

## chapter 3

## Extensions to Mendel's Laws

## Synopsis

In Chapter 3, we see that the relationship between genotype and phenotype can be more complicated than envisaged by Mendel. Alleles do not have to be completely dominant or recessive with respect to each other. Not all genotypes are equally viable. Genes can have more than two alleles in a population. One gene can govern more than one phenotype. A single phenotype can be influenced by more than one gene, and these genes can interact in a variety of ways.

Despite these complications, the alleles of individual genes still segregate according to Mendel's Law of Segregation, and different pairs of genes still usually behave as dictated by Mendel's Law of Independent Assortment.

## Key terms

**wild-type alleles** – alleles with a frequency of greater than 1% in the population. Colloquially, wild-type alleles are the *normal* alleles found most commonly in the population.

**mutant alleles** – rare alleles with a frequency of less than 1% in the population.

**monomorphic gene** – a gene with only one common, wild-type allele.

**polymorphic gene** – a gene with many wild-type alleles. The wild-type alleles of a polymorphic gene are often called **common variants**.

**incomplete dominance** and **codominance** – cases in which the phenotype of heterozygotes is different than that of either type of homozygote. Incomplete dominance describes alleles where the heterozygote has a phenotype in between that of either homozygote, while heterozygotes for codominant alleles have both of the phenotypes associated with each homozygote. Usually in incomplete dominance one allele is nonfunctional or only partially functional, while in codominance both alleles are fully functional.

**recessive lethal allele** – an allele (usually a loss-of-function allele) of an **essential gene** necessary to the survival of the individual. A zygote homozygous for a recessive lethal allele cannot survive and thus is not detected among the progeny of a cross.

**dominance series of multiple alleles** – Although each individual has only two alleles of a gene, many alleles of the gene may exist in the population. These alleles may be completely dominant, incompletely dominant, or codominant with respect to each other as determined by the phenotype of heterozygotes for the particular pair.

**pleiotropy** – A gene may affect more than one phenotype.

**epistasis** – An allele of one gene hides the effects of different alleles at a second gene.

**redundant genes** – Two or more genes provide the same function.

**penetrance** – the fraction of individuals with a particular genotype who display the genotype's characteristic phenotype.

**expressivity** – the degree to which an affected individual displays the phenotype associated with that individual's genotype. Expressivity of a genotype can vary due to environment, chance, and alleles of other genes (**modifier genes**).

**conditional mutation** – a change in the base sequence of a gene that affects gene function only under specific environmental conditions.

**continuous (quantitative) trait** – a trait whose phenotype varies over a wide range of values that can be measured. Continuous traits are **polygenic** – they are controlled by the combined activities of many genes.

**locus heterogeneity** – exhibited by a trait where mutation in any one of two or more genes results in the same mutant phenotype.

**complementation test** – method of discovering whether two mutations are in the same or separate genes. Two mutant strains with the same mutant phenotype are crossed. If the progeny are all wild type, **complementation** occurred and the strains had mutations in different genes. If instead the progeny of this cross are all mutant, no complementation occurred and the strains had mutations in the same gene.

## Exceptions to the 3:1 Mendelian monohybrid ratio

**1:2:1** – Ratio of progeny genotypes and phenotypes in a cross between hybrids when there is **incomplete dominance** or **codominance**:

$$(Aa \times Aa \rightarrow 1 AA : 2 Aa : 1aa)$$

Note that in incomplete dominance and codominance, a new (third) phenotype will appear in the hybrids ( $Aa$ ) of the  $F_1$  generation. In the  $F_2$  generation, this same phenotype must be the largest component of the 1:2:1 monohybrid ratio.

**2:1** – Ratio of progeny phenotypes observed in a cross between hybrids when one allele is a **recessive lethal allele** that has a dominant effect on a visible phenotype:

$$(Aa \times Aa \rightarrow 1 \text{ red } AA : 2 Aa : 1aa)$$

Note that in this case, homozygotes for the recessive lethal allele  $A$  die (*red color*), but  $Aa$  heterozygotes have a phenotype different from  $aa$  homozygotes.

## Interactions of two genes

You should be able to recognize traits influenced by two genes as variations on the 9:3:3:1 ratio of genotypic classes resulting from a dihybrid cross. For your convenience, an abbreviated version of [Table 3.2](#) summarizing these gene interactions is presented at the top of the next page. It is particularly useful to understand the concepts of additivity, epistasis, redundancy, and complementation.

If you are given the details of a biochemical pathway, you should be able to work out the ratios of phenotypes expected among the progeny of a cross. Note that you cannot go in the opposite direction: A particular ratio does not tell much about the underlying biochemistry. Thus, you should **NOT** try to memorize specific examples relating particular ratios to specific biochemical pathways. Instead, think about each problem from the ground up each time.

**F<sub>2</sub> Genotypic Ratios from an  
F<sub>1</sub> Dihybrid Cross**

Gene interaction	A- B-	A- bb	aa B-	aa bb	F <sub>2</sub> Phenotypic Ratio
<b>Additive:</b> Four distinct F <sub>2</sub> phenotypes	9	3	3	1	9:3:3:1
<b>Recessive epistasis:</b> Homozygous recessive allele of one gene masks both alleles of another gene	9	3	3	1	9:3:4
<b>Reciprocal recessive epistasis:</b> When homozygous, recessive alleles of each gene mask the dominant allele of the other gene	9	3	3	1	9:7
<b>Dominant epistasis I:</b> Dominant allele of one gene hides effects of both alleles of other gene	9	3	3	1	12:3:1
<b>Dominant epistasis II:</b> Dominant allele of one gene hides effects of dominant allele of other gene	9	3	3	1	13:3
<b>Redundancy:</b> Only one dominant allele of either of two genes is necessary to produce phenotype	9	3	3	1	15:1

## Problem Solving

In [Chapter 2](#), the major goal was to determine which allele of a gene is dominant and which is recessive, and then to ascribe genotypes to various individuals or classes of individuals based on the ratio of progeny types seen in a cross. The challenges become more difficult in this chapter, but the first step in problem solving remains the same: You need to **DIAGRAM THE CROSS in a consistent manner**. The next steps are to answer the following questions:

**How many genes are involved in determining the phenotype?**

**How many alleles of each gene are present?**

**What phenotypes are associated with which genotypic classes?** (The answer to this last question will help you understand the dominance relationships between the alleles of each gene and the interactions between alleles of traits determined by more than one gene.)

The points listed below will be particularly helpful in guiding your problem solving:

- To distinguish between one gene and two gene traits, look for the number of phenotypic classes in the F<sub>2</sub> generation and the ratios in the F<sub>2</sub>s among those classes. If a single gene is involved, there will be either two classes (3:1, or 2:1 if an allele is a recessive lethal) or three classes (1:2:1 in the cases of codominance or incomplete dominance). If two genes are involved, you could see two classes (9:7, 13:3, or 15:1) or three classes (9:3:4 or 12:3:1) or four classes (9:3:3:1). (*Note:* These ratios require that the P generation is true-breeding and that the F<sub>1</sub> crosses examined are between monohybrids or dihybrids.)
- Understand that when there is codominance or incomplete dominance, a novel phenotype will appear in the F<sub>1</sub> generation. In the F<sub>2</sub> generation, this same phenotype must be the largest component of the 1:2:1 monohybrid ratio.
- If you see a series of crosses involving different phenotypes for a certain trait like coat color, and each cross gives a monohybrid ratio (3:1 or 1:2:1), then all the phenotypes are controlled by one gene with many alleles that form an **allelic series**. You should write out the dominance hierarchy for this series (e.g.,  $a = b > c$ ) to keep track of the relationships among the alleles.
- Lethal alleles are almost always recessive because a zygote with a dominant lethal allele could not grow into an adult. (The only exceptions to this rule involve **conditional lethal alleles** that survive in some environments but not others.) On the basis of what you have learned in this chapter, you can recognize recessive lethal alleles if they are pleiotropic and show a dominant visible phenotype such that the monohybrid phenotypic ratio is 2 (dominant phenotype) : 1 (recessive phenotype).
- Remember that the 9:3:3:1 dihybrid ratio and its variants represent various combinations of the genotypic classes 9  $A- B-$  : 3  $A- bb$  : 3  $aa B-$  : 1  $aa bb$ , where the dash indicates either a dominant or recessive allele. Based on the observed ratios, you should be able to tell which genotypic classes correspond to which phenotypes. Although you should not memorize the table on the previous page displaying these variants of 9:3:3:1, you should be able to consider whether particular biochemical explanations fit the ratios seen.
- Don't forget to use the product rule of probability to determine the proportions of genotypes or phenotypes for independently assorting genes.
- Sometimes genotypes and phenotypes share similar symbols. To distinguish these usages, remember that in this book genotypes are always in italics (i.e.,  $Rb^+$ ), while phenotypes are written in Roman script (i.e., Rh<sup>+</sup>).

## Vocabulary

### 1.

- |                   |  |
|-------------------|--|
| a. epistasis      | 2. the alleles of one gene mask the effects of alleles of another gene |
| b. modifier genes | 5. genes whose alleles alter phenotypes produced by the                |

- |                          |   |
|--------------------------|---|
|                          | action of other genes   |
| c. conditional lethal    | 10. a genotype that is lethal in some situations (for example, high temperature) but viable in others                     |
| d. permissive conditions | 7. environmental condition that allows conditional lethals to live  |
| e. reduced penetrance    | 6. less than 100% of the individuals possessing a particular genotype express it in their phenotype                       |
| f. multifactorial trait  | 8. a trait produced by the interaction of alleles of at least two genes or from interactions between gene and environment |
| g. incomplete dominance  | 11. the heterozygote resembles neither homozygote   |
| h. codominance           | 3. both parental phenotypes are expressed in the F <sub>1</sub> hybrids   |
| i. mutation              | 4. a heritable change in a gene   |
| j. pleiotropy            | 1. one gene affecting more than one phenotype   |
| k. variable expressivity | 9. individuals with the same genotype have related phenotypes that vary in intensity                                      |

## Section 3.1

2. The problem states that the intermediate pink phenotype is caused by incomplete dominance for the alleles of a single gene. We suggest that you employ genotype symbols that can show the lack of complete dominance; the obvious  $R$  for red and  $r$  for white does not reflect the complexity of this situation. In such cases we recommend using a base letter as the gene symbol and then employing superscripts to show the different alleles. To avoid any possible misinterpretations, it is always advantageous to include a separate statement making the complexities of the dominant/recessive complications clear.

Designate the two alleles  $F^r$  = red and  $F^w$  = white, so the possible genotypes are  $F^rF^r$  = red;  $F^rF^w$  = pink; and  $F^wF^w$  = white. Note that the phenotypic ratio is the same as the genotypic ratio in incomplete dominance.

