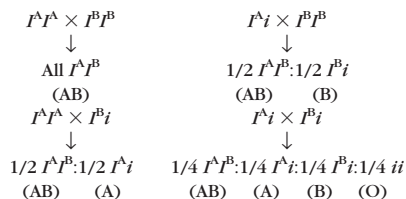


APPENDIX A

Brief Answers to Selected Exercises, Problems,
and Critical Thinking Questions

Chapter 2 Mendel's Principles

1. Dwarf F_2 ($1/4$ of total F_2), when selfed, produce all dwarf progeny (tt). Tall F_2 ($3/4$ of total F_2), when selfed, fall into two categories: $1/3$ (TT , $1/4$ of total F_2) produce all tall, and $2/3$ (Tt , $1/2$ of total F_2) produce tall and dwarf progeny in a 3:1 ratio. (The 3:1 ratio is from $1/2$ the F_2 , so the tall component is $3/8$ of the total F_3 [$3/4 \times 1/2$], and the dwarf is $1/8$ of the total F_3 [$1/4 \times 1/2$].) Overall, the F_3 are $3/8$ TT (tall), $2/8$ Tt (tall), and $3/8$ tt dwarf (see fig. 2.7).
3. Rule of segregation: adult diploid organisms possess two copies of each gene. Gametes get one copy. Fertilization restores the diploid number to the zygote. Rule of independent assortment: alleles of different genes segregate independently of each other.
5. Black is dominant, white is recessive, and both parents were heterozygous. The progeny are in an approximate 3:1 ratio. Since both parents had the same phenotype, the simplest cross is $Bb \times Bb$.
7. The disease is recessive at the individual level but incompletely dominant at the enzymatic level. Check the glossary for definitions.
9. $Ll \times Ll$. Let L = long ears and l = no ears. We see three phenotypes in an approximate 1:2:1 ratio. One of the phenotypes (short) is intermediate between long ears and no ears. Therefore, we have incomplete dominance.
11. Washed eye mutant, We ; wild-type, We^+ . (W is already the allelic designation for the wrinkled phenotype.)
13. All. Since the child was type A, it must have gotten the I^A allele from its mother. The other allele in the child is either I^A or i . A type A ($I^A I^A$ or $I^A i$), type B ($I^B I^B$ or $I^B i$), type O, or type AB man could have supplied either an I^A or i allele.
15. Universal donor, type O (no red-cell antigens); universal recipient, type AB (no serum antibodies).
17. All AB; or $1/2$ AB, $1/2$ A; or $1/2$ AB, $1/2$ B; or $1/4$ A: $1/4$ AB: $1/4$ B: $1/4$ O. Crosses can be $I^A I^A \times I^B I^B$, $I^A I^A \times I^B i$, $I^A i \times I^B I^B$, or $I^A i \times I^B i$.



19. Steve and his fiancé could be related. Both the dean and Steve's father must be I^i to produce O children, and each could have contributed M to produce M offspring. If the dean and Steve's father each contributed an S allele, the daughter would be SS . Note that if the daughter had B blood, she and Steve could not be related.

21. $RrTt \times$ self yields:

$1/16 RRTT$ red, tall
 $2/16 RRTt$ red, medium
 $1/16 RRtt$ red, dwarf
 $2/16 RrTT$ pink, tall
 $4/16 RrTt$ pink, medium
 $2/16 Rrtt$ pink, dwarf
 $1/16 rrTT$ white, tall
 $2/16 rrTt$ white, medium
 $1/16 rrtt$ white, dwarf

23. Choice (b) is preferred because although each will give the correct genotype, generally, testcrossing has the greatest probability of exposing the recessive allele in a heterozygote. For example, an Aa genotype, when selfed, produces aa offspring one-fourth of the time; when testcrossed, aa offspring appear one-half of the time; and when crossed with the Aa type (backcross), aa offspring occur one-fourth of the time. Thus, with a limited number of offspring examined per cross, testcrossing most reliably exposes the recessive allele.
25. The F_1 are tetrahybrids ($Aa Bb Cc Dd$). If selfed, an F_1 would form $2^4 = 16$ different types of gametes; 2^4 different phenotypes would appear in the F_2 , which would be made up of $3^4 = 81$ different genotypes; $1/(16)^2 = 1/256$ of the F_2 would be of the $aa bb cc dd$ genotype.
27. a. In the first cross, look at the yellow-to-green ratio, 120:43—almost exactly 3:1. Therefore, yellow is dominant and both parents must be heterozygous. Now look at the tall-to-short ratio, 122:41. Again, we see a 3:1 ratio, which indicates that tall is dominant and each parent is heterozygous. Thus, the cross is probably $YyTt \times YyTt$.
 b. In the second cross, there are no tall progeny. Therefore, either the short phenotypes are homozygous, or short is dominant and at least one parent is homozygous. In the absence of the first cross, we can't determine the mode of inheritance of height. We can, however, conclude that yellow is dominant (we got a 3:1 ratio) and that each parent is heterozygous. Based on the first cross, we can conclude that this cross is $Yytt \times Yytt$.
 c. In the third cross, we see 41 yellow:46 green, and 45 tall:42 short. Both of these ratios are 1:1, and all we can conclude is that these ratios result from matings between a heterozygote and a recessive homozygote. With only this cross, we can't determine dominance. However, we can if we use all three crosses. The cross is $yyTt \times Yytt$.

A-2 Appendix A Brief Answers to Selected Exercises, Problems, and Critical Thinking Questions

29. From the F_1 progeny, we can see that long and tan must be dominant, and the F_2 result confirms this assumption. We see a 3:1 ratio for both tan:dark and long:short. The total number of flies is 80. An ideal 9:3:3:1 ratio would be 45:15:15:5. Our results are very close to this. Therefore, we conclude that tan and long are dominant, and that the F_1 flies were heterozygous.

31. First list all possible genotypes for colored plants:

$AACcRR$
 $AACcRr$
 $AACcRR$
 $AACcRr$
 $AaCCRR$
 $AaCCRr$
 $AaCcRR$
 $AaCcRr$

The first genotype can be eliminated because all progeny should be colored, regardless of the tester strain. $AACcRr$ can be eliminated because the progeny of the first cross would have all been colored. $AACcRR$ can be eliminated because the progeny of the second cross would have all been colored. $AaCCRR$ can also be eliminated because the progeny of the third cross would have all been colored.

We are now left with $AACcRr$, $AaCCRr$, $AaCcRR$, and $AaCcRr$. Try $AACcRr \times aaccRR$ (cross 1), which will give 1/2 colored ($A-C-R$): 1/2 colorless ($A-ccR$) progeny. This could be the genotype, so try it in the second cross: $AACcRr \times aaCCrr$. This too will give 1/2 colored and 1/2 colorless offspring ($A-C-rr$). Since this does not fit the observed result, the unknown genotype is not $AACcRr$. Now try $AaCCRr \times aaccRR$ (cross 1). This fits the results. Try $AaCCRr \times aaccRR$. This will give 3/4 colorless ($aaccRR$, $A-ccRR$, or $aaC-RR$), which does not fit the results; therefore, the genotype is not $AaCCRr$. Now try $AaCcRr \times aaccRR$. This, too, will give 3/4 colorless ($aaccR$, $A-ccR$, or $aaC-R$), which is not seen. Therefore, the genotype must be $AaCCRr$. Confirm this with the other two crosses:

$AaCCRr \times aaCCrr \rightarrow 1/4$ colored : $3/4$ colorless, which fits.

$AaCCRr \times AACcrr \rightarrow 1/2$ colored : $1/2$ colorless, which fits.

33. a. all normal

b. 9 normal: 7 dark

c. 1/2 ebony: 1/2 normal; 1/2 black: 1/2 normal

If we let e = ebony, e^+ = wild-type, b = black, and b^+ = wild-type, the first cross is:

$ee b^+ b^+ \times e^+ e^+ bb$
 $\downarrow$
 $all e^+ e b^+ b$
 $\downarrow$ selfing

9/16 $e^+ - b^+ -$	wild-type
3/16 $ee b^+ -$	ebony
3/16 $e^+ - bb$	black
1/16 $ee bb$	ebony, black

Since it is difficult to distinguish black and ebony, 7/16 will be dark-bodied.

In c, the crosses are:

1. $e^+ e b^+ b \times ee b^+ b^+$
2. $e^+ e b^+ b \times e^+ e^+ bb$

In each case we have a testcross situation for only one gene.

35. 9 wild-type: 3 orange-1: 3 orange-2: 1 pink.

The first two crosses indicate that wild-type is dominant to both oranges, and the fourth indicates that orange-2 is dominant to pink. The fifth cross produces four phenotypes, indicating we are dealing with at least two genes. The presence of two genes is also suggested by orange-1 $\times$ orange-2. If these two traits were allelic, all the progeny should have been orange. The $F_1 \times$ pink produces progeny that resemble those from a double testcross. If $A-B-$ = wild-type, $A-bb$ = orange-1, $aaB-$ = orange-2, $aabb$ is probably pink. The crosses in question are then:

$AAbb$ $\times$ $aaBB$
 (orange-1) (orange-2)
 $\downarrow$
 $AaBb$
 (wild-type)
 $\downarrow$ self

9/16 $A-B-$	wild-type
3/16 $A-bb$	orange-1
3/16 $aaB-$	orange-2
1/16 $aabb$	pink

37. Two loci with epistasis. $AaBb \times$ self yields 9/16 $A-B-$: 6/16 $A-bb$ + $aaB-$: 1/16 $aabb$. Verify by testcrossing the various classes.

39. See for example figure 2.25. Other ratios are 10:3:3; 10:6; 12:3:1; 12:4.

41. 9 nonworkers: 7 hard workers. The F_1 indicates nonworker is dominant; therefore, a worker must be a recessive homozygote. If one gene is involved, the cross of the F_1 female $\times$ worker male ($Ww \times ww$) should produce 1 worker: 1 nonworker, for this is a testcross. This result is not seen, so we must have more than one gene involved. Perhaps a worker can result from more than one gene. Let $A-B-$ = nonworker, and $A-bb$, $aaB-$, or $aabb$ = workers. The original worker is $aabb$, and the cross is:

$aabb$ $\times$ $AABB$
 $\swarrow$
 $AaBb$ $\times$ $aabb$
 (nonworker) (worker)

1/4 $A-B-$	1/4 $A-bb$	1/4 $aaB-$	1/4 $aabb$
nonworker	worker	worker	worker

If $AaBb \times$ $AaBb$
 $\downarrow$

9/16 $A-B-$ nonworkers	
3/16 $aaB-$	workers
3/16 $A-bb$	
1/16 $aabb$	

43. 2 1 3 4
 $? \rightarrow$ indole $\rightarrow$ tryptophan $\rightarrow$ 3-hydroxyanthranilic $\rightarrow$ niacin
 or kynurenine acid

Accumulation: 1, indole; 2, ?; 3, tryptophan or kynurenine; 4, 3-hydroxyanthranilic acid. Without serine, the pathway would be blocked before the point of tryptophan production, after indole.

45. a. Maple sugar urine disease is recessive. If two individuals with the same phenotype produce offspring, some of whom have a different phenotype, both parents must be heterozygous. Since they are heterozygous, their phenotype must be dominant.

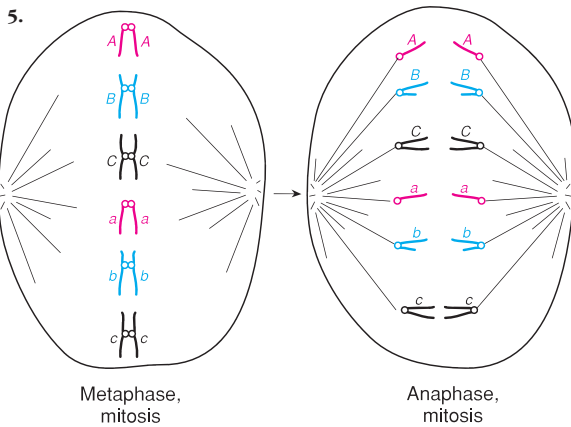
b. 3/4. Let M = normal and m = maple sugar urine alleles. The cross is $Mm \times Mm$. At each conception, the probability is three-fourths that each child is of the $M-$ genotype.

Critical Thinking Question:

1. The 15:1 ratio indicates that all genotypes except the recessive homozygote (*aabb*) produce a triangular capsule. The rounded capsule results from the recessive homozygote. One way to look at this is that the rounded form is a "default" form when neither locus has functional—dominant—alleles. However, a dominant allele at either of two loci is adequate to form the triangular seed capsule. The loci can be considered redundant in the pathway of seed capsule shape since a functional allele at one, the other, or both will provide a dominant (triangular) phenotype. At this point in time, it is impossible to know precisely what the enzymatic function of each dominant allele is.

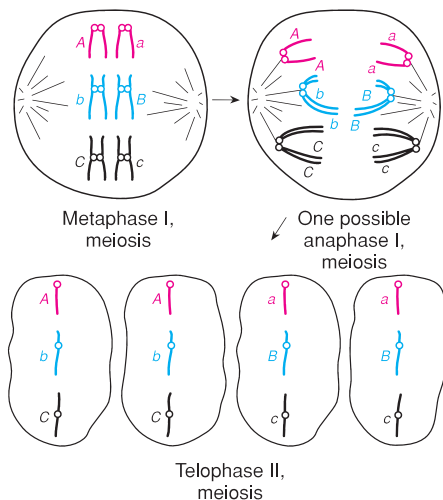
Chapter 3 Mitosis and Meiosis

1. See table 3.1 for a summary answer.
3. When a eukaryotic chromosome replicates during the S phase of the cell cycle, one chromosome becomes two chromatids, attached near the centromere. These are sister chromatids. Chromatids of different chromosomes are nonsister chromatids. Homologous chromosomes are members of a pair of essentially identical chromosomes. In diploid organisms, one member from each pair comes from each parent. Nonhomologous chromosomes do not share this relationship.



7. S phase

9. $2^3 = 8$ different gametes can arise. A crossover between the *A* locus and its centromere does not alter gametic combinations.



11. The intent of this problem is to make you think about the essential steps of meiosis, primarily the necessity to separate members of homologous pairs of chromosomes. Presumably, any method you devise will force you through that process.
13. Meiosis apportions homologous chromosomes the same way Mendel's rules apportion alleles. Each gamete gets one member of a homologous pair of chromosomes. Segregation predicts the same about alleles. The separation of homologues of one chromosome pair at meiosis is independent of the separation of other homologous pairs. Independent assortment makes the same prediction about alleles.
15. A gamete from wheat will have twenty-one chromosomes, and a gamete from rye will have seven chromosomes. Even if the seven rye chromosomes could pair with seven wheat chromosomes, a highly unlikely possibility, the remaining fourteen wheat chromosomes could not pair and would segregate randomly during meiosis. Almost every gamete would get an incomplete set; if fertilization did occur, the zygotes would have extra chromosomes (trisomic) or would be missing some chromosomes (monosomic or nullosomic).
17. 64. The number of combinations is 2^n where n = the number of different chromosomes in the set.

	DNA (Number of Chromatids)	Ploidy
Spermatogonium or Oogonium	2	$2n$
Primary Spermatocyte or Primary Oocyte	4	$2n$
Secondary Spermatocyte or Secondary Oocyte	2	n
Spermatid or Ovum	1	n
Sperm	1	n

21. a. 50 b. 50. The primary oocyte is diploid and will undergo meiosis, but only one functional ovum results from each primary oocyte. The secondary oocyte will divide to produce an ovum and a polar body.
23. Any possible genotype, from *AAABBB* through *aaabbb* can occur in the endosperm. If at a given locus the endosperm is homozygous, so is the embryo. If the locus is heterozygous (e.g., *Aaa* or *Aaa*), so is the embryo. Thus, an *AAabbb* endosperm is associated with an *Aabb* embryo.
25. A greater maternal influence in *Drosophila* and corn than in *Neurospora* means the sexes (mating types) do not show the disparity in size between male and female cells as in *Drosophila* and corn.
27. b. Homologous chromosomes will pair during meiosis. Each gamete gets one of each chromosome, A, B, C, D, and E. Fertilization fuses two cells with the chromosome complement given. Since root cells are somatic tissue, these cells will be diploid.
29. a. 2^{50} or about 1.1×10^{15} b. 2^2 . The number of gametes produced is 2^n , where n = number of independently behaving entities. If the genes are completely independent, we expect 2^{50} , and if they are completely linked, we expect 2^2 . In reality, the number falls between these extremes.

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Appendix A Brief Answers to Selected Exercises, Problems, and Critical Thinking Questions

Critical Thinking Question:

1. Both meiosis and mitosis are processes that initiate under certain circumstances of cell cycle and place. Neither is actually dependent on the chromosomal content of the cell. Thus, meiosis could begin in a haploid cell, but it would not be a successful process because there is no homologue for any chromosome to pair with. Mitosis would, however, be successful because there is no pairing (synapsis) required for successful completion of the process.

Chapter 4 Probability and Statistics

1. a. $(5!/3!2!)(1/2)^3(1/2)^2 = 0.3125$
b. $(1/2)^5 = 0.03125$ (SDSDS, in which S = son, D = daughter)
c. $2(1/2)^5 = 0.0625$ (SDSDS + DSDSD)
d. $(1/2)^5 = 0.03125$
e. $2(1/2)^5 = 0.0625$ (all sons + all daughters)
f. 4 daughters, 1 son + 5 daughters: $(5!/4!1!)(1/2)^4(1/2) + (1/2)^5 = 0.1875$
g. $(1/2)^2 = 0.25$ (DXXXS, in which X is either a daughter or a son, with $p = 1$; $P = [1/2][1][1][1][1/2]$)
3. Remember that albinos have blue eyes. Therefore, 7/16 of the offspring will have blue eyes. If we let B = brown, b = blue, C = normal color expression, and c = albinism, the following genotypes are blue-eyed: $C-bb$ and $cc-$.
a. $(1/4)^5 = 0.0009765$
b. $(1/8)^5 = 0.0000305$
c. $(5!/4!1!)(7/32)^4(9/32) = 0.00322$
d. $(4!/2!2!)(1/8)^2(1/8)^2 = 0.0014648$
5. a. $2(1/2)^4 = 0.125$
b. $(1/2)^4(1/2)^4 = 0.0039063$ (Probability that sperm and egg creating the zygote each had only paternal centromeres.)
7. One-half.
9. a. 81/256 b. 108/256 c. 9/256. In (a), since all children have the same phenotype, each child will have the same probability of having no molars. Therefore, $(3/4)^4 = 81/256$. In (b),

$$P = \frac{4!}{3!1!} (3/4)^3(1/4) = 108/256.$$

When order is given, we multiply the chance of each event, $1/4 \times 1/4 \times 3/4 \times 3/4 = 9/256$.

11. One-eighth. B must be heterozygous (Gg), as must A's father. We assume A's mother is GG , since there is no mention of the disease in her family. Therefore, A has a one-half chance of getting g from his father. If two heterozygotes mate, the chance of a recessive child is one-fourth, so $P = 1/4 \times 1/2$.
13. 1/512. The F_1 progeny are $Aa Bb Cc Dd Ee$. The chance of getting any individual with a particular homozygous genotype is $(1/4)^5$. Since we are looking for two different possibilities, we have $2(1/4)^5 = 2/1024 = 1/512$.
15. 0.049. Since the order is not specified, we use the multinomial formula. We have six mice, so $n = 6$. If p = chance of agouti, $p = 3/4 \times 1/2 = 3/8$; q (black coat color) = $1/4 \times 1/2 = 1/8$, and r (albino) = $1/2$. The equation becomes:

$$p = \frac{6!}{2!2!2!} (3/8)^2(1/8)^2(1/2)^2 =$$

$$\frac{6 \times 5 \times 4 \times 3 \times 2 \times 1 \times 9 \times 1 \times 1}{2 \times 2 \times 2 \times 64 \times 64 \times 4} = 0.049$$

17. Hypothesis: $RrYy \times RrYy$ produces $R-Y:R-yy:rrY:rryy$ in a 9:3:3:1 ratio. The critical chi-square, three degrees of freedom at probability of 0.05, = 7.815.

	<i>R-Y-</i>	<i>R-yy</i>	<i>rrY-</i>	<i>rryy</i>	Sum
Observed	315	108	101	32	556
Expected	9/16	3/16	3/16	1/16	
	312.75	104.25	104.25	34.75	556
$O - E$	2.25	3.75	-3.25	-2.75	
$(O - E)^2$	5.06	14.06	10.56	7.56	
$(O - E)^2/E$	0.016	0.135	0.101	0.218	$0.470 = \chi^2$

Since this chi-square, 0.470, is less than the critical chi-square, we fail to reject our hypothesis of two-locus genetic control with dominant alleles at each locus.

19. We reject the 3:1 ratio as an appropriate null hypothesis. If we calculate the chi-square using 3:1 as the expected ratio, we expect 72 and 24 ($3/4$ and $1/4$ of 96, respectively):

	<i>O</i>	<i>E</i>	<i>O-E</i>	$(O-E)^2$	$(O-E)^2/E$
Curly-winged flies	61	72	-11	121	$121/72 = 1.681$
Straight-winged flies	35	24	11	121	$121/24 = 5.042$

Chi-square = 6.723 (1.681 + 5.042). With one degree of freedom, $p > 0.05$ (critical chi-square = 3.841).

Critical Thinking Question:

1. You should change your choice because the box you chose originally has a 1/3 chance of containing the prize, whereas the remaining box has a 2/3 chance of containing the prize. The 1/3 chance of your choice is set by the fact that there were three equally likely choices at the beginning. When your friend eliminated an empty box, she left two choices: your original box and the third box. Since the probability of your original choice has not changed, the probability of the remaining box must be 2/3 to give a combined probability of 1.0 that a box contains a prize.

Chapter 5 Sex Determination, Sex Linkage,
and Pedigree Analysis

1. The differences are in terminology only, not in shape or size of the chromosomes. In species in which females have a homomorphic pair of sex chromosomes, the members of the pair are called X chromosomes. In species in which males have a homomorphic sex chromosome pair, the members of the pair are called Z chromosomes.
3. $3/8$ males, $3/8$ females, $2/8$ intersexes ($dsx dsx$ homozygotes). The $1/4 dsx dsx$ flies will be intersexes, whereas half of the $3/4$ will be normal males and half will be normal females.
5. The protein is probably a dimer, which, in the heterozygote, can be of fast-fast, fast-slow, or slow-slow subunit combinations. A female heterozygous for a sex-linked gene controlling a dimeric enzyme should show the pattern of lane 3 in whole blood (mixture of slow-slow and fast-fast dimers) and lanes 1 or 2 in individual cells.

Appendix A Brief Answers to Selected Exercises, Problems, and Critical Thinking Questions

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7. a. 0; human female, male fly
b. 1; human female, female fly
c. 0; human male, male fly
d. 1; human male, female fly
e. 2; human female, female fly
f. 4; human female, female fly
g. 1/0 mosaic; human male-female mosaic, male-female fly mosaic

9.

	Cross	Reciprocal
P_1 female	X^+X^+	$X^{Lz}X^{Lz}$
male	$X^{Lz}Y$	X^+Y
F_1 female	X^+X^{Lz}	X^+X^{Lz}
male	X^+Y	$X^{Lz}Y$
F_2 females	X^+X^+, X^+X^{Lz}	$X^+X^{Lz}, X^{Lz}X^{Lz}$
males	$X^+Y, X^{Lz}Y$	$X^+Y, X^{Lz}Y$

11. Exemptions should be made minimally for hemophilia in brother, sister's son, mother's brother, mother's sister's son, mother's father, and others more distantly related.

13. P_1 $f_1f_1 X^+X^+$ (female) $\times$ $f_1^+f_1^+ X^{ct}Y$ (male)
 F_1 $f_1^+f_1 X^+X^{ct}$ (female) $\times$ $f_1^+f_1 X^+Y$ (male)

	Male			
	$f_1^+ X^+$	$f_1 X^+$	$f_1^+ Y$	$f_1 Y$
Female				
$f_1^+ X^+$	$f_1^+f_1^+ X^+X^+$	$f_1^+f_1 X^+X^+$	$f_1^+f_1^+ X^+Y$	$f_1^+f_1 X^+Y$
$f_1^+ X^{ct}$	$f_1^+f_1^+ X^+X^{ct}$	$f_1^+f_1 X^+X^{ct}$	$f_1^+f_1^+ X^{ct}Y$	$f_1^+f_1 X^{ct}Y$
$f_1 X^+$	$f_1^+f_1 X^+X^+$	$f_1f_1 X^+X^+$	$f_1^+f_1 X^+Y$	$f_1f_1 X^+Y$
$f_1 X^{ct}$	$f_1^+f_1 X^+X^{ct}$	$f_1f_1 X^+X^{ct}$	$f_1^+f_1 X^{ct}Y$	$f_1f_1 X^{ct}Y$

F_2 : females, 3/4 wild-type, 1/4 fuzzy; males, 3/8 wild-type, 3/8 cut, 1/8 fuzzy, and 1/8 cut and fuzzy.

15. a. X linked

b. gray

c. 1/2 gray:1/2 yellow in both sexes

In both crosses, we see a difference in the phenotypes of the sexes, suggesting sex linkage. The F_1 offspring from the first cross indicate that gray is dominant to yellow. The F_1 females from this cross must be heterozygous, and the two phenotypes in the F_2 males result from each of the X chromosomes in the F_1 female being hemizygous in the F_2 males. The first cross is therefore (calling gray the wild-type):

X^+X^+	$\times$	X^yY	
	$\downarrow$		
X^+X^y	$\times$	X^+Y	
gray		gray	
	$\downarrow$		
X^+X^+	X^+X^y	X^+Y	X^yY
gray	gray	gray	yellow

Now diagram the second cross:

X^YX^Y	$\times$	X^+Y	
	$\downarrow$		
X^+X^Y	$\times$	$X^Y Y$	
gray		yellow	
	$\downarrow$		
X^+X^Y	X^YX^Y	X^+Y	$X^Y Y$
gray	yellow	gray	yellow

17. Yes. Begin by determining genotypes of the two individuals. The woman must be heterozygous $X^C X^c$. A man with normal vision must be $X^C Y$, and all his daughters must receive his X chromosome and should be normal, either $X^C X^C$ or $X^C X^c$. Since color blindness is recessive, the daughter must have two X^c chromosomes. A *very* rare possibility is that the man is the father and nondisjunction occurred in both parents: at meiosis I in the male and at meiosis II in the female (see chapter 8).

19. a. F_1 : wild-type females, white-eyed males.

b. F_2 : 3 wild-type:3 white-eyed:1 ebony:1 ebony, white-eyed in both sexes.

c. Reciprocal F_2 : 3 wild-type females:1 ebony female:3 wild-type males:3 white-eyed males:1 ebony male:1 ebony, white-eyed male. Let X^+ = red, X^w = white, e^+ = wild-type, e = ebony.

$X^wX^w e^+e^+$	$\times$	$X^{w+}Y ee$	P_1
$\downarrow$			
$X^+X^w e^+e$	$\times$	$X^{w+}Y e^+e$	F_1
(wild-type)		(white-eyed)	

Use probability for the F_2 generation rather than the Punnett square:

$$\begin{aligned}
 (1/2)X^+ \times (1/2)X^{w+} \times (3/4)e^+ &= 3/16 \text{ wild-type females} \\
 &\times (1/4)ee = 1/16 \text{ ebony females} \\
 \times (1/2)Y \times (3/4)e^+ &= 3/16 \text{ wild-type males} \\
 &\times (1/4)ee = 1/16 \text{ ebony males} \\
 (1/2)X^w \times (1/2)X^{w+} \times (3/4)e^+ &= 3/16 \text{ white-eyed females} \\
 &\times (1/4)ee = 1/16 \text{ white-eyed, ebony females} \\
 \times (1/2)Y \times (3/4)e^+ &= 3/16 \text{ white-eyed males} \\
 &\times (1/4)ee = 1/16 \text{ white-eyed, ebony males}
 \end{aligned}$$

For the reciprocal cross,

$X^+X^+ ee$	$\times$	$X^{w+}Y e^+e^+$
$\downarrow$		
$X^+X^{w+} e^+e$	$\times$	$X^+Y e^+e$
(wild-type)		(wild-type)

All F_2 females will get X^+ ; e^+e will be 3:1. The males will be as in the males shown.

