Chapter 14: Mendel and the Gene Idea

1. In the 1800s the most widely favored explanation of genetics was blending.

The explanation of heredity most widely in favor during the 1800s was the "blending" hypothesis, the idea that genetic material contributed by the two parents mixes in a manner analogous to the way blue and yellow paints blend to make green. This hypothesis predicts that over many generations, a freely mating population will give rise to a uniform population, which contradicts empirical evidence. The blending hypothesis also fails to explain other phenomena of inheritance, such as traits reappearing after skipping a generation.

According to the alternative "particulate" hypothesis of inheritance popular today, parents pass on discrete heritable units (genes) that retain their separate identities in offspring. An organism's collection of genes is more like a deck of cards than a pail of paint. Like playing cards, genes can be shuffled and passed along, generation after generation, in undiluted form.

2. Explain how using pea plants allowed Mendel to control mating.

Peas have a short generation time and generate a large number of offspring from each mating. The reproductive organs of a pea plant are its flowers, and each pea flower has both pollen-producing organs (stamens) and an egg-bearing organ (carpel). In nature, pea plants usually self-fertilize. To achieve cross-pollination, Mendel removed the immature stamens of a plant before they produced pollen and then dusted pollen from another plant onto the altered flowers. Each resulting zygote then developed into a plant embryo encased in a seed (pea). Mendel could thus always be sure of the parentage of new seeds.

3. The P generation is the parental generation and the F_1 generation is the first filial generation.

True-breeding parents are the P generation, and their hybrid offspring are the F_1 generation. Self- or cross-pollination of these F_1 hybrids with other F_1 hybrids produces an F_2 generation. If my mother's grandparents were the P generation, my grandparents would be the F_1 generation, my mother would be the F_2 generation, and I would be the F_3 generation.

4. What did Mendel's simple cross of purple and white followers accomplish?

Mendel's experiments refuted blending by demonstrating that the "heritable factor" for the white trait had not been destroyed, deleted, or "blended" in the F_1 generation, but was merely masked by the presence of the factor for purple flowers. Mendel determined that purple flower color was a dominant trait, while white flower color was a recessive trait. Moreover, his work reveals the merit of experiments that cover multiple generations since the basic patterns of inheritance would have escaped him if he had stopped his experiments with the F_1 generation.

5. Alternate versions of the same gene, like purple and white flower color…

…are termed traits.

7. In sexually reproducing organisms, why are there exactly two chromosomes in each homolog?

A somatic cell has two copies of each chromosome (forming a homologous pair) and thus two copies of each gene; the alleles may be identical or different. Each chromosome derives from one of the sexually reproducing organism's parents.

8. Describe each of Mendel's concepts and indicate which can be observed during meiosis by placing an asterisk (*) beside it.

First, alternative versions of genes account for variations in inherited characters. These alternative genes are called alleles. Each gene is a sequence of nucleotides at a specific place, or locus, along a particular chromosome. The DNA at that locus, however, can vary slightly in its nucleotide sequence and hence in its information content.

Second, for each character, an organism inherits two copies of a gene, one from each parent. Each somatic cell in a diploid cell has two sets of chromosomes, one set inherited from each parent. Thus, a genetic locus is actually represented twice in a diploid cell, once on each homolog of a specific pair of chromosomes. The two alleles at a particular locus may be identical, as in true-breeding plants of Mendel's P generation, or different, as in Mendel's F_1 hybrids.

Third, if the two alleles at a locus differ, then one, the dominant allele, determines the organism's appearance, while the other, the recessive allele, has no noticeable effect on the organism's appearance.

Fourth, the two alleles for a heritable character separate from each other during gamete formation and end up in different gametes. This part of Mendel's model is known as the law of segregation. An egg or a sperm gets only one of the two alleles that are present in the somatic cells of the organism making the gamete. In terms of chromosomes, this segregation corresponds to the distribution of the two members of a pair of homologous chromosomes to different gametes in meiosis.

9. Explain the notations for the figure.

The F_2 genotypic ratio is 50% heterozygous, 25% homozygous recessive, and 25% homozygous dominant. The F_2 phenotypic ratio is 75% dominant and 25% recessive. The F_1 generation is completely heterozygous, while the F_2 generation has both heterozygous and homozygous offspring.