- a. Diagram the cross:  $F^rF^w \times F^rF^w \rightarrow 1/4 F^rF^r$  (red) :  $1/2 F^rF^w$  (pink) :  $1/4 F^wF^w$  (white).
- b.  $F^wF^w \times F^rF^w \rightarrow 1/2 F^rF^w$  (pink) :  $1/2 F^wF^w$  (white).
- c.  $F^rF^r \times F^rF^r \rightarrow 1 F^rF^r$  (red).
- d.  $F^rF^r \times F^rF^w \rightarrow 1/2 F^rF^r$  (red) :  $1/2 F^rF^w$  (pink).
- e.  $F^wF^w \times F^wF^w \rightarrow 1 F^wF^w$  (white).

f.  $F^rF^r \times F^wF^w \rightarrow 1 F^rF^w$  (pink).

The cross shown in **part (f)** is the most efficient way to produce pink flowers, because all the progeny will be pink.

3. In Mendel's *Pp* heterozygotes, the amount of enzyme leading to purple pigment is sufficient to produce purple color as intense as the purple color in *PP* homozygotes. Presumably the heterozygote has enough enzyme P so that the maximal level of purple pigment is produced; more enzyme cannot make more purple pigment.

In the snapdragons in [Fig. 3.3](#), the amount of red pigment in the *Aa* heterozygote is less than that in the *AA* homozygote. Presumably here, the amount of enzyme A catalyzing the production of the red pigment in the heterozygotes is insufficient to produce the maximum level of the red pigment seen in the *AA* homozygote. That is, in the case of this snapdragon gene, **the intensity of the phenotype is proportional to the dosage of functional alleles (1 dose in the *Aa* heterozygote; 2 doses in the *AA* homozygote)**. In such cases of incomplete dominance, it is usually assumed that *AA* has twice as much of the protein product of the gene as does *Aa*, if *a* is a null allele.

4. Presumably, because *r* is nonfunctional, *RR* peas have twice the number of Sbe1 protein molecules as *Rr* peas, while *rr* peas have zero Sbe1 protein molecules. If we describe the phenotype as the number of Sbe1 molecules, the *R* and *r* alleles would exhibit **incomplete dominance** because the phenotype of the heterozygote is in between that of dominant and recessive homozygotes.

5. a. Diagram the cross:

$$e^+e^+ \times e^+e \rightarrow 1/2 e^+e^+ : 1/2 e^+e.$$

The trident marking is only found in the heterozygotes, so the probability is **1/2**.

- b. The offspring with the trident marking are  $e^+e$ , so the cross is  $e^+e \times e^+e \rightarrow 1/4 ee : 1/2 e^+e : 1/4 e^+e^+$ . Therefore, of 300 offspring, **75 should have ebony bodies, 150 should have the trident marking and 75 should have honey-colored bodies**.

6. Diagram the cross:

$$\text{yellow} \times \text{yellow} \rightarrow 38 \text{ yellow} : 22 \text{ red} : 20 \text{ white}$$

Three phenotypes in the progeny indicate that the yellow parents are not true-breeding. The ratio of the progeny is close to  $1/2 : 1/4 : 1/4$ . This is the result expected for crosses between individuals heterozygous for incompletely dominant alleles. Thus:

$$C^rC^w \times C^rC^w \rightarrow 1/2 C^rC^w (\text{yellow}) : 1/4 C^rC^r (\text{red}) : 1/4 CC^w (\text{white}).$$

7. A cross between individuals heterozygous for incompletely dominant alleles of a gene give a ratio of  $1/4$  (one homozygote) :  $1/2$  (heterozygote with the same phenotype as the parents) :  $1/4$  (other homozygote). Because the problem already states which genotypes correspond to which phenotypes, you know that the color gene will give a monohybrid phenotypic ratio of  $1/4$  red :  $1/2$  purple :  $1/4$  white, while the shape gene will give a monohybrid phenotypic ratio of  $1/4$  long :  $1/2$  oval :  $1/4$  round.

Because the inheritance of these two genes is independent, use the product rule to generate all the possible phenotype combinations (note that there will be  $3 \times 3 = 9$

classes) and their probabilities, thus generating the dihybrid phenotypic ratio for two incompletely dominant genes: **1/16 red long : 1/8 red oval : 1/16 red round : 1/8 purple long : 1/4 purple oval : 1/8 purple round : 1/16 white long : 1/8 white oval : 1/16 white round**. As an example, to determine the probability of red long progeny, multiply  $1/4$  (probability of red)  $\times$   $1/4$  (probability of long) =  $1/16$ . If you have trouble keeping track of the 9 possible classes, it may be helpful to list the classes in the form of a branch diagram (*not shown*) or a table as follows:

Phenotype	Probability of phenotype
red, long	$1/4 \times 1/4 = 1/16$
red, oval	$1/4 \times 1/2 = 1/8$
red, round	$1/4 \times 1/4 = 1/16$
purple, long	$1/2 \times 1/4 = 1/8$
purple, oval	$1/2 \times 1/2 = 1/4$
purple, round	$1/2 \times 1/4 = 1/8$
white, long	$1/4 \times 1/4 = 1/16$
white, oval	$1/4 \times 1/2 = 1/8$
white, round	$1/4 \times 1/4 = 1/16$

8. The cross is: white long  $\times$  purple short  $\rightarrow$  301 long purple : 99 short purple : 612 long pink : 195 short pink : 295 long white : 98 short white.

Deconstruct this dihybrid phenotypic ratio for two genes into separate constituent monohybrid ratios for each of the 2 traits: flower color and pod length. For flower color note that there are 3 phenotypes: 301 + 99 purple : 612 + 195 pink : 295 + 98 white = 400 purple : 807 pink : 393 white =  $1/4$  purple :  $1/2$  pink :  $1/4$  white. This is a typical monohybrid ratio for incompletely dominant alleles, so **flower color is caused by incompletely dominant alleles of a gene, with  $C^P$  giving purple when homozygous,  $C^W$  giving white when homozygous, and the  $C^P C^W$  heterozygotes giving pink**.

For pod length, the phenotypic ratio is (301 + 612 + 295) long : (99 + 195 + 98) short = 1208 long : 392 short =  $3/4$  long :  $1/4$  short. This 3:1 ratio is that expected for a cross between individuals heterozygous for a gene in which one allele is completely dominant to the other, so **pod shape is controlled by a single gene with the long allele ( $L$ ) completely dominant to the short allele ( $l$ )**.

9. Remember that the gene determining ABO blood groups has 3 alleles and that  $I^A = I^B > i$ .
- a. The O phenotype means the girl's genotype is  $ii$ . Each parent contributed an  $i$  allele, so her parents could be  $ii$  (O) or  $I^A i$  (A) or  $I^B i$  (B) in any combination.



- b. A person with the B phenotype could have either genotype  $I^B I^B$  or genotype  $I^B i$ . The mother is A and thus could not have contributed an  $I^B$  allele to this daughter. Instead, because the daughter clearly does not have an  $I^A$  allele, the mother must have contributed the  $i$  allele to this daughter. The mother must have been an  $I^A i$  heterozygote. The father must have contributed the  $I^B$  allele to his daughter, so he could be **either  $I^B I^B$ ,  $I^B i$ , or  $I^B I^A$ .**
- c. The genotypes of the girl and her mother must both be  $I^A I^B$ . The father must contribute either the  $I^A$  or the  $I^B$  allele, so there is only one phenotype and genotype which would exclude a man as her father – **the O phenotype (genotype  $ii$ ).**
10. To approach this problem, look at the mother/child combinations to determine what alleles the father must have contributed to each child's genotype.
- a. The father had to contribute  $I^B$ ,  $N$ , and  $Rb^-$  alleles to the child. The only male fitting these requirements is **male d** whose phenotype is B, MN, and  $Rh^+$  (note that the father must be  $Rb^+ Rb^-$  because the daughter is  $Rh^-$ ; the same is true of the mother).
- b. The father had to contribute  $i$ ,  $N$ , and  $Rb^-$  alleles. The father could be either male c (O MN  $Rh^+$ ) or male d (B MN  $Rh^+$ ). As we saw previously, male d is the only male fitting the requirements for the father in part (a). Assuming one child per male as instructed by the problem, the father in part (b) must be **male c**.
- c. The father had to contribute  $I^A$ ,  $M$ , and  $Rb^-$  alleles. Only **male b** (A M  $Rh^+$ ) fits these criteria. (Note that the father must be  $Rb^+ Rb^-$ .)
- d. The father had to contribute either  $I^B$  or  $i$ ,  $M$ , and  $Rb^-$ . Three males have the alleles required: these are male a, male c, and male d. However, of these three possibilities, only **male a** remains unassigned to a mother/child pair.
11. Designate the alleles:  $p^m$  (marbled)  $>$   $p^s$  (spotted)  $=$   $p^d$  (dotted)  $>$   $p^c$  (clear).
- a. Diagram the crosses:
- $p^m p^m$  (homozygous marbled)  $\times$   $p^s p^s$  (spotted)  $\rightarrow$   $p^m p^s$  (marbled  $F_1$ )
  - $p^d p^d$   $\times$   $p^c p^c$   $\rightarrow$   $p^d p^c$  (dotted  $F_1$ )
  - $p^m p^s$   $\times$   $p^d p^c$   $\rightarrow$   $1/4 p^m p^d$  (marbled) :  $1/4 p^m p^c$  (marbled) :  $1/4 p^s p^d$  (spotted dotted) :  $1/4 p^s p^c$  (spotted) =  **$1/4$  spotted dotted :  $1/2$  marbled :  $1/4$  spotted.**
- b. The  $F_1$  from cross 1 are **marbled ( $p^m p^s$ )** from the first cross **and dotted ( $p^d p^c$ )** from the second cross as shown in part (a).
12. Suppose, as maintained by your fellow student, that spotting is due to the action of one gene with alleles  $S$  (spotting) and  $s$  (no spots), and that dotting is due to the action of a second gene with alleles  $D$  (dotting) and  $d$  (no spots). The cross series shown in Fig.