21. The female is heterozygous for an X-linked color gene (one of the X chromosomes in the cells of female cats is inactivated, leading to the black and yellow spots). We immediately deduce X-linkage because of the different phenotypes in the sexes. Finding two types of males indicates that the female was heterozygous. The patches of yellow and black come from X-inactivation. The cross is:

X^bX^o		$\times$	X^bY	
		$\downarrow$		
X^bX^b	X^bX^o		X^bY	X^oY
black	black and yellow		black	yellow

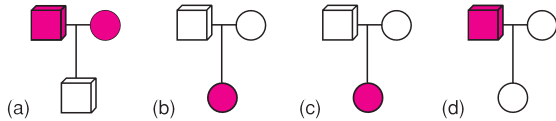
23. Penetrance is the proportion of individuals of a particular genotype that shows the appropriate phenotype; expressivity is the degree to which a trait is expressed.

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Appendix A Brief Answers to Selected Exercises, Problems, and Critical Thinking Questions

25. a. The phenotype is the propensity to have twin offspring. It could be caused by a recessive or dominant, sex-linked or autosomal allele.
b. Autosomal dominant or possibly autosomal recessive inheritance.
c. Autosomal, or sex-linked, recessive inheritance.
d. Autosomal recessive inheritance.

27. Assuming 100% penetrance:



Critical Thinking Question:

1. The immediate effect of a null allele is to make heterozygous genotypes appear to be homozygotes. That is, in the simplest system, we expect one band in a homozygote and two bands in a heterozygote. If we see only one band, we assume that the individual is homozygous for that allele, when in fact that individual might be heterozygous for the allele that produces the particular band and the allele that produces no band (null allele). The null allele can be verified by both the absence of any bands in the null homozygote and the results of breeding experiments when the null allele is suspected.

Chapter 6 Linkage and Mapping in Eukaryotes

1. a.

P ₁ groucho	×	rough
<i>grogro ro⁺ro⁺</i>		<i>gro⁺gro⁺ roro</i>
F ₁ female <i>gro⁺gro ro⁺ro</i>	×	male <i>grogro roro</i>
F ₂ <i>grogro ro⁺ro</i>		518
<i>gro⁺gro roro</i>		471
<i>grogro roro</i>		6
<i>gro⁺gro ro⁺ro</i>		5
(6+5)/1,000 = 0.011 = 1.1% recombination		
= 1.1 map units apart		

- b. Given the map units, F₁ gametes are produced on the average by females as follows: *gro ro⁺*, 49.45% = 0.4945 (98.9%/2); *gro⁺ro*, 49.45% = 0.4945; *gro ro*, 0.55% (1.1%/2) = 0.0055; and *gro⁺ro⁺*, 0.55% = 0.0055. Males, lacking crossing over, produce only two gamete types: *gro ro⁺* and *gro⁺ro*, each 50% = 0.50. Summing from the Punnett square following, the phenotypes of the offspring would be as follows: wild-type, 50%; groucho, rough, 0%; groucho, 25%; and rough, 25%.

Female	Male	
	<i>gro ro⁺ (0.5)</i>	<i>gro⁺ro (0.5)</i>
<i>gro ro⁺</i>	groucho	wild-type
(0.4945)	0.24725	0.24725
<i>gro⁺ro</i>	wild-type	rough
(0.4945)	0.24725	0.24725
<i>gro ro</i>	groucho	rough
(0.0055)	0.00275	0.00275
<i>gro⁺ro⁺</i>	wild-type	wild-type
(0.0055)	0.00275	0.00275

3. A dihybrid female is testcrossed (with a hemizygous male having both recessive alleles). Each recombinant class will make up about 5% of the offspring. Each parental class will make up about 45% of

the offspring. Phenotypic classes will be equally distributed between the two sexes. The same results will be found for an autosomal locus if the dihybrids are females (no crossing over in males). A reciprocal cross cannot be done for X-linked genes because males cannot be dihybrid. Males dihybrid for an autosomal gene produce only two classes of offspring when testcrossed—parentals.

5. a. The hotfoot locus is in the middle (compare, for example, hotfoot, a double crossover, with the wild-type, a parental); there are 16.0 map units from hotfoot to either end locus: 74 + 66 + 11 + 9 recombinants between hotfoot and waved and 79 + 61 + 11 + 9 recombinants between hotfoot and obese.
b. The trihybrid parent was *o b wa/o⁺b⁺wa⁺*.
c. The coefficient of coincidence is 20/25.6 (20/[0.16 × 0.16 × 1,000]); interference is 1 - (20/25.6) = 0.22, or 22%.
7. a. Work backward from the 0.61% double recombinants (0.100 × 0.061 × 100). Thus, there would be 6 of 1,000 double recombinants. In the *an-sple* region, we need the total of single + double recombinants = 100 of 1,000 (10 map units). Thus, 100 - 6 = 94; divided by 2 (two phenotypes) is 47 each. For the *sple-at* region, the total of single and double recombinants = 61 (6.1 map units). Thus, 61 - 6 = 55; divided by 2 is 27 and 28. The parentals make up the remainder for a total of 1,000.
b. With a coefficient of coincidence of 0.60, only 0.366% (0.61 × 0.60) of the expected double recombinants will occur, that is, 4 instead of 6. Thus:

Coefficient of Coincidence

	1.0	0.6
ancon, spiny, arctus oculus	422	421
wild-type	423	422
ancon, spiny	27	28
arctus oculus	28	29
ancon	47	48
spiny, arctus oculus	47	48
ancon, arctus oculus	3	2
spiny	3	2
Total	1,000	1,000

9. a. linked; b. *trans*; c. 28.7%. The cross is a testcross. If the genes were not linked, we would expect a 1:1:1:1 ratio of offspring; we don't see that. The alleles that are linked will appear as the majority classes, which are Trembling, long-haired and normal, Rex. Therefore, Trembling and Rex are in the *trans* position. If we let *T* = Trembling, and *R* = Rex, the cross is

$$\frac{Tr}{tR} \times \frac{tr}{tr}$$

Recombinants are Trembling, Rex and normal, long-haired;

$$\frac{42 + 44}{300} \times 100 = 28.7\%$$

11. a. $\frac{k e^{+} cd}{k^{+} e cd^{+}}$ b. $\frac{k}{k^{+}} \frac{e}{e^{+}} \frac{cd}{cd^{+}}$

The initial cross is $\frac{k e^{+} cd}{k e^{+} cd} \times \frac{k^{+} e cd^{+}}{k^{+} e cd^{+}}$

Producing a trihybrid F₁ female: $\frac{k e^{+} cd}{k^{+} e cd^{+}}$, in any order.

Appendix A Brief Answers to Selected Exercises, Problems, and Critical Thinking Questions

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The last two classes (3 + 4 offspring in the F_2) are double crossovers and allow us to determine order by comparison with the parents (880 + 887). If the order is $k\ cd\ e$, a double crossover in the F_1 females yields $k\ cd^+ e^+$ and $k^+ cd\ e$. If the order is $cd\ k\ e$, a double crossover yields $cd\ k^+ e^+$ and $cd^+ k\ e$. Therefore, the order must be $k\ e\ cd$, which gives the correct double recombinants in the F_2 generation. After reconstructing the trihybrid

$$\frac{k\ e^+ cd}{k^+ e\ cd^+}$$

and scoring each of the offspring for crossovers in the $k\text{-}e$ and $e\text{-}cd$ regions: map units, $k\text{-}e = [(64 + 67 + 4 + 3)/2000] \times 100 = 6.9$ and map units, $e\text{-}cd = [(49 + 46 + 4 + 3)/2000] \times 100 = 5.1$.

13. 0.0125. This problem requires the manipulation of equations. We know that interference = 1 - coefficient of coincidence, so coefficient of coincidence = 1 - interference = 1 - (-1.5) = 2.5. Since coefficient of coincidence =

$$\frac{\text{observed double crossovers}}{\text{expected double crossovers}}$$

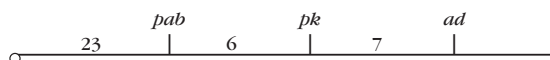
observed double crossovers = (coefficient of coincidence) $\times$ (expected double crossovers). The expected double crossover frequency is (2.5)(0.005) = 0.0125.

15. PD, 1, 2, 4, 6, 8-10; NPD, 3; TT, 5, 7. Map units = $[(\text{NPD} + \{1/2\}\text{TT})/\text{Total}] \times 100 = [(1 + \{1/2\}2)/10] \times 100 = 20$. The loci are 20 map units apart.
17. FDS, 3-5, 8, 10; SDS, 1, 2, 6, 7, 9. The distance between the *arg* locus and its centromere is $(1/2)\%\text{SDS} = (1/2)50\% = 25$ map units.
19. For example, the only variant of the type 2 pattern of table 6.7 is a^+b , a^+b , a^+b , a^+b , ab^+ , ab^+ , ab^+ , ab^+ . Other patterns that are variants of the remaining five categories are derived by inverting the eight spores of a pattern (bottom to top) or by switching spores 1 and 2 with spores 3 and 4 or spores 5 and 6 with spores 7 and 8.
21. $a \times a^+ \rightarrow a/a^+$, which undergoes meiosis. Twelve map units means that the SDS pattern makes up 24% of the asci.
23. For example: 1, 408; 2, 42; 3, 250; 4, 250; 5, 30; 6, 5; 7, 15. To make the numbers work out, classes 4-7 must equal 300, as must classes 3 + 5-7, in order to make $(1/2)\%\text{SDS} = 150$. Thus, if classes 3 and 4 are 250 each, 50 must be spread out among classes 5-7. NPD + $(1/2)\text{TT}$ should equal 300 to confirm the arrangement (*a-b* distances). These numbers will give an *a-b* distance of 30.
25. a. yes; PD > > NPD (actually, no NPD). b. a: 2.5 map units; b: 7.5 map units. Classify each ascus—I: PDT, FDS for both; II: TT, FDS for *a*, SDS for *b*; III: TT, FDS for *a*, SDS for *b*; IV: TT, FDS for *a*, SDS for *b*; V: TT, FDS for *a*, SDS for *b*; VI: PDT, SDS for *a* and *b*; VII, PDT, SDS for *a* and *b*; VIII: PDT, SDS for *a* and *b*. We see no NPDs, so genes are linked. For gene to centromere distances, use the formula

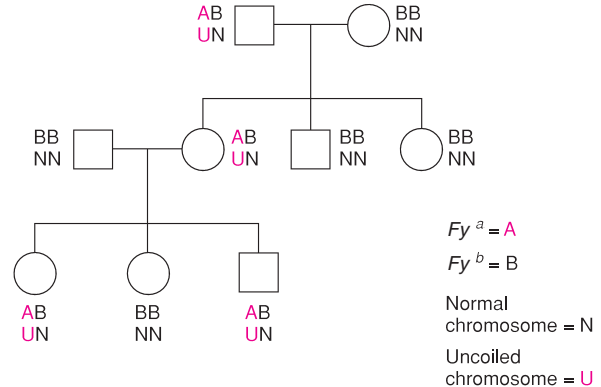
$$\frac{1/2 (\text{number of SDS asci})}{100} \times 100:$$

a to centromere = $(1/2)(3 + 1 + 1)\% = 2.5$ map units; *b* to centromere = $(1/2)(2 + 3 + 2 + 3 + 3 + 1 + 1)\% = 7.5$ map units.

27. 12.5 map units. The only genotype that grows on minimal medium is $arg^+ ade^+$. If the two genes were unlinked, 1/4 of the progeny should have this genotype; this is not seen. The genes must be linked; wild-type results from recombination between these two genes. The reciprocal class, $arg^- ade^-$, which has not been selected for, should be equally frequent, so: map units = $(2 \times 25)/400 \times 100 = 12.5$.



29. Construct a pedigree of the Duffy alleles (Fy^a , Fy^b). Arbitrarily assign one allele to a normal chromosome 1 and the other allele to the coiled chromosome. Then, accompanying the pedigree, the alleles and their morphologically proper chromosomes would be associated.



31. Twenty map units apart (two of ten recombinant sons according to the "grandfather method").
33. Thirty-three map units. The woman is heterozygous in *trans* configuration for color blindness and hemophilia: bc^+/b^+c . Recombination between these two markers yields b^+c^+ and bc . We can only detect recombinants in sons, so

$$\frac{\# \text{ normal sons} + \# \text{ double mutant sons}}{\text{total sons}} \times 100 = \text{map distance} = 2/6 = 0.33$$

35. Enzyme A on 11; B on 15; C on 18; D on 3; E on 7. Enzyme A is present in clones X and Y, and chromosome 11 is common to these two clones. Enzyme B is present only in X, and 15 is the only chromosome unique to X. Similar logic allows the assignment of the other genes.

Critical Thinking Question:

1. Three-point crosses capture (allow us to see) double crossovers that have taken place in the two regions defined by the three loci. However, any double crossovers that occur within one region or any crossovers involving more than two events will not be indicated correctly by random-strand analysis.

Chapter 7 Linkage and Mapping in Prokaryotes and Bacterial Viruses

1. The prokaryotic chromosome is a double-stranded DNA circle that is small compared with most eukaryotic chromosomes. Viral chromosomes can be DNA or RNA. Viruses are obligate intracellular parasites. Whether they are alive depends on the definition of the term *alive*.
3. A colony is a visible mass of cells derived usually from a single progenitor. A plaque is the equivalent growth of phages on a bacterial lawn, producing a cleared area lacking intact bacteria.
5. The bacterium could have survived and produced a colony if it was on a λ -free area, it became lysogenic (and thus resistant to further phage attack), or it was genetically resistant to phage λ .
7. 1, $bis^- arg^-$; 2, leu^- ; 3, lys^- ; 4, $bis^- met^-$ or $bis^+ met^-$; 5, arg^- .
9. Where phages cannot grow: *E. coli ton^r*, phage h^+ . Where phage can grow: *E. coli ton^s*, phage b^+ or *h*, or *E. coli ton^r*, phage *b*.

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11. Far. If the selective locus is near, it passes into the F^- cell very early during conjugation. Consequently, there is a great reduction in the recovery of loci distal to the selective marker because both the Hfr and F^- members of a conjugation event can be killed by the selective agent (e.g., an antibiotic such as streptomycin).

13. See figures 7.8, 7.9, 7.15, 7.17, 7.18, and 7.26.

15. 9 min 1 min 8 min 7 min
Origin ——— *az* ——— *ton* ——— *lac* ——— *galB*

17. For example, use an Hfr that is wild-type but str^s with the F factor integrated at minute 20. Use an F^- strain that is $pyrD^-$, $purB^-$, man^- , $uvrC^-$, bis^- , and str^r . Interrupt mating at one-minute intervals; plate cells on complete medium with streptomycin to kill Hfr cells and grow recombinant and nonrecombinant F^- cells. The next day, after colonies have grown up, replica-plate onto selective media. The following data would be generated:

Colony Growth on Media Selective for

	<i>pyrD</i> ⁺	<i>purB</i> ⁺	<i>man</i> ⁺	<i>uvrC</i> ⁺	<i>bis</i> ⁺
Minute 0	—	—	—	—	—
1	+	—	—	—	—
5	+	+	—	—	—
16	+	+	+	—	—
22	+	+	+	+	—
24	+	+	+	+	+

19. The order is $a\ c\ b$, and c is close to a . Genes c and a are cotransformed 76% of the time, suggesting that these two genes are very close and b is far away (a - b cotransduction is 10%, or 0.1). Two orders are possible: a - c ———— b and c - a ———— b . If the first order is correct, $a^+ b^+ c^-$ results from a double crossover; this class should be the least frequent. If the second order is correct, a single exchange between a and c would yield $a^+ b^+ c^-$, but this frequency should be similar to $a^+ b^- c^-$, and it is not.

21. *thr leu pro bis*. We see that cells that are thr^+ are the most frequent. The chance of interruption in the conjugation increases with the length of time for the mating. Therefore, genes farther from the origin of transfer appear less frequently. We can order the genes based merely upon the frequency of genotypes seen. The order must be *thr leu pro bis*. Since we see no bis^+ , and since we stopped the mating at 25 minutes, *bis* must be after minute 25 on the map of this Hfr strain.

23. a and c are close; b is farther away; c is probably in the middle. The numbers of the first three transformant classes indicate that each gene, by itself, is readily transformed. We notice that classes with b and any other gene are quite rare, a situation that indicates b is far from a and c . We notice that a and c are cotransformed about 13% of the time, b and c about 3% of the time, and a and b about 2% of the time.

25. a. $lys^+ bis^+ val^+$
 $lys^+ bis^+ val^-$
 $lys^+ bis^- val^+$
 $lys^+ bis^- val^-$

Since there is no lysine in the medium, lys^+ must be present to allow growth.

- b. $lys^+ val^+ bis^+$
 $lys^+ val^+ bis^-$

Both lys^+ and val^+ must be present to allow growth.

- c. $lys^+ val^+ bis^+$
 $lys^+ val^- bis^+$

Both lys^+ and bis^+ must be present to allow growth.

- d. $lys^+ val^+ bis^-$
 $lys^+ val^- bis^+$

We see no $lys^+ val^+ bis^+$ cells.

- e. lys^+ and val^+ are close together; they are cotransformed 75% of the time. Order could be *lys val bis* or *val lys bis*.

- f. *val lys bis*. If the order is *val lys bis*, $val^+ lys^- bis^+$ should be rare, since this genotype results from a double exchange; and indeed, this class is the least frequent.

27. Mix the phages together with bacteria with increasing quantities of the two phages. Knowing the numbers of each in a particular case, it is possible to predict the proportion of cells doubly infected (product of probabilities). The recovery of recombinants should increase with that probability. In other words, recombination should occur only in doubly infected cells.

29. $\frac{0.6}{a} \quad \frac{0.2}{b} \quad \frac{1.2}{d} \quad \frac{1.2}{c}$

Small recombination frequencies should be approximately additive. Note that recombination distances are twice the value of wild-type plaques since the double mutant recombinants were not counted. Thus, the data table should be:

Cross	Percent Wild-Type Plaques	Percent Recombinants
$a \times b$	0.3	0.6
$a \times c$	1.0	2.0
$a \times d$	0.4	0.8
$b \times c$	0.7	1.4
$b \times d$	0.1	0.2
$c \times d$	0.6	1.2

The largest distance is between a and c ; therefore a and c must be at opposite ends. Since a - b = 0.6, b must be 0.6 units to the right of a . This position gives b - c as 1.4, the observed distance. We now have the following map

$$\frac{0.6}{a} \quad \frac{1.4}{b} \quad \frac{1.4}{c}$$

If d is to the left of b , then d - c should be greater than 1.4, a result not seen. Therefore, d is 0.2 units to the right of b .

31. $\frac{0.04}{2} \quad \frac{0.02}{1} \quad \frac{0.02}{3}$

To calculate map distance, you must have the number of recombinations and the total number of progeny. Since all phages grow on strain B, this number must equal the total number of progeny; this is 250×10^7 . Since only wild-type phages grow on K12, and since wild-types result from recombination between two genes,

$$\frac{a \quad b^+}{a^+ \quad b} \quad \frac{X}{\quad} \quad \longrightarrow \quad a \quad b \quad \text{and} \quad a^+ \quad b^+,$$

the number that grow on K12 must be recombinants. But this number represents only half of the recombinants, for the double mutant will not grow on K12. Total recombinants are:

Appendix A Brief Answers to Selected Exercises, Problems, and Critical Thinking Questions

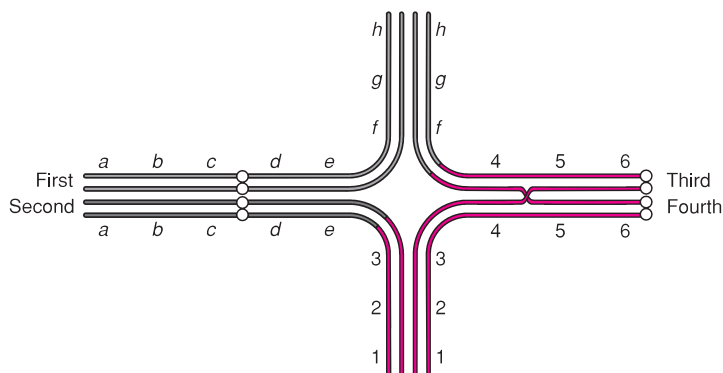
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$$\begin{array}{ll} 1 \times 2 & (2 \times 50)(10^4) = 10^6 \\ 1 \times 3 & (2 \times 25)(10^4) = 5 \times 10^5 \\ 2 \times 3 & (2 \times 75)(10^4) = 1.5 \times 10^6 \end{array}$$

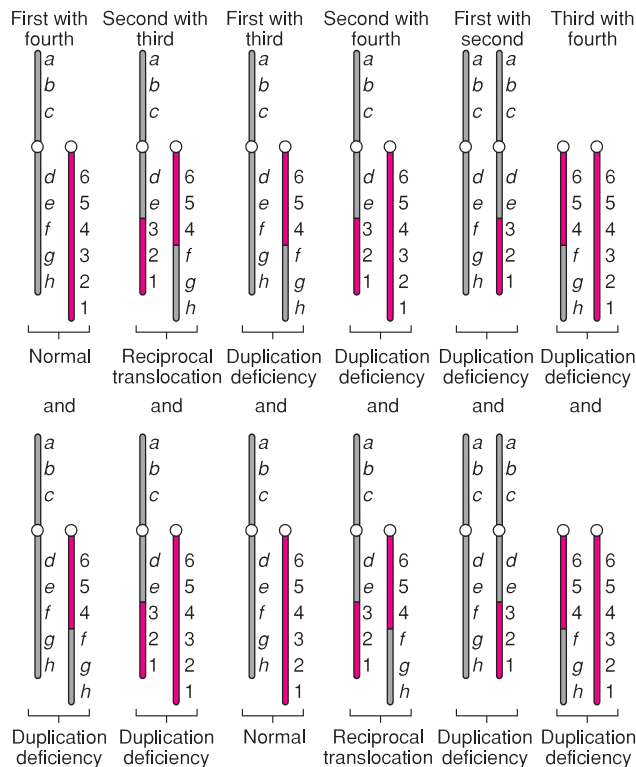
$$\begin{aligned} \text{map distance } 1-2 &= \frac{(100 \times 10^4)}{(250 \times 10^7)} \times 100 \\ &= (0.4 \times 10^{-3})(100) = 4 \times 10^{-2} = 0.04 \end{aligned}$$

$$\begin{aligned} \text{map distance } 1-3 &= \frac{(50 \times 10^4)}{(250 \times 10^7)} \times 100 \\ &= (0.2 \times 10^{-3})(100) = 2 \times 10^{-2} = 0.02 \end{aligned}$$

$$\begin{aligned} \text{map distance } 2-3 &= \frac{(150 \times 10^4)}{(250 \times 10^7)} \times 100 \\ &= (0.6 \times 10^{-3})(100) = 6 \times 10^{-2} = 0.06 \end{aligned}$$



Alternate segregation Adjacent-1 segregation Adjacent-2 segregation



33. Use replica-plating on selective media with arabinose as the sole carbon source, thus selecting for ara^+ cells. Although all three loci can be cotransduced, the rarity of $ara^+ leu^- ilvH^+$ indicates *leu* is the middle locus (*ara leu ilvH*). Cotransductance frequencies:

$$ara \text{ to } leu = (9 + 340)/(9 + 340 + 32) = 0.92$$

$$ara \text{ to } ilvH = 340/(340 + 32 + 9) = 0.89$$

35. $azi^f leu^+ thr^+$. The cotransduction frequency of *leu* and *azi* indicated *leu* is closer to *azi* than to *thr*. Thus, two orders are possible: *leu azi thr* or *azi leu thr*. If the first order is correct, and *leu^+* is selected, $leu^+ azi^f > leu^+ thr^+$; this prediction fits. However, with the same gene order, and selecting for *thr^+*, $thr^+ azi^f > thr^+ leu^+$; this result is not seen, and the order must be *azi leu thr*. The second order predicts $thr^+ leu^+ > thr^+ azi^f$.

Critical Thinking Question:

- At first, with much smaller data sets, scientists came up with branching models of the bacterial chromosome. However, with the very large data set of table 7.4, it is almost impossible to come up with a reasonable mechanism other than a circular bacterial chromosome.

Chapter 8 Cytogenetics

- All chromosomes form linear bivalents. The cross-shaped figure is seen only in heterozygotes.
- No, there are no inversion loops formed in homozygotes.
- A diagram will show that a crossover between a centromere and the center of the cross can change the consequences of the pattern of centromere separation. For example, in the figure at left, we diagram a crossover between loci 4 and 5, as in figure 8.11.
- Reciprocal translocation (some effects occur only in the heterozygous condition). Look for the cross-shaped figure at meiosis or in salivary gland chromosomes.
- Assume crossovers as shown (following page, top left).
- All females should get a wild-type X chromosome from their father. Irradiation produces chromosomal breaks, so a deletion of part of the X is possible, producing a situation of pseudodominance. (Alternatively, the offspring could have gotten a mutant X^w from the father.)
 - Diagram the crosses (a “/” represents a deleted part of the chromosome and “—” = X chromosome)

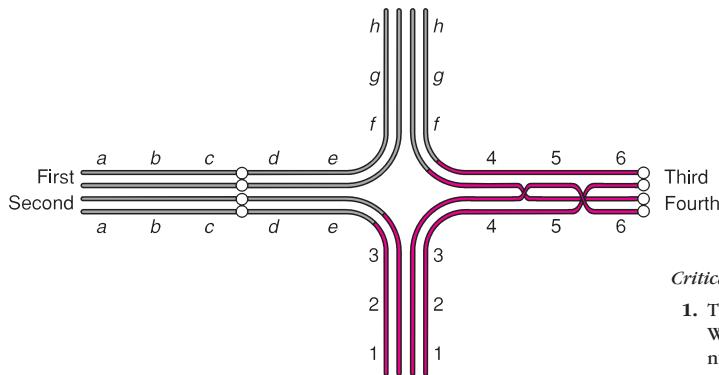
$$\begin{array}{ccc} w & \times & + \\ \text{////} & & Y \\ & \downarrow & \\ w & \text{////} & w \quad \text{////} \\ + & + & Y \quad Y \\ \text{wild-type} & \text{wild-type} & \text{white} \quad \text{dead} \end{array}$$

We expect all wild-type females and all white-eyed males, but in a ratio of 2 female:1 male.

- We see that the F_2 offspring from cross A $\times$ B yields fewer progeny than the other crosses. Something unusual must be involved. One explanation is that one of the strains is homozygous for a reciprocal translocation. The translocation, when heterozygous, results in some inviable gametes or progeny, and thus reduces the number of progeny.
- We expect to see about 32% recombination between these two genes, but we see only 2%. The most likely explanation is that an inversion occurred so that these two genes came to lie close to each other; the stocks are homozygous. Since semisterility is not reported, we are probably not dealing with crossover suppression in inversion heterozygotes.