10. In pea plants, T *is the allele for tall plants, while* t *is the allele for dwarf plants. Demonstrate with a test cross how you could determine if a tall plant is homozygous tall or heterozygous tall.*

We can cross the mystery plant (*T?*) with a homozygous recessive short plant (*tt*), which will make only gametes with the recessive allele (*t*). The allele in the gamete contributed by the mystery plant will therefore determine the appearance of the offspring. If all the offspring of the cross are tall, then the mystery plant must be homozygous dominant (*TT*). But if both the short and tall phenotypes appear among the offspring, then the mystery plant must be heterozygous (*Tt*).

11. Explain the difference between a monohybrid cross and a dihybrid cross.

While monohybrids are heterozygous for one particular character being followed in a cross, dihybrids are heterozygous for *two* characters being followed in a cross.

12. Explain how the gametes are derived for the cross YyRr x YyRr.

According to the principle of independent assortment, the two pairs of alleles (*Yy* and *Rr*) segregate independently of each other. Genes are packaged into gametes in all possible allelic combinations, as long as each gamete has one allele for each gene. Thus, the F_1 plant in this example produces four classes of gametes in equal quantities: *YR*, *Yr*, *yR*, and *yr*.

13. Place the gametes from the cross in #12 in a Punnett square. Then provide the phenotypic ratio of the offspring.

If the sperm of the four classes fertilize eggs of the four classes in #12, there will be 16 equally probable ways in which the alleles can combine in the F_2 generation. These combinations result in four phenotypic categories with a ratio of 9:3:3:1.

14. Explain Mendel's law of independent assortment.

The law of independent assortment states that each pair of alleles segregates independently of each other pair of alleles during gamete formation. This law applies only to genes located on different chromosomes, or very far apart on the same chromosome.

15. An event that is certain to occur…

…has a probability of 1, while an event that is certain not to occur has a probability of 0.

16. What is an independent event?

An independent event, such as a coin toss, is unaffected by what has happened on previous trials.

17. What is the multiplication rule?

The multiplication rule calculates the probability of two or more independent events occurring together in some specific combination by multiplying the probability of one event by the probability of the other event. If you have a bag of 6 red and 3 green marbles, the probability of successively randomly choosing 2 red marbles out of the bag (if you replace a marble after removing one) would be $(6/9)^2 = 0.5 = 50\%$.

18. What is the addition rule?

The addition rule calculates the probability that any one of two or more mutually exclusive events will occur by adding their individual properties. If you have a bag of 6 red, 3 green and 1 blue marbles, the probability of randomly choosing a red or blue marble out of the bag would be $(6/10) + (1/10) = 0.7 = 70\%$.

19. What is the probability that a couple will have a girl, a boy, a girl, and a boy in this specific order?

$(1/2)^4 = 1/16 = 6.25\%$

20. Explain how incomplete dominance is different from complete dominance, and give an example of incomplete dominance.

When one allele in a pair shows complete dominance over the other, the phenotypes of the heterozygote and the dominant homozygote are indistinguishable. For some genes, however, neither allele is completely dominant, and the F_1 hybrids have a phenotype somewhere between those of the two parental varieties. This phenomenon, called incomplete dominance, is seen when red snapdragons are crossed with white snapdragons: all the F_1 hybrids have pink flowers. This third, intermediate phenotype results from flowers of the heterozygotes having less red pigment than the red homozygotes. This is unlike the case of Mendel's pea plants, where the *Pp* heterozygous make enough pigment for the flowers to be purple, indistinguishable from those of the *PP* plants.

