# 8

### Inheritance, Genes, and Chromosomes

#### **Chapter Outline**

- 8.1 Genes Are Particulate and Are Inherited According to Mendel's Laws
- 8.2 Alleles and Genes Interact to Produce Phenotypes
- 8.3 Genes Are Carried on Chromosomes
- 8.4 Prokaryotes Can Exchange Genetic Material

Genetic inheritance is explainable and predictable. Once you are familiar with the field's specialized vocabulary you will be able to accurately describe how the inherited characteristics of organisms (e.g., round versus wrinkled garden peas studied by Gregor Mendel in the 19<sup>th</sup> century) are based on what the parent generation provided to their offspring.

In the simple case of seed appearance in peas, there are two alleles, or alternate forms of the gene. One of the seed-appearance alleles codes for a protein that results in a round and smooth appearance on the surface of the seed. Another codes for a protein that results in a wrinkled and irregular appearance. By interbreeding "true-breeding" round-seed-producing plants with "true-breeding" wrinkled-seed-producing plants to produce F<sub>1</sub> offspring, Mendel found that *all* of the seeds in the F<sub>1</sub> offspring were round and smooth. "True-breeding" means that, after many generations, the offspring of members of the strain always have the same appearance as the parent and are assumed to be genetically identical. (Mendel did not know about DNA and proteins.)

An important detail from Mendel's studies is that the appearance (phenotype) of the pea seeds in his crosses was either round or wrinkled, not something in between. This shows that even if an individual pea plant inherited the allele for wrinkled seeds from one parent, receiving an allele for round and smooth seeds from the other parent results in all of the offspring's seeds being round and smooth. Thus, we say that the allele for round seed appearance is dominant over the recessive allele for wrinkled seed appearance. Such knowledge gives us predictability for what seeds will look like in offspring, provided we know about their parents and the inheritance patterns of the genes they have.

Geneticists utilize several systems for symbols of genes; we will utilize a simple system by describing the alleles for seed appearance in peas in the following way. The uppercase "S" represents the dominant allele for round and the lowercase "s" represents the recessive allele for wrinkled. By custom, most alleles are abbreviated by the first letter(s) of the dominant allele. Here, "S" indicates "smooth."

The greatest breakthrough Mendel made was not in the demonstration of simple dominant and recessive traits of garden pea phenotypes. Rather, it was what happened when he bred together the round F<sub>1</sub> offspring. These F<sub>1</sub> seeds and the plants that grew from them must have received one allele for round and one allele for wrinkled. Today we know that this is true because when producing gametes (pollen and ova in plants), only one of the two alleles for a gene will be present in any given gamete. Thus, there will be some pollen with a round allele for the gene for coat appearance and other pollen with the wrinkled allele, with the same either/or situation being true for the ova. When breeding the F<sub>1</sub> plants together to make the F<sub>2</sub> generation, Mendel kept track of how many smooth seeds were produced among the F<sub>2</sub> plants and noted that 75 percent of the F, plants were smooth and round while the other 25 percent were wrinkled. Mendel examined tens of thousands of peas to reach his results.

The possible genotypes for seed appearance in the  $F_2$  offspring described above is limited to this set: SS, Ss, sS, and ss. Recall that each parent contributes one of the alleles in its offspring. Any  $F_2$  plant with a single "S" will produce round seeds, the dominant trait, and we see that there are three such combinations: SS, Ss, and sS. By the same rule, there is only one combination of alleles, ss, that will result in wrinkled seeds. Therefore, there are three times as many round seeds (75 percent) as there are wrinkled seeds (25 percent) in the  $F_2$  generation.

In addition to explaining straightforward genetics, there are some tricks of nature presented in this chapter. For example, mitochondria and chloroplasts are organelles that almost exclusively come from the maternal parent because the gametes of females, the ova, are much larger cells than sperm. The larger cells are the ones that provide mitochondria and chloroplasts, as pollen and spermatozoa are generally too small to carry any cargo other than the primary genetic information. Since mitochondria and chloroplasts have some genes (and therefore, DNA) of their own, the "mother" is typically the sole source of mitochondria and chloroplast genes.

Chapter 8 covers aspects of **Big Idea 3** and **Big Idea 4**. **Big Idea 3**, stating that living systems store, retrieve, transmit, and respond to information essential to life processes, is the dominant theme of the chapter. Specific parts of the AP Biology curriculum that are covered in Chapter 8 include:

- 3.A.3: The chromosomal basis of inheritance provides an understanding of the pattern of passage (transmission) of genes from parent to offspring.
- 3.A.4: The inheritance pattern of many traits cannot be explained by simple Mendelian genetics.
- **3.C.2:** Biological systems have multiple processes that increase genetic variation.
- 3.C.3: Viral replication results in genetic variation, and viral infection can introduce genetic variation into the hosts.

**Big Idea 4** states that biological systems interact in complex ways. Specifically, this chapter includes:

• **4.C.2**: Environmental factors influence the expression of the genotype in an organism.

#### Chapter Review

heterozygous, dihybrid cross, phenotype, genotype, law of segregation, Punnett square, law of independent assortment, and pedigree analysis.