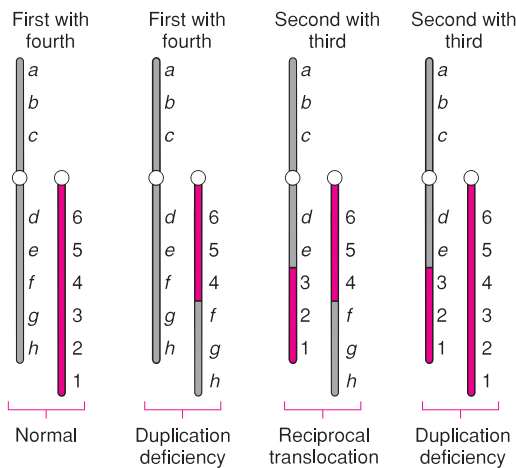
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Appendix A Brief Answers to Selected Exercises, Problems, and Critical Thinking Questions



Critical Thinking Question:

- The numbers are all multiples of 14 (1, 2, 3, 4, 5, 6, 7, and 8 copies). We can thus hypothesize that the original diploid chromosome number ($2n$) is 14. The other species would be polyploids, multiples of the original 14 (tetraploid, hexaploid, etc.). These are all the even ploids up to 112 chromosomes. As we saw, even ploids have the potential to succeed in meiosis, whereas odd ploids rarely do.

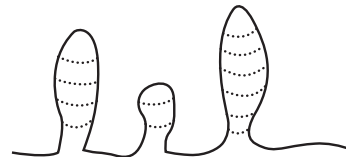


Chapter 9 Chemistry of the Gene

- The genetic code would somehow be read in number of tetranucleotide units, in which each unit consists of one each of the four bases (G, C, T, A). For example, one unit might be the amino acid alanine, two units might be the amino acid arginine, and so on.
- Sugars: DNA has deoxyribose, RNA has ribose; and bases: DNA has thymine in place of uracil, RNA has uracil in place of thymine.
- See figures 9.18 and 9.19.
- 28% G; 28% C; 22% A; 22% T.
 - Same percentages except 22% U, 0% T. Chargaff's rule states that the quantity of A = T and the quantity of G = C. If G = 28%, then C = 28% and G + C = 56%. The sum of all bases must equal 100%. Therefore, (A + T) = 100 - 56 = 44. Since A = T, $1/2(44) = 22\%$. This is the amount of both A and T. For an RNA molecule, proceed the same way, except remember that U replaces T, so we have 22% U.
- There must be regions of complementarity within the single-stranded regions. A melting temperature indicates some regions that are double-stranded. We can envision at least two different possible configurations:
 - Whole molecule complementary



- Fragment complementary



In fact, most single-stranded molecules have some regions that are complementary.

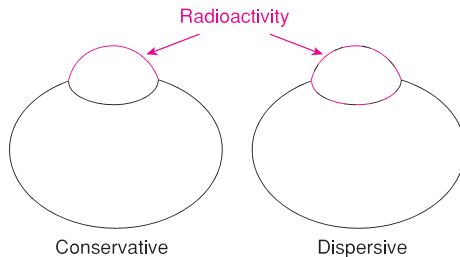
- 19.1 billion base pairs. One base takes up 3.4 Å. Since 1 Å is 1/10,000,000 millimeter, there are $10,000,000/3.4 = 2.94$ million bases per millimeter. Multiply by 1,000 to get to meters, and finally by 6.5 for 6.5 meters: $= 1.91 \times 10^{10}$ bases.

- A translocation from the tip of the normal X in the male to the Y. We expect all males to receive an X chromosome with the white-eye allele from the female. For the male to be wild-type, we still must have part of the wild-type X chromosome. To test, cross this wild-type male with white-eyed females. All the female progeny should be white-eyed and all the male progeny red-eyed. Cytological examination of the chromosomes should reveal the translocation.
- $4n = 92; 2n - 1 = 45$.
- $n = 8 + n = 6$ equals $14 \times 2 = 28; 20 + 20 = 40$
- An XO/XXY mosaic can occur by nondisjunction of the Y chromosome in a cell during early cleavage in an XY individual. An XX/XXY mosaic can come about if one of the cells during early cleavage in an XX zygote is fertilized again by a Y-bearing sperm. Trisomy 21 usually comes about from an egg with two copies of the chromosome; the egg had two copies because of meiotic nondisjunction.
- The father. The allele for color blindness can only come from the mother. If meiosis in her is normal, an egg could get the X chromosome carrying the mutant allele. The daughter has only one X chromosome, so the sex chromosomes failed to separate in the man, and a sperm with neither X nor Y fertilized the egg.
- The first meiotic division in the father is normal, producing cells with either two X or two Y chromatids. During the second meiotic division in the cell with the two Y chromatids, both Y chromatids move to the same pole and end up in the same sperm cell.

Appendix A Brief Answers to Selected Exercises, Problems, and Critical Thinking Questions

A-11

13.

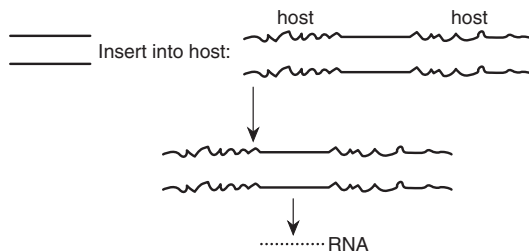
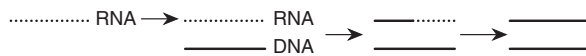


15. A primosome is a helicase plus a primase; it opens the DNA and creates RNA primers on lagging strands and is part of the replisome. A replisome includes a primosome plus two copies of DNA polymerase III; it coordinates replication on both the leading and lagging strands at the Y-junction.

17. See figure 9.28.

19. At one time molecular swivels, presumably protein in nature, located periodically along the DNA, were suggested.

21.



This question really asks how double-stranded DNA can be formed from single-stranded RNA. First, we could synthesize a complementary DNA strand, then begin to make a second DNA strand complementary to the first. While synthesizing the second DNA strand, we begin to degrade the RNA. The double-stranded DNA molecule now inserts into the host DNA. In order to get more viruses, a single-stranded RNA molecule must be made from the DNA. We would need, at a minimum, an enzyme to make a single-stranded DNA to form a hybrid with the RNA; an enzyme to degrade the RNA and to make the second, complementary DNA strand; an enzyme to cut the host DNA to allow the viral DNA to insert itself; an enzyme to ligate the two molecules; and an enzyme to make more viral RNA. The earlier functions are performed by one viral enzyme, reverse transcriptase.

23. Finding small pieces or fragments of DNA suggests the Okazaki pieces are only slowly, if at all, joined, a function of DNA ligase. The fact that not many long DNA molecules are seen also suggests that the DNA is being broken, implicating a nuclease as well.

25. It is unlikely that bases are added faster in developing embryos. So we must look for another mechanism. If there are more replicons, and hence more origins of DNA replication, each replicon will be shorter and be able to duplicate faster. Alternatively, and more likely, the process is regulated to slow down adult division.

Critical Thinking Question:

- One way to study mutations that are generally lethal is by isolating temperature-sensitive mutations. These mutations involve amino acids that disrupt the functioning of the enzymes at some critical temperature but are phenotypically normal at other temperatures. Thus, the mutant organisms can be kept alive by growing them at one temperature (the *permissive* temperature) but their mutant effect can be studied at the temperature in which the protein function is disabled (the *restrictive* temperature). (For additional discussion of these mutations, see chapter 12.)

Chapter 10 Gene Expression: Transcription

- See figure 10.3. Complementarity is achieved between messenger RNA and ribosomal RNA and between messenger RNA and transfer RNA.
- Transcription has higher error rates. Errors of DNA polymerase tend to become permanent, whereas errors of RNA polymerase do not.
- A consensus sequence is made up of the nucleotides that appear in a significant proportion of cases when similar sequences are aligned. A conserved sequence consists of nucleotides found in all cases when similar sequences are aligned. For example, the Pribnow box (fig. 10.6) is the consensus sequence TATAAT.
- See figure 10.8 for a promoter and figure 10.10 for a terminator. The transcript starting from the promoter would be 5'-CUUAUACGGU.... The transcript from the terminator is shown in figure 10.10.
- A stem-loop structure can form when a single strand of DNA or RNA has a double helical section (see fig. 10.10). An inverted repeat is a sequence read outward on both strands of a double helix from a central point (see fig. 10.10). A tandem repeat is a segment of nucleic acid repeated consecutively; that is, the same sequence repeats in the same direction on the same strand:

5'-TCCGGTCCGGTCCGG-3'

3'-AGGCCAGGCCAGGCC-5'

A DNA sequence with a seven-base inverted repeat is

5'-ATTACGCGGTAAT-3'

3'-TAATGGCGCCATTA-5'

- Footprinting is a technique in which DNA in contact with a protein is exposed to nucleases; only DNA protected by the protein is undigested. Promoters could be isolated by protection with RNA polymerase in the absence of ribonucleotides—the polymerase will not move—and then sequenced.
- The superscripts of the sigma factors refer to their molecular weights (e.g., σ^{70} is 70,000 daltons). Different sigma factors usually recognize different prokaryotic promoters.
- 3'-G G T A G T A C T G T C T G G G A A C G A T T G C G -5' ←
 5'-C C A T C A T G A C A G A C C C T T G C T A A C G C -3'

Begin by writing the strand that is complementary to the RNA. This will be the transcribed strand. Remember, U in RNA pairs with A in DNA. Since transcription proceeds 5' → 3', the 5' end of the RNA is opposite the 3' end of the DNA.

- The bottom strand is transcribed and the molecule is arranged as



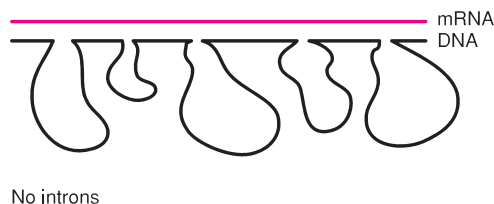
Begin by writing the RNA that could be transcribed from each strand. Since the DNA represents the beginning portion of the gene, the RNA must have an AUG to start protein synthesis. Unfortunately,

A-12

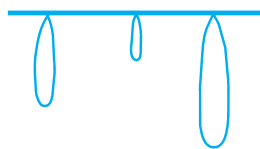
Appendix A Brief Answers to Selected Exercises, Problems, and Critical Thinking Questions

both strands yield RNAs with one or more AUGs. The RNA from the top strand has AUGs in both directions, but in each case the AUG is followed by a termination signal, UAA, or UAG. This RNA could not make a protein. The bottom strand produces an RNA with only one AUG. Since transcription and protein synthesis both proceed $5' \rightarrow 3'$, the left end of the bottom strand must be $3'$.

19. If transcription of the genes is rho-dependent, the RNAs made at 40°C will be longer than those made at 30°C . Since the rho cannot function at the high temperature, RNA polymerase will read past the termination region. If transcription is rho-independent, a rho mutant will have no effect on transcription, and hence, the size of the RNAs.
21. The double helix must unwind in order for transcription to occur. A-T pairs, because they have only two H-bonds, are more easily disrupted than G-C pairs.
23. Removing one base too many or too few would result in a shift in the reading frame during translation (see chapter 11), thus radically altering the protein product.
25. Five introns



27. Group I introns are self-splicing introns that require a guanine-containing nucleotide for splicing. Group II introns are similar but do not require an external nucleotide for splicing. Group I and II introns are released as linear and lariat-shaped molecules, respectively.
29. Spliceosomes are composed of small nuclear ribonucleoproteins (see fig. 10.37).
- 31.



There are three introns, so we expect three single-stranded DNA loops. The coding regions (exons) form RNA-DNA hybrids and appear thicker.

33. Enhancers [E] bind activator proteins that also bind proteins of the polymerase at the promoter [P]. In some eukaryotic genes, there are many enhancers, allowing numerous levels of control and forcing enhancers further and further upstream. For activators bound to enhancers to bind polymerase proteins, the DNA must loop around.

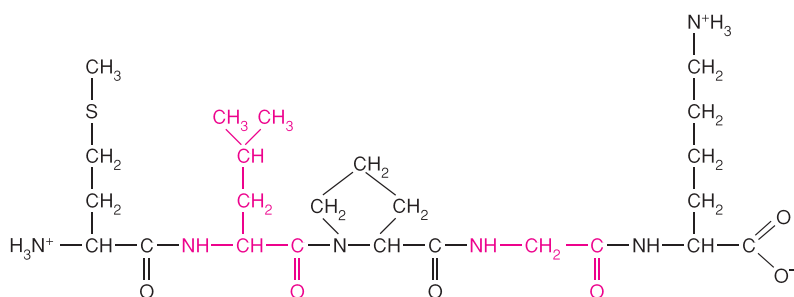


Critical Thinking Question:

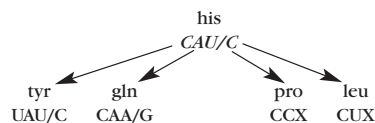
1. Given that a gene controls the production of a protein, there are both realistic and theoretical limits to the size of a gene. From what we know about the functioning of the centromere, a gene would have to occupy no more than the length of a chromosomal arm. However, given that human chromosomes must contain about fifty thousand genes, it is unlikely that any gene is that large. We also know that many functioning proteins are made up of subunits, each controlled by its own gene. Thus, large proteins tend to be conglomerates of smaller ones rather than large functional units. Finally, the larger the protein the more time it takes to transcribe and translate it, making very large size inefficient. As we mentioned before, the average protein is about 300 to 500 amino acids; with introns, and control elements, the gene for an average protein could be quite large. The largest known gene is the human dystrophin gene that codes for a cytoskeletal protein. It is 2,300,000 bases long, has 79 exons, and takes 16 hours to be transcribed.

Chapter 11 Gene Expression: Translation

1. The messenger RNA is $5'\text{-AUGUUACCGGGAAAAUAG-}3'$; the anticodons are $3'\text{-UAC-}5'$, $3'\text{-AAU-}5'$, $3'\text{-GGC-}5'$, $3'\text{-CCU-}5'$, $3'\text{-UUU-}5'$; the amino acids are methionine, leucine, proline, glycine, lysine (see the figure below).
3. See figure 11.16. Use the messenger RNA of problem 1 and be sure to include EF-Tu and EF-Ts.
5. There are approximately twenty aminoacyl-tRNA synthetases in an *E. coli* cell, one for each amino acid. Recognition signals can occur at any point on a given transfer RNA, although the anticodon figures prominently in most.
7. See figure 11.7.
9. Three; see figure 11.29.
11. $5'\text{-UAA-}3' \rightarrow 5'\text{-UUA-}3'$ (leucine). The consequence is the failure to terminate the particular protein leading to continued chain elongation to the next nonsense codon or to the end of the messenger RNA. The result is probably a nonfunctioning enzyme or protein.



13. EF-Tu brings a charged transfer RNA to the A site at the ribosome. EF-Ts is involved in recharging EF-Tu (see fig. 11.14). The eukaryotic equivalents are eEF1 α and eEF1 β .
15. A signal peptide is a sequence of amino acids at the amino-terminal end of a protein that signals that the protein should enter a membrane (see fig. 11.25). Although the concept is the same, the situation in eukaryotes is somewhat more complex because there are many different membrane-bound organelles, each having their own membrane-specific requirements. Signal peptides are usually cleaved off the protein after the protein enters or passes through the membrane.
17. NH₂-FGKICABHLNOEDJM-COOH
19. a. 5'-AUG AUU GAA UGC GAG CGG AGU-3'
b. N-met-ile-glu-cys-glu-arg-ser
- First determine the sequence of the RNA complementary to the given DNA strand. Don't forget about polarity; as the strand is written, the 5' end of the RNA will be on the left. Blocking off successive groups of three bases allows the determination of the codons. Use the code to determine the amino acid sequence.
21. 12/27 phenylalanine (UUU, [2/3]³; UUC, [2/3]²[1/3]); 6/27 serine (UCU, [2/3]²[1/3]; UCC, [2/3][1/3]²); 6/27 leucine (CUU, [2/3]²[1/3]; CUC, [2/3][1/3]²); 3/27 proline (CCU, [2/3][1/3]²; CCC, [1/3]³).
23. The table could look the same (see table 11.4) except that the position would be left side = first position (5' end); top = third position (3' end); right side = second position. For example, the codons for valine (currently 5'-GUU-3', 5'-GUC-3', 5'-GUA-3', and 5'-GUG-3') would be 5'-GUU-3', 5'-GCU-3', 5'-GAU-3', and 5'-GGU-3'.
25. We are mixing two RNA strands that are complementary; these strands will form a double-stranded RNA molecule. Since we observed the incorporation of no amino acids, the ribosome must not be able to read a double-stranded molecule.
27. If we write out (GUA)_n as GUA GUA GUA GUA ..., we see that we could use any of three different reading frames: GUA, UAG, or AGU. Since we see only two amino acids incorporated, either two of the possible codons code for the same amino acid, or one of the codons is a stop codon. If you look at the code, you will see that UAG is a stop codon.
29. The stop codon has probably mutated to give a codon for the amino acid leucine. The longer-than-normal protein suggests that the original stop was not read. Numerous possibilities exist. If the second letter of a stop codon were changed to a U, we would have UUA or UUG leucine codons. Alternatively, the insertion of a C before the U would yield CUA, CUG, or CUA as leucine codons. Similarly, an insertion of a U next to the first G yields UUA or GUG as leucine codons. Since the next amino acid is phenylalanine, the next codon must be UUC or UUU. If a base were added, as above, the next codon would have to begin with A or G, and phenylalanine does not begin with A or G. Therefore, the most likely explanation is a change of the second letter from an A to a U.
31. Either CAU or CAC. Write down all possible codons for each amino acid.



For *leu*, note that UUA/G cannot result from a single change in the *bis* codon. Therefore, leucine must be CUX. All of the other amino acids could result from single changes in either the first or second base, and we are left with either codon being the one for *bis*.

Critical Thinking Question:

- In general, transcriptional and translational signals are independent. We could look at this by asking the question, how does changing one of the signals affect the other process? In other words, if we changed a translational signal such as a start or stop codon, would that affect the transcription of that gene? In general, the answer is no.

Chapter 12 DNA: Its Mutation, Repair, and Recombination

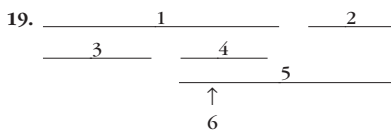
- For example, if the twenty individual cultures of table 12.1 had values of 15, 13, 15, 20, 17, 14, 21, 19, 16, 13, 27, 14, 15, 26, 12, 21, 14, 17, 12, 14, then the mutation theory would not have been supported because the variation between the individual and bulk cultures would not have been different.
 - Reading across each row, we gather more and more information.
Row 1: 1, 6, and 7 are part of one complementation group.
Row 2: 2 and 5 are part of one complementation group.
Row 3: 3 and 4 are part of one complementation group.
Row 4: no new information.
Row 5: no new information.
Row 6: reinforces that 6 and 7 are part of the same complementation group.
Row 7: no new information.
- Thus, we conclude that there are three complementation groups present: 1, 6, and 7 are mutually noncomplementing, as are 2 and 5, and 3 and 4. The half-table missing is a mirror image because a cross of 1 and 3 is the same as a cross of 3 and 1 (reciprocity). The diagonal always contains negative elements because every mutant is a functional allele of itself.
- All mutants should be crossed in pairwise combinations yielding heterozygote daughters. (Presumably, earlier crosses indicated that these are X-linked loci.) All F₁ daughters will be wild-type: The mutations complement and therefore are not alleles. When eosin flies are crossed in a similar fashion, daughters will be wild-type except when the parents were eosin and white. In that case, daughters will be mutant, showing the lack of complementation and hence that the mutations are alleles.

- Prokaryotic and phage genes generally do not have intervening sequences. Benzer and Yanofsky worked at a time when introns were unknown and it was assumed that the length of a gene was transcribed and then translated. If the genes had introns, Benzer and Yanofsky would have been unaware of them; introns would not affect colinearity or mapping. Introns would affect physical measures of the lengths of DNA, with which Benzer and Yanofsky were not involved.
- The *rex* gene of phage λ represses growth of phage T4 *rII* mutants.
- In replicating DNA, a transition mutation can occur by tautomerization of a base in the template strand (template transition) or entering the progeny strand (substrate transition).
- 5-bromouracil (pyrimidine analogue) and 2-aminopurine (purine analogue) are incorporated into DNA as thymine and adenine, respectively. However, each undergoes tautomeric shifts more frequently than the normal base. Both cause transitions. Nitrous acid also promotes transitions by converting cytosine into uracil, which acts like thymine, and adenine into hypoxanthine, which acts like guanine. Proflavin induces insertions and deletions by intercalating and buckling DNA. Ethyl ethane sulfonate removes purine rings and thus promotes transitions and transversions.

- See figure 12.25.

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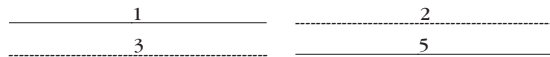
17. Three genes. Gene *A*: mutants 1, 4, 8; gene *B*: mutants 2, 5; gene *C*: mutants 6, 7. Mutant 3 probably contains a deletion that spans genes *A* and *C*. Begin by finding mutants that do not complement. These should have mutations in the same gene. Mutants 2 and 5 are in the same gene. Initially, we suspect that mutants 1, 3, 4, and 8 are in the same gene, which is different from the gene that contains 6 and 7. If mutant 3 is in gene *A*, it should complement 6 and 7, and it does not. One explanation is that 3 is a deletion spanning genes *A* and *C*. Alternatively, mutant 3 could be in gene *A* but be a polar mutation. Either possibility implies that the order is *B A C*.



Begin with deletions that yield mostly “–”s. These must be large deletions that cover most of the other deletions. Mutations 1 and 5 are such mutations. Since they give no wild-type, they must overlap:



Now look at mutant 2. It gives wild-type recombinants with 1 but not 5. Therefore it must overlap the region deleted in 5. Mutant 3, by similar logic, must cover part of deletion 1. We can draw these results as follows. Broken lines indicate we do not know yet how long the deletion is:



Now look at 4. It gives “–” (no wild-type recombinants) with both 1 and 5 and therefore must be in the region that 1 and 5 overlap. If 4 extended to and overlapped either 2 or 3, we expect to see “–” with them. Since this prediction is not met, 4 must be a small deletion spanning at least part of the overlap of 1 and 5. Since mutant 6 gives no wild-type with 1, 4, or 5, it must be within the common region deleted in all three strains.

21. We know that the anticodon pairs with the codon, and we expect nonsense suppressors to contain an altered anticodon. The fact that the nonsense codon can be read by a transfer RNA with a normal anticodon but altered dihydrouridine loop suggests that the way in which this loop interacts with the ribosome causes the anticodon sequence to be misread.
23. $x^+ \rightarrow x$: $AT \rightarrow GC$. $y^+ \rightarrow y$: $GC \rightarrow AT$. This problem requires logic and a knowledge of how mutagens work. For x , the key is the response to HA, which only causes $GC \rightarrow AT$ transitions. Mutant x is reverted by HA, therefore, x must be GC , and the normal x^+ was AT . AP-induced mutations can also be reverted by AP. Since y is not reverted by HA, y must be AT . Therefore, y^+ must be GC .
25. A deletion that spans regions of more than one gene. Consider the following two genes:



By definition, mutations in gene 1 will complement mutations in gene 2 but not other mutations in gene 1. If we have a deletion, x , that covers part of both 1 and 2 (slashes),



presumably gene 1 will be nonfunctional because it is missing the last part of the protein. Gene 2 will be nonfunctional because the

beginning portion of the gene is missing. The following genotypes will give no complementation:

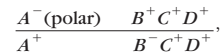


No functional product of gene 1 No functional product of gene 2

We could also get a lack of complementation when mutants are in two genes if we have a bacterial operon in which one of the genes contains a polar mutation creating a transcription stop signal. Such a mutation eliminates all distal functions. Thus, if the operon is



and we construct the partial diploid



we will get no complementation because the top DNA is effectively $A^- B^- C^- D^-$.

27. The auxotroph probably contains a deletion. If a few bases are missing, nothing is available to cause transitions or transversions. It is highly unlikely that the correct number of missing bases could be spontaneously and correctly inserted.
29. Excision repair endonucleases can recognize dimerizations, mismatched bases, and apurinic-apyrimidinic sites.
31. See figure 12.38 for a diagram of recombination. Branch migration is shown in figure 12.39.

Critical Thinking Question:

1. The gene is a linear entity that specifies the linear order of amino acids in a protein in a colinear fashion. Although scientists in the 1960s were convinced of colinearity, there were other alternatives possible. For example, DNA could be a branching structure. Or, transcription of DNA could take place such that the beginning, middle, and end of the gene were not in order. Thus colinearity supported our understanding of the shape and functioning of DNA.

Chapter 13 Genomics, Biotechnology, and Recombinant DNA

1. Type II endonucleases are valuable because they cut DNA at specific points and many leave overlapping or “sticky” ends.
3. In DNA with a random sequence, a four-cutter will find sites approximately once in 4^4 bases ($= 1/256 = 0.0039$). A six-cutter will find sites approximately once in 4^6 bases ($= 1/4096 = 0.0002$). An eight-cutter will find sites approximately once in 4^8 bases ($= 1/65,536 = 0.000015$).
5. DNA can be joined by having compatible ends to begin with or by blunt-end ligation (linkers combine these methods). The appropriateness of a method depends on what DNA is to be cloned and how that DNA can be obtained. Having DNA with “sticky” ends created by the same restriction enzyme would be easiest but sometimes is not available. Adding linkers by blunt-end ligation with a particular restriction site is usually the best compromise.
7. A plasmid is a self-replicating circle of DNA found in many cells. Foreign DNA inserted into a vector forms an expression vector if that foreign DNA produces a protein product. Cosmids are plasmids that contain *cos* sites and are useful for cloning large segments of DNA (up to 50 kb). YACs, yeast artificial chromosomes, have the loci to replicate in yeast (centromere, replication origin, and telomeres). They can be used to clone very large pieces of DNA, upwards of one million bases.

Appendix A Brief Answers to Selected Exercises, Problems, and Critical Thinking Questions

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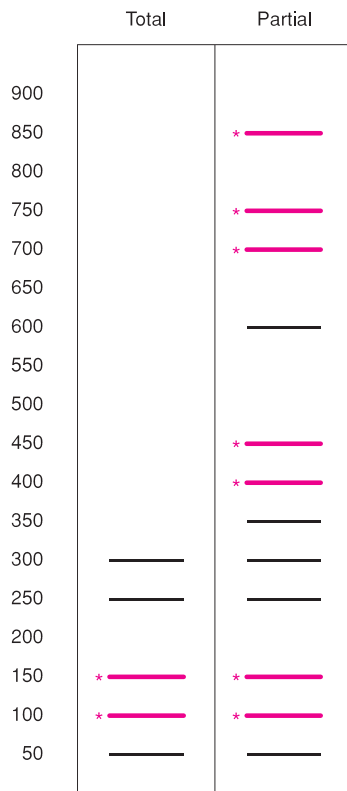
9. Chromosome walking is a technique for cloning overlapping chromosomal regions starting from an arbitrary point (see fig. 13.38). It is useful for determining relative locations of genes in uncharted regions as well as cloning regions too big to fit in a single vector.
11. Southern and northern blotting are gel transfer techniques used to probe for DNA and RNA sequences, respectively. Western blotting is a technique used for locating a protein by antibody recognition. Dot blotting is a probing technique for cloned DNA that eliminates the electrophoretic separation step.
13. Plasmids of *E. coli* origin survive in yeast when a yeast centromere (CEN region) is added, allowing them to replicate within the yeast cell. Inactivated SV40 viruses can function in the presence of intact "helper" viruses that allow them to complete their life cycles. Phage λ has parts of its chromosome that can be removed while still allowing it to complete its life cycle.

15. Partial digestion of molecule 2 leads to the following molecule:

AAAAAAAA
TTTTTTTT

Some of these molecules will form a circle with the single-stranded Ts paired with single-stranded As. The circle eliminates the free 5' phosphate, and the enzyme can no longer work.

17. We must insert DNA that has no introns into bacterial plasmids. This DNA can be obtained by isolating mature, cytoplasmic messenger RNA and then using reverse transcriptase to make double-stranded cDNA. Plasmids with cDNA inserted can then be used to produce human proteins (expression vectors).
19. Electrophoretic bands of the total digest are (* indicates end label) 50, 100*, 150*, 250, 300 bp. Bands of the partial digest are 50, 100*, 150*, 250, 300(×2), 350, 400*(×2), 450*(×2), 600, 700*, 750*, 850* bp.



