Since each pea plant contains genes half from the mother and half from the father, each plant should have two genes for flower color. If the two genes are alike – for instance, both having come from white-flowered parents (*rr*) – the plant is termed a **homozygote**.

If the two genes are different – for instance, one having come from a white-flowered parent (*rr*) and the other from a purple-flowered parent (RR) – the plant is termed **heterozygote** (Rr). Since the gene *R*, for purple, is dominant over *r*, for white, the F_1 generation hybrids will show purple flowers. They are phenotypically purple, but their genotype contains both *R* and *r* genes, and these alternative genes (alleles or allelic genes) do not blend or contaminate each other.

- **Allelic genes** – alternative forms of a gene (AKA **Alleles**)
- **Homozygous** – allelic genes from each parent are the same (e.g. AA or aa).
- **Heterozygous** – allelic genes from each parent are different (e.g. Aa or aA).

Mendel inferred that when a heterozygote forms its sex cells, the allelic genes segregate and pass to different gametes. This is expressed in the first law of Mendel, the **law of segregation** of unit genes.

Mendel also crossbred varieties of peas that differed in two or more easily distinguishable traits. When a variety with yellow round seed was crossed to a green wrinkled-seed variety, the F_1 generation hybrids produced yellow round seed. Evidently, yellow (*A*) and round (*B*) are dominant traits, and green (*a*) and wrinkled (*b*) are recessive. By allowing the F_1 plants (genotype *AaBb*) to self-pollinate, Mendel obtained an F_2 generation of 315 yellow round, 101 yellow wrinkled, 108 green round, and 32 green wrinkled seeds, a ratio of approximately 9 : 3 : 3 : 1. The important point here is that the segregation of the trait of color (*A* – *a*) is independent of the segregation of the trait of seed texture (*B* – *b*).

Yellow (*A*) and Round (*B*) are dominant traits
Green (*a*) and Wrinkled (*b*) are recessive traits

Table 3. Self-pollination of F_1 hybrid pea plants with yellow and round seeds to produce second-generation hybrids (F_2).

Color and texture of seeds	(AB)	(Ab)	(aB)	(ab)
(AB)	(AABB) Yellow/Round	(AABb) Yellow/Round	(AaBB) Yellow/Round	(AaBb) Yellow/Round
(Ab)	(AABb) Yellow/Round	(AAbb) Yellow/Wrinkled	(AaBb) Yellow/Round	(Aabb) Yellow/Wrinkled
(aB)	(AaBB) Yellow/Round	(AaBb) Yellow/Round	(aaBB) Green/Round	(aaBb) Green/Round
(ab)	(AaBb) Yellow/Round	(Aabb) Yellow/Wrinkled	(aaBb) Green/Round	(aabb) Green/Wrinkled

Yellow/Round = 9: Yellow/Wrinkled = 3: Green/Round = 3: Green/Wrinkled = 1

From these results and other similar results, Mendel derived his second law: the **law of recombination**, or independent assortment of genes. Ironically, the genes for the traits Mendel examined are located on separate chromosomes. If the genes for traits are located on the same chromosome, the law of recombination does not apply.