1. What can be deduced from the observation that all offspring have the phenotype of only one of the parents when two true-breeding animals with different traits are bred?

2. Predict and explain the expected result on the F<sub>1</sub> from breeding one type of wheat that is homozygous for the dominant allele of a particular trait with another strain that is homozygous for the recessive allele of that trait.

3. Explain the importance of Mendel's observations of the F<sub>2</sub> generation after he completed his careful observations of the F<sub>1</sub> generation.

**Concept 8.1** describes how genes are particulate and are inherited according to Mendel's laws. Key vocabulary terms in genetics that you need to be familiar with include: character, trait, parental generation (P), first filial generation ( $F_1$ ), second filial generation ( $F_2$ ), test cross, monohybrid cross, dominant, recessive, allele, homozygous,

Concept 8.2 explains how alleles and genes interact to produce phenotypes. Over the long term, it is clear that there have been many changes in the appearance of plants and animals. For example, the phenotype of today's human beings is likely quite different than the human phenotype of 200,000 years ago. We don't need to wait for 10,000 generations of breeding, however, before we see changes in phenotypes. A single gene with two or more alleles can result in two or more phenotypes. An allele present in 99 percent or more of the phenotypes seen in nature is labeled as the "wild" type allele of the gene, with alternate, uncommon alleles labeled as "mutants." In addition, the phenotype might be under the control of several genes that act in different patterns of expression when present in different combinations of alleles.

Match one of the following to demonstrates that phenotype		the examples in Q	uestions 4 through 7. Then explain how that
		codominance	incomplete dominance
4. True-breeding round peas	bred with true-	breeding wrinkled	peas produced offspring that were all round
Matching term:		Explanation:	·
5. True-breeding white-flow produced offspring that wer	ered snapdrago e all pink-flowe	ns bred with true- ered.	breeding red-flowered snapdragons
Matching term:		Explanation:	
· · · · · · · · · · · · · · · · · · ·			
6. A man with blood type A	and a woman w	vith blood type B p	roduce a daughter of blood type O.
Matching term:		Explanation:	
7. A male Labrador retriever produce a puppy that has a		at and a female La	brador retriever with a chocolate coat
Matching term:		Explanation:	

**Concept 8.3** examines how genes are carried on chromosomes. The seven phenotypic traits selected by Mendel for detailed study are examples of the simple dominant/recessive pattern of trait characterization. His idea that there are two "determinants" of each trait mesh well with later observations that chromosomes exist as pairs in cells (except in sperm and ova). Thus it was recognized, and later demonstrated, that one determinant can be on one chromosome, while the other member of the chromosome pair might have the same or another determinant.

We now know that DNA, the genetic code, is arranged in the form of the chromosomes that are found in the nucleus of eukaryotic cells. The "law of independent assortment" most directly applies to genes that reside on different chromosomes, as genes that are on different chromosomes are inherited independently of one another.

Not all genes are on different chromosomes, of course, and in general, two different genes that are located on the same chromosome are more likely to be inherited together than are two different genes that are located on different chromosomes. Thus, genes on the same (autosomal) chromosome are said to be "linked" in their pattern of inheritance. Linkage by virtue of sharing space on the same chromosome does not forever bind two genes to be inherited together. In the production of gametes, it is possible for genes to undergo recombination when exchanged between homologous chromosomes, in a process called "crossing over." Crossing over does not occur with sex chromosomes.

For the special case of genes located on the X and Y (sex) chromosomes, it is impossible for exchange between X and Y chromosomes. Among flies and humans, each male offspring (XY sex chromosomes) will have a Y chromosome identical to its paternal parent's Y chromosome. Each male offspring must also receive an X chromosome from its maternal parent. Although the Y chromosome has only a very limited number of genes, which are related to the differentiation of the testes, the X chromosome contains many, many more genes, and therefore the maternal parent determines more of the male offspring's traits than does the paternal parent, especially if the allele is recessive. Note that a male with a mutation or rare allele on the X chromosome can indeed pass along that mutation or rare allele, but only to his female offspring.

8. Plant scientists studying inheritance in sweet peas developed two true-breeding strains of peas, one with purple flowers and the other with red flowers. When these two strains were crossed, all of the F<sub>1</sub> were purple. Another trait of interest was pollen grain appearance. For this, two true-breeding strains were produced, one strain yielding long (tube-like) pollen grains and the other, round pollen grains. When these two strains were crossed, all of the offspring had long pollen grains.

Predict the flower color and pollen shape of 1,000 members of the F<sub>1</sub> generation for a mating between true-breeding purple-flowered, long-grained pollen peas and true-breeding red-flowered, round-grained pollen peas.

Now predict the flower color and pollen shape of 1,000 members of the F<sub>2</sub> generation, resulting from corssing F<sub>1</sub> plants above, and assuming Mendel's law of independent assortment applies to the flower and pollen traits.