21. Mutant A: elimination of site between the 300- and 50-bp segments. Mutant B: elimination of site between 100- and 300-bp segments. Mutant C: creation of a new site within the 300-bp segment, dividing it into 75- and 225-bp segments.
23. This problem begins as a trial and error attempt to overlay two restriction maps, made more difficult by the fact that one enzyme, *Bam*HI, has made three cuts that are unordered, leaving many possibilities. However, a bit of thought beforehand makes this problem much easier. If you compare the double digest with the *Bam*HI digest, they share 200-, 250-, and 400-bp segments. The double digest has 50- and 100-bp segments replacing the 150-bp segment in the *Bam*HI digest. The inference is that there is an *Eco*RI cut in the 150-bp segment leading to the 50- and 100-bp segments, with all other segments of the *Bam*HI digestion left uncut. That leaves only two possibilities, as shown below; the data are insufficient to distinguish between the two choices.

<i>Eco</i> RI				<i>Eco</i> RI			
300	↓	700		300	↓	700	
200	↑	150	↑	250	↑	150	↑
[any order]				[any order]			
<i>Bam</i> HI				<i>Bam</i> HI			

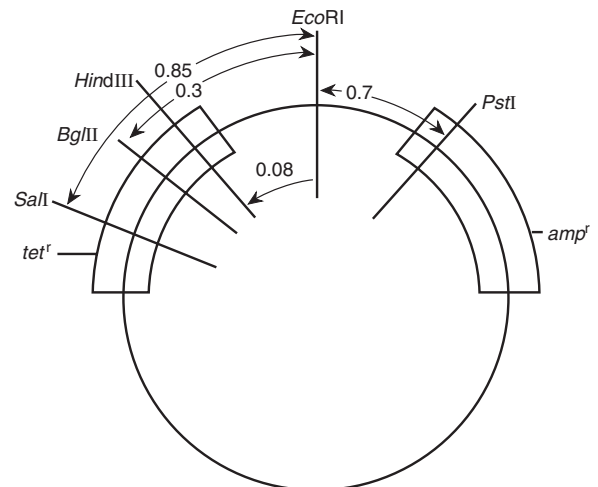
<i>Eco</i> RI	6.2	2.8	4.6	7.4	8.0
<i>Bam</i> HI	10.0		13.0		6.0

We know that the 6.2 and 8.0 kb *Eco*RI fragments are at opposite ends, and that the 10.0 and 6.0 kb *Bam*HI fragments are at the ends. Therefore, the *Bam*HI 13.0 kb fragment must be in the middle. If the 6.2 and 6.0 kb fragments are at the same end, a double digest should produce a fragment of 0.2 kb. This is not seen, so they are at opposite ends:

6.2	(2.8, 4.6, 7.4)	8.0
10.0	13.0	6.0

If 7.4 is next to 6.2, we should see a 3.8 fragment. Similarly, if 4.6 is next to 6.2, we should see a 3.8 fragment. We see neither of these fragments, so 2.8 is next to 6.2. The 4.6 fragment must be next.

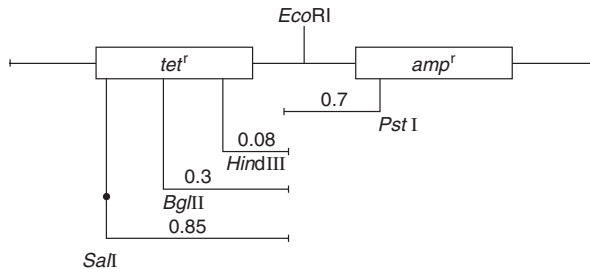
27. Since *Eco*RI does not eliminate either resistance, its site must be between the *tet*^r and *amp*^r genes. The *Pst*I site must be within the *amp*^r gene, since insertion of DNA into this site eliminates ampicillin resistance. By similar logic, the other sites must be in the *tet*^r gene. In the double digests, the smaller fragment must represent the distance from the *Eco*RI site to the other site. We can draw part of the plasmid as:



(a)

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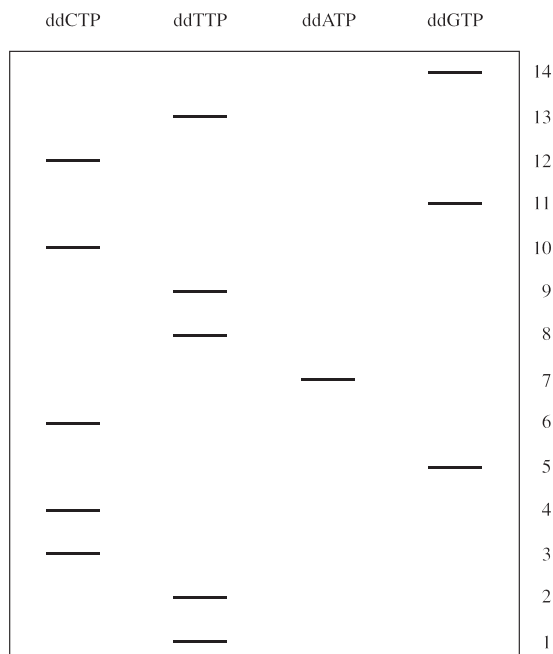
Appendix A Brief Answers to Selected Exercises, Problems, and Critical Thinking Questions



(b)

If this arrangement is correct, digestion with *BglII*, *EcoRI*, and *PstI* should yield 0.3 kb (*BglII* + *EcoRI*), 0.7 kb (*EcoRI* + *PstI*) + 2.0 kb (*PstI* + *BglII*), and this is, in fact, observed.

29. The second possibility predicts that a 1.5-kb fragment should be seen in the double digestion. This fragment would result from digestion of the 3.5-kb *EcoRI* fragment with *BglII*. Since we don't see a 1.5-kb fragment in the double digest, the second possibility does not agree with the observed results.
31. Two of the three number 21 chromosomes present in the child came from the father, not the mother. Since the probe produces different bands in the mother and the father, all of these bands must also be present in the child. The intensity of a band is proportional to the amount of DNA present. The bands that are of paternal origin are more intense than the maternal bands. The father contributed two number 21 chromosomes.
33. For the steps in the dideoxy sequencing method, see figures 13.32 and 13.33. Use of fluorescent dyes has allowed for the automation of the process and the elimination of radioactive tags.
35. The DNA can be inserted into the M13 general sequencing vector.
37. What will appear in the gel are fragments of the newly synthesized strand. Since DNA synthesis proceeds 5' → 3', the 5' base will be T in the new strand. Proceed up the gel by indicating the base complementary to the sequence given.



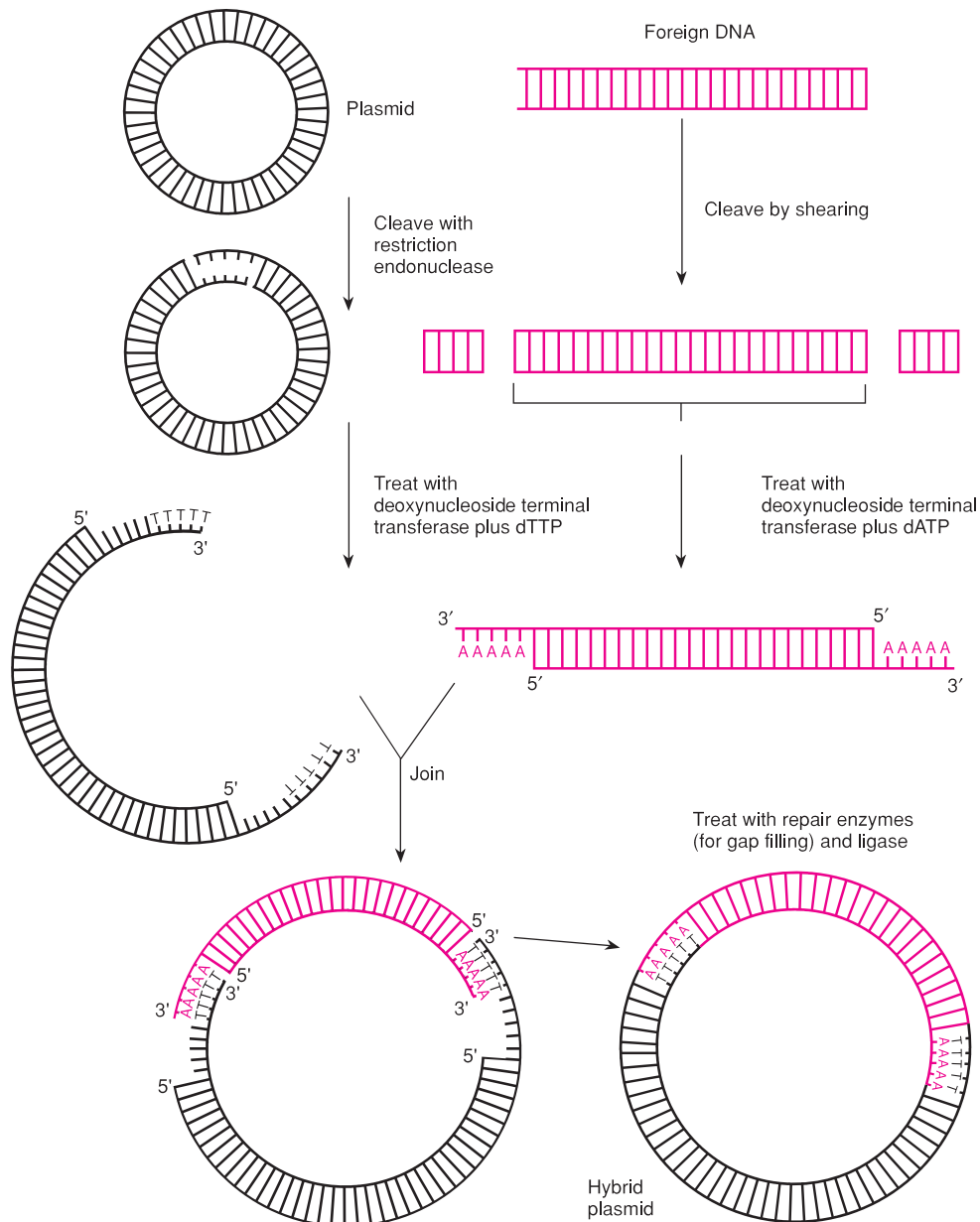
39. Hypervariable DNA is DNA showing a great deal of interindividual variation. A RFLP (restriction fragment length polymorphism) is a polymorphism (variation) that shows up after Southern blotting and probing of restriction digests. A VNTR (variable number of tandem repeats) locus is one that is hypervariable due to unequal crossing over among the tandem repeats. VNTR loci are a hypervariable subset of RFLPs. Sequence-tagged sites are unique sites in the genome that can be amplified with polymerase chain reaction. Microsatellite DNA, repeats of very short segments such as CA, forms VNTR loci that are usually examined by polymerase chain reaction (PCR) if primer sequences are known.

Critical Thinking Question:

1. Sticky ends would exist if the plasmid DNA had a 3' overhang of one nucleotide residue, while the foreign DNA had a 3' overhang of its complement. The exact number of bases would not need to be the same because repair enzymes plus ligase could close the gap. For example, the plasmid could have a 3' tail of thymines added, whereas the foreign DNA could have a 3' tail of adenines added. This method (see figure next page) was called the poly-dA/poly-dT technique.

Chapter 14 Gene Expression: Control in Prokaryotes
and Phages

1. a. inducible (wild-type); b–d. constitutive; e. neither, super-repressed; f. inducible
3. One mutant could fail to bind to operator DNA but could still bind the inducer (*op⁻, in⁺*). This mutant would have constitutive transcription of the operon. The reverse situation could also be true; the repressor could bind the operator but not the inducer (*op⁺, in⁻*). This mutant would be off all the time. A third mutant could fail to bind both (*op⁻, in⁻*), being constitutive.
5. a. Operator and repressor. b. Make a partial diploid with wild-type; the operator mutant will make β -galactosidase constitutively, and the repressor mutant will make it only in the presence of lactose. Four mutations are possible in the *lac* operon: mutations in the *z* gene or the promoter never make the enzyme. Mutations in the repressor always make the enzyme because the repressor cannot bind DNA. In operator mutations, a good repressor can never bind DNA. In a partial diploid, *i⁻ o⁺/i⁺ o⁺*, the wild-type repressor is *trans* acting and can bind to both operators, creating an inducible situation. In *i⁺ o⁻/i⁺ o⁺*, repressor cannot bind to *o⁻*, and this DNA is always on, even though the wild-type DNA is off in the absence of lactose.
7. Cyclic AMP, combined with CAP protein, attaches to CAP sites enhancing transcription of nonglucose, sugar-metabolizing operons in *E. coli*. Glucose inhibits its formation by inhibiting adenylcyclase.
9. We must think about how these operons are controlled. Not only do they need inducer, but they also require the catabolite repression-activation system. These mutants could be unable to make cAMP because the adenylcyclase gene is defective. Alternatively, they could be making a defective catabolite activating protein (CAP).
11. b = tryptophan synthetase gene, a = operator, c = repressor. Look first for the single mutation that never gives enzyme; this genotype will give the letter of the structural gene. The genotype *a⁺ b⁻ c⁺* fits this requirement, so b is the structural gene, and a and c represent control regions. Genotypes 4 and 5 tell us nothing. Look at genotype 6. The right DNA will never make the enzyme. If c is the operator, the left DNA should always make the enzyme, and this is not seen. Therefore, a must be the operator. Check these assignments with genotype 7. The right DNA will never make the enzyme. If a is the operator, the left DNA is always on. If a is the repressor, the right DNA makes a good repressor that will bind to the top DNA and regulate it.



13. The *E. coli trp* operon functions as a normal repressible operon. In addition, attenuation, based on secondary structure and stalling of the ribosome on the leader transcript, can further prevent transcription (see fig. 14.16). Attenuator control can be exerted based on other amino acids if their codons appear in the leader transcript, causing ribosome stalling.
15. Assuming that the mutants produce inactive proteins: *cl*, *cII*, and *cIII*, lytic response; *N*, neither lytic nor lysogenic responses possible; *cro*, no lytic response possible; *att*, no lysogenic response possible; *Q*, no lytic response possible.
17. The λ chromosome has one circular and two linear forms. The circular form is the infective cellular form. A break at one point (*cos* site) takes place during packaging into the phage head, and

a break at another point forms the linear integrative prophage (see fig. 14.18).

19. The prophage region of the Hfr chromosome enters the F^- cell with no repressor present. The situation is thus similar to regular phage infection, which can go either way (lysogenic or lytic cycles).
21. The cells that form colonies do not contain a prophage. The initial heat shock inactivated the repressor and allowed the prophage to excise. The lower temperature activated the repressor, and the repressor bound to the excised phage DNA, preventing gene expression and reintegration of some of the phages. These "cytoplasmic" phages failed to replicate and were lost during cell division. The cells that retained the virus were lysed when the temperature was raised because the phage could now make RNA.

A-18 *Appendix A Brief Answers to Selected Exercises, Problems, and Critical Thinking Questions*

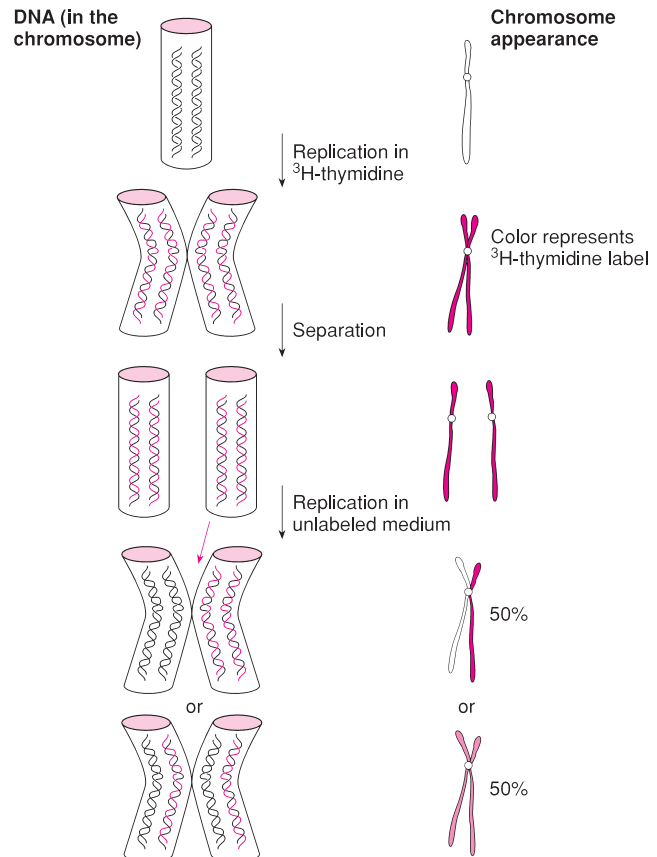
23. An IS element is a simple transposon, which is a segment of DNA that can make a copy of itself to be inserted at another place in the genome. An intron is an intervening sequence, a region excised from messenger RNA before expression. A plasmid is an autonomous, self-replicating genetic particle. A cointegrate is an intermediate structure in transposition.
25. See figure 14.29.
27. See figure 14.35.
29. Transcription is the level at which most control mechanisms work. These include sigma factors, efficiency of promoter recognition, catabolite repression, operon-repressor systems, attenuation, and transposition. Translational control mechanisms include polarity placement, antisense RNA, differences in the efficiency of processes due to nucleotide sequence differences, codon preference, and the stringent response. Posttranslational mechanisms include feedback inhibition and differential rates of protein degradation.
31. Heat shock proteins are normally induced by the presence of a specific sigma factor, which itself is induced by heat shock.
33. The stringent response is the response of a prokaryotic cell to amino acid starvation. The idling reaction of the ribosome results in production of 3'-ppGpp-5', whose appearance is associated with the cessation of transcription, especially of transfer RNAs and ribosomal RNAs, through an unknown mechanism.
35. Feedback inhibition: allosteric enzymes in some synthetic pathways can be inhibited by the end product of that pathway. In addition, many repressor proteins are allosteric.

Critical Thinking Question:

- One might think that a bacterium should metabolize any sugar in its environment. However, glucose is the most efficient sugar to metabolize and if it is present, it should be metabolized first. Even under maximal growth, it takes an *E. coli* cell about 20 minutes to grow and divide. If the cell were to begin to take up and metabolize other sugars, there would be a cost for the manufacture of new enzymes and the inefficiency of the initial steps of the sugar metabolism. In fact, the growth of the cell would slow down under these circumstances and be at an evolutionary disadvantage.

Chapter 15 The Eukaryotic Chromosome

- In general, prokaryotes are small, have a relatively small circular chromosome, and have little internal cellular structure compared to eukaryotes. Most prokaryotic messenger RNAs are polycistronic, under operon control; eukaryotic messenger RNAs are highly processed, monocistronic, and usually not under operon control. Prokaryotes are mostly single-celled organisms, whereas eukaryotes are mostly multicellular. Eukaryotes have repetitive DNA, absent for the most part in prokaryotes. Prokaryotic chromosomes are not complexed with protein to anywhere near the same extent that eukaryotic chromosomes are.
- Assume that each chromosome contained two complete copies of the same DNA. Following the protocol of figure 15.1, the final results would be chromosomes, before separation, that consisted of either two labeled chromatids or only one labeled chromatid, in a 1:1 ratio (barring sister chromatid exchanges). The labeled chromatid in chromosomes with just one chromatid labeled will have twice the label of each chromatid in the chromosomes in which both chromatids are labeled (see the following figure).



- The length of DNA associated with nucleosomes was determined by footprinting, in which free DNA was digested, leaving only those segments protected by nucleosomes. Nucleosome hypersensitive sites are sites not in a nucleosomal state; they seem to be sites involved in the initiation of replication, transcription, and other DNA activities.
- See figures 15.10 and 15.11 for the relationship of the 110, 300, and 2,400 Å chromosome fibers.
- See figure 15.19.
- Polytene chromosomes are chromosomes that underwent endomitosis: They consist of numerous copies of the same chromatid (e.g., in the *Drosophila* salivary glands). Regions of active transcription in polytene chromosomes form diffuse areas called puffs or Balbiani rings (see fig. 15.14). Lampbrush chromosomes occur in amphibian oocytes (see fig. 15.18).
- Satellite DNA differs in its base sequence from the main quantity of DNA and thus forms a satellite band during buoyant density analysis. It is usually centromeric heterochromatin, composed of a highly repetitive DNA.
- Telomeres are repetitive DNA sequences at chromosomal ends. They are repetitions of a five- to eight-base sequence. Most telomeres are G-rich. Telomeres protect the ends of chromosomes and probably provide signals on senescence of cells.
- Highly repetitive DNA usually makes up the centromeric and telomeric regions of the chromosome. Unique DNA, making up the bulk of structural genes, has a large component that is transcribed. Repetitive DNA is composed of dispersed DNA (e.g., short and long interspersed elements—SINES and LINES), multiple copies of

transcribed DNA (e.g., ribosomal RNA, histones), and diverged copies of ancestral genes (e.g., globin family genes).

19. The most direct method of determining the direction of transcription of the histone genes would be to clone and sequence the region, from which transcriptional information can be ascertained.
21. Cloning and then sequencing the region would provide the answer. Analysis would show genes of similar sequence to the active genes but lacking the sequences for transcription.
23. It is very rich in A-T sequences. Density is proportional to G-C content. Since the molecule has a low density, it has low G-C, and therefore high A-T.
25. Spaces between the nucleosomes must contain many promoter sequences. For the DNA to be digested, it must be unprotected. Since we see little transcription, the promoters must be missing and must have been destroyed by the nucleases.
27. Highly repetitive DNA must be located in these regions. Since most highly repetitive DNA is not transcribed, the results suggest that centromeric and telomeric regions are not transcribed.
29. The C-value paradox involves the issues of the excessive amounts of DNA in eukaryotic cells and the difference between eukaryotic species that seem to have similar complexity. It is explained by the large amount of structural DNA in chromosomes as well as the large amounts of short and long interspersed elements (SINEs and LINEs).

Critical Thinking Question:

1. Comparative DNA studies can be helpful in understanding the roles of the various types of DNA in the eukaryotic chromosomes if there are cases in which there are remarkably large differences in the amount of DNA in similar species. It can then be inferred that the basic developmental plan of an organism is contained in the one with the lower amount of DNA, and the extra DNA in the species with more DNA may be superfluous. We do have cases in which amphibians differ by as much as one hundred times the amount of DNA found in similar species. The puffer fish has only one-sixth the amount of DNA as other higher eukaryotes.

Chapter 16 Gene Expression: Control in Eukaryotes

1. See figure 16.5.
3. Since 5-azacytidine prevents methylation, the observed increase in transcription suggests that the presence of methyl groups inhibits transcription.
5. Genomic equivalence means that all of the cells of a multicellular eukaryotic organism are genetically identical. Yet, cells in different tissues and different regions of the organism are phenotypically different, expressing different suites of genes. Explaining differences in gene expression among cells that are genetically identical is a major question of eukaryotic genetics.
7. The three classes of segmentation genes in *Drosophila* are gap, pair-rule, and segment polarity. Mutations in gap genes leave gaps of missing segments. Mutations in pair-rule genes leave gaps of even or odd sets of segments. Mutations of segment polarity genes cause changes in all segments, generally the change in anterior or posterior portions of each.
9. In the development of the early *Drosophila* embryo, a syncytial blastoderm stage is achieved after thirteen cell divisions. The nuclei are near the surface of the embryo but not surrounded by cell membranes. Thereafter, membranes form, creating a cellular blastoderm.
11. Maternal-effect genes determine four regions of the developing embryo (major gene in parentheses): anterior (*bicoid*), posterior (*nanos*), dorso-ventral (*Toll*), and terminal (*torso*).
13. The helix-turn-helix motif (see box 16.1, fig. 1) consists of two alpha helices separated by a short turn within the protein, providing the structure to interact with DNA. Two other motifs are the zinc finger and the leucine zipper. Another motif, a combination of helix-turn-helix and leucine zipper, is shown in box 16.1, figure 4.
15. Amphibians have very large eggs, development is external to the female, a ready supply of zygotes is available, and they are easily manipulated experimentally.
17. Assuming that each cancer might be controlled by a single locus and assuming that breast cancer appears only in women and prostate cancer appears only in men (sex limited), pancreatic and prostate cancer are probably controlled by autosomal recessive genes; colon cancer is probably controlled by a dominant gene (autosomal or sex linked); and breast cancer by a recessive gene, either autosomal or sex linked.
19. The protein product of the retinoblastoma gene, p105, binds with oncogene proteins. Thus, the protein may somehow suppress transformation; when bound by oncogene proteins, p105 may be rendered ineffective. Hence, p105 seems to act to suppress transformation and thus the gene is called an anti-oncogene.
21. Animal viruses can have DNA or RNA, either single- or double-stranded. They can be enveloped or nonenveloped. They can have simple or complex protein coats.
23. The following are translation mechanisms: normal translation; read-through translation; and splice and then translation.
25. The v and c refer to viral and cellular, respectively. A proto-oncogene is a cellular oncogene within a nontransformed cell.
27. The *v-src* gene has no introns and the virus can function without the gene.
29. We see one band that is common to both cell lines; this band must represent the normal oncogene. The fact that this band is present in both lines indicates that the insertion of a virus has occurred in only one of the two copies of the gene present in the clone 1 cell. If it had inserted within both genes, we should not have seen the normal band. We see a larger fragment in clone 1, indicating that the DNA of the virus does not contain a site for the restriction enzyme used and that the virus has inserted within the restriction sites that define the band, lengthening the region probed. Alternatively, the virus could contain a restriction site and has still inserted in such a way as to lengthen the band probed by inserting between the sequence probed and the original restriction site.
31. The components of an immunoglobulin light chain are V, J, and C regions; the components of an immunoglobulin heavy chain are V, D, J, and C regions.
33. The V-J joining recognition signal is a heptamer and nonamer separated by twenty-three and twelve base pairs; see figure 16.35.
35. A T-cell receptor is an immunoglobulinlike molecule located on the surfaces of T cells, enabling them to identify infected host cells.
37. One explanation is a defect in the maturation process of B cells. Another explanation is a defect in the process of V(D)J joining, which could involve five or more genes.
39. The simplest interpretation of these results is that something is different in the organization of antibody genes in embryonic cells and in B lymphocytes. If the genes were in the same place in both cases, we should have seen identical patterns for the two types of cells. The probe recognizes both variable and constant regions of the antibody gene, since it is made from the mature mRNA. In the B lymphocyte, the variable and constant regions are adjacent, but in the embryonic cells, there is some extra DNA between these two genes. This result led to the notion that variable genes are rearranged during the development of the immune system.

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Appendix A Brief Answers to Selected Exercises, Problems, and Critical Thinking Questions

Critical Thinking Question:

- Adenovirus attacks normal cells by binding to p53; it also attacks cancerous cells that lack p53. If we remove the gene for the protein that binds p53, the *E1B* gene, then the modified adenovirus will not be able to attack normal cells but will be able to attack cancerous ones lacking p53. This modification of adenovirus as a potential tool in treating cancer was published in 1996.