3.4a, starting with true-breeding spotted and true-breeding dotted strains, could then be diagrammed as:

$SS\ dd \times ss\ DD \rightarrow Ss\ Dd$  (spotted and dotted  $F_1$ )  $\rightarrow F_2$  consisting of 9  $S-\ D-$  (spotted and dotted) : 3  $S-\ dd$  (spotted, not dotted) : 3  $ss\ D-$  (not spotted, dotted) : 1  $ss\ dd$  (not spotted, not dotted)

Thus, the alternative hypothesis suggested by your fellow student would predict that some lentils would be found in the  $F_2$  generation that would be neither spotted nor dotted. The results shown in Fig. 3.4 do not include any such lentils. **If you counted a large number of  $F_2$  individuals and you failed to see lentils that were neither spotted nor dotted, you would be able to exclude the hypothesis that two genes were involved.**

13. a. All the crosses have results that can be explained by one gene – either a 3:1 phenotypic monohybrid ratio showing that one allele is completely dominant to the other (crosses 1, 3, and 5); or a 1:1 ratio showing that a testcross was done for a single gene (crosses 2, 7, and 9); or all progeny with the same phenotype as one or both parents (crosses 4, 6, and 8); or a 1:2:1 phenotypic monohybrid ratio (cross 10). You can thus conclude that all the coat colors are controlled by the alleles of one gene, with chinchilla ( $C$ ) > himalaya ( $c^h$ ) > albino ( $c^a$ ).

- b.
1.  $c^h c^a \times c^h c^a$
  2.  $c^h c^a \times c^a c^a$
  3.  $Cc^h \times C(c^h \text{ or } c^a)$
  4.  $CC \times c^h (c^h \text{ or } c^a)$
  5.  $Cc^a \times Cc^a$
  6.  $c^h c^h \times c^a c^a$
  7.  $Cc^a \times c^a c^a$
  8.  $c^a c^a \times c^a c^a$
  9.  $Cc^h \times c^h (c^h \text{ or } c^a)$  or  $Cc^a \times c^h c^h$
  10.  $Cc^a \times c^h c^a$  (Note that the 1:2:1 ratio among the progeny in this special case does not reflect incomplete dominance or codominance, but instead results from the fact that the cross involved three alleles with a particular dominance relationship.)
- c. Two answers are possible depending on the genotype of the chinchilla parents in cross 9. Alternative 1:  $Cc^h$  (from cross 9)  $\times Cc^a$  (from cross 10)  $\rightarrow 1/4 CC$  (chinchilla) :  $1/4 Cc^a$  (chinchilla) :  $1/4 Cc^h$  (chinchilla) :  $1/4 c^h c^a$  (himalaya) = **3/4 chinchilla : 1/4 himalaya**. Alternative 2:  $Cc^a$  (cross 9)  $\times Cc^a$  (cross 10)  $\rightarrow 3/4 C-$  chinchilla : **1/4  $c^a c^a$  albino**.

14. Designate the gene  $p$  (for pattern). Seven alleles exist,  $p^1$ - $p^7$ , with  $p^7$  being the allele that specifies absence of pattern and  $p^1 > p^2 > p^3 > p^4 > p^5 > p^6 > p^7$ .

- a. **Seven different patterns** are possible. These are associated with the following genotypes:  $p^1-$ ,  $p^2 p^a$  (where  $p^a = p^2, p^3, p^4 \dots p^7$ ),  $p^3 p^b$  (where  $p^b = p^3, p^4, p^5 \dots p^7$ ),

$p^4p^c$  (where  $p^c = p^4, p^5, p^6$ , and  $p^7$ ),  $p^5p^d$  (where  $p^d = p^5, p^6$ , and  $p^7$ ),  $p^6p^e$  (where  $p^e = p^6$  and  $p^7$ ), and  $p^7p^7$ .

- b. The phenotype dictated by the allele  $p^1$  has the greatest number of genotypes associated with it = 7 ( $p^1p^1, p^1p^2, p^1p^3$ , etc.). **The absence of pattern** is caused by just one genotype,  $p^7p^7$ .
  - c. This finding suggests that **the allele determining absence of pattern ( $p^7$ ) is very common** in these clover plants so that the  $p^7p^7$  genotype is the most frequent in the population. The other alleles are present, but are much less common in this population.
15. a. This ratio is approximately **2/3 Curly : 1/3 normal**.
- b. The expected result for this cross is:  $Cy^+Cy \times Cy^+Cy \rightarrow 1/4 CyCy (?) : 1/2 Cy^+Cy$  (Curly) :  $1/4 Cy^+Cy^+$  (normal). If the  **$Cy Cy$  genotype is lethal**, then the expected ratio will match the observed data.
  - c. The cross is  $Cy^+Cy \times Cy^+Cy^+ \rightarrow 1/2 Cy^+Cy : 1/2 Cy^+Cy^+$ , so there would be approximately **90 Curly-winged and 90 normal-winged flies**. (Note that because  $Cy Cy$  adults are not found, all curly-winged flies must be  $Cy^+Cy$  heterozygotes.)
16. Two keys to this problem exist: (1) The sperm in pollen grains and ovules are gametes that have only one copy of the  $S$  incompatibility gene, while the stigma (the part of the female plant on which the pollen grains land has two copies of this gene. (2) Sperm with a particular  $S$  gene allele cannot fertilize any ovules in a plant whose stigma has the same  $S$  allele, because the pollen will not grow a tube allowing it to fertilize an ovule.
- a. In the cross  $S^1S^2 \times S^1S^2$  all the pollen grains (whether they are  $S^1$  or  $S^2$ ) will land on the stigmas of plants that have the same alleles, and therefore **no progeny would be produced at all**.
  - b. The pollen grains would be  $S^1$  or  $S^2$ . The  $S^2$  pollen could not fertilize the female plant, but the  $S^1$  pollen could. **The progeny would thus be  $S^1S^2$  and  $S^1S^3$  (in a 1:1 ratio)**.
  - c. All pollen grains would be able to fertilize all ovules, because the pollen grains do not share any alleles with the female parent. As a result, **four types of progeny would be produced in equal numbers:  $S^1S^3, S^1S^4, S^2S^3$ , and  $S^2S^4$** .
  - d. **This mechanism would prevent plant self-fertilization because any pollen grain produced by any plant would land on a stigma sharing the same allele.** For example, if an  $S^1$  pollen grain produced by an  $S^1S^2$  plant lands on a stigma from the same plant, the stigma would have the same allele and no pollen tube would be able to grow to allow fertilization. The same would be true for a  $S^2$  pollen grain from the same plant. (Of interest, tomato plants in the wild cannot self-fertilize because of this incompatibility mechanism; they proliferate only through cross-fertilization. However, many domesticated cultivars of tomatoes can self-fertilize

because they were selected for varieties that have mutations causing the failure of the incompatibility mechanism.)

- e. **Plants with functioning incompatibility systems must be heterozygotes because a pollen grain cannot fertilize a female plant sharing the same allele of the *S* incompatibility gene.** For example, an *S'* pollen grain cannot fertilize successfully any female plant that also has an *S'* allele. No way thus exists to create *S'S'* homozygous progeny.
  - f. **Peas cannot be governed by this mechanism because you already saw in Chapter 2 that Gregor Mendel routinely self-fertilized his peas in the  $F_1$  generation to produce the  $F_2$  generation.**
  - g. **The larger the number of different alleles of the *S* gene that are present in the population, the more likely it is that any given pollen grain of any genotype would land on the stigma of a flower that did not share the same allele, and the less likely that the pollen will interact unproductively with flowers that share the same allele.** Within the population, the proportion of matings that could produce progeny would increase with a greater variety in *S* gene alleles; this would clearly increase the fertility (and thus the average evolutionary fitness) of the population.
17. a. The 2/3 montezuma : 1/3 wild type phenotypic ratio, and the statement that montezumas are never true-breeding, together suggest that there is a **recessive lethal allele** of this gene. When a recessive lethal exists, crossing two heterozygotes results in a 1:2:1 genotypic ratio, but one of the 1/4 classes of homozygotes do not survive. **The result is the 2:1 phenotypic ratio as seen in this cross. Both the montezuma parents were therefore heterozygous, *Mm*.** The *M* allele must confer the montezuma coloring in a dominant fashion, but homozygosity for *M* is lethal.
- b. Designate the alleles: *M* = montezuma, *m* = greenish; *F* = normal fin, *f* = ruffled. Diagram the cross: ***Mm FF* × *mm ff* →** expected monohybrid ratio for the *M* gene alone: 1/2 *Mm* (montezuma) : 1/2 *mm* (wild type); expected monohybrid ratio for the *F* gene alone: all *Ff*. The expected dihybrid ratio = **1/2 *Mm Ff* (montezuma, normal fin) : 1/2 *mm Ff* (greenish, normal fin).**
- c. ***Mm Ff* × *Mm Ff* →** expected monohybrid ratio for the *M* gene alone: 2/3 montezuma (*Mm*) : 1/3 greenish (*mm*); expected monohybrid ratio for the *F* gene alone: 3/4 normal fin (*F*−) : 1/4 ruffled (*ff*). The expectations when considering both genes together is: **6/12 montezuma, normal fin : 2/12 montezuma, ruffled fin : 3/12 greenish, normal fin : 1/12 greenish, ruffled fin.**
18. The answer for the phenotype of viability is straightforward. **The mutant allele is clearly recessive to the wild-type allele for the phenotype of viability:** Heterozygotes are viable, and thus have the same phenotype as homozygotes for the wild-type allele (viability) and *not* the phenotype of homozygotes for the recessive allele (lethality).

For the phenotype of having fingerprints, the answer is more complicated. The reason is that aside from the fact that they become inviable at some point during development, we don't know the fingerprint phenotype of the *SMARCAD1* mutant homozygotes. Remember that the definition of dominance is that the heterozygote has the phenotype associated with the homozygote for the dominant allele, but this definition is problematic if we do not know the homozygote's phenotype.

This problem suggests two interesting possibilities for the function of the *SMARCAD1* gene, and each scenario suggests a different kind of dominance relationship. One possibility is that the gene is pleiotropic – that is, *SMARCAD1* has distinct roles in early development and in fingerprint development later. Suppose that some method exists to supply *SMARCAD1* function early in development and then to remove the function later. Homozygotes for the mutant allele could then be born, and we could then see if they have fingerprints. **If homozygotes for nonfunctional *SMARCAD1* alleles would lack fingerprints, then for the phenotype of having no fingerprints, the nonfunctional *SMARCAD1* allele would be dominant to the normal allele.** (This experiment of supplying gene function early in development and then removing it later is not possible in humans, but you will learn in later chapters that such genetic manipulations can be done with organisms like mice and fruit flies.)

**A second possibility is that *SMARCAD1* is required for skin development generally,** and not only the formation of fingerprints. In this case, homozygosity for nonfunctional *SMARCAD1* alleles is lethal because the skin cannot develop properly without SMARCAD1 protein. When the level of the SMARCAD1 protein is half of normal (in heterozygotes), the skin can develop more-or-less normally except that fingerprints cannot form. **If this is the case, then the *SMARCAD1* nonfunctional mutant alleles and normal *SMARCAD1* alleles display incomplete dominance for the phenotype of skin development:** no fingerprints is a phenotype between normal skin and skin formed so improperly that the person cannot be born.