21. Compare and contrast codominance with incomplete dominance.

In codominance, the two alleles each affect the phenotype in separate, distinguishable ways. For example, the human *MN* blood group is determined by codominant alleles for two specific molecules located on the surface of red blood cells, the *M* and *N* molecules. A single gene locus, at which two allelic variations are possible, determines the phenotype of this blood group. Individuals homozygous for the *M* allele (*MM*) have red blood cells with only *M* molecules; individuals homozygous for the *N* allele (*NN*) have red blood cells with only *N* molecules. But *both M* and *N* molecules are present on the red blood cells of individuals heterozygous for the *M* and *N* alleles (*MN*). The *MN* phenotype is *not* intermediate between the *M* and *N* phenotypes,

22. Why are dominant alleles not necessarily more common than recessive alleles in the gene pool?

An allele is dominant because it is seen in the phenotype, not because it somehow subdues a recessive allele. For any character, the observed dominant/recessive relationship of alleles depends on the level (organismal, biochemical, or molecular) at which we examine the phenotype. Dominant alleles are less likely to be more frequent than recessive alleles if they lead to conditions that are not evolutionarily selected for, such as some forms of polydactyly.

23. Explain what is meant when a gene is said to have multiple alleles.

Most genes exist in more than two allelic forms. The ABO blood groups in humans, for instance, are determined by three alleles of a single gene: I^A, I^B, and i.

24. Complete this ABO blood type chart.

Genotype	**Phenotype (Blood Group)**
I^AI^A I^Ai	A
I^BI^B I^Bi	B
I^AI^B	AB
ii	O

25. If a man with type AB blood marries a woman with type O, what blood types would you expect in their children?

Half the children would have blood type A (genotype I^Ai) and half would have blood type B (genotype I^Bi).

26. What is pleiotropy?

Most genes have pleiotropy, i.e. multiple phenotypic effects. In humans, for example, pleiotropic alleles are responsible for the multiple symptoms associated with certain hereditary diseases, such as cystic fibrosis and sickle-cell disease.

27. Explain epistasis.

In epistasis, the phenotypic expression of a gene at one locus alters that of a gene at a second locus. In Labrador retrievers, black coat color (*B*) is dominant to brown (*b*). For a Lab to have brown fur, its genotype must be *bb*; these dogs are called chocolate Labs. However, a second gene determines whether or not pigment will be deposited in the hair. The dominant allele (*E*) results in the deposition of either black or brown pigment, depending on the genotype at the first

locus. But if the Lab's genotype is *ee*, then the coat is yellow, regardless of the genotype at the black/brown locus. In this case, the gene for pigment deposition is said to be epistatic to the gene that codes for black or brown pigment.

28. Why does the dihybrid cross in Fig. 14.12 have four yellow Labs instead of the three predicted by Mendel's work?

As a result of epistasis, the phenotypic ratio among the F_2 offspring is 9 black to 3 brown to 4 yellow. Other types of epistatic interactions produce different ratios, but all are modified versions of 9:3:3:1.

29. Why his height a good example of polygenic inheritance?

For many characters, such as human skin color and height, an either-or classification is impossible because the characters vary in the population in gradations along a continuum. These are called quantitative characters.

30. Quantitative variation usually indicates…

…polygenetic inheritance.

31. Explain the potential influence of the environment on phenotypic expression.

A genotype generally is not associated with a rigidly defined phenotype, but rather with a range of phenotypic possibilities due to environmental influences. This phenotypic range is called the norm of reaction for a genotype. For some characters, such as the ABO blood group system, the norm of reaction has no breadth whatsoever; that is, a given genotype mandates a very specific phenotype. Other characteristics, such as a person's blood count of red and white cells, vary quite a bit, depending on such factors as the altitude, the customary level of physical activity, and the presence of infectious agents. Generally, norms of reaction are broadest for polygenic characters. Environment contributes to the quantitative nature of these characters, as we have seen in the continuous variation of skin color. Geneticists refer to such characters as multifactorial, meaning that many factors, both genetic and environmental, collectively influence phenotype.

32. What is the mode of inheritance for the pedigree in Fig. 14.15b?

The first-born daughter in the third generation has attached earlobes, although both of her parents lack that trait. Such a pattern is easily explained if the attached-lobe phenotype is due to a recessive allele. If it were due to a dominant allele, then at least one parent would also have had the trait.

33. Explain why you know the genotype of one female in the third generation, but are unsure of the other.

The cross of her heterozygous parents could result in either a homozygous dominant or a heterozygous genotype for the daughter exhibiting the free earlobe phenotype.