Chapter 17 Non-Mendelian Inheritance

- Persistence of an environmentally induced trait into later generations is known as dauermodification and does not imply genetic control. After a suitable number of generations, the phenotype returns to normal, indicating an environmental rather than genetic response.
- Maroon-like must affect the cytoplasm of the egg. The first cross is unusual and alerts us to maternal inheritance. A true Mendelian factor should produce one-half maroon-like males and females. The genotypes of the F_1 females are $ma-1^+/ma-1$ and $ma-1/ma-1$; half of the females should be of each genotype. If the wild-type allele produces wild-type cytoplasm for the progeny, regardless of the genotype of the progeny, any female that is $ma-1^+/ma-1$ will produce all wild-type progeny.
- Although the genetic scheme predicts shell coiling perfectly, one could do experiments involving the injection of cytoplasm into eggs to test the viral hypothesis.
- Female parent: *Dd*. Male parent: *d-*. (A): *dd*. Since selfing produces only sinistral snails, individual (A) must be homozygous for sinistral coiling, *dd*. The individual must get one *d* allele from each parent, so each parent must be at least heterozygous. Since (A) has dextral coiling, the mother must be heterozygous. The father could be either *D/d* or *d/d*.
- By looking at different species, it is clear that few genes for oxidative phosphorylation are found in all mitochondrial genomes.
- The rule of thumb is that suppressive petite mitochondria will dominate a cell, whereas neutral petite mitochondria will be lost in a competitive situation. Therefore
 segregational petite $\times$ segregational petite $\rightarrow$ segregational petites
 segregational petite $\times$ neutral petite $\rightarrow$ segregational petites
 segregational petite $\times$ suppressive petite $\rightarrow$ suppressive petites
 neutral petite $\times$ neutral petite $\rightarrow$ neutral petites
 neutral petite $\times$ suppressive petite $\rightarrow$ suppressive petites
 suppressive petite $\times$ suppressive petite $\rightarrow$ suppressive petites
- In 0.02% of the offspring cells, the mt^- allele of the streptomycin locus is inherited. In essence, these cells seem to have inherited chloroplast genes from the mt^- parent. These cells thus provide us with a window on the possibility of having chloroplast genotypes from both parents viable within the same cell. Thus, interaction among other chloroplast genes can be looked for in this class of offspring (0.02% of total). If recombination occurs, map distances can be calculated by the usual methods, keeping in mind that we are taking data only from within this 0.02% of offspring.
- Mitochondria and chloroplasts have prokaryotic affinities. They both have circular chromosomes, and their metabolism is affected by prokaryotic inhibitors (e.g., antibiotics). Certain prokaryotic messenger RNAs will hybridize with the organelle's DNA. There are numerous other aspects of biochemistry, morphology, and physiology that help to demonstrate affinities.
- One way to determine that two loci are involved is to look at the proportion of offspring that lose mu particles after autogamy. In some strains, one-half the offspring will lose mu particles, indicating one locus was initially segregating (autogamy of $M_1m_1M_2m_2$

yields $M_1M_1m_2m_2$ or $m_1m_1m_2m_2$ in a 1:1 ratio). In other strains, one-fourth of the offspring will lose mu after autogamy, indicating that two unlinked loci were segregating (autogamy in $M_1m_1M_2m_2$ yields $M_1M_1M_2M_2$, $M_1M_1m_2m_2$, $m_1m_1M_2M_2$, or $m_1m_1m_2m_2$ in a 1:1:1:1 ratio).

- Human mitochondrial DNA does not have introns. Finding an intron would suggest that the mitochondria had acquired a nuclear gene.
- a. All type 1, 1.5 and 3.7 kilobases. b. All type 2, 2.5 and 6.0 kilobases. Recall that the chloroplast DNA from mt^- cells does not appear in progeny.
- Conjugation produces exconjugants with the same genotype, but which haploid micronucleus survives in each cell is random. Therefore, one-fourth of the time, the genotypes of the exconjugants are expected to be *KK*, one-half of the time *Kk*, and one-fourth of the time *kk*. The sensitive exconjugant remains sensitive, regardless of which of the genotypes is present. Autogamy does not affect the genotype of homozygous exconjugants, but it does affect the heterozygote. Among the killer exconjugants we expect:

Killer Exconjugants	Autogamous Products	Phenotypic Ratio
1/4 <i>KK</i>	all <i>KK</i>	1/4 killer
1/2 <i>Kk</i>	1/2 <i>KK</i> 1/2 <i>kk</i>	1/4 killer 1/4 sensitive (Kappa are lost)
1/4 <i>kk</i>	all <i>kk</i>	1/4 sensitive (Kappa are lost)

- Use the striped plant as the egg parent, and get pollen from a plant with the following genotype: *IjIj jj*. If the striped plant is *iojap (ijij JJ)*, all progeny will be heterozygous for both genes and will also contain *iojap* cytoplasm. The F_1 plants will segregate green, striped, and white sections within the plant. If the original plant is *IjIj jj* (and thus *japonica*), all F_1 plants will have striped leaves.
- a. 2 normal:2 petite b. 0 normal:4 petite c. 4 normal:0 petite. A nuclear gene should segregate 2:2 for each allele. Cytoplasmic factors will produce four spores with identical cytoplasm.

Critical Thinking Question:

- We believe there are two mechanisms to ensure the distribution of cellular organelles during cytokinesis: stochastic and ordered inheritance. Stochastic inheritance simply means that no real mechanism exists; rather, the cell depends on the large number of the organelles to ensure an even distribution during the dividing of the cell. Ordered inheritance requires the even distribution of organelles in small numbers. This can be accomplished by special structures that divide a large organelle (e.g., a single, large chloroplast), or by other mechanisms that insert part of the mitochondrial system into new buds in budding yeast.

Chapter 18 Quantitative Inheritance

- Three in two hundred is approximately 1 in $64 = 1/(4)^3$; therefore, three loci (see table 18.1). Each effective allele contributes about 1/2 pound over the 2-pound base (3-pound difference divided by six effective alleles: $AA BB CC = 5$ pounds, $aa bb cc = 2$ pounds).
- Independent assortment: for example, $Aa Bb Cc Dd$ parents can have $AA BB CC DD$ offspring.
- Individuals of intermediate color can produce both lighter and darker offspring by independent assortment. That is, $Aa Bb Cc Dd$

parents can produce $AA\ BB\ CC\ DD$ and $aa\ bb\ cc\ dd$ offspring. However, if each effective allele adds color, then individuals with the base color (white) who mate with each other ($aabbccdd$) presumably cannot have children with darker skin.

7. The marker stock is exposed to DDT over many generations, selecting for DDT resistance. It is then crossed with the wild-type and F_1 offspring are backcrossed to generate flies with various combinations of selected chromosomes. These flies are then tested for their DDT resistance. For example:

$P_1\ Cy/Pm\ H/S\ Ce/M(4) \times +$ (wild-type)
 F_1 (male) $Cy/+ \ H/+ \ Ce/+ \times$ (backcross) $+ \ (wild-type\ female)$
 $F_2\ Cy/+ \ +/+ \ +/+ \ +/+ \ H/+ \ +/+ \ +/+ \ +/+ \ Ce/+ \ +/+ \ M(4)/M(4)$, etc.

or,

F_1 (male) $Cy/+ \ H/+ \ Ce/+ \times$ (backcross) $Cy/Pm\ H/S\ Ce/M(4)$
 (female)
 $F_2\ Cy/Pm\ H/+ \ Ce/+ \ +/Pm \ +/S \ +/M(4)$; etc.

9. 50 cm. The difference between the two heights is 20 cm. This difference must result from the presence of effective (uppercase) alleles. The tall plant has four effective alleles, so each effective allele contributes an average of 5 cm to the height of the plant. The heterozygote has two effective alleles; $2 \times 5 = 10$ cm above the base of 40 cm, or a total of 50 cm.
11. Eight. The frequency of individuals in the F_2 that resemble one parent is $1/4^n$, where n = the number of genes involved. In this case, 100 of 6,200,000 were like one parent. $100/6,200,000$ is approximately $1/64,000$, which approximates $1/4^8$. So we probably have eight genes involved.
13. $r = 0.43$; $H_N = 0.43/0.50 = 0.86$ (narrow-sense heritability); environmental variance, a component of the total phenotypic variance appears in the denominator of the heritability equations (18.11 and 18.12). Thus, environmental factors that lower environmental variance (more uniform environments or environmental effects) increase heritability. And, factors that raise the environmental variance (less uniform environments or environmental effects) decrease heritability.
15. $H = (4 - 0.9)/(5 + 5) = 0.31$; H (high line) $= (4 - 3)/5 = 0.2$; H (low line) $= (3 - 0.9)/5 = 0.42$. Part of the difference may be due to the number of alleles available for selection in each direction and nonadditive factors.
17. First, "outstanding athletic ability" must be defined. It can be defined subjectively by accomplishment or more objectively with a physiological measure. Then genetic effects must be assessed through heritability analyses such as twin studies and correlations among relatives.
19. Uniformly good nutrition should increase the heritability of height by eliminating some of the environmental variance; it affects only the denominator in a heritability equation.
21. Thorax length: $V_D + V_I = (100 - [43 + 51]) = 6$; $H_N = 43/100 = 0.43$; $H_B = 49/100 = 0.49$. Eggs laid: $V_D + V_I = 44$; $H_N = 0.18$; $H_B = 0.62$.
23. 4.5 g.

$$H_N = \frac{\text{gain}}{\text{selection differential}}$$

Thus (H_N) (selection differential) = gain. Since $H_N = 0.5$ and selective differential = 9, $(0.5)(9.0) = 4.5$ g.

25. 176 pounds. To solve this problem, use the formula for realized heritability:

$$H = \frac{\text{gain}}{\text{selection differential}} = \frac{(Y_0 - \bar{Y})}{(Y_p - \bar{Y})}$$

$$0.4 = \frac{(Y_0 - \bar{Y})}{(Y_p - \bar{Y})} = \frac{Y_0 - \bar{Y}}{185 - 170}$$

$$Y_0 - \bar{Y} = (0.4)(15) = 6.0$$

$$6.0 = Y_0 - 170$$

$$Y_0 = 176 \text{ lbs}$$

Critical Thinking Question:

1. The simplest cause for retraction of a study is the failure of that study to be replicated by others. That would come about when the conclusion isn't generally supportable, a phenomenon that could have two major causes. First, the study might have been done well but the phenomenon was specific to that study, due possibly to small sample sizes or a "private mutation," an effect found in one family or a small group of individuals but not a general phenomenon. Second, there could have been an inadvertent error in the study. For example, a size difference in a small region of the brain of homosexual and heterosexual males was reported but has not been verified. One investigator, who is following up the study, suggested that the difference could be an effect of differences in the way the brains were preserved and thus represent no real (biological) effect.

Chapter 19 Population Genetics: The Hardy-Weinberg Equilibrium and Mating Systems

1. The frequencies of the three genotypes are $f(MM) = 41/100 = 0.41$; $f(MN) = 38/100 = 0.38$; $f(NN) = 21/100 = 0.21$. The frequency of M , p , is the frequency of MM homozygotes plus half the frequency of heterozygotes:

$$p = f(M) + (1/2)f(MN) = 0.41 + (1/2)(0.38) = 0.41 + 0.19 = 0.60$$

$$q = 1 - p = 1 - 0.60 = 0.40$$

Alternatively

$$p = f(M) = \frac{2(\#MM) + \#MN}{2 \times \text{total}} = \frac{2(41) + 38}{200}$$

$$= \frac{120}{200} = 0.60$$

$$q = 1 - p = 0.40$$

We do the following chi-square test:

	<i>MM</i>	<i>MN</i>	<i>NN</i>	Total
Observed	41	38	21	100
Expected	$p^2 \times 100$ 36	$2pq \times 100$ 48	$q^2 \times 100$ 16	100
Chi-square	0.694	2.083	1.563	4.340

The critical chi-square (0.05, one degree of freedom) = 3.841. We thus reject the null hypothesis that this population is in Hardy-Weinberg proportions.

3. Here we must assume Hardy-Weinberg equilibrium because of dominance. The $f(tt) = 65/215 = 0.302$. Thus $q = f(t) = \sqrt{f(tt)} = \sqrt{0.302} = 0.55$; and $p = f(T) = 1 - 0.55 = 0.45$. Since there are zero degrees of freedom (number of phenotypes - number of alleles = $2 - 2 = 0$), we cannot do a chi-square test

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to determine if the population is mating at random (in Hardy-Weinberg proportions).

5. **a.** Most human populations should be in Hardy-Weinberg proportions at the taster locus regardless of what the allelic frequencies are. 3:1 is a family ratio when the parents are heterozygotes or a population ratio when $p = q = 0.5$, given dominance.
- b.** With no other information, it is probably safest to assume $p = q = 0.5$. At equal frequencies of alleles, the dominant phenotype (tasting) occurs in 75% of people ($p^2 + 2pq$).
7. $p = 0.99$, $q = 0.01$. If the population is in equilibrium, there should be p^2 of AA + $2pq$ of Aa + q^2 of aa individuals. Since 1/10,000 shows the recessive trait, this is q^2 . Therefore,

$$q = \sqrt{1/10,000} = \sqrt{0.0001} = 0.01$$

Since $p + q = 1$, $p = 1 - 0.01 = 0.99$.

9. The expected frequency of brown-eyed individuals will depend on the allelic frequencies of the original population. If we assume that mating is random with respect to eye color, and there is no selection, allelic frequencies will not change with time. We can, for example, calculate the frequencies of brown-eyed individuals for two populations at equilibrium. Let p = frequency of the brown-eye allele and q = frequency of the blue-eye allele.

Population	p	q	Frequency of brown ($p^2 + 2pq$)
1.	0.7	0.3	0.91
2.	0.5	0.5	0.75

We see that the original premise will be met only if the alleles are equally frequent.

11. 0.187 MM , 0.491 MN , 0.321 NN . The next generation will achieve equilibrium and there will be $(0.43)^2 MM + 2(0.43)(0.57)MN + (0.57)^2 NN$.
13. $f(A) = 0.792$; $f(B) = 0.208$. The easiest way to calculate frequencies is to do it empirically. We have three hundred people, so we have six hundred alleles.

$$f(A) = \frac{2 \times 200(AA) + 75(AB)}{600} = \frac{475}{600} = 0.792$$

$$f(B) = \frac{2 \times 25(BB) + 75(AB)}{600} = \frac{125}{600} = 0.208$$

15. **a.** Hardy-Weinberg proportions are achieved in one generation of random mating (multiple-allelic extension) if the locus is autosomal. If the locus is autosomal but there are different frequencies in the two sexes, then Hardy-Weinberg proportions are achieved in two generations. If the locus is sex linked, with different initial frequencies in the two sexes, then approach to equilibrium is gradual.
- b.** If the loci are not in equilibrium to begin with (linkage disequilibrium), then equilibrium is achieved asymptotically.
17. 0.63 type A, 0.08 type B, 0.28 type AB, 0.01 type O. Since the population is in equilibrium, the genotypic frequencies can be calculated as $(p + q + r)^2 = p^2 + 2pq + q^2 + 2pr + 2qr + r^2$. Let $p = 0.7$, $q = 0.2$, and $r = 0.1$. Blood types will be represented by the following:

$$\begin{array}{llll} A: p^2 + 2pr & B: q^2 + 2qr & AB: 2pq & O: r^2 \\ = 0.49 + 0.14 & = 0.04 + 0.04 & = 0.28 & = 0.01 \end{array}$$

19. Inbreeding is disadvantageous when it causes recessive deleterious alleles to become homozygous. This occurs in normally outbred populations of diploids that have built up these harmful alleles. In species that normally inbreed, these deleterious alleles are probably no longer present; they were either removed by selection long ago or cannot build up in the population because of the regular pattern of inbreeding.

21. There are four paths passing through A, the only common ancestor ($F_A = 0.01$). All have ABCI as one side of the path. The second legs of the four other paths are ADEHI, ADGHI, AFGHI, and AFEHI. (A path such as IHEDAFGHI is invalid, passing through H twice.) Since each path has six ancestors, the inbreeding coefficient is $F_1 = 4(1/2)^6(1.01) = 0.063$.

23. Using the formula $F = (2pq - H)/2pq$, we calculate that $2pq = 0.48$, and $H = 38/100 = 0.38$. Therefore, $F = (0.48 - 0.38)/0.48 = 0.208$.

$$25. 0.375. F = (2pq - H)/2pq = \frac{(0.32 - 0.20)}{0.32} = \frac{0.12}{0.32} = 0.375$$

Critical Thinking Question:

1. Let us use the superscripts m and p for male and female, respectively. Then, we can construct the following Punnett square creating the next generation:

	Males, $A; f(A) = p^m$	Males, $a; f(a) = q^m$
Females, $A; f(A) = p^f$	$f(AA) = p^m p^f$	$f(Aa) = p^f q^m$
Females, $a; f(a) = q^f$	$f(Aa) = p^m q^f$	$f(aa) = q^m q^f$

Since the distribution of offspring in the table is independent of sex, it is the same in both sexes. The frequency of the A allele, p (in both sexes), will be the sum of the frequencies of the homozygotes and half the heterozygotes, or:

$$p = p^m p^f + (1/2)(p^m q^f + p^f q^m)$$

We then substitute $(1 - p)$ for all q 's:

$$p = p^m p^f + (1/2)p^m (1 - p^f) + (1/2)p^f (1 - p^m)$$

which simplifies to:

$$p = (1/2)(p^m + p^f)$$

In other words, after one generation of random mating, the allelic frequencies in each sex are the averages for both sexes. Now the frequency is the same in both sexes, and a second generation of random mating will achieve Hardy-Weinberg proportions. The population is not in equilibrium after one generation because the proportion of genotypes is not p^2 , $2pq$, and q^2 if p^m does not equal p^f . In other words, $p^m p^f$ does not equal p^2 .

Chapter 20 Population Genetics: Processes That Change Allelic Frequencies

1. **a.** The equilibrium frequency of a is $\hat{q} = \frac{\mu}{(\mu + \nu)}$. Therefore

$$\hat{q} = (6 \times 10^{-5}) / (6 \times 10^{-5} + 7 \times 10^{-7}) = 0.00006 / 0.0000607 = 0.988$$

- b.** If q = 0.90 at generation n , then

$$\begin{aligned} q_{n+1} &= q_n + \mu p_n - \nu q_n = \\ 0.90 &+ (6 \times 10^{-5})(0.10) - (7 \times 10^{-7})(0.90) = 0.9000054 \end{aligned}$$

3. 0.714.

$$\hat{q} = \frac{\mu}{(\mu + \nu)} = \frac{5 \times 10^{-5}}{5 \times 10^{-5} + 2 \times 10^{-5}} = \frac{5}{7} = 0.714$$

5. We use equation 20.12 to calculate the migration rate, m :

$$m = (q_C - q_N)/(q_M - q_N)$$

In this case, $q_C = 0.45$, $q_N = 0.62$, and $q_M = 0.03$. Thus

$$m = (0.45 - 0.62)/(0.03 - 0.62) = (-0.17)/(-0.59) = 0.288$$

7. Fast: 0.56; slow: 0.44. With 900 butterflies we have 1,800 alleles. $0.6(1,800) = 1,080$ fast alleles, and $0.4(1,800) = 720$ slow alleles. In the migrant population, $(0.8)(180) = 144$ slow and $(0.2)(180) = 36$ fast alleles. Therefore, the frequency of the fast allele is $(1,080 + 36)/1,980 = 0.56$. The frequency of the slow allele is $1 - f(\text{fast allele}) = 1 - 0.56 = 0.44$.

9. 0.47. We again use equation 20.12:

$$m = \frac{q_C - q_N}{q_M - q_N}$$

where $m = 0.1$, $q_C = 0.45$, and $q_M = 0.25$.

$$0.1 = \frac{0.45 - q_N}{0.25 - q_N}$$

$$0.1(0.25 - q_N) = 0.45 - q_N$$

$$0.025 - 0.1q_N = 0.45 - q_N$$

$$0.9q_N = 0.425$$

$$q_N = \frac{0.425}{0.9} = 0.47$$

11. In stabilizing selection, extremes of a distribution are selected against. In directional selection, one extreme is favored over the other. In disruptive selection, both extremes are favored over the middle of the distribution.

13. Heterozygote disadvantage:

	AA	Aa	aa	Total
Before selection	p^2	$2pq$	q^2	1
Fitnesses (W)	1	$1 - s$	1	
Frequencies after selection	$p^2/\bar{W}$	$2pq(1 - s)/\bar{W}$	$q^2/\bar{W}$	$\bar{W} = 1 - 2pqs$

Then

$$q_{n+1} = (pq[1 - s] + q^2)/\bar{W}$$

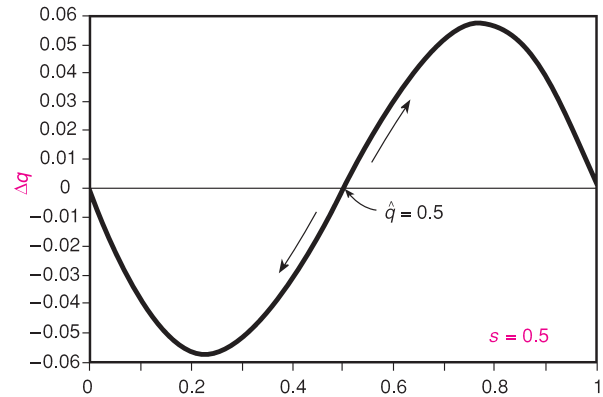
$$\Delta q = q_{n+1} - q = (pq[1 - s] + q^2)/\bar{W} - q\bar{W}/\bar{W}$$

$$= (pq[1 - s] + q^2 - q[1 - 2pqs])/\bar{W}$$

which simplifies to

$$\Delta q = spq(2q - 1)/\bar{W}$$

At $\Delta q = 0$, $\hat{q} = 0, 1$, or 0.5 ($2q - 1 = 0$, therefore $\hat{q} = 0.5$). The equilibrium points of zero and one are stable—if perturbed slightly (less than 0.5), the population will return to these values. The value $\hat{q} = 0.5$ is, however, unstable—if perturbed, it will continue away from the equilibrium point. This can be seen by either substituting into or graphing the Δq equation.

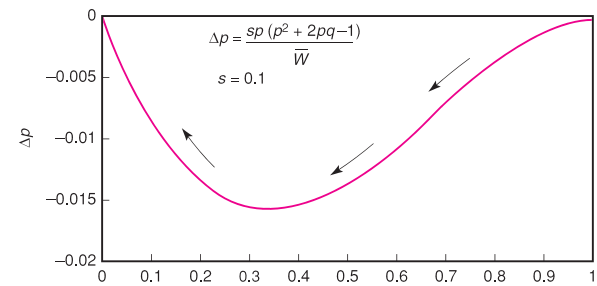


15.

	AA	Aa	aa	Total
Before selection	p^2	$2pq$	q^2	1
Fitnesses (W)	$1 - s$	$1 - s$	1	$\bar{W} = 1 - p^2s - 2pqs$
Frequencies after selection	$p^2(1 - s)/\bar{W}$	$2pq(1 - s)/\bar{W}$	$q^2/\bar{W}$	1

$$p_{n+1} = (p^2[1 - s] + pq[1 - s])/\bar{W}$$

$$\begin{aligned} \Delta p &= p_{n+1} - p = (p^2[1 - s] + pq[1 - s])/\bar{W} - p\bar{W}/\bar{W} \\ &= sp(p^2 + 2pq - 1)/\bar{W} = sp(-p^2 + 2p - 1)/\bar{W} \end{aligned}$$

And $\hat{p} = 0$ or 1 (from the root of the quadratic). Only zero is stable.

17. Since only heterozygotes survive, $\hat{q} = 0.5$. This can also be derived from equation 20.24:

$$\hat{q} = s_1/(s_1 + s_2)$$

$$\text{If } s_1 = s_2 = 1, \text{ then } \hat{q} = 1/2$$

19. The *ST* inversion seems to do best at lower elevations and the *AR* at higher elevations. *CH* (and others) do not appear to be affected by altitude. To test this hypothesis, we would grow caged populations of flies with different initial frequencies of the various inversions at different simulated elevations, simulated by temperature, pressure, oxygen content, or other. We predict that, regardless of initial conditions, they would eventually equilibrate at the values in the table for the given parameter of the altitude (temperature, pressure, oxygen content, or other) that is acting as a selective agent. We would thus identify the selective agent. Since the inversions isolate various allelic combinations, our next step (a potentially long-term step) would be to determine which loci the selective agent is acting on.

A-24 *Appendix A Brief Answers to Selected Exercises, Problems, and Critical Thinking Questions*

21. 0.575. We can use the formula in equation 20.15, which is a simplified form of the sum of homozygotes + one-half the proportion of heterozygotes of the *b* allele.

$$q_{n+1} = \frac{q - sq^2}{1 - sq^2}$$

Since the relative fitness, $W = 0.4$, $s = 1 - W = 0.6$.

$$q_{n+1} = \frac{0.7 - (0.6)(0.49)}{1 - (0.6)(0.49)} = \frac{0.406}{0.706} = 0.575$$

23. 0.33, 0.25. Since the fitness is zero, $s = 1$, and we can use equation 20.17: $q_1 = q_0/(q_0 + 1) = 0.5/1.5 = 0.33$. For the second generation, we substitute the first generation numbers: $q_2 = 0.33/1.33 = 0.248$.

Critical Thinking Question:

- There could be several reasons why these systems are in existence. First, they could be in selection-mutation equilibrium. However, that would not account for the high frequencies of both alleles in human populations in the Rh system. Second, the polymorphism could be relatively new, somehow maintaining both alleles as the human population increased in recent times with natural selection not having enough time to eliminate one of the alleles. Third, although the Rh blood system could follow the heterozygous disadvantage model, selection could also be acting in other ways that might maintain the polymorphism. That is, aside from Rh incompatibility eliminating heterozygotes, other genotypic combinations could be favored under other circumstances. Finally, although one or the other allele is being eliminated in any one population due to heterozygous disadvantage, the constant mixing of human populations could be reintroducing the rarer allele.

Chapter 21 Evolution and Speciation

- Neo-Darwinism is the application of population genetics to Darwinian evolution. Darwinian evolution works as natural selection favors the most fit organisms in competition among the overproduced young of any species.
- Each process lets reproductive isolating mechanisms evolve while some barrier to breeding arises (see fig. 21.3).
- Constraint* refers to the limitations on changes that can take place. Some changes result in nonfunctional proteins and enzymes and thus cannot be in a successful lineage. For example, many base changes that lead to new amino acids in enzyme active sites disrupt enzymatic activity. If these mutations take place, they are eliminated by natural selection.
- Yes. Two distinct species should not yield fertile progeny. We see a great reduction in the numbers of offspring from hybrids, indicating that hybrid inviability is one isolating mechanism operating.
- Punctuated equilibrium proposes that species remain unchanged for long periods of time and that major changes occur only periodically. If species A existed for ten million years and suddenly (geologically speaking) changed dramatically to species X and Y, there would be few fossils because of the relatively short time in which intermediate forms were present. Another argument is, barring an incredibly detailed and complete sequence (of which there are almost none), there will always be gaps in the fossil record.
- Genetic variability can be maintained by heterozygote advantage (e.g., sickle-cell anemia in people); frequency-dependent selection

(rare-male mating advantage in *Drosophila*); transient polymorphism (industrial melanism in moths during an increase or decrease in industrialization); life-stage selection, which often happens when comparing egg, larval, pupal, and adult mortalities in *Drosophila*; differential selection in heterogeneous environments, common in some land snails; and neutrality.

- Presumably, in mammals, selection could involve types of substrates acted upon by electrophoretic variants; functioning at different pHs and ionic strengths in various cellular compartments; resistance to enzyme inhibitors; interaction with other proteins and membrane components; and others.

- Use the formula $K = -\ln(1 - d/n)$, in which d is the number of amino acid differences and n is the total number of amino acid sites. Thus:

$$\text{human being-dog: } K = -\ln(1 - 1/8) = 0.133$$

$$\text{human being-chicken: } K = -\ln(1 - 3/8) = 0.470$$

$$\text{and dog-chicken: } K = -\ln(1 - 3/8) = 0.470$$

These values place human beings and dogs very close and both equally far from chickens, which is consistent with the known evolutionary relationships.