19. a. The wild-type allele and the mutant allele display **incomplete dominance** because the heterozygotes have a phenotype (blue lips and phenotypes) between that of the two homozygotes (normal skin or blue skin).
- b. Polly Ritchie is a heterozygote, but neither of her parents is recorded as a heterozygote, although both parents have heterozygous ancestors. Thus, **either James Ritchie (and if so, his father Martin Ritchie also) or Hannah Fugate** must have been a heterozygote. Similarly, **one of Manuel Fugate's parents (Zachariah Fugate or Polly Campbell), one of Richard Smith's parents (William Smith or Betty Ritchie), and one of Eleanor Fugate's parents (William Fugate or Juda Campbell)** must have been heterozygous.
- c. Mary is likely a Ritchie or a Smith because **she's a carrier of a rare methemoglobinemia allele** known to be present in those families.
- d. **Richard Smith and Martin Fugate's wife (Mary?)** are the earliest people in the pedigree recorded as having a blue phenotype (heterozygotes). The two of them introduced the mutant allele(s) into the family. There is no indication in the pedigree diagram that Richard Smith and Mary were related. If they are not related, then two

different mutant *NADH diaphorase* alleles were introduced into this complex pedigree.

## Section 3.2

**20.** The cross is: walnut  $\times$  single  $\rightarrow$  F<sub>1</sub> walnut  $\times$  F<sub>1</sub> walnut  $\rightarrow$  93 walnut : 29 rose : 32 pea : 11 single

- a.** How many genes are involved? The four F<sub>2</sub> phenotypes means that 2 genes are involved, *A* and *B*. Both genes affect the same structure, the comb. The F<sub>2</sub> phenotypic dihybrid ratio among the progeny is close to 9:3:3:1, so there is no epistasis. Because walnut is the most abundant F<sub>2</sub> phenotype, it must be the phenotype due to the *A-* *B-* genotype. Single combs are the least frequent class, and are thus *aa bb*. Now assign genotypes to the cross. If the walnut F<sub>2</sub> are *A-* *B-*, then the original walnut parent must have been *AA BB*:

***AA BB*  $\times$  *aa bb*  $\rightarrow$  *Aa Bb* (walnut)  $\rightarrow$  9/16 *A-* *B-* (walnut) : 3/16 *A-* *bb* (rose) : 3/16 *aa B-* (pea) : 1/16 *aa bb* (single).**

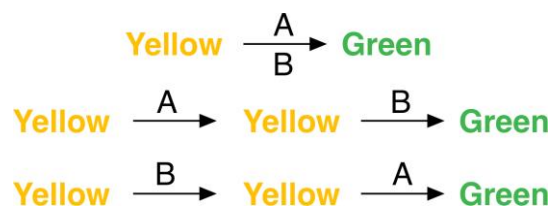
- b.** Diagram the cross, recalling that the problem states the parents are homozygous:  
***AA bb* (rose)  $\times$  *aa BB* (pea)  $\rightarrow$  *Aa Bb* (walnut)  $\rightarrow$  9/16 *A-* *B-* (walnut) : 3/16 *A-* *bb* (rose) : 3/16 *aa B-* (pea) : 1/16 *aa bb* (single).** Notice that this F<sub>2</sub> is in identical proportions as the F<sub>2</sub> generation in part (a).
- c.** Diagram the cross: *A-* *B-* (walnut)  $\times$  *aa B-* (pea)  $\rightarrow$  12 *A-* *B-* (walnut) : 11 *aa B-* (pea) : 3 *A-* *bb* (rose) : 4 *aa bb* (single). Because pea and single progeny exist, you know that the walnut parent must be *Aa*. The 1 *A-* : 1 *aa* monohybrid ratio in the progeny also tells you the walnut parent must have been *Aa*. Because some of the progeny are single, you know that both parents must be *Bb*. In this case, the monohybrid ratio for the *B* gene is 3 *B-* : 1 *bb*, so both parents were *Bb*. The original cross must have been ***Aa Bb*  $\times$  *aa Bb***. You can verify that this cross would yield the observed ratio of progeny by multiplying the probabilities expected for each gene alone. For example, you anticipate that 1/2 the progeny would be *Aa* and 3/4 of the progeny would be *Bb*, so  $1/2 \times 3/4 = 3/8$  of the progeny should be walnut; this is close to the 12 walnut chickens seen among 30 total progeny.
- d.** Diagram the cross: *A-* *B-* (walnut)  $\times$  *A-* *bb* (rose)  $\rightarrow$  all *A-* *B-* (walnut). The progeny are all walnut, so **the walnut parent must be *BB***. No pea progeny are seen, so **both parents cannot be *Aa*, so one of the two parents must be *AA***. This could be either the walnut or the rose parent or both.

**21.** black  $\times$  chestnut  $\rightarrow$  F<sub>1</sub> bay  $\rightarrow$  F<sub>2</sub> black : bay : chestnut : liver

Four phenotypes in the F<sub>2</sub> generation means two genes determine coat color. The F<sub>1</sub> bay animals produce four phenotypic classes, so they must be doubly heterozygous, *Aa Bb*. Crossing a liver-colored horse to either of the original parents resulted in the parent's phenotype. The liver horse's alleles do not affect the phenotype, suggesting the recessive genotype *aa bb*. Though it is probable that the original black mare was *AA bb* and the chestnut stallion was *aa BB*, each of these

animals produced only 3 progeny, so it cannot be concluded definitively that these animals were homozygous for the dominant allele they carry. Thus, the black mare was  $A- bb$ , the chestnut stallion was  $aa B-$ , and the  $F_1$  bay animals are  $Aa Bb$ . The  $F_2$  horses were: bay ( $A- B-$ ), liver ( $aa bb$ ), chestnut ( $aa B-$ ), and black ( $A- bb$ ).

22. a. Because unaffected individuals had affected children, the **trait is recessive**. From affected individual II-1, you know the mutant allele is present in this generation. The trait was passed on through II-2 who was a carrier. All children of affected individuals III-2  $\times$  III-3 are affected, as predicted for a recessive trait. However, generation V seems inconsistent with recessive inheritance of a single gene. This result is consistent with two different genes involved in hearing with a defect in either gene leading to deafness: **The trait is *heterogeneous*, meaning that two family lines shown contain mutations in two separate genes. Furthermore, the mutant alleles of both genes determining deafness are recessive.**
- b. Individuals in generation V are doubly heterozygous ( $Aa Bb$ ), having inherited a dominant and recessive allele of each gene from their parents ( $aa BB \times AA bb$ ). The people in generation V are unaffected because **the product of the dominant allele of each gene is sufficient for normal function**. This is an example of *complementation*: The gamete from each parent provided the dominant allele that the gamete from the other parent lacked.
23. green  $\times$  yellow  $\rightarrow F_1$  green  $\rightarrow F_2$  9 green : 7 yellow
- a. The 9:7 ratio is a variant of the 9:3:3:1 phenotypic dihybrid ratio, suggesting that **two genes are controlling color**. The genotypes are:  
 $AA BB$  (green)  $\times$   $aa bb$  (yellow)  $\rightarrow F_1 Aa Bb$  (green)  $\rightarrow F_2$  9/16  $A- B-$  (green) : 3/16  $A- bb$  (yellow) : 3/16  $aa B-$  (yellow) : 1/16  $aa bb$  (yellow).
- b.  $Aa Bb \times aa bb \rightarrow 1/4 Aa Bb$  (green) :  $1/4 aa Bb$  (yellow) :  $1/4 Aa bb$  (yellow) :  $1/4 aa bb$  (yellow) = 1/4 green : 3/4 yellow.
- c. This is an example of reciprocal recessive epistasis. That is,  $aa$  is epistatic to  $B$ , while  $bb$  is epistatic to  $A$ .
- d. One simple model is that the proteins made by the  $A$  and  $B$  genes work together or in succession to generate a green pigment from a yellow precursor in any of the following three ways:



Note that the genetic interactions *do not* distinguish between these three pathways. Nor do the results guarantee that any one of these pathways is correct—many more-complicated models are possible.



- e. Zucchini that are  $AA\ bb$  or  $aa\ BB$  are both pure-breeding yellow, and crossing them results in  $Aa\ Bb$  progeny that are green.
- f. **Complementation** occurred in part (e). Note that this interaction can be described as complementation only if the recessive alleles are nonfunctional (or have lost some function) and the dominant alleles are functional. In this case, the green phenotype is the wild type.
24. a. white  $\times$  white  $\rightarrow$  F<sub>1</sub> white  $\rightarrow$  F<sub>2</sub> 126 white : 33 purple

At first glance this cross seems to involve only one gene, as true-breeding white parents give white F<sub>1</sub>s. However, if this were true, then the F<sub>2</sub> MUST be totally white as well. The purple F<sub>2</sub> plants show that this cross is NOT controlled by 1 gene.

These results may instead be due to 2 genes. To determine if this is the case, it makes sense to ask: Does a ratio of 126 : 33 represent a variant of the 9:3:3:1 dihybrid ratio? Usually when you are given raw numbers of individuals for the classes, you divide through by the smallest number, yielding in this case 3.8 white : 1 purple. This is neither a recognizable monohybrid nor dihybrid ratio. Dividing through by the smallest class is NOT the correct way to convert raw numbers to a ratio, if it is possible that the smallest class in the ratio is not 1.

The correct method for this problem is as follows. Assuming that the F<sub>1</sub> in this case are dihybrids, 16 different equally likely fertilization events must have produced the F<sub>2</sub> progeny (16 boxes in the 4  $\times$  4 Punnett square), even though the phenotypes may not be distributed in the usual 9/16 : 3/16 : 3/16 : 1/16 ratio. If the 159 F<sub>2</sub> progeny are divided equally into 16 fertilization types, then 159/16 = ~10 F<sub>2</sub> plants exist for each fertilization type. The 126 white F<sub>2</sub>s therefore represent 126/10 = ~13 of these fertilizations. Likewise, the 33 purple plants represent 33/10 = ~3 fertilization types. **The F<sub>2</sub> phenotypic ratio is thus approximately 13 white : 3 purple. The data fit the hypothesis that two genes control color, and that the F<sub>1</sub> are dihybrids.**

You can now assign genotypes to the parents in the cross. Because the parents are homozygous (true-breeding) and 2 genes control the phenotypes, you can set up the genotypes of the parents in two different ways so that the F<sub>1</sub> dihybrids are heterozygous for dominant and recessive alleles of each gene. One option is:  **$AA\ BB$  (white)  $\times$   $aa\ bb$  (white)  $\rightarrow$   $Aa\ Bb$  (white, same as  $AA\ BB$  parent)  $\rightarrow$  9  $A-\ B-$  (white) : 3  $A- bb$  (unknown phenotype) : 3  $aa B-$  (unknown phenotype) : 1  $aa\ bb$  (white, same as  $aa\ bb$  parent).** If you assume that  $A- bb$  is white and  $aa B-$  is purple (or *vice versa*), then this is a match for the observed data presented in the cross above [(9 + 3 + 1) = 13 white : 3 purple].