34. Describe what you think is important to know medically about the behavior of recessive alleles.

A recessively inherited disorder shows up only in the homozygous recessive individuals who inherit one recessive allele from each parent. Although phenotypically normal with regard to that disorder, heterozygotes may transmit the recessive allele to their offspring and thus are called carriers. When a disease-causing recessive allele is rare, it is relatively unlikely that two carriers of the same harmful allele will meet and mate. However, if the man and woman are close relatives, the probability of passing on recessive traits increases greatly. These are called consanguineous matings. Many deleterious alleles have such severe effects that a homozygous embryo spontaneously aborts long before birth, and stillbirths and birth defects are more common when parents are closely related.

35. Discuss the pattern of inheritance and the common symptoms of four genetic disorders.

Among people of European descent, one out of 25 (4%) are carriers of the cystic fibrosis allele. The normal allele for this gene codes for a membrane protein that functions in the transport of chloride ions between certain cells and the extracellular fluid. These chloride transport channels are defective or absent in the plasma membranes of children who inherit two recessive alleles for cystic fibrosis. The result is an abnormally high concentration of extracellular chloride, which causes the mucus that coats certain cells to become thicker and stickier than normal. The mucus builds up in the pancreas, lungs, digestive tract, and other organs, leading to pleiotropic effects, including chronic bronchitis and recurrent bacterial infections. If untreated, most children with cystic fibrosis die before their fifth birthday.

Sickle-cell disease is caused by the substitution of a single amino acid in the hemoglobin protein of red blood cells; in homozygous individuals, all hemoglobin is of the sickle-cell variety. When the oxygen content of an affected individual's blood is low, the red blood cells deform into a sickle shape. Sickled cells may clump and clog small blood vessels, often leading to other symptoms throughout the body, including physical weakness, organ damage, and even paralysis. Although two sickle-cell alleles are necessary for an individual to manifest full-blown sickle-cell disease, the presence of one sickle-cell allele can affect the phenotype. Thus, at the organismal level, the normal allele is

incompletely dominant to the sickle-cell allele. A the molecular level, the two alleles are codominant; both normal and abnormal hemoglobins are made in heterozygotes.

In achondroplasia, a form of dwarfism, heterozygous individuals have the dwarf phenotype. Therefore, all people who are not achondroplastic dwarfs are homozygous for the recessive allele. Dominant alleles that cause a lethal disease are much less common than recessive alleles that have lethal effects. However, a lethal dominant allele is able to be passed on if it causes death at a relatively advanced age. In Huntington's disease, a degenerative disease of the nervous system, the lethal dominant allele has no obvious phenotypic effect until the individual is about 35 to 45 years old. Once the deterioration of the nervous system begins, it is irreversible and inevitably fatal. As with other dominant traits, a child born to a parent with the Huntington's disease allele has a 50% chance of inheriting the allele and the disorder.

36. Explain the three main steps in amniocentesis and the two main steps of CVS.

In amniocentesis, a sample of amniotic fluid can be taken starting at the 14^{th} to 16^{th} week of pregnancy. Biochemical and genetic tests can be performed immediately on the amniotic fluid or later on the cultured cells. Fetal cells must be cultured for several weeks to obtain sufficient numbers for karyoptyping. In CVS, a sample of chronic villus tissue can be taken as early as the 8^{th} to 10^{th} week of pregnancy. Karyotyping and genetic tests can be performed on the fetal cells immediately, providing results within a day or so.

37. What are the strengths and weaknesses of each fetal test?

The rapid analysis offered by CVS is faster than amniocentesis, in which cells must be cultured for several weeks before karyotyping. Another advantage of CVS is that it can be performed at least four weeks earlier.

38. Explain the symptoms of phenylketonuria.

Some genetic disorders can be detected at birth by simple biochemical tests that are now routinely performed in most hospitals in the United States. One common screening program is for phenylketonuria (PKU), a recessively inherited disorder. Affected individuals cannot properly metabolize the amino acid phenylalanine, causing the compound and its byproduct to accumulate to toxic levels in the blood, causing severe intellectual disability. However, if PKU is detected in the newborn, a special diet low in phenylalanine will usually allow normal development.