- 0.33 *AA*, 0.49 *Aa*, 0.18 *aa*. The mean fitness of the population after selection is $0.5 + 2pq = 0.98$ (table 21.2). The new frequency of a genotype is its original frequency times its fitness, all divided by the mean fitness of the population. Or:

$$f(AA) = \frac{(0.36)(1.5 - 0.6)}{0.98} = 0.33$$

$$f(Aa) = 0.48/0.98 = 0.49$$

$$f(aa) = (0.16)(1.5 - 0.4)/0.98 = 0.18$$

- 0.023. $K = -\ln(1 - p)$

$$K = -\ln(1 - [23/1,000]) = -\ln(0.977) = 0.023$$

- The third. For many amino acids, the third position can be any of the four bases. In addition, wobble allows for some variation here as well. Changing the first or second base almost always produces a new amino acid.

- Eight first cousins, on average, carry the complete genome of an individual. Therefore, from an evolutionary point of view, an individual and his or her eight cousins include the same alleles.

Critical Thinking Question:

- Given that the gene for peppering is recessive and that one year equals one generation, the moths should be following a selection model in which natural selection acts against the recessive homozygote. In that case, we have already developed a selection model for this in chapter 20. Equation 20.15 relates the new allelic frequency to the old, given a particular selection coefficient:

$$q_{n+1} = q(1 - sq)/(1 - sq^2)$$

We can solve this equation for the selection coefficient:

$$s = (q - q_{n+1})/(q^2 - q^2q_{n+1})$$

Here, $q = 0.6$ and $q_{n+1} = 0.5$. When we solve the equation, we get: $s = 0.56$, or $W = 1 - s = 0.44$, which is the fitness of the peppered moth.



APPENDIX B

Suggestions for Further Reading

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GLOSSARY

acentric fragment A chromosomal piece without a centromere.

acrocentric chromosome A chromosome whose centromere lies very near one end.

acron The anterior end of the arthropod embryo from which eyes and antennae develop.

activation energy See free energy of activation ($\Delta G^\ddagger$).

activator A eukaryotic specific transcription factor that binds to an enhancer, often far upstream of a promoter.

active site The part of an enzyme where the actual enzymatic function is performed.

adaptive mutation See directed mutation.

adaptive value See fitness.

additive model A mechanism of quantitative inheritance in which alleles at different loci either add an amount to the phenotype or add nothing.

adenine See purines.

adjacent-1 segregation A separation of centromeres during meiosis in a reciprocal translocation heterozygote so that unbalanced zygotes are produced.

adjacent-2 segregation Separation of centromeres during meiosis in a translocation heterozygote so that homologous centromeres are pulled to the same pole.

A DNA The form of DNA with high water content; it has tilted base pairs and more base pairs per turn than does B DNA.

affected Individuals in a pedigree that exhibit the specific phenotype under study.

allele Alternative form of a gene.

allelic exclusion A process whereby only one immunoglobulin light chain and one heavy chain gene are transcribed in any one cell; the other genes are repressed.

allopatric speciation Speciation in which the evolution of reproductive isolating mechanisms occurs during the physical separation of the populations.

allopolyploidy Polyploidy produced by the hybridization of two species.

allosteric protein A protein whose shape is changed when it binds a particular molecule. In the new shape, the protein's ability to react to a second molecule is altered.

allotype Mutant of a nonvariant part of an immunoglobulin gene that follows the rules of simple Mendelian inheritance.

allozygosity Homozygosity in which the two alleles are alike but unrelated. See autozygosity.

allozymes Forms of an enzyme, controlled by alleles of the same locus, that differ in electrophoretic mobility. See isozymes.

alternate segregation A separation of centromeres during meiosis in a reciprocal translocation heterozygote so that balanced gametes are produced.

alternative splicing Various ways of splicing out introns in eukaryotic premessenger RNAs so that one gene produces several different messenger RNA and protein products.

altruism A form of behavior in which an individual risks lowering its fitness for the benefit of another.

Alu family A dispersed, intermediately repetitive DNA sequence found in the human genome about 300,000 times. The sequence is about 300 bp long. The name *Alu* comes from the restriction endonuclease that cleaves it.

aminoacyl-tRNA synthetases Enzymes that attach amino acids to their proper transfer RNAs.

amphidiploid An organism produced by hybridization of two species, followed by somatic doubling. It is an allotetraploid that appears to be a normal diploid.

anagenesis The evolutionary process whereby one species evolves into another without any splitting of the phylogenetic tree. See cladogenesis.

anaphase The stage of mitosis and meiosis in which sister chromatids or homologous chromosomes are separated by spindle fibers.

anaphase A The stage of anaphase in which chromatids are separated by the shortening of kinetochore microtubules.

anaphase B The stage of anaphase in which chromatids are separated by the general elongation of the spindle.

anaphase-promoting complex (APC) Protein complex that breaks down cyclin B and promotes anaphase among its various roles in controlling the cell cycle. (Also called the cyclosome.)

aneuploids Individuals or cells exhibiting aneuploidy.

aneuploidy The condition of a cell or of an organism that has additions or deletions of whole chromosomes.

angiosperms Plants whose seeds are enclosed within an ovary. Flowering plants.

antibody A protein produced by a B lymphocyte that protects the organism against antigens.

anticoding strand The DNA strand that forms the template for both the transcribed messenger RNA and the coding strand.

anticodon The three-base sequence on transfer RNA complementary to a codon on messenger RNA.

antigen A foreign substance capable of triggering an immune response in an organism.

antimutator mutations Mutations of DNA polymerase that decrease the overall mutation rate of a cell or of an organism.

anti-oncogene A gene that represses malignant growth and whose absence results in malignancy (e.g., *retinoblastoma*).

antiparallel strands Strands, as in DNA, that run in opposite directions with respect to their 3' and 5' ends.

antisense RNA RNA product of *mic* (mRNA-interfering complementary RNA) genes that regulates another gene by base pairing with, and thus blocking, its messenger RNA.

antisense strand See anticoding strand.

anti-sigma factor A protein that interferes with the action of a sigma factor.

antiterminator protein A protein that, when bound at its normal attachment sites, lets RNA polymerase read through normal terminator sequences (e.g., the N- and Q-gene products of phage λ).

AP endonucleases Endonucleases that initiate excision repair at apurinic and apyrimidinic sites on DNA.

apoptosis Programmed cell death.

archaea Highly specialized, bacterially like organisms that make up the third kingdom of life on earth along with the bacteria and the eukaryotes. Identified by Carl Woese in 1977 based on ribosomal RNA sequences. Most are thermophilic, halophilic, or methanogenic.

ascospores Haploid spores found in the asci of Ascomycete fungi.

ascus The sac in Ascomycete fungi that holds the ascospores.

A (aminoacyl) site The site on the ribosome occupied by an aminoacyl-tRNA just prior to peptide bond formation.

assignment test A test that determines whether a locus is on a specific chromosome by observing the concordance of the locus and the specific chromosome in hybrid cell lines.

assortative mating The mating of individuals with similar phenotypes.

G-2

Glossary

- aster** Configuration at the centrosome with microtubules radiating out in all directions.
- ataxia-telangiectasia** A disease in human beings caused by a defect in X-ray-induced repair mechanisms.
- attenuator region** A control region at the promoter end of repressible amino acid operons that exerts transcriptional control based on the translation of a small leader peptide gene.
- attenuator stem** See terminator stem.
- autogamy** Nuclear reorganization in a single *Paramecium* cell similar to the changes that occur during conjugation.
- autonomously replicating sequence (ARS)** Eukaryotic site of the initiation of DNA replication consisting of an 11 base-pair consensus sequence and several other sequences covering 100–200 base pairs.
- autopolyploidy** Polyploidy in which all the chromosomes come from the same species.
- autoradiography** A technique in which radioactive molecules make their locations known by exposing photographic plates.
- autosomal set** A combination of nonsex chromosomes consisting of one from each homologous pair in a diploid species.
- autosomes** The nonsex chromosomes.
- autotrophs** Organisms that can utilize carbon dioxide as a carbon source.
- autozygosity** Homozygosity in which the two alleles are identical by descent (i.e., they are copies of an ancestral gene).
- auxotrophs** Organisms that have specific nutritional requirements.
- bacillus** A rod-shaped bacterium.
- backcross** The cross of an individual with one of its parents or with an organism with the same genotype as a parent.
- back mutation** The process that causes reversion. A change in a nucleotide pair in a mutant gene that restores the original sequence and hence the original phenotype.
- bacterial artificial chromosomes (BACs)** Artificial chromosomes used for sequencing that are derived from bacterial fertility factors (F plasmids).
- bacterial lawn** A continuous cover of bacteria on the surface of a growth medium.
- bacteriophages** Bacterial viruses.
- Balbani rings** The larger polytene chromosomal puffs. Generally synonymous with *puffs*. See chromosome puffs.
- Barr body** Heterochromatic body (X chromosome) found in the nuclei of normal female mammals but absent in the nuclei of normal males.
- basal bodies** Microtubule organizing center for cilia and flagella, composed of centrioles.
- base excision repair** The DNA excision repair mechanism that replaces a nucleotide lacking its base with a complete nucleotide. Glycosylases or the environment create the AP (apurinic and apyrimidinic) nucleotides.
- base flipping** A process whereby enzymes gain access to bases within the DNA double helix by first flipping the bases out of the interior to the outside.
- basic/helix-loop-helix/leucine zipper** A motif of proteins that bind DNA that consists of a series of basic amino acids followed by a helix-loop-helix domain and then a leucine zipper. See helix-turn-helix motif; leucine zipper.
- Batesian mimicry** Form of mimicry in which an innocuous model gains protection by resembling a noxious or dangerous host.
- B DNA** The right-handed, double-helical form of DNA described by Watson and Crick.
- β -galactosidase** The enzyme that splits lactose into glucose and galactose (coded by a gene in the *lac* operon).
- β -galactoside acetyltransferase** An enzyme that is involved in lactose metabolism and encoded by a gene in the *lac* operon.
- β -galactoside permease** An enzyme involved in concentrating lactose in the cell (coded by a gene in the *lac* operon).
- binary fission** Simple cell division in single-celled organisms.
- binomial expansion** The terms generated when a binomial expression is raised to a particular power.
- binomial theorem** The theorem that gives the terms of the expansion of a binomial expression raised to a particular power.
- biochemical genetics** The study of the relationships between genes and enzymes, specifically the role of genes in controlling the steps in biochemical pathways.
- bioinformatics** The science of storing, retrieving, and analyzing genomic data.
- biolistic** A method (*biological ballistic*) of transfecting cells by bombarding them with microprojectiles coated with DNA.
- biological species concept** The idea that organisms are classified in the same species if they are potentially capable of interbreeding and producing fertile offspring.
- bivalents** Structures, formed during prophase of meiosis I, consisting of the synapsed homologous chromosomes. Equivalent to a tetrad of chromatids.
- blastoderm** The outer layer of cells in an insect embryo after cleavage but before gastrulation. A syncytial blastoderm gives way to a cellular blastoderm when cell walls form.
- blunt-end ligation** The ligating or attaching of blunt-ended pieces of DNA by, for example, T₄ DNA ligase. Used in creating hybrid vectors.
- bottleneck** A brief reduction in the size of a population, which usually leads to random genetic drift.
- bouquet stage** A stage during zygonema in which chromosome ends, attached to the nuclear membrane, come to lie near each other.
- branch migration** The process in which a crossover point between two duplexes slides along the duplexes.
- breakage and reunion** The general mode by which recombination occurs. DNA duplexes are broken and reunited in a crosswise fashion according to the Holliday model.
- breakage-fusion-bridge cycle** Damage that a dicentric chromosome goes through during each cell cycle.
- buoyant density of DNA** A measure of the density or size of DNA determined by the equilibrium point reached by DNA after density gradient centrifugation.
- calculus of the genes** Apparent calculation by the genes to determine when a particular altruistic behavior is beneficial to inclusive fitness and hence worth doing.
- cancer** An informal term for a diverse class of diseases marked by abnormal cell proliferation.
- cancer-family syndromes** Pedigree patterns in which unusually large numbers of blood relatives develop certain kinds of cancers.
- cap** A methylated guanosine added to the 5' end of eukaryotic messenger RNA.
- capsid** The protein shell of a virus.
- capsomere** Protein clusters making up discrete subunits of a viral protein shell.
- carcinoma** Tumor arising from epithelial tissue (e.g., glands, breasts, skin, linings of the urogenital and respiratory systems).
- cassette mechanism** The mechanism by which homothallic yeast cells alternate mating types. The mechanism involves two silent transposons (cassettes) and a region where these cassettes can be expressed (cassette player).
- catabolite activator protein (CAP)** A protein that, when bound with cyclic AMP, can attach to sites on sugar-metabolizing operons to enhance transcription of these operons.
- catabolite repression** Repression of certain sugar-metabolizing operons in favor of glucose utilization when glucose is present in the environment of the cell.
- cdNA** See complementary DNA.
- cell cycle** The cycle of cell growth, replication of the genetic material, and nuclear and cytoplasmic division.
- cell-free system** A mixture of cytoplasmic components from cells, lacking nucleic acids and membranes. Used for in vitro protein synthesis and other purposes.
- cellular immunity** Immunity controlled by killer and helper T cells, which recognize infected body cells and either cause the infected cells to destroy internal invaders or destroy the infected cells directly.
- centimorgan** A chromosome-mapping unit. One centimorgan equals 1% recombinant offspring.

central dogma The original postulate of the way that information can be transferred between DNA, RNA, and protein, barring any transfer originating in protein.

centric fragment A piece of chromosome containing a centromere.

centrioles Cylindrical organelles, found in eukaryotes (except in higher plants), that reside in the centrosome. Also called basal bodies when they organize flagella or cilia.

centromere Constrictions in eukaryotic chromosomes on which the kinetochore lies. Also, the DNA sequence within the constriction that is responsible for appropriate function.

centromeric fission Creation of two chromosomes from one by splitting at the centromere.

centrosome The spindle-microtubule organizing center in eukaryotes except for those, such as fungi, that use spindle pole bodies to organize the spindles.

chaperone See molecular chaperone.

Chargaff's rule Chargaff's observation that in the base composition of DNA, the quantity of adenine equals the quantity of thymine, and the quantity of guanine equals the quantity of cytosine (equal purine and pyrimidine content).

Charon phages Phage lambda derivatives used as vehicles in DNA cloning.

checkpoint Used to describe points in the cell cycle that can be stopped if certain conditions are not met.

chemiluminescent techniques Techniques in which various molecules are made visible when exposed to ultraviolet or laser light if the molecules have a fluorescent tag.

chiasmata X-shaped configurations seen in tetrads during the later stages of prophase I of meiosis. They represent physical crossovers (singular: *chiasma*).

chimeras Individuals made up of two or more cell lines in which the cells originated in different zygotes. See mosaics.

chimeric plasmid Hybrid, or genetically mixed, plasmid used in DNA cloning.

chi site Sequence of DNA at which the RecBCD protein cleaves one of the DNA strands during recombination.

chi-square distribution The sampling distribution of the chi-square statistic. A family of curves whose shapes depend on degrees of freedom.

chloroplast The organelle that carries out photosynthesis and starch grain formation.

chromatids The subunits of a chromosome prior to anaphase of meiosis or mitosis. At anaphase of meiosis II or mitosis, when the sister chromatids separate, each chromatid becomes a chromosome.

chromatin The nucleoprotein material of the eukaryotic chromosome.

chromatin assembly factors Proteins involved in the construction of nucleosomes.

chromatin remodeling The change in the structure or positioning of nucleosomes, usually to allow transcription.

chromatosome The core nucleosome plus the H1 protein, a unit that includes approximately 168 base pairs of DNA.

chromomeres Dark regions of chromatin condensation in eukaryotic chromosomes at meiosis, mitosis, or endomitosis.

chromosomal painting A variant of the technique known as fluorescent in situ hybridization. Fluorescent dyes, attached to numerous nucleotide probes, give each human chromosome a different fluorescent signature.

chromosomal theory of inheritance

The theory that chromosomes are linear sequences of genes.

chromosome The form of the genetic material in viruses and cells. A circle of DNA in most prokaryotes; a DNA or an RNA molecule in viruses; a linear nucleoprotein complex in eukaryotes.

chromosome jumping A technique for isolating clones from a genomic library that are not contiguous but skip a region between known points on the chromosome. This is usually done to bypass regions that are difficult or impossible to "walk" through or regions known not to be of interest.

chromosome puffs Diffuse, uncoiled regions in polytene chromosomes where transcription is actively taking place.

chromosome walking A technique for studying segments of DNA, larger than can be individually cloned, by using overlapping cloned DNA.

cis Meaning "on the near side of"; refers to geometric configurations of atoms or mutants on the same chromosome.

cis-dominant Mutants (e.g., of an operator) that control the functioning of genes on the same piece of DNA.

cis-trans complementation test A mating test to determine whether two mutants on opposite chromosomes complement each other; a test for allelism.

cistron Term Benzer coined for the smallest genetic unit that exhibits the *cis-trans* position effect; synonymous with *gene*.

cladogenesis The evolutionary process whereby one species splits into two or more species. See anagenesis.

classical linkage map A chromosomal map, measured in centimorgans, based on genetic crosses to locate the relative distances between genes and their relative locations on chromosomes.

clinal selection Selection that changes gradually along a geographic gradient.

clonal evolution theory The theory that cancer develops from sequential changes (mutations) in the genome of a single cell.

clone A group of cells arising from a single ancestor.

coccus A spherical bacterium.

coding strand The DNA strand with the same sequence as the transcribed messenger RNA (given U in RNA and T in DNA). Compare with "anticoding strand."

codominance The relationship of alleles in a heterozygote that shows the individual expression of each allele in the phenotype.

codon preference The idea that for amino acids with several codons, one or a few are preferred and are used disproportionately. They would correspond with abundant transfer RNAs.

codons The sequences of three RNA or DNA nucleotides that specify either an amino acid or termination of translation.

coefficient of coincidence The number of observed double crossovers, divided by the number expected based on the independent occurrence of crossovers.

coefficient of relationship, *r* The proportion of alleles held in common by two related individuals.

cohesin Proteinaceous complex that holds sister chromatids together until anaphase of mitosis.

cointegrate A fusion of two elements. An intermediate structure in transposition.

colicinogenic factors See col plasmids.

col plasmids Plasmids that produce antibiotics (colicinogens) that the host uses to kill other strains of bacteria.

combinatorial control Transcriptional control in eukaryotes, which involves a large number of polypeptides, many of which recognize specific DNA sequences.

common ancestry The shared genetic inheritance of two individuals who are blood relatives. When two parents have a common ancestor, their offspring will be inbred.

compensasome The multisubunit dosage compensation complex in *Drosophila* with attendant RNAs.

competence factor A surface protein that binds extracellular DNA and enables the bacterial cell to be transformed.

complementarity The correspondence of DNA bases in the double helix so that adenine in one strand is opposite thymine in the other strand and cytosine in one strand is opposite guanine in the other. This relationship explains Chargaff's rule.

complementary DNA (cDNA) DNA synthesized by reverse transcriptase using RNA as a template.

complementation The production of the wild-type phenotype by a cell or an organism that contains two mutant genes. If complementation occurs, the mutants are almost certainly nonallelic.

complementation group Cistron (determined by the *cis-trans* complementation test).

complete medium A culture medium that is enriched to contain all of the growth requirements of a strain of organisms.

G-4

Glossary

- component of fitness** A particular aspect in the life cycle of an organism upon which natural selection acts.
- composite transposon** A transposon constructed of two IS elements flanking a control region that frequently contains host genes.
- concordance** The amount of phenotypic similarity between individuals.
- condensin** A protein complex including SMC (structural maintenance of chromosomes) proteins needed for the condensation of interphase chromosomes to mitotic chromosomes.
- conditional-lethal mutant** A mutant that is lethal under one condition but not lethal under another.
- confidence limits** A statistical term for a pair of numbers that predicts the range of values within which a particular parameter lies.
- conjugation** A process whereby two cells come in contact and exchange genetic material. In prokaryotes, the transfer is a one-way process.
- consanguineous** Meaning “between blood relatives”; usually refers to inbreeding or incestuous matings.
- consensus sequence** A sequence of the common nucleotides found in many different DNA or RNA samples of homologous regions (e.g., promoters).
- conservative replication** A postulated mode of DNA replication in which an intact double helix acts as a template for a new double helix; known to be incorrect.
- conserved sequence** A sequence found in many different DNA or RNA samples (e.g., promoters) that is invariant in the sample.
- constitutive heterochromatin** Heterochromatin that surrounds the centromere. *See* satellite DNA.
- constitutive mutant** A mutant whose transcription is no longer under regulatory control.
- contigs** Genomic libraries of overlapping, contiguous clones that cover complete regions of a chromosome.
- continuous replication** In DNA, uninterrupted replication in the 5' to 3' direction using a 3' to 5' template.
- continuous variation** Variation measured on a continuum rather than in discrete units or categories (e.g., height in human beings).
- corepressor** The metabolite that when bound to the repressor (of a repressible operon) forms a functional unit that can bind to the operator and block transcription.
- correlation coefficient** A statistic that gives a measure of how closely two variables are related.
- cosmid** A hybrid plasmid that contains *cos* sites at each end. *Cos* sites are recognized during head filling of lambda phages. Cosmids are useful for cloning segments of foreign DNA up to 50 kb.
- cotransduction** The simultaneous transduction of two or more genes.
- coupling** Allelic arrangement in which mutants are on the same chromosome and wild-type alleles are on the homologue.
- covariance** A statistical value measuring the simultaneous deviations of x and y variables from their means.
- CpG islands** Stretches of CG repeats (in which CpG indicates sequential bases on the same strand of DNA, rather than a C-G base pair). These repeats, found in imprinting centers, are important in regulation.
- crisscross pattern of inheritance** The phenotypic pattern of inheritance controlled by X-linked recessive alleles.
- critical chi-square** A chi-square value for a given degree of freedom and probability level, to which we can compare an experimental chi-square.
- crossbreed** To facilitate fertilization between separate individuals.
- cross-fertilization** *See* crossbreed.
- crossing over** A process in which homologous chromosomes exchange parts by a breakage-and-reunion process.
- crossovers** *See* chiasmata.
- crossover suppression** The apparent lack of crossing over within an inversion loop in heterozygotes. Usually due to mortality of zygotes carrying defective crossover chromosomes rather than to actual suppression.
- C-value paradox** Structural and junk DNA create large eukaryotic genomes and large differences in DNA content between eukaryotic species.
- cyclic AMP** A form of AMP (adenosine monophosphate) used frequently as a second messenger in eukaryotic hormone nets and in catabolite repression in prokaryotes.
- cyclin** Family of proteins involved in cell cycle control.
- cyclin-dependent kinase (CDK)** Family of kinases (phosphorylating enzymes) that, when combined with cyclin, are active in controlling checkpoints in the cell cycle.
- cyclosome** *See* anaphase-promoting complex (APC).
- cytogenetics** The study of cells from the perspective of genetics. In practice, the study of changes in the gross structure and number of chromosomes in cells.
- cytokinesis** The division of the cytoplasm of a cell into two daughter cells. *See* karyokinesis.
- cytoplasmic inheritance** Extrachromosomal inheritance controlled by nonnuclear genomes.
- cytosine** *See* pyrimidines.
- cytotoxic T lymphocytes** T cells responsible for attacking host cells that have been infected with an invading bacterium or virus.
- dauermodification** The persistence for several generations of an environmentally induced trait.
- degenerate code** A code in which several code words have the same meaning. The genetic code is degenerate because many different codons may specify the same amino acid.
- degrees of freedom** An estimate of the number of independent categories in a particular statistical test or experiment.
- deletion chromosome** A chromosome with part deleted.
- deme** A locally interbreeding population.
- denatured** Loss of natural configuration (of a molecule) through heat or other treatment. Denatured DNA is single-stranded.
- denominator elements** Genes on the autosomes of *Drosophila* that regulate the sex switch (*sxl*) to the off condition (maleness). Refers to the denominator of the X/A genic balance equation.
- density-gradient centrifugation** A method of separating molecular entities by their differential sedimentation in a centrifugal gradient.
- depauperate fauna** A fauna, especially common on islands, lacking many species found in similar habitats.
- derepressed** The condition of an operon that is transcribing because repressor control has been lifted.
- deterministic** Referring to events that have no random or probabilistic aspects but proceed in a fixed, predictable fashion.
- development** The process of orderly change an individual goes through in the formation of structure.
- diakinesis** The final stage of prophase I of meiosis.
- dicentric chromosome** A chromosome with two centromeres.
- dictyotene** A prolonged diplonema of primary oocytes that can last many years.
- dideoxy method** A method of DNA sequencing that uses chain-terminating (dideoxy) nucleotides.
- dihybrid** An organism heterozygous at two loci.
- dimerization** The chemical union of two similar molecules.
- diploid** Having each chromosome in two copies per nucleus or cell.
- diplonema (diplotene stage)** The stage of prophase of meiosis I in which chromatids appear to repel each other.
- directed mutation** A form of mutation that appears to respond to the needs of the cell but may, in fact, be due to the cell's hypermutable state under duress.
- directional selection** A type of selection that removes individuals from one end of a phenotypic distribution and thus causes a shift in the distribution.
- disassortative mating** The mating of two individuals with dissimilar phenotypes.

discontinuous replication In DNA, the replication in short 5' to 3' segments, using the 5' to 3' strand as a template while going backward, away from the replication fork.

discontinuous variation Variation that falls into discrete categories (e.g., the green and yellow color of garden peas).

discrete generations Generations that have no overlapping reproduction. All reproduction takes place between individuals of the same cohort.

dispersive replication A postulated mode of DNA replication combining aspects of conservative and semiconservative replication; known to be incorrect.

disruptive selection A type of selection that removes individuals from the center of a phenotypic distribution and thus causes the distribution to become bimodal.

D-loop Configuration found during DNA replication of chloroplast and mitochondrial chromosomes wherein the origin of replication is different on the two strands. The first structure formed is a displacement loop, or D-loop.

DNA cloning See gene cloning.

DNA-DNA hybridization The process of taking DNA from the same or different sources and heating and then cooling it, causing double helices to re-form at homologous regions. This technique is useful for determining sequence similarities and degrees of repetitiveness among DNAs.

DNA fingerprint A pattern of bands created on an electrophoretic gel of a DNA digest probed for a variable locus.

DNA glycosylases Endonucleases that initiate excision repair at the sites of various damaged or improper bases in DNA.

DNA gyrase A topoisomerase that relieves supercoiling in DNA by creating a transient break in the double helix.

DNA ligase An enzyme that closes nicks or discontinuities in one strand of double-stranded DNA by creating an ester bond between adjacent 3'-OH and 5'-PO₄ ends on the same strand.

DNA polymerase One of several classes of enzymes that polymerize DNA nucleotides using single-stranded DNA as a template.

DNA-RNA hybridization The process of heating and then cooling a mixture of DNA and RNA so that the RNA can hybridize (form a double helix) with DNA with a complementary nucleotide sequence.

docking protein Responsible for attaching (docking) a ribosome to a membrane by interacting with a signal particle attached to a ribosome destined to be membrane bound.

dominant An allele that expresses itself even when heterozygous. Also, the trait controlled by that allele.

dosage compensation A mechanism by which species with sex chromosomes ensure that one sex does not have differential activity of alleles on the sex chromosomes.

dot blotting A blotting technique, used on DNA already cloned, that eliminates the electrophoretic separation step.

Autoradiographs reveal dots rather than bands on a gel, indicating a probed sequence.

double digest The product formed when two different restriction endonucleases act on the same segment of DNA.

double helix The normal structural configuration of DNA consisting of two helices rotating about the same axis.

downstream A convention on DNA related to the position and direction of transcription by RNA polymerase (5'→3'). Downstream (in the 3' direction) is in the direction of transcription, whereas upstream (in the 5' direction) is in the direction from which the polymerase has come.

downstream promoter element (DPE)

A consensus sequence at about +28 to +34 of RNA polymerase II promoters that have initiator elements but not TATA boxes.

dyad Two sister chromatids attached to the same centromere.

dynein Microtubule motor protein.

dysplasia Excessive cell growth that involves pathological changes to the cells and their nuclei.

electrophoresis The separation of molecular entities by electric current.

electroporation A technique for transfecting cells by applying a high-voltage electric pulse.

elongation complex The form of RNA polymerase II that actively carries out basal transcription.

elongation factors (EF-Ts, EF-Tu, EF-G)

Proteins necessary for the proper elongation and translocation processes during translation at the ribosome in prokaryotes.