Alternatively, you could try to diagram the cross as  $AA\ bb$  (white)  $\times$   $aa\ BB$  (white)  $\rightarrow$   $Aa\ Bb$  (whose phenotype is unknown as this is NOT a genotype seen in the parents)  $\rightarrow$  9  $A- B-$  (same unknown phenotype as in the F<sub>1</sub>) : 3  $A- bb$  (white like the  $AA\ bb$  parent) : 3  $aa B-$  (white like the  $aa\ BB$  parent) : 1  $aa\ bb$  (unknown phenotype). Such a cross cannot give an F<sub>2</sub> phenotypic ratio of 13 white : 3 purple. The only F<sub>2</sub> classes that could be purple are  $A- B-$ , but this is impossible because (i) this class (9/16) is much larger than the number of purple plants observed



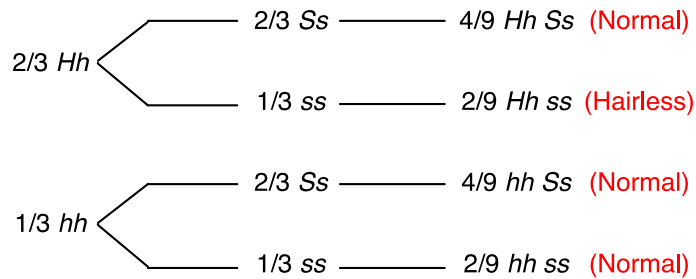
( $\sim 3/16$ ); and (ii) the  $F_1$  plants must then have been purple (which was not the case). Therefore, the first set of possible genotypes (written in bold above) is the best fit for the observed data.

Assume that  $A- bb$  plants are white, and  $aa B-$  plants are purple. Our model above states that to be purple, a plant must have a  $B$  allele and no  $A$  allele. Thus, we can say that  $A$  is epistatic to  $B$ . This phenomenon is a form of *dominant epistasis*. [Table 3-2 (reproduced on p. 3-3 of the Solutions Manual) calls this *dominant epistasis II*, although the Roman numeral is arbitrary and only included to facilitate discussion.]

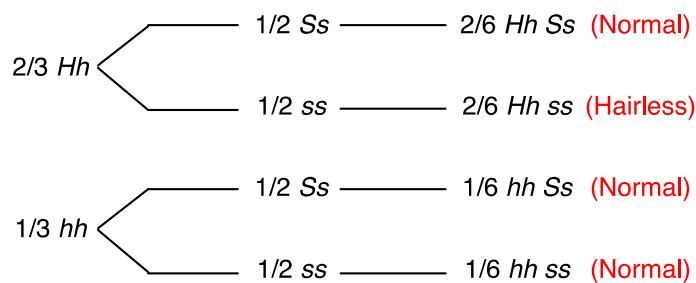
- b. white  $F_2 \times$  white  $F_2$  (self-fertilization)  $\rightarrow$  3/4 white : 1/4 purple. Assume again that the  $aa B-$  class is purple in part (a) above. A 3:1 monohybrid ratio means the parents are both heterozygous for one gene with purple due to the recessive allele. The second gene is not affecting the ratio, so both parents must be homozygous for the same allele of that gene. Thus the self-fertilization must be:  **$Aa BB$**  (white)  $\times$   **$Aa BB$**  (white)  $\rightarrow$  3/4  $A- BB$  (white) : 1/4  $aa BB$  (purple).
  - c. purple  $F_2 \times$  purple  $F_2$  (self-fertilization)  $\rightarrow$  3 purple : 1 white. Again, the selfed parent must be heterozygous for one gene and homozygous for the other gene. Because purple is  $aa B-$ , the genotypes of the purple  $F_2$  plants must be  **$aa Bb$** .
  - d. white  $F_2 \times$  white  $F_2$  (a cross, not a self-fertilization)  $\rightarrow$  1/2 purple : 1/2 white. The 1:1 monohybrid ratio means a testcross was done for one of the genes. The second gene is not altering the ratio in the progeny, so the parents must be homozygous for that gene. If purple is  $aa B-$ , then the genotypes of the parents must be  **$aa bb$**  (white)  $\times$   **$Aa BB$**  (white)  $\rightarrow$  1/2  $Aa Bb$  (white) : 1/2  $aa Bb$  (purple).
25. a. The  $F_2$  would be 9  $A- B-$  (purple) : 3  $A- bb$  (blue) : 3  $aa B-$  (white) : 1  $aa bb$  (white). The phenotypic ratio would therefore be **9 purple : 4 white : 3 blue**.
- b. From Table 3.2 (reproduced on p. 3-3 of this Answer Book), the 9:4:3 ratio indicates recessive epistasis. In this case,  **$aa$  is epistatic to  $B$  and  $bb$** . The reason is that  $aa$  flowers are white regardless of the gene  $B$  genotype, even though gene  $B$  contributes otherwise to the same phenotype.
26. Dominance relationships are between alleles of the same gene. **Only one gene is involved when considering dominance relationships. Epistasis involves two genes**. The alleles of one gene affect the phenotypic expression of the second gene.
27. a. The cross is between two normal flies that carry  $H$  and  $S$ . These individuals cannot be homozygous for  $H$  or for  $S$ , because we are told that both are lethal in homozygotes. Thus, the mating described is a dihybrid cross:  $Hb Ss \times Hb Ss$ . The genotypic classes among the progeny zygotes should be 9  $H- S-$ , 3  $H- ss$ , 3  $hh S-$ , and 1  $hh ss$ . However, the results are complicated by the fact that all zygotes that are  $HH$  or  $SS$  or both will die before they hatch into adult flies.

One approach is to do this problem as the branched-line diagram shown in the following figure, in which the progeny should be 2/3  $Hb$  and 1/3  $bb$  (considering the  $H$  gene alone) and 2/3  $Ss$  and 1/3  $ss$  (considering the  $S$  gene alone). As can be

seen from the diagram,  $7/9$  of the adult progeny will be normal, and  $2/9$  will be hairless.



- b. As just seen in the diagram, the hairless progeny of the cross in part (a) are  $Hh ss$ , and these are mated with parental flies that are  $Hh Ss$ . You could again portray the results of this cross as a branched-line diagram. For the  $H$  gene, again  $2/3$  of the viable adult progeny will be  $Hh$  and  $1/3$  will be  $hh$ . The cross involving the  $S$  gene is a testcross, and all the progeny will be viable, so  $1/2$  the progeny will be  $Ss$  and  $1/2$  will be  $ss$ . As seen in the diagram that follows,  $2/6 = 1/3$  of the progeny will be hairless and the remaining  $2/3$  will be normal.



28.  $I^A I^B Ss \times I^A I^A Ss \rightarrow$  expected ratio for the  $I$  gene of  $1/2 I^A I^A : 1/2 I^A I^B$ ; expected ratio for the  $S$  gene considered alone of  $3/4 S- : 1/4 ss$ . Use the product rule to generate the phenotypic ratio for both genes considered together and then assign phenotypes, remembering that all individuals with the  $ss$  genotype look like type O. The phenotypic ratio for both genes is:  $3/8 I^A I^A S- : 3/8 I^A I^B S- : 1/8 I^A I^A ss : 1/8 I^A I^B ss = 3/8 A : 3/8 AB : 1/8 O : 1/8 O = 3/8 \text{ Type A} : 3/8 \text{ Type AB} : 2/8 \text{ Type O}$ .
29. You would first self the mutant plant. If the mutant traits are dominant and the plant is heterozygous for the genes involved, it is possible that you might see some progeny displaying different combinations of the recessive wild-type traits. Such a result would suggest that different genes are responsible for the different traits.

If the mutant plant is pure-breeding, you should cross it (or its self-fertilized descendants) with a pure-breeding wild-type strain, and then self-fertilize the  $F_1$  progeny. If several genes were involved, the  $F_2$  would have several different combinations of the petal color, markings, and stem position traits. If all 3 traits were determined by an allele of one gene, the three non-wild-type or three wild-type traits would always be inherited together.

30. If ABO blood type were controlled only by the  $I$  gene, then her husband would have a reason to be concerned. In that case, he is  $ii$ , and she is either  $I^B i$  or  $I^B I^B$ ; their child could not have blood type A because neither of them has an  $I^A$  allele. However, we know that the  $H$  gene is also involved in ABO blood type, and that  $hh$  is epistatic to  $I$  such that all  $hh$  genotypes are blood type O. This means that **the husband's O blood type could be due to an  $I^A-hh$  genotype, and his wife's genotype could be  $I^B i H-$ ; they could easily have had an  $I^A i Hh$  (type A) child.**
31. a. blood types: I-1 AB; I-2 A; I-3 B; I-4 AB; II-1 O; II-2 O; II-3 AB; III-1 A; III-2 O.
- b. genotypes: I-1  $Hh I^A I^B$ ; I-2  $Hh I^A i$  (or  $I^A I^A$ ); I-3  $H- I^B I^B$  (or  $I^B i$ ); I-4  $H- I^A I^B$ ; II-1  $H- ii$ ; II-2  $hh I^A I^A$  (or  $I^A i$  or  $I^A I^B$ ); II-3  $Hh I^A I^B$ ; III-1  $Hh I^A i$ ; III-2  $hh I^A I^A$  (or  $I^A I^B$  or  $I^A i$  or  $I^B i$  or  $I^B I^B$ )

At first glance, you find inconsistencies between expectations and what could be inherited from a parent. For example, I-1 (AB)  $\times$  I-2 (A) could not have an O child (II-2). The epistatic  $h$  allele (which causes the Bombay phenotype) could explain these inconsistencies. If II-2 has an O phenotype because she is  $hh$ , her parents must both have been  $Hh$ . The Bombay phenotype would also explain the second seeming inconsistency of two O individuals (II-1 and II-2) having an A child. II-2 could have received an  $I^A$  allele from one of her parents and passed this on to III-1 together with one  $h$  allele. Parent II-1 would have to contribute the  $H$  allele so that the  $I^A$  allele would be expressed; the presence of  $H$  means that II-1 must also be  $ii$  to be type O. A third inconsistency is that individuals II-2 and II-3 could not have an  $ii$  child since II-3 has the  $I^A I^B$  genotype, but III-2 has the O phenotype. This could also be explained if II-3 is  $Hh$  and III-2 is  $hh$ .