Predict the flower color and pollen shape of 1,000 members and period assortment did <i>not</i> apply to the flower and period proposition and period as a previous prediction.	ers of the $F_2$ generation if Mendel's law of collen traits. This is considered to be the alternative $F_2$	nate
9. Steroid hormones (gonadal androgens), such as testoste reproductive system. However, if the receptors for the howing the gene for the androgen-receptor protein, a condition likely to develop and result in a female-like external pheral Y chromosome that is typically normal. The syndrome is Speculate on the chromosomal location of the mutation the explain your answer fully.	ormones are non-functional due to a mutation in called "complete androgen insensitivity" is motype. Note that all affected individuals have is not seen in genetic (XX) females, however.	е
10. Thomas Hunt Morgan reported on many different genseveral scenarios of eye color inheritance. Red eye color i all possible genotypes, including sex, that can be formed a red-eyed male (X <sup>R</sup> Y), and give an explanation for the ey	is dominant (R) over white eye color (r). Show by the cross of a white-eyed female (X <sup>r</sup> X <sup>r</sup> ) wi	V
11. The karyotype figure at the right shows the chromosomes of a person with Down syndrome, a trisomy in which there are more than the usual number of chromosome copies.  Circle the trisomy in the karyotype, and describe the errant inheritance process that resulted in its presence. ( <i>Hint</i> : Consider whether the trisomy more		9
likely results from an error in mitosis or an error in meisosis.)	10 11 12 13 14 15 16 17	18
	19 20 21 22 X X	X
	Courtesy: National Human Genome Research Institute	e

Concept 8.4 examines how prokaryotes can exchange genetic material. Most prokaryotes reproduce asexually by cloning (binary fission) and have only a single chromosome that is located in the cytosol of their cells. Evolution of prokaryotes is strongly driven by mutational changes in DNA, but there exist a limited number of ways that gene exchange between individuals can occur, even without sex. Bacteria can form a connection between two organisms, called a sex pilus. After DNA is moved through pili or conjugation tubes, segments of the DNA can be interchanged between the two genomes, via bacterial conjugation and genetic recombination. In a second variation of gene exchange, plasmid DNA of bacteria, a small circle of DNA independent of the larger segment of DNA in the chromosome, can move between bacteria and result in DNA transfer between individuals.

## Science Practices & Inquiry

In the AP Biology Curriculum Framework, there are seven Science Practices. In this chapter, we focus on Science Practice 1: The student can use representations and models to communicate scientific phenomena and solve scientific problems; Science Practice 2: The student can use mathematics appropriately; Science Practice 3: The student can engage in scientific questioning to extend thinking or to guide investigations within the context of the AP course; Science Practice 6: The student can work with scientific explanations and theories; and Science Practice 7: The student is able to connect and relate knowledge across various scales, concepts, and representations in and across domains.

Questions 14 and 15 ask you to analyze the empirical outcomes from genetic crosses in order to extract knowledge about the pattern of inheritance of genes. Question 16 provides data you will use to estimate the proximity of two genes.

In a completely hypothetical case, assume that "yogurt flies" were recently discovered in the International Space Station, and that these flies complete their life cycle in only 90 hours, making them ideal for genetic experiments. The astronauts observe variations in fly phenotypes. With lots of time on their hands, the astronauts decide to run some genetic crosses to see if inheritance patterns in space match those back on Earth.

14. First, they cross a true-breeding female with a purple abdomen to a true-breeding male with a pink abdomen. Here are the results of that cross:

	Females	Males
Purple abdomen	787	774
Pink abdomen	0	0

Being careful scientists, they arrange another cross, a true-breeding female with a pink abdomen to a true-breeding male with a purple abdomen. Here are the results of that cross:

	Females	Males
Purple abdomen	646	702
Pink abdomen	0	0

Discuss the inheritance pattern sh	iown.	

15. The scientists test another trait by crossing strains. First, they cross a true-breeding female with four antennae to a true-breeding male with two antennae. Here are the results of that cross:

	Females	Males
Four antennae	827	904
Two antennae	0	0

Seeking balance, they do the reciprocal cross: mating a true-breeding female with two antennae to a true-breeding male with four antennae. Here are the results of that cross:

	Females	Males
Four antennae	757	0
Two antennae	0	690

Discuss this second example of an inheritance pattern and compare it with the results on abdomen color.
*

16. A fruit scientist conducted genetic experiments to breed a pomegranate that is juicier and that stays fresh longer on the grocery shelf. She isolated two genes, F and J. She developed and then crossed these two lines of pomegranate shrubs:  $FfJj \times ffjj$ .

Predict what the cross would yield in percentages of offspring type if the genes are not linked (i.e., they are subject to Mendel's law of independent assortment). Write your percentages in the table.

%	offspring of the FJ type
%	offspring of the Fj type
%	offspring of the fJ type
%	offspring of the fj type

a. Explain why you chose these percentages.
The actual results of her cross were:
518 offspring of the FJ type (38%)
175 offspring of the <i>Fj</i> type (13%)
168 offspring of the fJ type (13%)
491 offspring of the fj type (36%)
1,352 total offspring
b. Discuss what these data suggest about the linkage and the distance between genes $F$ and $J$ .