Replaced by eEF1 α and eEF1 β in eukaryotes.

endogenote Bacterial host chromosome.

endomitosis Chromosomal replication without nuclear or cellular division that results in cells with many copies of the same chromosome, such as in the salivary glands of *Drosophila*.

endonucleases Enzymes that make nicks internally in the backbone of a polynucleotide. They hydrolyze internal phosphodiester bonds.

enhancer A eukaryotic DNA sequence that increases transcription of a gene by binding specific transcription factors.

enriched medium See complete medium.

enzyme Protein catalyst.

epigenetic effect An environmentally induced change in the genetic material that does not cause a change in base pairs.

Generally, a phenomenon of differential expression of alleles of a locus depending on the parent of origin. Also applied to an effect in proteins.

epistasis The masking of the action of alleles of one gene by allelic combinations of another gene.

equational division A division, such as the second meiotic division, that does not reduce chromosomal numbers.

E (exit) site Site on the ribosome that depleted transfer RNAs pass through during ejection.

euchromatin Regions of eukaryotic chromosomes that are diffuse during interphase. Presumably the actively transcribing DNA of the chromosomes.

eugenics A social movement designed to improve humanity by encouraging those with beneficial traits to breed and discouraging those with undesirable traits from breeding.

eukaryotes Organisms with true nuclei.

euploidy The condition of a cell or organism that has one or more complete sets of chromosomes.

evolution A change in phenotypic frequencies in a population.

evolutionary rates The rate of divergence between taxonomic groups, measurable as number of amino acid substitutions per million years.

excision repair A process whereby cells remove part of a damaged DNA strand and replace it through DNA synthesis, using the undamaged strand as a template.

exconjugant Each of the two cells that separate after conjugation has taken place.

exogenote DNA that a bacterial cell has taken up through one of its sexual processes.

exon In a gene that has intervening sequences (introns), a region that is actually exported from the nucleus to be expressed or become part of a transfer or ribosomal RNA.

exon shuffling The hypothesis put forward by Walter Gilbert that exons code for the functional units of a protein, and that the evolution of new genes proceeds by recombination or the exclusion of exons.

exonucleases Enzymes that digest nucleotides from the ends of polynucleotide molecules. They hydrolyze phosphodiester bonds of terminal nucleotides.

experimental design A branch of statistics that attempts to outline the way in which experiments should be carried out so the data gathered has statistical value.

expression vector A hybrid vector (plasmid) that expresses its cloned genes.

expressivity The degree of expression of a genetically controlled trait.

F₁ See filial generation.

factorial The product of all integers from the specified number down to one (unity).

Fanconi's anemia A disease in human beings with a syndrome of congenital malformations; associated with various cancers.

fate map A map of the developmental fate of a zygote or early embryo showing the adult organs that will develop from a given position on the zygote or early embryo.

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Glossary

F-duction See sexduction.

fecundity selection The forces causing one genotype to be more fertile than another genotype.

feedback inhibition A posttranslational control mechanism in which the end product of a biochemical pathway inhibits the activity of the first enzyme of the same pathway.

fertility factor The plasmid that allows a prokaryote to engage in conjugation with, and pass DNA into, an F^- cell.

F factor See fertility factor.

filial generation Offspring generation. F_1 is the first offspring, or filial, generation; F_2 is the second; and so on.

fimbriae See pili.

first-division segregation (FDS) The allelic arrangement of ordered spores that indicates the lack of recombination between a locus and its centromere.

fitness, W The relative reproductive success of a genotype as measured by survival, fecundity, or other life-history parameters.

5' untranslated region (5' UTR) See leader.

floral meristem The shoot apical meristem sets aside this tissue that gives rise to flowers.

floral-meristem identity genes At least five genes known to establish the identity of the floral meristem.

fluctuation test An experiment by Luria and Delbrück that compared the variance in number of mutations among small cultures with that among subsamples of a large culture to determine the mechanism of inherited change in bacteria.

fluorescent in situ hybridization (FISH) A technique in which a fluorescent dye is attached to a nucleotide probe that then binds to a specific site on a chromosome and makes itself visible by its fluorescence.

Fokker-Planck equation An equation that describes diffusion processes. It is used by population geneticists to describe random genetic drift.

footprinting A technique to determine the length of nucleic acid in contact with a protein. While in contact, the free DNA is digested. The remaining DNA is then isolated and characterized.

founder effect Genetic drift observed in a population founded by a small, nonrepresentative sample of a larger population.

F-pili Sex pili. Hairlike projections on an F^+ or Hfr bacterium involved in anchorage during conjugation.

fragile site A chromosomal region that has a tendency to break.

fragile-X syndrome The most common form of inherited mental retardation. Named for its association with an X chromosome with a tip that breaks or appears uncondensed. Inheritance involves imprinting.

frameshift A mutation in which there is an addition or deletion of nucleotides that causes the codon reading frame to shift.

free energy of activation ($\Delta G^\ddagger$) Energy needed to initiate a chemical reaction.

frequency-dependent selection A selection whereby a genotype is at an advantage when rare and at a disadvantage when common.

functional alleles Mutations that fail to complement each other in a *cis-trans* complementation test.

fundamental number (NF) The number of chromosome arms in a somatic cell of a particular species.

gamete A germ cell having a haploid chromosomal complement. Gametes from parents of opposite sexes fuse to form zygotes.

gametic selection The forces acting to cause differential reproductive success of one allele over another in a heterozygote.

gametophyte The haploid stage of a plant life cycle that produces gametes (by mitosis). It alternates with a diploid, sporophyte generation.

G-bands Eukaryotic chromosomal bands produced by treatment with Giemsa stain.

gene Inherited determinant of the phenotype. See *cistron*; *locus*.

gene amplification A process or processes by which the cell increases the number of repeats of a particular gene within the genome.

gene cloning Production of large numbers of a piece of DNA after that piece of DNA is inserted into a vector and taken up by a cell. Cloning occurs as the vector replicates.

gene conversion In Ascomycete fungi, a 2:2 ratio of alleles is expected after meiosis, yet a 3:1 ratio is sometimes observed. The gene conversion mechanism is explained by repair of heteroduplex DNA produced by recombination.

gene family A group of genes that has arisen by duplication of an ancestral gene. The genes in the family may or may not have diverged.

gene flow The movement of genes from one population to another by interbreeding between individuals in the two populations.

gene pool All of the alleles available among the reproductive members of a population from which gametes can be drawn.

generalized transduction Form of transduction in which any region of the host genome can be transduced. See *specialized transduction*.

general transcription factors Eukaryotic proteins that form part of the RNA polymerase holoenzymes.

genetic code The linear sequences of nucleotides that specify the amino acids during the process of translation at the ribosome.

genetic engineering Popular term for recombinant DNA technology. See *recombinant DNA technology*.

genetic load The relative decrease in the mean fitness of a population due to the presence of genotypes that have less than the highest fitness.

genetic polymorphism The occurrence together in the same population of more than one allele at the same locus, with the least frequent allele occurring more frequently than can be accounted for by mutation.

genic balance theory Bridges's theory that the sex of a fruit fly is determined by the relative number of X chromosomes and autosomal sets.

genome The entire genetic complement of a prokaryote or virus or the haploid genetic complement of a eukaryote.

genomic equivalence The concept that differentiated cells in a eukaryotic organism have identical genetic contents.

genomic library A set of cloned fragments making up the entire genome of an organism or species.

genomics The study of the mapping and sequencing of genomes. Bioinformatics is the science of mining the data from these DNA sequences obtained from sequencing.

genophore The chromosome (genetic material) of prokaryotes and viruses.

genotype The genes that an organism possesses.

Giemsa stain A complex of stains specific for the phosphate groups of DNA.

Goldstein-Hogness box See *TATA box*.

green fluorescent protein A reporter system that uses the gene from a jellyfish that specifies a protein that fluoresces green when ultraviolet light is shined on it, indicating the success of a transfection experiment.

group I introns Self-splicing introns that require a guanine-containing nucleotide for splicing; the intron is released in a linear form.

group II introns Self-splicing introns that do not require an external nucleotide for splicing; the intron is released in a lariat form.

group selection Selection for traits that would be beneficial to a population at the expense of the individual possessing the trait.

G-tetraplex A structure of four guanines that can base pair to form a planar structure that may be involved in novel structures at the end of eukaryotic chromosomes.

guanine See *purines*.

guide RNA (gRNA) RNA that guides the insertion of uridines (RNA editing) into messenger RNAs in trypanosomes. Found in transcripts from minicircles and maxicircles of DNA in kinetoplasts.

gynandromorphs Mosaic individuals having simultaneous aspects of both the male and the female phenotype.

hammerhead ribozyme A catalytic RNA, shaped like a hammerhead, capable of splitting other RNA molecules with appropriate complementary sequences.

haplodiploidy The sex-determining mechanism found in some insect groups among which males are haploid and females are diploid.

haploid The state of having one copy of each chromosome per nucleus or cell.

HAT medium A selection medium for hybrid cell lines; contains hypoxanthine, aminopterin, and thymidine. HPRT⁺ TK⁺ cell lines can survive in this medium.

heat shock proteins Proteins that appear in a cell after the cell has been subjected to elevated temperatures.

helicase A protein that unwinds DNA, usually at replicating Yjunctions.

helix-turn-helix motif Configuration found in DNA-binding proteins consisting of a recognition helix and a stabilizing helix, separated by a short turn.

hemizygous The condition of loci present in only one copy in a diploid organism, such as loci on the X chromosome of the heterogametic sex of a diploid species.

heritability A measure of the degree to which the variance in the distribution of a phenotype is due to genetic causes. In the broad sense, it is measured by the total genetic variance divided by the total phenotypic variance. In the narrow sense, it is measured by the genetic variance due to additive genes divided by the total phenotypic variance.

hermaphrodite An individual with both male and female genitalia.

heterochromatin Chromatin that remains tightly coiled (and darkly staining) throughout the cell cycle.

heteroduplex DNA See hybrid DNA.

heterogametic The sex with heteromorphic sex chromosomes; during meiosis, it produces different kinds of gametes in accordance with these sex chromosomes.

heterogeneous nuclear mRNA (hnRNA) The original RNA transcripts found in eukaryotic nuclei before posttranscriptional modifications.

heterokaryon A cell that contains two or more nuclei from different origins.

heteromorphic chromosome pair Members of a homologous pair of chromosomes that are not morphologically identical (e.g., the sex chromosomes).

heteroplasmy The existence within an organism of genetic heterogeneity within the populations of mitochondria or chloroplasts.

heterothallic A botanical term used for organisms in which the two sexes reside in different individuals.

heterotrophs Organisms that require an organic form of carbon as a carbon source.

heterozygote A diploid or polyploid with different alleles at a particular locus.

heterozygote advantage A selection model in which heterozygotes have the highest fitness.

heterozygous DNA See hybrid DNA.

Hfr High frequency of recombination. A strain of bacteria that has incorporated an F factor into its chromosome and can then transfer the chromosome during conjugation.

histone acetyl transferases (HATs)

Proteins that remodel chromatin by acetylating histones.

histones Arginine- and lysine-rich basic proteins making up a substantial portion of eukaryotic nucleoprotein.

hnRNA See heterogeneous nuclear mRNA.

Hogness box See TATA box.

holandric trait Trait controlled by a locus found only on the Y chromosome. Involves father-to-son transmission.

Holliday junction A junction point between two cross-linked DNA double helices. It is an intermediate stage in DNA recombination.

holoenzyme The complete enzyme, including all subunits. Often used in reference to RNA and DNA polymerases.

homeo box A consensus sequence of about 180 base pairs discovered in homeotic genes in *Drosophila*. Also found in other developmentally important genes from yeast to human beings.

homeo domain The sixty amino acid polypeptide translated from the homeo box.

homeotic gene Gene that controls the developmental fate of a cell type; mutations of the homeotic gene cause one cell type to follow the developmental pathway of another cell type.

homogametic The sex with homomorphic sex chromosomes; it produces only one kind of gamete in regard to the sex chromosomes.

homologous chromosomes Members of a pair of essentially identical chromosomes that synapse during meiosis.

homologous recombination Breakage and reunion between homologous lengths of DNA mediated by RecA and RecBCD.

homomorphic chromosome pairs Morphologically identical members of a homologous pair of chromosomes.

homoplasmy The existence within an organism of only one type of plastid; usually referring to the genetic identity of mitochondria or chloroplasts.

homothallic A botanical term used for groups whose individuals are not of different sexes.

homozygote A diploid or a polyploid with identical alleles at a locus.

humoral immunity Immunity due to antibodies in the serum and lymph.

H-Y antigen The histocompatibility Yantigen, a protein found on the cell surfaces of male mammals.

hybrid Offspring of unlike parents.

hybrid DNA DNA whose two strands have different origins.

hybridoma A cell resulting from the fusion of a spleen cell and a multiple myeloma cell. These cells can be maintained indefinitely in cell culture, in which they produce monoclonal antibodies.

hybrid plasmid A plasmid that contains an inserted piece of foreign DNA.

hybrid vector See hybrid vehicle.

hybrid vehicle A plasmid or phage containing an inserted piece of foreign DNA.

hybrid zone Geographical region in which previously isolated populations that have evolved differences come into contact and form hybrids.

hyperplasia Excessive cell growth that does not involve pathological changes to the cells.

hypervariable loci Loci with many alleles; especially those whose variation is due to variable numbers of tandem repeats.

hypostatic gene A gene whose expression is masked by an epistatic gene.

identity by descent The state of two alleles when they are identical copies of the same ancestral allele (autozygous).

idiogram A photograph or diagram of the chromosomes of a cell arranged in an orderly fashion. See karyotype.

idiotypic variation Variation in the variable parts of immunoglobulin genes.

idling reaction The production of guanosine tetraphosphate (3'-ppGpp-5') by the stringent factor when a ribosome encounters an uncharged transfer RNA in the A site.

immunity The ability of an organism to resist infection.

immunoglobulins (Igs) Specific proteins produced by derivatives of B lymphocytes that protect an organism from antigens.

imprinting See molecular imprinting.

imprinting center (IC) A region responsible for the control of imprinting. The imprinting mark is almost certainly DNA methylation, which is able to turn off gene transcription.

inbreeding The mating of genetically related individuals.

inbreeding coefficient, F The probability of autozygosity.

inbreeding depression A depression of vigor or yield due to inbreeding.

incestuous A mating between blood relatives who are more closely related than the law of the land allows.

inclusive fitness The expansion of the concept of the fitness of a genotype to include benefits accrued to relatives of an individual since relatives share parts of their genomes. An apparently altruistic act toward a relative may thus enhance the fitness of the individual performing the act.

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Glossary

incomplete dominance The situation in which both alleles of the heterozygote influence the phenotype. The phenotype is usually intermediate between the two homozygous forms.

independent assortment, rule of Mendel's second rule, describing the independent segregation of alleles of different loci.

inducible system A system (a coordinated group of enzymes involved in a catabolic pathway) is inducible if the metabolite it works upon causes transcription of the genes controlling these enzymes. These systems are primarily prokaryotic operons.

induction In regard to temperate phages, the process of causing a prophage to become virulent.

informatics See bioinformatics.

initiation codon The messenger RNA sequence AUG, which specifies methionine, the first amino acid used in the translation process. (Occasionally, GUG is recognized as an initiation codon.)

initiation complex The complex formed for initiation of translation. It consists of the 30S ribosomal subunit, messenger RNA, N-formyl-methionine transfer RNA, and three initiation factors.

initiation factors (IF1, IF2, IF3) Proteins (prokaryotic with eukaryotic analogues) required for the proper initiation of translation.

initiator element (inr) A CT-rich area found in RNA polymerase II promoters without TATA boxes.

initiator proteins Proteins that recognize the origin of replication on a replicon and take part in primosome construction.

insertion mutagenesis Change in gene action due to an insertion event that either changes a gene directly or disrupts control mechanisms.

insertion sequences (IS) Small, simple transposons. See transposable genetic element.

inside marker The middle locus of three linked loci.

intercalary heterochromatin

Heterochromatin, other than centromeric heterochromatin, dispersed throughout eukaryotic chromosomes.

intergenic suppression A mutation at a second locus that apparently restores the wild-type phenotype to a mutant at a first locus.

interkinesis The abbreviated interphase that occurs between meiosis I and II. No DNA replication occurs here.

internal ribosome entry site Sequence in eukaryotic messenger RNAs that allows ribosomes to initiate translation at a point other than the 5' cap.

interphase The metabolically active, nondividing stage of the cell cycle.

interpolar microtubules Microtubules extending from one pole of the spindle and overlapping spindle fibers from the other pole, but not in contact with kinetochores.

interrupted mating A mapping technique in which bacterial conjugation is disrupted after specified time intervals.

intersex An organism with external sexual characteristics of both sexes.

intervening sequences (introns) DNA sequences within a gene that are transcribed but removed prior to translation.

intra-allelic complementation The restoration of activity in an enzyme made of subunits in a heterozygote of two mutants that, when homozygous, are not active. Caused by the interaction of the subunits in the protein.

intragenic suppression A second change within a mutant gene that results in an apparent restoration of the original phenotype.

intron See intervening sequences.

inversion The replacement of a section of a chromosome in the reverse orientation.

inverted repeat sequence A nucleotide sequence read in opposite orientations on the same double helix.

in vitro Biological or chemical work done in the test tube (literally, "in glass") rather than in living systems.

IS elements See insertion sequences.

isochromosome A chromosome with two genetically and morphologically identical arms.

isozymes Different electrophoretic forms of the same enzyme. Unlike allozymes, isozymes are due to differing subunit configurations rather than allelic differences.

junctional diversity Variability in immunoglobulins caused by variation in the exact crossover point during V-J, V-D, and D-J joining.

kappa particles The bacteriophage particles that give a *Paramecium* the killer phenotype.

karyokinesis The process of nuclear division. See cytokinesis.

karyotype The chromosome complement of a cell. See idiogram.

kinesin Microtubule motor protein.

kinetochore The chromosomal attachment point for the spindle fibers, located on the centromere.

kinetochore microtubules Microtubules radiating from the centrosome and attached to kinetochores of chromosomes during mitosis and meiosis.

kin selection The mode of natural selection that acts on an individual's inclusive fitness.

Klenow fragment Proteolytic fragment—obtained by treatment with a protease, or protein-cleaving enzyme—of *E. coli* DNA polymerase I with both 5'→3' polymerase activity and 3'→5' exonuclease activity. (It has been studied extensively because it has been easier for X-ray crystallographers and biochemists to work with this fragment than with the whole enzyme.)

knockout mice Transgenic mice that have been made homozygous for a nonfunctioning allele at a particular locus.

lac operon The inducible operon, including three loci involved in the uptake and breakdown of lactose.

ladder gel See stepladder gel.

lagging strand Strand of DNA being replicated discontinuously.

lampbrush chromosomes Chromosomes of amphibian oocytes having loops that are suggestive of a lampbrush.

leader The length of messenger RNA from the 5' end to the initiation codon, AUG.

leader peptide gene A small gene within the attenuator control region of a repressible amino acid operon. Translation of the gene tests the concentration of amino acids in the cell.

leader transcript The messenger RNA transcribed by the attenuator region of a repressible amino acid operon. The transcript is capable of several alternative stem-loop structures, depending on the translation of a short leader peptide gene.

leading strand Strand of DNA being replicated continuously.

leptonema (leptotene stage) The first stage of prophase I of meiosis, in which chromosomes become distinct.

lethal-equivalent alleles Alleles whose summed effect is that of lethality—for example, four alleles, each of which would be lethal 25% of the time (or to 25% of their bearers), are equivalent to one lethal allele.

leucine zipper Configuration of a DNA-binding protein in which leucine residues on two helices interdigitate, in zipper fashion, to stabilize the protein.

leukemia Cancer of the bone marrow resulting in excess production of leukocytes.

level of significance The probability value in statistics used to reject the null hypothesis.

linkage The association of loci on the same chromosome.

linkage disequilibrium The condition among alleles at different loci such that allelic combinations in a gamete do not follow the product rule of probability.

linkage equilibrium The condition among alleles at different loci such that any allelic combination in a gamete occurs as the product of the frequencies of each allele at its own locus.

linkage groups Associations of loci on the same chromosome. In a species, there are as many linkage groups as there are homologous pairs of chromosomes.

linkage number The number of times one strand of a helix coils about the other.

linker A small segment of DNA that contains a restriction site. It can be added to blunt-ended DNA to give that DNA a particular restriction site for cloning.

liposomes Transfecting DNA is delivered to target cells by way of these membrane-bound vesicles.

locus The position of a gene on a chromosome (plural: *loci*).

lod score method A technique (*logarithmic odds*) for determining the most likely recombination frequency between two loci from pedigree data.

long interspersed elements (LINEs) Sequences of DNA, up to seven thousand base pairs in length, interspersed in eukaryotic chromosomes in many copies.

lymphoma Cancer of the lymph nodes and spleen that causes excessive production of lymphocytes.

Lyon hypothesis The hypothesis that suggests that the Barr body is an inactivated X chromosome.

lysate The contents released from a lysed cell.

lysis The breaking open of a cell by the destruction of its wall or membrane.

lysogenic The state of a bacterial cell that has an integrated phage (prophage) in its chromosome.

major histocompatibility complex A group of highly polymorphic genes whose products appear on the surfaces of cells, imparting to them the property of "self" (belonging to that organism). Some other functions are also involved.

mapping The process of locating the positions of genes on chromosomes.

mapping function The mathematical relationship between measured map distance in a given experiment and the actual recombination frequency.

map unit The distance equal to 1% recombination between two loci.

mate-killer A *Paramecium* phenotype induced by intracellular bacteriophage particles.

maternal effect The effect of the maternal parent's genotype on the phenotype of her offspring.

maternal-effect gene A gene expressed in maternal tissue that influences a developing embryo.

mating type In many species of microorganisms, individuals can be divided into two mating types. Mating can take place only between individuals of opposite mating types due to the interaction of cell surface components.

maturation-promoting factor (MPF)

A protein complex of cyclin B and p34^{cdc2} that initiates mitosis during the cell cycle.

Also called the mitosis-promoting factor.

mean The arithmetic average; the sum of the data divided by the sample size.

mean fitness of the population, $\bar{W}$ The sum of the fitnesses of the genotypes of a population weighted by their proportions; hence, a weighted mean fitness.

meiosis The nuclear process in diploid eukaryotes that results in gametes or spores with only one member of each original homologous pair of chromosomes per nucleus.

meiotic drive See gametic selection.

merozygote A bacterial cell having a second copy of a particular chromosomal region in the form of an exogenote.

messenger RNA (mRNA) A complementary copy of a gene that is translated into a polypeptide at the ribosome.

metacentric chromosome A chromosome whose centromere is located in the middle.

metafemale A fruit fly with an X/A ratio greater than unity.

metagon An RNA necessary for the maintenance of mu particles in *Paramecium*.

metamale A fruit fly with X/A ratio below 0.5.

metaphase The stage of mitosis or meiosis in which spindle fibers are attached to kinetochores, and the chromosomes are positioned in the equatorial plane of the cell.

metaphase plate The plane of the equator of the spindle into which chromosomes are positioned during metaphase.

metastasis The migration of cancerous cells to other parts of the body.

metrical variation See continuous variation.

microsatellite DNA Repeats of very short sequences of DNA, such as CACACACA, dispersed throughout the eukaryotic genome. The loci can be studied by polymerase chain reaction amplification.

microtubule organizing center Active center from which microtubules are organized. The spindle is organized by the centrosome, which may or may not contain a centriole.

microtubules Hollow cylinders made of the protein tubulin (α and β subunits) that make up, among other things, the spindle fibers.

mimicry A phenomenon in which an individual gains an advantage by looking like the individuals of a different species.

minimal medium A culture medium for microorganisms that contains the minimal necessities for growth of the wild-type.

mismatch repair A form of excision repair initiated at the sites of mismatched bases in DNA.

missense mutation Mutations that change a codon for one amino acid into a codon for a different amino acid.

mitochondrion The eukaryotic cellular organelle in which the Krebs cycle and electron transport reactions take place.

mitosis The nuclear division producing two daughter nuclei identical to the original nucleus.

mitosis-promoting factor See maturation-promoting factor (MPF).

mitotic apparatus See spindle.

mixed families Groups of four codons sharing their first two bases and coding for more than one amino acid.

modern linkage map A chromosomal map based on the positions of RFLP markers along its length.

molecular chaperone A protein that aids in the folding of a second protein. The chaperone prevents proteins from forming structures that would be inactive.

molecular evolutionary clock

A measurement of evolutionary time in nucleotide substitutions per year.

molecular imprinting The phenomenon in which there is differential expression of a gene depending on whether it was maternally or paternally inherited.

molecular mimicry The situation in which one type of molecule resembles another type in order to function. For example, the prokaryotic ribosomal release factors, RF1 and RF2, mimic the structure of a transfer RNA.

monocistronic Usually referring to a messenger RNA that carries the information for only one gene (cistron).

monoclonal antibody The antibody from a clone of cells producing the same antibody. An individual with multiple myeloma usually produces monoclonal antibodies.

monohybrids Offspring of parents that differ in only one genetic characteristic. Usually implies heterozygosity at a single locus under study.

monosomic A diploid cell missing a single chromosome.

monovalent A single chromosome composed of two sister chromatids. Equivalent to a dyad.

morphogen A substance transported into or produced in a developing embryo that diffuses to form a gradient that helps determine cell differentiation.

morphological species concept The idea that organisms are classified in the same species if they appear similar.

mosaicism The condition of being a mosaic. See mosaics.

mosaics Individuals made up of two or more cell lines in which the cells originated in the same zygote.

mRNA See messenger RNA (mRNA).

Müllerian mimicry A form of mimicry in which noxious species evolve to resemble each other.

multihybrid An organism heterozygous at numerous loci.