32. a. Diagram one of the crosses:

white-1  $\times$  white-2  $\rightarrow$  red  $F_1 \rightarrow$  9 red : 7 white

Even though only 2 phenotypes are present in the  $F_2$ , color is not controlled by one gene. Instead, the 9:7 ratio is a variation of 9:3:3:1, so 2 genes control the colors in this cross. Individuals must have at least one dominant allele of each gene to get the red color; this is an example of *reciprocal recessive epistasis* (refer to [Table 3.2](#) reproduced on p. 3-3 of this Answer Book). Thus, the genotypes of the two pure-breeding white parents in this cross are  $aa BB \times AA bb$ . The same conclusions hold for the other 2 crosses.

If white-1 is  $aa BB$  and white-2 is  $AA bb$ , then white-3 must be  $AA BB \alpha$ . The reason is that if white-3 had the same genotype as white-1 or white-2, then one of the three crosses would have produced an all-white  $F_1$ . Because none of the crosses had an all-white  $F_1$ , we can conclude that **three genes are involved**.

- b. White-1 is  $aa BB CC$ ; white-2 is  $AA bb CC$  and white-3 is  $AA BB cc$ .
- c.  $aa BB CC$  (white-1)  $\times$   $AA bb CC$  (white-2)  $\rightarrow$   $Aa Bb CC$  (red)  $\rightarrow$  9/16  $A- B- CC$  (red) : 3/16  $A- bb CC$  (white) : 3/16  $aa B- CC$  (white) : 1/16  $aa bb CC$

(white). Red color requires a dominant, functional allele of each of the three genes ( $A- B- C-$ ).

33. Diagram the cross. Figure out an expected monohybrid ratio for each gene separately, then apply the product rule to generate the expected dihybrid ratio.

$A^yA Cc \times A^yA cc \rightarrow$  monohybrid ratio for the  $A$  gene alone:  $1/4 A^yA^y$  (dead) :  $1/2 A^yA$  (yellow) :  $1/4 AA$  (agouti) =  $2/3 A^yA$  (yellow) :  $1/3 AA$  (agouti);  
monohybrid ratio for the  $C$  gene:  $1/2 Cc$  (non-albino) :  $1/2 cc$  (albino).

Overall there will be  $2/6 A^yA Cc$  (yellow) :  $2/6 A^yA cc$  (albino) :  $1/6 AA Cc$  (agouti) :  $1/6 AA cc$  (albino) =  **$2/6 A^yA Cc$  (yellow) :  $3/6 -- cc$  (albino) :  $1/6 AA Cc$  (agouti)**. Note that the  $A^yA cc$  animals must be albino because the albino parent had exactly the same genotype; this indicates that  $cc$  is epistatic to all alleles of gene  $A$ . Although you were not explicitly told that the  $AA cc$  animals are also albino, this makes sense because  $cc$  albino color must be epistatic to alleles of all genes that confer color given that no pigments are produced.

34. a. No, a single gene cannot account for this result. While the 1:1 ratio seems like a testcross, **the fact that the phenotype of one class of offspring (linear) is not the same as either of the parents argues against this being a testcross.**
- b. The appearance of four phenotypes suggests that **two genes** control the phenotypes.
- c. The 3:1 ratio suggests that **two alleles of one gene** determine the difference between the wild-type and scattered patterns.
- d. The true-breeding wild-type fish are homozygous by definition, and the scattered fish have to be homozygous recessive according to the ratio seen in part (c), so the cross is:  **$bb$  (scattered)  $\times BB$  (wild type)  $\rightarrow F_1 Bb$  (wild type)  $\rightarrow F_2 3/4 B-$  (wild type) :  $1/4 bb$  (scattered).**
- e. The inability to obtain a true-breeding nude stock suggests that the nude fish are heterozygous ( $Aa$ ) and that the  **$AA$  genotype dies**. Thus  **$Aa$  (nude)  $\times Aa$  (nude)  $\rightarrow 2/3 Aa$  (nude) :  $1/3 aa$  (scattered).**
- f. Going back to the linear cross from part (b), the fact that four phenotypes appeared led us to propose two genes were involved. The 6:3:2:1 ratio looks like an altered 9:3:3:1 ratio in which some genotypes may be missing, as predicted from the result in part (e) that  $AA$  animals do not survive. The 9:3:3:1 ratio results from crossing double heterozygotes, so **the linear parents are doubly heterozygous  $Aa Bb$ . The lethal phenotype associated with the  $AA$  genotype produces the 6:3:2:1 ratio. The phenotypes and corresponding genotypes of the progeny of the linear  $\times$  linear cross are: 6 linear,  $Aa B-$  : 3 wild type,  $aa B-$  : 2 nude,  $Aa bb$  : 1 scattered,  $aa bb$ .** Note that the  $AA BB$ ,  $AA Bb$ ,  $AA Bb$ , and  $AA bb$  genotypes are missing due to lethality.
35. This problem shows that gene interactions producing variations of the 9:3:3:1 ratio in addition to those shown in Table 3.2 (reproduced on p. 3-3 of the Solutions Manual) are possible.

- a. Using the information provided, one of the pure-breeding white strains must be homozygous for recessive alleles of gene *A* and the other pure-breeding white strain must be homozygous for recessive alleles of gene *B*. That is, the cross was ***AA bb* (white) × *aa BB* (white) → F<sub>1</sub> *Aa Bb* (all blue)**.
- b. In the F<sub>2</sub> generation produced by selfing of the F<sub>1</sub> plants, you would find a genotypic ratio of 9 *A- B-* : 3 *A- bb* : 3 *aa B-* : 1 *aa bb*. The *A- B-* plants would have blue flowers because colorless precursor 1 would be converted into blue pigment. (Colorless precursor 2 would not produce blue pigment in these flowers because the second pathway is suppressed by the proteins specified by the dominant alleles of the two genes. However, the color would still be blue because the pigment produced by the first pathway is sufficient for the blue phenotype.) The *A- bb* plants would be white because the first pathway could not produce blue pigment in the absence of the protein specified by *B*, while the second pathway would be shut off by the protein specified by *A*. The *aa B-* plants would be white because the first pathway could not produce blue pigment in the absence of the protein specified by *A*, while the second pathway would be shut off by the protein specified by *B*. Interestingly, the *aa bb* plants would be blue because even though the first pathway would not function, the second would as it is not suppressed. **You would thus expect in the F<sub>2</sub> generation a ratio of 10 blue (9 *A- B-* + 1 *aa bb*) : 6 white (3 *A- bb* + 3 *aa B-*).**
36. The answers are presented in the table below. Different colors in the table represent different phenotypes; these colors are chosen arbitrarily and do not signify anything. The numbers in parentheses indicate the compounds that are present to produce the colors.

Part	9 <i>A- B-</i>	3 <i>A- bb</i>	3 <i>aa B-</i>	1 <i>aa bb</i>	Ratio
a	(2 + 4)	(2 + 3)	(1 + 4)	(1 + 3)	9:3:3:1
b	(2)	(2)	(2)	(1)	15:1
c	(3)	(2)	(1)	(1)	9:3:4
d	(2)	(1)	(1)	(1)	9:7
e	(2 + 3)	(2)	(3)	(1)	9:3:3:1
f	(2 + 4) = (2)	(2 + 3) = (2)	(1 + 4)	(1 + 3)	12:3:1
g	(3)	(2) = (1)	(1) = (2)	(1) = (2)	9:7
h	(2)	(1)	(2)	(2)	13:3

37. A particular phenotypic ratio does not allow you to infer the operation of a specific biochemical mechanism because as can be seen from the answers to Problem 36, different biochemical mechanisms can produce the same ratio of phenotypes [for example, the pathways in parts (d) and (g) are different yet both yield 9:7 ratios]. The ratio seen in a cross may nonetheless provide information about types of biochemical pathways you could exclude from consideration because those pathways could not produce the observed ratio.

In contrast, if you know the biochemical mechanism behind a gene interaction and you also know the dominance relationships of the alleles, you can then

trace out the consequences of each genotypic class and thus **you can predict the ratios of phenotypes you would see among the F<sub>2</sub> progeny.**

## Section 3.3

- 38. a.** The yellow parent must have an  $A^Y$  allele, but we don't know the second allele of the  $A$  gene ( $A^Y-$ ). We don't know at the outset what alleles this yellow mouse has at the  $B$  gene, so we'll leave these alleles for the time being as  $??$ . Because this mouse does show color we know it is not  $cc$  (albino), so it must have at least one  $C$  allele ( $C-$ ). The brown agouti parent has at least one  $A$  allele ( $A-$ ); it must be  $bb$  at the  $B$  gene; and as there is color it must also be  $C-$ . The mating between these two can thus be represented as  $A^Y- ?? C- \times A- bb C-$ .

Now consider the progeny. Because one pup was albino ( $cc$ ), the parents must both be  $Cc$ . A brown pup ( $bb$ ) indicates that both parents had to be able to contribute a  $b$  allele, so we now know the first mouse (the yellow parent) must have had at least one  $b$  allele. The fact that this brown pup was non-agouti means both parents carried an  $a$  allele. The black agouti progeny tells us that the first mouse must have also had a  $B$  allele. This latter fact also clarifies that  $A^Y$  is epistatic to  $B$  because this parent was yellow rather than black. The complete genotypes of the mice are therefore:  $A^Ya Bb Cc \times Aa bb Cc$ .

- b.** Think about each gene individually, then consider the effect of the other genes in combination with that phenotype.  $C-$  leads to a phenotype with color;  $cc$  gives albino (which is epistatic to all colors determined by the other genes because no pigments are produced). The possible genotypes of the progeny of this cross for the  $A$  gene are  $A^YA$ ,  $A^Ya$ ,  $Aa$  and  $aa$ , giving yellow, yellow, agouti and non-agouti phenotypes, respectively. Since yellow ( $A^Y$ ) is epistatic to  $B$ , non-albino mice with  $A^Y$  will be yellow regardless of the genotype of the  $B$  gene.  $Aa$  is agouti; with the  $aa$  genotype there is no yellow on the hair (non-agouti). The type of coloration depends on the  $B$  gene. For  $B$  the offspring could be  $Bb$  (black) or  $bb$  (brown). In total, **six different coat color phenotypes are possible: albino ( $-- -- cc$ ), yellow ( $A^Y(A \text{ or } a) -- C-$ ), brown agouti ( $A- bb C-$ ), black agouti ( $A- B- C-$ ), brown ( $aa bb C-$ ), and black ( $aa B- C-$ ).** [Note: Although  $A^Y$  (yellow color) is in fact epistatic to  $B$  (black) or  $bb$  (brown) colors governed by the  $B$  gene, you were not explicitly told this. Thus, based on the information provided, you might have included an additional color phenotype if you assumed that  $A^Y(A \text{ or } a) bb C-$  confers a lighter color than the yellow of  $A^Y(A \text{ or } a) Bb C-$  animals.]

