5B–Basic Genetics

Gregor Mendel was born in 1822 on a farm in Heinzendorf, Austria. At age 21 he entered the Augustinian order of the Roman Catholic church. As a monk he studied science at the University of Vienna and became an excellent mathematician. Later, as a schoolteacher, he engaged in many scientific activities. He recorded sun spots, read Darwin (with whom he disagreed), and maintained 50 beehives in which he tried to mate various European, American, and Egyptian types of bees. In 1857 Mendel began a program of selective breeding of peas in a small plot in the vegetable garden of Konigster monastery near Brunn, Moravia.

Mendel's experience in breeding and raising plants and animals on his father's farm, along with his mathematical and experimental science background from the university, equipped him well to investigate heredity. After 8 years of raising thousands of pea plants and recording and classifying many pages of notes, Mendel wrote a paper which presented a set of conclusions now called **Mendelian genetics.** In 1868 Mendel became the abbot of the monastery, and political problems forced him to give up most of his scientific work. In 1884 he died of a kidney disorder.

Gregor Mendel's paper on heredity in peas was published in 1865 but lay unnoticed in libraries for about 35 years. In 1900, after scientists had learned much about the cell, they rediscovered the paper and recognized its worth. Mendel became known as "the Father of Genetics." To understand modern genetic theories, you must understand what Mendel discovered about heredity.

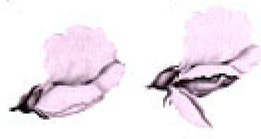
Mendelian Genetics

Mendel ordered 34 varieties of pea seeds, planted them, and observed their characteristics. From those varieties he chose 7 *sets of opposing characteristics*. For example, he noted that peas are either about 6 ft. tall or about 2 ft. tall. Tall and short are a set of opposing characteristics. Pod color is either green or yellow, another set of opposing characteristics. Peas are round or wrinkled.

5B-1 *Gregor Mendel (left), and a summary of the sets of characteristics he observed in peas (below).*

dominant trait	tall plants	axial flowers	green pods	inflated pods	yellow peas	round peas	colored seed coat
X	X	X	X	X	X	X	X
recessive trait	short plants	terminal flowers	yellow pods	constricted pods	green peas	wrinkled peas	white seed coat
F_1 generation	all tall plants	all axial flowers	all green pods	all inflated pods	all yellow peas	all round peas	all colored seed coat
F_2 generation	787 tall: 277 short (2.84:1)	651 axial: 207 terminal (3.14:1)	428 green: 152 yellow (2.82:1)	882 inflated: 229 constricted (2.95:1)	6022 yellow: 2001 green (3.01:1)	5474 round: 1850 wrinkled (2.96:1)	705 colored: 224 white (3.15:1)

The flower of the pea plant made it ideal for genetic experimentation. The petals are arranged so that the pollen (which contains the male gamete) naturally fertilizes the pistil (which contains the female gamete) of the same flower. This is called **self-pollination.** If Mendel wanted to **cross-pollinate** it, he had to tear open the petals and remove the pollen sacs before they matured. When the pistil was to be fertilized, he could supply pollen from another pea flower.



5B-2 *The pea flower: (left) closed, resulting in self-pollination; (right) opened, ready to be cross-fertilized*

Mendel began his experiments with peas that had been self-pollinating and breeding *true;* in other words, the tall plants always produced tall plants, the short plants always produced short plants, and so forth. He called these the parent plants and used the symbol P_1 to represent them. One of Mendel's experiments involved cross-pollinating a tall pea plant with a short pea plant. He called the offspring of this cross the **first filial* generation** (F_1). All the F_1 plants were tall. He allowed F_1 plants to self-pollinate and produce the **second filial generation** (F_2). Of the 1,064 plants in Mendel's F_2 generation, 787 of them were tall, and 277 of them were short.

Mendel's concepts

To explain the outcome of this experiment and similar results he obtained when he crossed peas with other sets of opposing characteristics, Mendel proposed several concepts. His concepts have been validated as scientists have observed similar results in other organisms and the cellular structures responsible for heredity. The following concepts are illustrated in the crosses described in figure 5B-3.

❑ **The concept of unit characteristics** Mendel stated that an organism's characteristics are caused by units which he called *factors* (now called *genes*), which occur in pairs. In pea plants the tall parent has two genes for being tall, which are represented by *TT;* and the short parent has two genes for being short, *tt*. Remember that genes (made of DNA) are responsible for inherited characteristics, that genes are located on chromosomes, and that most organisms have homologous pairs of chromosomes. Thus most normal organisms have *pairs* of genes in their cells.

❑ **The concept of dominant and recessive** Since the short plant of the P_1 could give only a short gene (*t*), and the tall plant only a tall gene (*T*), the F_1 generation was *Tt*. But rather than being medium-sized as expected, all the F_1 generation were tall. Mendel called a trait that expressed itself when factors for two opposing traits are present the **dominant trait** (caused by a *dominant gene*). The trait that is masked (hidden) when two genes for opposing traits are present is the **recessive trait** (caused by a *recessive gene*).