- 39. In Fig. 3.28b, the  $A^1$  and  $B^1$  alleles each have the same effect on the phenotype** (plant height in this example), while the  $A^0$  and  $B^0$  alleles are non-functional. The shortest plants are  $A^0A^0B^0B^0$ , and the tallest plants are  $A^1A^1B^1B^1$ . **The phenotypes are determined by the total number of  $A^1$  and  $B^1$  alleles in the genotype.** Thus,  $A^0A^1B^0B^0$  plants are the same phenotype as  $A^0A^0B^0B^1$ . In total, there will be five different phenotypes: four '0' alleles (total  $A^1 + B^1$  alleles = 0); one 1 allele + three 0



alleles (total = 1); two 1 alleles + two 0 alleles (total = 2); three 1 alleles + one 0 allele (total = 3); and four 1 alleles (total = 4).

In Fig. 3.21, the  $a$  allele =  $b$  allele = no function (in this case no color = white). If the  $A$  allele has the same level of function as a  $B$  allele then you would see 5 phenotypes as was the case for Fig. 3.28b. But as a total of 9 phenotypes exist, this cannot be true so  $A \neq B$ . Notice that  $aa Bb$  is lighter than  $Aa bb$  even though both genotypes have the same number of dominant alleles. Thus, in Fig. 3.21 an  $A$  allele has more effect on coloration than a  $B$  allele, so 16 genotypes lead to 9 phenotypes.

40. a.  $Aa Bb Cc \times Aa Bb Cc \rightarrow 9/16 A- B- \times 3/4 C- : 9/16 A- B- \times 1/4 cc : 3/16 A- bb \times 3/4 C- : 3/16 A- bb \times 1/4 cc : 3/16 aa B- \times 3/4 C- : 3/16 aa B- \times 1/4 cc : 1/16 aa bb \times 3/4 C- : 1/16 aa bb \times 1/4 cc = 27/64 A- B- C- \text{ (wild type)} : 9/64 A- B- cc : 9/64 A- bb C- : 3/64 A- bb cc : 9/64 aa B- C- : 3/64 aa B- cc : 3/64 aa bb C- : 1/64 aa bb cc = 27/64 \text{ wild type} : 37/64 \text{ mutant.}$

b. Diagram the crosses:

1. unknown male  $\times AA bb cc \rightarrow 1/4$  wild type ( $A- B- C-$ ) :  $3/4$  mutant
2. unknown male  $\times aa BB cc \rightarrow 1/2$  wild type ( $A- B- C-$ ) :  $1/2$  mutant
3. unknown male  $\times aa bb CC \rightarrow 1/2$  wild type ( $A- B- C-$ ) :  $1/2$  mutant

The 1:1 ratio in test crosses 2 and 3 is expected if the unknown male is heterozygous for one of the genes that are recessive in the testcross parent. The 1 wild type : 3 mutant ratio arises when the male is heterozygous for two of the genes that are homozygous recessive in the testcross parent. (If you apply the product rule to  $1/2 B- : 1/2 bb$  and  $1/2 C- : 1/2 cc$  in the first cross, then you find  $1/4 B- C-$ ,  $1/4 B- cc$ ,  $1/4 bb C-$ , and  $1/4 bb cc$ . Only  $B- C-$  will be wild type, the other 3 classes will be mutant). Thus, the unknown male must be  $Bb Cc$ . In testcross 1 the male could be either  $AA$  or  $aa$ . Crosses 2 and 3 show that the male is only heterozygous for one of the recessive genes in each case: gene  $C$  in testcross 2 and gene  $B$  in testcross 3. To get wild-type progeny in both crosses, the male must be  $AA$ . **Therefore, the genotype of the unknown male is  $AA Bb Cc$ .**

41. a. For all five crosses, determine the number of genes involved in the trait and the dominance relationships between the alleles. Cross 1: One gene, red>blue. Cross 2: One gene, lavender>blue. Cross 3: One gene, codominance/incomplete dominance (1:2:1), bronze is the phenotype of the heterozygote. Cross 4: Two genes with recessive epistasis (9 red : 4 yellow : 3 blue). Cross 5: Two genes with recessive epistasis (9 lavender : 4 yellow : 3 blue). **In total there are two genes. One gene controls blue ( $c^b$ ), red ( $C^r$ ) and lavender ( $C^l$ ) where  $C^r = C^l > c^b$ . A second gene controls the yellow phenotype:  $Y$  seems to have no effect on color, so in the presence of  $Y$  the phenotype is determined by the alleles of the  $C$  gene. The  $y$  allele makes the flower yellow, and  $yy$  is epistatic to all alleles of the  $C$  gene.**

- b. cross 1:  $C^r C^r YY$  (red)  $\times c^b c^b YY$  (blue)  $\rightarrow C^r c^b YY$  (red)  $\rightarrow 3/4 C^r- YY$  (red) :  $1/4 c^b c^b YY$  (blue)  
 cross 2:  $C^l C^l YY$  (lavender)  $\times c^b c^b YY$  (blue)  $\rightarrow C^l c^b YY$  (lavender)  $\rightarrow 3/4 C^l- YY$  (lavender) :  $1/4 c^b c^b YY$  (blue)



cross 3:  $C^l C^l YY$  (lavender)  $\times C^r C^r YY$  (red)  $\rightarrow C^l C^r YY$  (bronze)  $\rightarrow 1/4 C^l C^l YY$  (lavender) :  $1/2 C^l C^r YY$  (bronze) :  $1/4 C^r C^r YY$  (red)

cross 4:  $C^r C^r YY \times c^b c^b yy$  (yellow)  $\rightarrow C^r c^b Yy$  (red)  $\rightarrow 9/16 C^- Y^-$  (red) :  $3/16 C^- yy$  (yellow) :  $3/16 c^b c^b Y^-$  (blue) :  $1/16 c^b c^b yy$  (yellow)

cross 5:  $C^l C^l yy$  (yellow)  $\times c^b c^b YY$  (blue)  $\rightarrow C^l c^b Yy$  (lavender)  $\rightarrow 9/16 C^- Y^-$  (lavender) :  $3/16 C^- yy$  (yellow) :  $3/16 c^b c^b Y^-$  (blue) :  $1/16 c^b c^b yy$  (yellow)

- c.  $C^r C^r yy$  (yellow)  $\times C^l C^l YY$  (lavender)  $\rightarrow C^r C^l Yy$  (bronze)  $\rightarrow$  monohybrid ratio for the  $C$  gene is  $1/4 C^r C^r$  :  $1/2 C^r C^l$  :  $1/4 C^l C^l$  and monohybrid ratio for the  $Y$  gene is  $3/4 Y^-$  :  $1/4 yy$ . Using the product rule, these generate a dihybrid ratio of  **$3/16 C^r C^r Y^-$  (red) :  $3/8 C^r C^l Y^-$  (bronze) :  $3/16 C^l C^l Y^-$  (lavender) :  $1/16 C^r C^r yy$  (yellow) :  $1/8 C^r C^l yy$  (novel genotype) :  $1/16 C^l C^l yy$  (yellow).** (Note: You expect the  $C^r C^l yy$  genotype to be yellow as  $yy$  is normally epistatic to the  $C$  gene. However, you have no direct evidence from the data in any of these crosses that this assumption is true, so it is possible that this genotype could cause a different and perhaps completely new phenotype.)

42. a. Analyze each cross by determining how many genes are involved in the coloration phenotypes as well as the relationships between the alleles of these genes. In cross 1, there are 2 genes because 3 classes in the  $F_2$  show a modified 9:3:3:1 ratio (12:1:3), and LR is the doubly homozygous recessive class. In cross 2, only 1 gene is involved because 2 phenotypes occur in a 3:1 ratio;  $WR > DR$ . In cross 3, again only 1 gene is involved (2 phenotypes in a 1:3 ratio);  $DR > LR$ . In cross 4, 1 gene is involved (2 phenotypes, with a 3:1 ratio);  $WR > LR$ . In cross 5, there are again 2 genes (and as in cross 1, there is a 12:1:3 ratio of three classes); LR is the double homozygous recessive. In total, **2 genes control these phenotypes in foxgloves.**
- b. Remember that all four starting strains are true-breeding. In cross 1 the parents can be assigned the following genotypes:  **$AA BB$  (WR-1)  $\times aa bb$  (LR)  $\rightarrow Aa Bb$  (WR)  $\rightarrow 9 A^- B^-$  (WR) :  $3 A^- bb$  (WR; this class displays the epistatic interaction) :  $3 aa B^-$  (DR) :  $1 aa bb$  (LR).** The results of cross 2 suggested that DR differs from WR-1 by one gene, so **DR is  $aa BB$** ; cross 3 confirms these genotypes for DR and LR. Cross 4 introduces WR-2, which differs from LR by one gene and differs from DR by 2 genes, so **WR-2 is  $AA bb$** . Cross 5 would then be  $AA bb$  (WR-2)  $\times aa BB$  (DR)  $\rightarrow Aa Bb$  (WR)  $\rightarrow 9 A^- B^-$  (WR) :  $3 A^- bb$  (WR) :  $3 aa B^-$  (DR) :  $1 aa bb$  (LR) = 12 WR : 3 DR : 1 LR.
- c. WR from the  $F_2$  of cross 1 LR  $\rightarrow 253$  WR : 124 DR : 123 LR. Remember from part (b) that LR is  $aa bb$  and DR is  $aa B^-$  while WR can be either  $A^- B^-$  or  $A^- bb = A^- ??$ . The experiment is essentially a testcross for the WR parent. The observed monohybrid ratio for the  $A$  gene is  $1/2 Aa$  :  $1/2 aa$  (253  $Aa$  : 124 + 123  $aa$ ), so the WR parent must be  $Aa$ . The DR and LR classes of progeny show that the WR parent is also heterozygous for the  $B$  gene (DR is  $Bb$  and LR is  $bb$  in these progeny). Thus, the cross is  **$Aa Bb$  (WR)  $\times aa bb$  (LR).**

43. The hairy  $\times$  hairy  $\rightarrow$  2/3 hairy : 1/3 normal cross described in the first paragraph of the problem tells us that the hairy flies are heterozygous, that the hairy phenotype is dominant to normal, and that the homozygous hairy progeny are lethal (that is, hairy is a recessive lethal). Thus, hairy is  $Hh$ , normal is  $hh$ , and the lethal genotype is  $HH$ . Normal flies therefore should be  **$hh$  (normal-1)** and a cross with hairy ( $Hh$ ) would be expected to always give 1/2  $Hh$  (hairy) : 1/2  $hh$  (normal) as seen in cross 1.