❑ **The concept of segregation** Mendel reasoned that when a cell forms gametes, the genes separate (segregate) so that there is only one gene for each characteristic in each gamete. Our knowledge of the behavior of *chromosomes* in meiosis indicates that Mendel's description of gamete formation is accurate.

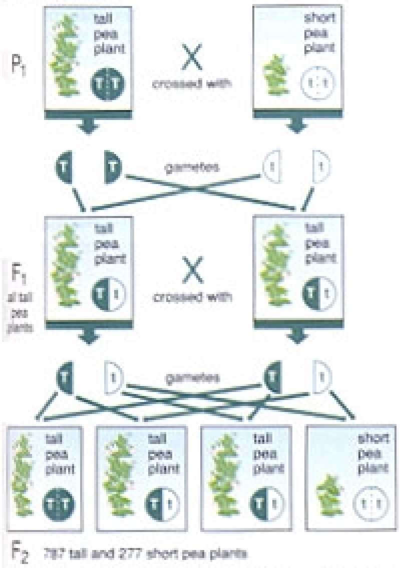
These concepts are also illustrated by the self-pollination which produced Mendel's F_2 generation of tall and short plants. Since the F_1 generation were all *Tt*, half of their pollen should contain a *T* gene and the other half a *t* gene. Half of the ova they produce should contain a *T* gene and the other half a *t* gene. This illustrates the concept of segregation.

The recessive gene (*t*) was not destroyed or altered when it was masked by the dominant gene in the F_1 generation; therefore, in the F_2 generation there are some short pea plants. Whenever both parent plants gave the recessive gene, the offspring expressed the recessive trait. This illustrates the concept of unit characteristics.

Notice the possible unions of the various gametes of the F_1 as illustrated in figure 5B-3. Three-fourths of the possible gamete combinations in the F_2 have at least one dominant gene; only 1/4 of them have two recessive genes. This, combined

filial (L. FILIUS, son)

with the fact that Mendel's F_2 generation results were 787 tall to 277 short (which is about 3/4 to 1/4), lends support to the concept of unit characteristics.



5B-3 A summary of Mendel's crosses of tall and short pea plants.

Genetic terms

Knowing the following genetic terms will help in our discussion of genetic principles:

❏ **Phenotype*** (FEE nuh TYPE) The expression of an organism's genes; what an organism is like (tall, green, constricted); not all genes result in a visible trait. For example, you inherited genes for digestive enzymes which are not seen but must work for you to be healthy. Expressing these genes is part of your phenotype.);

❏ **Genotype*** (JEN uh TYPE) The genes that the organism has (often expressed by letters such as *TT*, *Tt*, *tt*, and so forth);

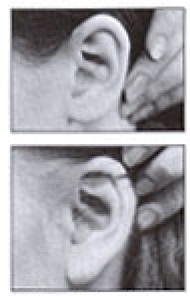
❏ **Allele** (uh LEEL) One pair of genes which has equivalent positions on homologous (paired) chromosomes; these are often expressed by letters–for example, *T* or *t;*

❏ **Homozygous*** (HOH moh ZYE gus) The condition of both alleles in a cell (organism) being the same–for example, *TT* or *tt;*

❏ **Heterozygous*** (HET uhr oh ZYE gus) The condition of both alleles in a cell (organism) *not* being the same–for example, *Tt;*

❏ **Monohybrid*** (MAHN oh HYE brid) **Cross** A cross that deals with only 1 set of alleles–that is, with 1 set of opposing characteristics.

Suppose a man with the dominant phenotype (free earlobes) knows that he is homozygous for the trait. His genotype is *FF*. His wife has the recessive phenotype. What kind of earlobes does she have? The only genotype a person with the recessive phenotype can have is *ff*. (If she had a dominant allele, her phenotype would be different.) All the man's sperm contain the dominant allele (*F*), and all the woman's ova the recessive allele (*f*). All their children, therefore, would have the heterozygous genotype (*Ff*). What would be the phenotype of these children?



5B-4 Attached (top) and free (bottom) earlobes.

Review Questions 5B-1

1. Who is "the Father of Genetics?" What did he do to earn this title?
2. Describe the (a) concept of unit characteristics, (b) the concept of segregation, and (c) the concept of dominance.
3. The gene for yellow peas is dominant over the gene for green peas. What is the difference between homozygous yellow peas and heterozygous yellow peas?
4. Compare and contrast the genotype and phenotype of an organism.
5. What is the difference between a gene and an allele?

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phenotype: pheno- (to show) + -type (Gk. TUPOS, type)
genotype: geno- (beginning) + -type (type)
homozygous: homo- (Gk. HOMOS, same) + -zygous (ZUGOS, yoked)
heterozygous: hetero- (other) + -zygous (yoked)
monohybrid: mono- (single) + -hybrid (L. HYBRIDA, offspring of different parents)