In cross 2, the progeny MUST for the same reasons be 1/2  $Hh$  : 1/2  $hh$ , yet they ALL appear normal. This suggests the normal-2 stock has another mutation that suppresses the hairy wing phenotype in the  $Hh$  progeny. The hairy parent must have the recessive alleles of this suppressor gene ( $ss$ ), while the normal-2 stock must be homozygous for the dominant allele ( $SS$ ) that suppresses the hairy phenotype. Thus cross 2 is  **$hh$   $SS$  (normal-2)**  $\times$   $Hh$   $ss$  (hairy)  $\rightarrow$  1/2  $Hh$   $Ss$  (normal because hairy is suppressed) : 1/2  $hh$   $Ss$  (normal).

In cross 3, the normal-3 parent is heterozygous for the suppressor gene:  **$hh$   $Ss$  (normal-3)**  $\times$   $Hh$   $ss$  (hairy)  $\rightarrow$  the expected ratios for each gene alone are 1/2  $Hh$  : 1/2  $hh$  and 1/2  $Ss$  : 1/2  $ss$ , so the expected ratio for the two genes together is 1/4  $Hh$   $Ss$  (normal) : 1/4  $Hh$   $ss$  (hairy) : 1/4  $hh$   $Ss$  (normal) : 1/4  $hh$   $ss$  (normal) = 3/4 normal : 1/4 hairy.

In cross 4 you see a 2/3 : 1/3 ratio again, as if you were crossing hairy  $\times$  hairy. After a bit of trial-and-error examining the remaining possibilities for these two genes, you will be able to demonstrate that this cross was  **$Hh$   $Ss$  (normal-4)**  $\times$   $Hh$   $ss$  (hairy)  $\rightarrow$  expected ratio for the individual genes are 2/3  $Hh$  : 1/3  $hh$  and 1/2  $Ss$  : 1/2  $ss$ , so the expected ratio for the two genes together from the product rule is 2/6  $Hh$   $Ss$  (normal) : 2/6  $Hh$   $ss$  (hairy) : 1/6  $hh$   $Ss$  (normal) : 1/6  $hh$   $ss$  (normal) = 2/3 normal : 1/3 hairy.

44. a. The mutant plant lacks the function of all three genes, so its genotype must be  **$aa$   $bb$   $cc$** .
- b. Considering each gene separately, 3/4 of the  $F_2$  progeny will have at least one dominant allele, whereas 1/4 will be homozygous for the recessive allele. As just seen in part (a), mutant plants must be triply homozygous recessive. The chance that a plant will have the  $aa$   $bb$   $cc$  genotype is  $1/4 \times 1/4 \times 1/4 = 1/64$ . All other  $F_2$  plants will be normal for this phenotype, so the fraction of normal plants =  $1 - 1/64 = 63/64$ .
- c. The most likely explanation for redundant gene function is that **in the relatively recent past, a single gene became duplicated (or in this case, triplicated)**. The three copies of the *SEP* gene are nearly identical to each other and thus fulfill the same function. Only if the functions of all three genes are lost does a mutant phenotype result. In fact, these kinds of gene duplication events occur often enough in nature that redundant gene function is a common phenomenon.
45. a. The split-hand deformity shows a **dominant** inheritance pattern; affected people occur in every generation.
- b. The penetrance is **at most 5/6  $\approx$  83%**. The father of the proband must have the allele for the deformity although he does not display it. Because the pedigree does not contain enough information to know if several other unaffected people in the family have the allele, the penetrance could be lower than 83%.

- c. As the proband is heterozygous for the deformity allele, the chance that the child inherited it is  $1/2$ . If the child is also a heterozygote, the likelihood that he or she would express the defect is  $\sim 83\%$ . Thus, the chance that the child would be affected by the deformity is  $1/2 \times 83\% \approx 42\%$ .
- d. **Four people in the pedigree diagram (III-1, III-2, III-4, and III-7) might have the mutant allele (because their parents had the mutant allele), but no information in the pedigree allows us to know one way or the other.** If any of them do have the mutant allele, then the penetrance is lower than 83%. For example, if we somehow determined that all of those four people had the mutant allele, then the penetrance would be  $5/10 = 50\%$ , and the answer to part (c) would be  $1/2 \times 50\% = 25\%$ . This would be the lowest possible likelihood consistent with the data given.
46. a. The genotypic ratio would be **9 purple ( $A- B-$ ) : 7 white ( $A- bb, aa B-, aa bb$ ).**
- b. If the penetrance of the purple phenotype in  $A- B-$  plants is 75%, then 25% of the ( $A- B-$ ) progeny would be white, and only 75% of them would be purple. This means that the purple plants in the 9/16 class would be  $9/16 \times 3/4 = 27/64$  of the total progeny. All the remaining  $F_2$  progeny  $[(64/64) - (27/64) = 37/64]$  will be white. Therefore, the genotypic ratio would be **27 purple : 37 white**.
- c. **If you crossed two different pure-breeding white strains, and some but not all the  $F_1$  were purple, one possible explanation is incomplete penetrance of the purple phenotype. In addition, you would never be able to make a pure-breeding purple strain, because even in an  $AA BB$  strain, in every generation, not all the plants will be purple.**
47. a. Two different phenotypes are mentioned in this problem. One phenotype is the shape of the erythrocytes. **All people with the genotype  $SPH^+ SPH^-$  have spherical erythrocytes. Therefore, this phenotype is fully penetrant and shows no variation in expression.** The second phenotype is anemia. **The expressivity among anemic patients varies from severe to mild. In fact, some people with the  $SPH^+ SPH^-$  genotype (150/2400) have no symptoms of anemia at all. Thus, the penetrance of the anemic phenotype is 2250/2400 or 0.94.**
- b. The severity of the anemia is greatly reduced when the spleen functions poorly and does not recognize the spherical erythrocytes as defective cells that must be eliminated from the bloodstream. Therefore, **treatment might involve removing the spleen (an organ which is not essential to survival). The more efficiently the spleen functions the earlier in a patient's life it should be removed.** Note that  $SPH^+ SPH^-$  with no symptoms of anemia should not be subjected to this drastic treatment
48. a. The most likely mode of inheritance is a **single gene with incomplete dominance** such that  $F^n F^n$  = normal ( $<250$  mg/dl),  $F^n F^a$  = intermediate levels of serum cholesterol (250-500 mg/dl) and  $F^a F^a$  homozygotes = elevated levels ( $>500$

mg/dl). Some of the individuals in the pedigrees do not fit this hypothesis. In two of the families (Families 2 and 4), two normal parents have a child with intermediate levels of serum cholesterol. **One possibility is that in each family, at least one of these normal parents (I-3 and/or I-4 in Family 2; I-1 and/or I-2 in Family 4) was actually a  $F^nF^a$  heterozygote who did not have elevated cholesterol in excess of 250 mg/dl. In this scenario, familial hypercholesterolemia is a trait with incomplete penetrance**, so that some unaffected people have a genotype that causes the disease in other people. It is also possible that the affected children of these parents do not have an  $F^a$  allele associated with elevated serum cholesterol, but they show the trait for other reasons such as diet, level of exercise, or other genes. This explanation is reasonable, but perhaps less likely because multiple children would have to have the trait but not the  $F^a$  allele.

- b. Familial hypercholesterolemia also shows variable expressivity**, meaning that people with the same genotype have the condition, but to different extents. This suggests that factors other than just the genotype are involved in the expression of the phenotype. Such factors could again include diet, level of exercise, and other genes.
- 49. a.** The pattern in both families is similar since unaffected individuals have affected progeny and the trait skips generations. It is highly unlikely that this trait is recessive. If that were the case, in the Smiths the unrelated people I-2, II-4, and II-7 must be carriers. Given that the trait is rare, a much more likely hypothesis is that **the trait is dominant but not 100% penetrant**.
  - b.** Assuming this is a dominant but not completely penetrant trait, **individuals II-3 and III-6 in the Smiths' pedigree individual and II-6 in the Jeffersons' pedigree** must carry the dominant allele but not express it in their phenotypes.
  - c.** If the trait were common, **recessive inheritance** must also be considered a possible, or even likely, mode of inheritance.
  - d. None;** in cases where two unaffected parents have an affected child, both parents would be carriers of the recessive trait.
- 50.** Several scenarios are possible. (i) **Perhaps the trait is incompletely penetrant.** That is, one parent could be  $Pp$  but not show the disease phenotype, and then the child could inherit the  $P$  disease allele. (ii) **Both parents could be  $pp$  yet the  $P$  allele inherited by the child was due to a spontaneous mutation during the formation of the gamete in one of the parents;** we will discuss this topic in [Chapter 7](#). (iii) **It is also possible that the father of the child is not the male parent of the couple.** In this case, the biological father must have the disease.
- 51.** While the general pattern of fingerprints is determined by genes, every detail of the pattern is not. Chance events that occur during skin development affect this trait.
- 52. The Black Lab:** A solid black dog must: make eumelanin ( $E-$ ); not be brown ( $B-$ ); have no pheomelanin striping in the hairs [ $(K^b-$  and any  $A$  gene alleles) or ( $aa$  and any  $K$

gene alleles)]; must not be diluted to gray ( $D-$ ); no spotting ( $S-$ ); and no merle ( $M^2M^2$ ). Because Labs always breed true for solid colors (black, brown, or some light yellow color), the black Lab cannot be heterozygous at any gene for recessive alleles that specify non-solid colors. So, the **Black Lab is most likely:  $EE$  or  $Ee$ ,  $BB$ , ( $K^bK^b$  and any gene  $A$  alleles) or ( $K^bK^b aa$ ),  $DD$ ,  $SS$ ,  $M^2M^2$ .**

**The Chocolate Lab:** A solid chocolate brown dog would be the same genotype as the solid black animal, except  $bb$ .

**The Yellow Lab:** A solid yellow dog must not make eumelanin ( $ee$ ). Any alleles of gene  $B$  are possible, and as above, the dog must be  $DD$ ,  $SS$ , and  $M^2M^2$ . The same considerations for genes  $A$  and  $K$  apply as for the other Labs above. The yellow dog pictured cannot be  $aa$ , however, or it would be white. Therefore, **yellow labs are  $ee$ ,  $B-$  or  $bb$ ,  $K^bK^b$ , any gene  $A$  alleles except  $aa$ ,  $DD$ ,  $SS$ ,  $M^2M^2$ .**