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Glossary

- multinomial expansion** The terms generated when a multinomial is raised to a power.
- mu particles** Bacterially like particles found in the cytoplasm of *Paramecium* that impart the mate-killer phenotype.
- mutability** The ability to change.
- mutants** Alternative phenotypes to the wild-type; the phenotypes produced by alternative alleles.
- mutation** The process by which a gene or chromosome changes structurally; the end result of this process.
- mutation rate** The proportion of mutations per cell division in bacteria or single-celled organisms or the proportion of mutations per gamete in higher organisms.
- mutator mutations** Mutations of DNA polymerase that increase the overall mutation rate of a cell or of an organism.
- muton** A term Benzer coined for the smallest mutable site within a cistron.
- natural selection** The process in nature whereby one genotype leaves more offspring than another genotype because of superior life history attributes such as survival or fecundity.
- negative interference** The phenomenon whereby a crossover in a particular region facilitates the occurrence of other apparent crossovers in the same region of the chromosome.
- N-end rule** The life span of a protein is determined by its amino-terminal (N-terminal) amino acid.
- neo-Darwinism** The merger of classical Darwinian evolution with population genetics.
- neoplasm** New growth of abnormal tissue.
- neutral gene hypothesis** The hypothesis that most genetic variation in natural populations is not maintained by natural selection.
- NF** See fundamental number.
- nickase** See DNA gyrase.
- noncoding strand** See anticoding strand.
- nondisjunction** The failure of a pair of homologous chromosomes to separate properly during meiosis.
- nonhistone proteins** The proteins remaining in chromatin after the histones are removed.
- nonparental ditype (NPD)** A spore arrangement in Ascomycete fungi that contains only the two recombinant-type ascospores (assuming two segregating loci).
- nonparentals** See recombinants.
- nonrecombinants** In mapping studies, offspring that have alleles arranged as in the original parents.
- nonsense codon** One of the messenger RNA sequences (UAA, UAG, UGA) that signals the termination of translation.
- nonsense mutations** Mutations that change a codon for an amino acid into a nonsense codon.
- normal distribution** Any of a family of bell-shaped frequency curves whose relative positions and shapes are defined on the basis of the mean and standard deviation.
- northern blotting** A gel transfer technique used for RNA. See Southern blotting.
- N segments** Sequences of nucleotides added in a template-free fashion at the joining junctions of heavy-chain antibody genes.
- nuclease-hypersensitive site** A region of a eukaryotic chromosome that is specifically vulnerable to nuclease attack because it is not wrapped as nucleosomes.
- nucleolar organizer** The chromosomal region around which the nucleolus forms; site of tandem repeats of the major ribosomal RNA gene.
- nucleolus** The globular, nuclear organelle formed at the nucleolar organizer. Site of ribosome construction.
- nucleoprotein** The substance of eukaryotic chromosomes consisting of proteins and nucleic acids.
- nucleoside** A sugar-base compound that is a nucleotide precursor. Nucleotides are nucleoside phosphates.
- nucleosomes** Arrangements of DNA and histones forming regular spherical structures in eukaryotic chromatin.
- nucleotide** Subunits that polymerize into nucleic acids (DNA or RNA). Each nucleotide consists of a nitrogenous base, a sugar, and one or more phosphate groups.
- nucleotide excision repair** The DNA excision repair mechanism responsible for repairing thymine dimers and other lesions. Enzymes excise a short segment of one of the DNA strands and then repair and ligate the DNA.
- null hypothesis** The statistical hypothesis that there are no differences between observed data and those data expected based on the assumption of no experimental effect.
- nullisomic** A diploid cell missing both copies of the same chromosome.
- numerator elements** Genes on the X chromosome in *Drosophila* that regulate the sex switch gene (*sex*) to the on condition (femaleness). Refers to the numerator of the X/A genic balance equation.
- nutritional-requirement mutants** See auxotrophs.
- Okazaki fragments** Segments of newly replicated DNA produced during discontinuous DNA replication.
- oncogene** Genes capable of transforming a cell. They are found in the active state in retroviruses and transformed cells and in the inactive state in nontransformed cells, in which they are called proto-oncogenes.
- one-gene-one-enzyme hypothesis** Hypothesis of Beadle and Tatum that states that one gene controls the production of one enzyme. Later modified to the concept that one cistron controls the production of one polypeptide.
- oogenesis** The process of ovum formation in female animals.
- oogonia** Cells in females that produce primary oocytes by mitosis.
- open reading frames (ORFs)** Sequence of codons between the initiation and termination codons in a gene.
- operator** A DNA sequence recognized by a repressor protein or repressor-corepressor complex. When the operator is complexed with the repressor, transcription is prevented.
- operon** A sequence of adjacent genes all under the transcriptional control of the same operator.
- origin recognition complex (ORC)** A complex of six proteins that bind to the eukaryotic autonomously replicating sequences (ARS). Needed for the initiation of DNA replication in concert with other proteins.
- outbreeding** The mating of genetically unrelated individuals.
- ovum** Egg. The one functional product of each meiosis in female animals.
- pachynema (pachytene stage)** The stage of prophase I of meiosis in which chromatids are first distinctly visible.
- palindrome** A sequence of words, phrases, or nucleotides that reads the same regardless of the direction from which one starts; the sites of recognition of type II restriction endonucleases.
- panmictic** Referring to unstructured (random mating) populations.
- paracentric inversion** A chromosomal inversion that does not include the centromere.
- paramycin** A toxin liberated by a "killer" *Paramecium*.
- parameters** Measurements of attributes of a population; denoted by Greek letters.
- parapatric speciation** Speciation in which reproductive isolating mechanisms evolve when a population enters a new niche or habitat within the range of the parent species.
- parasegment** The first series of segments that form in a developing insect embryo; they form after about 5.5 hours in the developing *Drosophila* embryo.
- parental ditype (PD)** A spore arrangement in Ascomycete fungi that contains only the two nonrecombinant-type ascospores.
- parental imprinting** See molecular imprinting.
- parentals** See nonrecombinants.
- parthenogenesis** The development of an individual from an unfertilized egg that did not arise by meiotic chromosomal reduction.
- partial digest** A restriction digest that has not been allowed to go to completion, and thus contains pieces of DNA that have restriction endonuclease sites that have not been cleaved.

partial dominance See incomplete dominance.

Pascal's triangle A triangular array of numbers made up of the coefficients of the binomial expansion.

path diagram A modified pedigree showing only the direct line of descent from common ancestors.

pedigree A representation of the ancestry of an individual or family; a family tree.

penetrance The normal appearance of genetically controlled traits in the phenotype.

peptidyl transferase The enzymatic center responsible for peptide bond formation during translation at the ribosome.

pericentric inversion A chromosomal inversion that includes the centromere.

permissive temperature A temperature at which temperature-sensitive mutants are normal.

PEST hypothesis Degradation of a protein in less than two hours is signaled by a region within the protein rich in proline (P), glutamic acid (E), serine (S), or threonine (T).

petite mutations Mutations of yeast that produce small colonies, like those grown under anaerobic conditions.

phages See bacteriophages.

phenocopy A phenotype that is not genetically controlled but looks like a genetically controlled phenotype.

phenotype The observable attributes of an organism.

pheromone A chemical signal, analogous to a hormone, that passes information between individuals.

phosphodiester bond A diester bond linking two nucleotides together (between phosphoric acid and sugars) to form the nucleotide polymers DNA and RNA.

photocrosslinking A technique used to determine which moieties (proteins, DNA) are in close proximity during a particular process.

photoreactivation The process whereby dimerized pyrimidines (usually thymines) in DNA are restored by an enzyme (deoxyribodipyrimidine photolyase) that requires light energy.

phyletic evolution See anagenesis.

phyletic gradualism The process of gradual evolutionary change over time.

phylogenetic tree A diagram showing evolutionary lineages of organisms.

physical map Chromosomal map in which distances are in physical units of base pairs. These maps can be of microsatellite markers or of sequence-tagged sites.

pili (fimbriae) Hairlike projections on the surface of bacteria; Latin for "hair."

plaques Clear areas on a bacterial lawn caused by cell lysis due to viral attack.

plasmid An autonomous, self-replicating genetic particle, usually of double-stranded DNA.

plastid A chloroplast prior to the development of chlorophyll.

pleiotropy The phenomenon whereby a single mutant affects several apparently unrelated aspects of the phenotype.

point centromere The type of centromere, such as that found in *Saccharomyces cerevisiae*, that has defined sequences large enough to accommodate one spindle microtubule.

point mutations Small mutations that consist of a replacement, addition, or deletion of one or a few bases.

polar bodies The small cells that are the by-products of meiosis in female animals. One functional ovum and as many as three polar bodies result from meiosis of each primary oocyte.

polarity Meaning "directionality" and referring either to an effect seen in only one direction from a point of origin or to the fact that linear entities (such as a single strand of DNA) have ends that differ from each other.

polar mutant An organism with a mutation, usually within an operon, that prevents the expression of genes distal to itself.

pollen grain The male gametophyte in higher plants.

poly-A tail A sequence of adenosine nucleotides added to the 3' end of eukaryotic messenger RNAs.

polycistronic Referring to prokaryotic messenger RNAs that contain several genes within the same messenger RNA transcript.

polygenic inheritance See quantitative inheritance.

polymerase chain reaction (PCR) A method to amplify DNA segments rapidly in temperature-controlled cycles of denaturation, primer binding, and replication.

polymerase cycling The process by which a DNA polymerase III enzyme completes an Okazaki fragment, releases it, and begins synthesis of the next Okazaki fragment.

polymerized Formed into a complex compound by linking together smaller elements.

polynucleotide phosphorylase An enzyme that can polymerize diphosphate nucleotides without the need for a primer. The function of this enzyme *in vivo* is probably in its reverse role as an RNA exonuclease.

polyploids Organisms with greater than two chromosome sets.

polyribosome See polysome.

polysome The configuration of several ribosomes simultaneously translating the same messenger RNA. Shortened form of the term *polyribosome*.

polytene chromosome Large chromosome, seen, for example, in *Drosophila* salivary glands, consisting of many chromatids formed by rounds of endomitosis. Synapsis of homologous chromosomes occurs during the process.

population A group of organisms of the same species relatively isolated from other groups of the same species. See deme.

position effect An alteration of phenotype caused by a change in the relative arrangement of the genetic material.

positive interference When the occurrence of one crossover reduces the probability that a second will occur in the same region.

postreplicative repair A DNA repair process initiated when DNA polymerase bypasses a damaged area.

posttranscriptional modifications Changes in eukaryotic messenger RNA made after transcription has been completed. These changes include additions of caps and tails and removal of introns.

preemptor stem A configuration of leader transcript messenger RNA that does not terminate transcription in the attenuator-controlled amino acid operons.

pre-initiation complex (PIC) The form of the RNA polymerase II enzyme with general transcription factors bound equivalent to the *E. coli* holoenzyme. Phosphorylation of the enzyme then allows transcription to begin.

Pribnow box Consensus sequence of TATAAT in prokaryotic promoters centered at the position -10 .

primary oocytes The cells that undergo meiosis in female animals.

primary spermatocytes The cells that undergo meiosis in male animals.

primary structure The sequence of polymerized amino acids in a protein.

primary transcript The product of eukaryotic transcription before posttranscriptional modification takes place.

primase An enzyme that creates a messenger RNA primer for Okazaki fragment initiation.

primer In DNA replication, a length of double-stranded DNA that continues as a single-stranded template in the 3' to 5' direction.

primosome A complex of two proteins, a primase and helicase, that initiates RNA primers on the lagging DNA strand during DNA replication.

prion Infectious agent responsible for several neurological diseases (scrapie, kuru, Creutzfeld-Jakob syndrome, mad-cow disease). It is a protein that lacks DNA or RNA.

probability The expectation of the occurrence of a particular event.

probability theory The conceptual framework concerned with quantification of probabilities. See probability.

proband See propositus.

probe In recombinant DNA work, a radioactive nucleic acid complementary to a region being searched for in a restriction digest or genomic library.

processivity The ability of an enzyme to repetitively continue its catalytic function without dissociating from its substrate.

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Glossary

- product rule** The rule that states that the probability that two independent events will both occur is the product of their separate probabilities.
- progeny testing** Breeding of offspring to determine their genotypes and those of their parents.
- prokaryotes** Organisms that lack true nuclei.
- promoter** A DNA region that RNA polymerase binds to in order to initiate transcription.
- proofread** Technically, to read for the purpose of detecting errors for later correction. DNA polymerase has 3' to 5' exonuclease activity, which it uses during polymerization to remove incorrect nucleotides it has recently added.
- prophage** A temperate phage replicating with the host that can later initiate the lytic cycle.
- prophase** The initial stage of mitosis or meiosis in which chromosomes become visible and the spindle apparatus forms.
- proplastid** Mutant plastids that do not grow and develop into chloroplasts.
- propositus (proposita)** The person through whom a pedigree was discovered.
- proteasome** A barrel-shaped cellular organelle for protein breakdown involving the ubiquitin pathway.
- proteome** From *proteins* of the *genome*; the complete set of proteins from a particular genome. It is the protein analogue to "genome."
- proteomics** The study of the complete set of proteins from a particular genome. It is the protein analogue to "genomics."
- proto-oncogene** A cellular oncogene in an untransformed cell.
- prototrophs** Strains of organisms that can survive on minimal medium.
- pseudoalleles** Genes that are functionally but not structurally allelic. Within gene families, pseudoalleles are alleles that are not expressed.
- pseudoautosomal gene** A gene that occurs on both sex-determining heteromorphic chromosomes.
- pseudodominance** The phenomenon in which a recessive allele shows itself in the phenotype when only one copy of the allele is present, as in hemizygous alleles or in deletion heterozygotes.
- P (peptidyl) site** The site on the ribosome occupied by the peptidyl-tRNA just before peptide bond formation.
- punctuated equilibrium** The evolutionary process involving long periods without change (stasis) punctuated by short periods of rapid speciation.
- Punnett square** A diagrammatic representation of a particular cross used to predict the progeny of the cross.
- purines** Nitrogenous bases of which guanine and adenine are found in DNA and RNA.
- pyrimidines** Nitrogenous bases of which thymine is found in DNA, uracil in RNA, and cytosine in both.
- quantitative inheritance** The mechanism of genetic control of traits showing continuous variation.
- quantitative trait loci** Chromosomal regions contributing to the inheritance of a quantitative trait. These regions may contain one or more polygenes that contribute to the phenotype.
- quantitative variation** *See* continuous variation.
- quaternary structure** The association of polypeptide subunits to form the final structure of a protein.
- random genetic drift** Changes in allelic frequency due to sampling error.
- random mating** The mating of individuals in a population such that the union of individuals with the trait under study occurs according to the product rule of probability.
- random strand analysis** Mapping studies in organisms that do not keep all the products of meiosis together.
- read-through** Transcription or translation beyond the normal termination signals in DNA or RNA, respectively.
- realized heritability** Heritability measured by a response to selection.
- recessive** An allele (or phenotype) that does not express itself in the heterozygous condition.
- reciprocal cross** A cross with the phenotype of each sex reversed as compared with the original cross. Made to test the role of parental sex on inheritance pattern.
- reciprocal translocation** A chromosomal configuration in which the ends of two nonhomologous chromosomes are broken off and become attached to the nonhomologous.
- recombinant DNA technology** Techniques of gene cloning. Recombinant DNA refers to the hybrid of foreign and vector DNA. *See* gene cloning.
- recombinant plasmid** A plasmid that contains an inserted piece of foreign DNA.
- recombinants** In mapping studies, offspring with allelic arrangements made up of a combination of the original parental alleles.
- recombination** The nonparental arrangement of alleles in progeny that can result from either independent assortment or crossing over.
- recombination nodule** Proteinaceous nodules found on bivalents during zygonema and pachynema associated with crossing over.
- recon** A term Benzer coined for the smallest recombinable unit within a cistron.
- reductional division** The first meiotic division. It reduces the number of chromosomes and centromeres to half that in the original cell.
- regional centromere** The type of centromere found in higher eukaryotes that can accommodate several spindle microtubules.
- regulator gene** A gene primarily involved in control of the production of another gene's product.
- reinitiation** The initiation of translation by a ribosome that has just completed translation of a region of the messenger RNA upstream of the current point of initiation.
- relative Darwinian fitness** *See* fitness.
- relaxed mutant** A mutant that does not exhibit the stringent response under amino acid starvation.
- release factors (RF1 and RF2)** Proteins in prokaryotes responsible for termination of translation and release of the newly synthesized polypeptide when a nonsense codon appears in the A site of the ribosome. Replaced by eRF in eukaryotes.
- repetitive DNA** DNA made up of copies of the same nucleotide sequence.
- replica-planting** A technique to rapidly transfer microorganism colonies to numerous petri plates.
- replication-coupling assembly factor** A protein complex in fruit flies that assembles new nucleosomes.
- replicons** A replicating genetic unit including a length of DNA and its site for the initiation of replication.
- replisome** The DNA-replicating structure at the Y-junction, consisting of two DNA polymerase III enzymes and a primosome (primase and DNA helicase).
- reporter systems** Genetic constructs that allow an investigator to determine that a specific locus is active by measuring the phenotypic expression of an associated locus, such as the luciferase reporter, which glows if watered with luciferin.
- repressible system** A coordinated group of enzymes involved in a synthetic pathway (anabolic) is repressible if excess quantities of the end product of the pathway lead to the termination of transcription of the genes for the enzymes. These systems are primarily prokaryotic operons.
- repressor** The protein product of a regulator gene that acts to control transcription of inducible and repressible operons.
- reproductive isolating mechanisms** Environmental, behavioral, mechanical, and physiological barriers that prevent two individuals of different populations from producing viable progeny.
- reproductive success** The relative production of offspring by a particular genotype.
- repulsion** Allelic arrangement in which each homologous chromosome has mutant and wild-type alleles.
- resistance transfer factor** Infectious transfer part of R plasmids.

restricted transduction See specialized transduction.

restriction digest The results of the action of a restriction endonuclease on a DNA sample.

restriction endonucleases Endonucleases that recognize certain DNA sequences, then cleave them. They protect cells from viral infection; they are useful in recombinant DNA work.

restriction fragment length

polymorphism (RFLP) Variations (among individuals) in banding patterns of electrophoresed restriction digests.

restriction map A physical map of a piece of DNA showing recognition sites of specific restriction endonucleases separated by lengths marked in numbers of bases.

restriction site The sequence of DNA recognized by a restriction endonuclease.

restrictive temperature A temperature at which temperature-sensitive mutants display the mutant phenotype.

retinoblastoma A childhood cancer of retinoblast cells caused by the inactivation of an anti-oncogene.

retrotransposons Transposable genetic elements found in eukaryotic DNA that move through the reverse transcription of an RNA intermediate.

reverse transcriptase An enzyme that can use RNA as a template to synthesize DNA.

reversion The return of a mutant to the wild-type phenotype by way of a second mutational event.

R factors See R plasmids.

rho-dependent terminator A DNA sequence signaling the termination of transcription; termination requires the presence of the rho protein.

rho-independent terminator A DNA sequence signaling the termination of transcription; the rho protein is not required for termination.

rho protein A protein involved in the termination of transcription.

ribosomal RNA (rRNA) RNA components of the subunits of the ribosomes.

ribosome recycling factor (RRF) A protein needed to prepare ribosomal subunits that have just finished translating a messenger RNA for another cycle of translation.

ribosomes Organelles at which translation takes place. They are made up of two subunits consisting of RNA and proteins.

ribozyme Catalytic or autocatalytic RNA.

RNA editing The insertion of uridines into messenger RNAs after transcription is completed; controlled by guide RNA. May also involve insertion of cytidines in some organisms or possible deletions of bases.

RNA phages Phages whose genetic material is RNA. They are the simplest phages known.

RNA polymerase The enzyme that polymerizes RNA by using DNA as a template. (Also known as *transcriptase* or *RNA transcriptase*.)

RNA replicase A polymerase enzyme that catalyzes the self-replication of single-stranded RNA.

Robertsonian fusion Fusion of two acrocentric chromosomes at the centromere.

rolling-circle replication A model of DNA replication that accounts for a circular DNA molecule producing linear daughter double helices.

R plasmids Plasmids that carry genes that control resistance to various drugs.

rRNA See ribosomal RNA (rRNA).

rule of independent assortment See independent assortment, rule of.

rule of segregation See segregation, rule of.

sampling distribution The distribution of frequencies with which various possible events could occur, or a probability distribution defined by a particular mathematical expression.

sarcoma Tumor of tissue of mesodermal origin (e.g., muscle, bone, cartilage).

satellite DNA Highly repetitive eukaryotic DNA primarily located around centromeres. Satellite DNA usually has a different buoyant density than the rest of the cell's DNA.

scaffold The eukaryotic chromosomal structure that remains when DNA and histones have been removed.

scanning hypothesis Proposed mechanism by which the eukaryotic ribosome recognizes the initiation region of a messenger RNA after binding the 5' capped end of it. The ribosome scans the messenger RNA for the initiation codon.

scientific method A procedure scientists use to test hypotheses, making predictions about the outcome of an observation or experiment before the experiment is performed. The results provide support or refutation of the hypothesis.

screening technique A technique to isolate a specific genotype or phenotype of an organism.

secondary oocytes The cells formed by meiosis I in female animals.

secondary spermatocytes The products of the first meiotic division in male animals.

secondary structure The flat or helical configuration of the polypeptide backbone of a protein.

second-division segregation (SDS) The allelic arrangement in the spores of *Ascomycete* fungi with ordered spores that indicates a crossover between a locus and its centromere.

securin An inhibitory protein that prevents separin from acting on cohesin to separate sister chromatids.

segmentation genes Genes of developing embryos that determine the number and fate of segments.

segregation, rule of Mendel's first principle, which describes how genes pass from one generation to the next.

segregational load Genetic load caused when a population is segregating less fit homozygotes because of heterozygote advantage.

segregation distortion See gametic selection.

selection See natural selection.

selection coefficients, s , t The sum of forces acting to lower the relative reproductive success of a genotype.

selection-mutation equilibrium

An equilibrium allelic frequency resulting from the balance between selection against an allele and mutation re-creating this allele.

selective medium A culture medium enriched with a particular substance to allow the growth of particular strains of organisms.

selfed See self-fertilization.

self-fertilization Fertilization in which the two gametes come from the same individual.

selfish DNA A segment of the genome with no apparent function, although it can control its own copy number.

semiconservative replication The mode by which DNA replicates. Each strand acts as a template for a new double helix. See template.

semisterility Nonviability of a proportion of gametes or zygotes.

sense strand See coding strand.

separin An enzyme that breaks down cohesin and allows sister chromatids to separate at the start of anaphase of mitosis.

sequence-tagged sites (STSs) DNA lengths of 100–500 base pairs that are unique in the genome. They are created by polymerase chain reaction amplification of primers that are then tested to be sure the sequence is unique.

sex chromosomes Heteromorphic chromosomes whose distribution in a zygote determines the sex of the organism.

sex-conditioned traits Traits that appear more often in one sex than in another.

sex-determining region Y (SRY) The sex switch, or testis-determining factor, in human beings, located on the Y chromosome (*Sry* in mice).

sexduction A process whereby a bacterium gains access to and incorporates foreign DNA brought in by a modified F factor during conjugation.

sex-influenced traits See sex-conditioned traits.

Sex-lethal A gene in *Drosophila*, located on the X chromosome, that is a sex switch, directing development toward femaleness when in the “on” state. It is regulated by numerator and denominator elements that act to influence the genic balance ratio (X/A).

sex-limited traits Traits expressed in only one sex. They may be controlled by sex-linked or autosomal loci.

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Glossary

sex-linked The inheritance pattern of loci located on the sex chromosomes (usually the X chromosome in XY species); also refers to the loci themselves.

sex-ratio phenotype A trait in *Drosophila* whereby females produce mostly, if not only, daughters.

sex switch A gene in mammals, normally found on the Y chromosome, that directs the indeterminate gonads towards development as testes.

sexual selection The forces, determined by mate choice, that act to cause one genotype to mate more frequently than another genotype.

Shine-Dalgarno hypothesis A proposal that prokaryotic messenger RNA is aligned at the ribosome by complementarity between the messenger RNA upstream from the Initiation codon and the 3' end of the 16S ribosomal RNA.

shoot apical meristem The major meristematic tissue of the plant; surrounds the shoot.

short interspersed elements (SINEs) Sequences of DNA interspersed in eukaryotic chromosomes in many copies. Alu, a three-hundred base-pair sequence, is found about half a million times in human DNA.

shotgun cloning The random cloning of pieces of the DNA of an organism without regard to the genes or sequences present in the cloned DNA.

shunting Process in which the first initiation codon on a messenger RNA is bypassed for an initiation codon further down the messenger. The process is probably guided by secondary structure in the messenger.

siblings (sibs) Brothers and sisters.

sigma factor The protein that gives promoter-recognition specificity to the RNA polymerase core enzyme of bacteria.

signal hypothesis The major mechanism whereby proteins that must insert into or across a membrane are synthesized by a membrane-bound ribosome. The first thirteen to thirty-six amino acids synthesized, termed a *signal peptide*, are recognized by a *signal recognition particle* that draws the ribosome to the membrane surface. The signal peptide may be removed later from the protein.

signal peptide See signal hypothesis.

signal recognition particle See signal hypothesis.

signal transduction pathway A pathway in which the action of kinase enzymes that free transcription factors, or some other action, translates an environmental signal into some form of gene action.

single-nucleotide polymorphisms (SNPs) Differences between individuals involving single base pairs that are located about every 1,000 bases along the human genome. SNPs are useful for mapping disease genes.

single-strand binding proteins Proteins that attach to single-stranded DNA, usually near the replicating Y-junction, to stabilize the single strands.

sister chromatids See chromatids.

site-specific recombination A crossover event, such as the integration of phage λ , that requires homology of only a very short region and uses an enzyme specific for that recombination.

small nuclear ribonucleoproteins

(**snRNPs**) Components of the spliceosome, the intron-removing apparatus in eukaryotic nuclei. See snRNPs.

small nucleolar ribonucleoprotein

particles (snoRNPs) Particles composed of RNA and protein found in the nucleolus that modify ribosomal RNAs, particularly by converting some uridines to pseudouridines and methylating some ribose sugars.

small nucleolar RNAs (snoRNAs) RNAs found in small nucleolar ribonucleoprotein particles (snoRNPs) that take part in modifying ribosomal RNA in the nucleolus.

SMC proteins For structural maintenance of chromosomes; proteins that aid mitotic segregation, sister-chromatid adhesion, dosage compensation, recombination, and other chromosomal activities.

snRNPs See small nuclear ribonucleoproteins.

sociobiology The study of the evolution of social behavior in animals.

somatic doubling A disruption of the mitotic process that produces a cell with twice the normal chromosome number.

somatic hypermutation The occurrence of a high level of mutation in the variable regions of immunoglobulin genes.

SOS box The region of the promoters of various genes that the LexA repressor recognizes. Release of repression results in the induction of the SOS response.

SOS response Repair systems (RecA, Uvr) induced by the presence of single-stranded DNA that usually occurs from postreplicative gaps caused by various types of DNA damage. The RecA protein, stimulated by single-stranded DNA, is involved in the inactivation of the LexA repressor, thereby inducing the response.

Southern blotting A method, first devised by E. M. Southern, used to transfer DNA fragments from an agarose gel to a nitrocellulose gel for the purpose of DNA-DNA or DNA-RNA hybridization during recombinant DNA work.

specialized transduction Form of transduction based on faulty looping out by a temperate phage. Only neighboring loci to the attachment site can be transduced. See generalized transduction.

speciation A process whereby, over time, one species evolves into a different species (anagenesis) or one species diverges to become two or more species (cladogenesis).

species A group of organisms capable of interbreeding to produce fertile offspring.

specific transcription factors Proteins needed for activation of transcription at specific eukaryotic promoters. Also, negative factors that can inhibit transcription at a specific eukaryotic promoter.

spermatids The four products of meiosis in males that develop into sperm.

spermatogenesis The process of sperm production.

spermatogonium A cell type in the testes of male vertebrates that gives rise to primary spermatocytes by mitosis.

sperm cells The gametes of males.

spermiogenesis The process by which spermatids mature into sperm cells.

spindle The microtubule apparatus that controls chromosomal movement during mitosis and meiosis.

spindle pole body Spindle microtubule organizing center found in fungi.

spiral cleavage The cleavage process in mollusks and some other invertebrates whereby the spindle is tipped at mitosis in relation to the original egg axis.

spirillum A spiral bacterium.

spliceosome Protein-RNA complex that removes introns in eukaryotic nuclear RNAs.

sporophyte The stage of a plant life cycle that produces spores by meiosis and alternates with the gametophyte stage.

stabilizing selection A type of selection that removes individuals from both ends of a phenotypic distribution, thus maintaining the same distributional mean.

standard deviation The square root of the variance.

standard error of the mean The standard deviation divided by the square root of the sample size. It is the standard deviation of a sample of means.

statistics Measurements of attributes of a sample from a population; denoted by Roman letters. See parameters.

stem-loop structure A lollipop-shaped structure formed when a single-stranded nucleic acid molecule loops back on itself to form a complementary double helix (stem), topped by a loop.

stepladder gel A DNA-sequencing gel. The numerous bands in each lane give the appearance of a stepladder.

stochastic A process with an indeterminate or random element as opposed to a deterministic process with no random element.

stringent factor A protein that catalyzes the formation of an unusual nucleotide (guanosine tetraphosphate) during the stringent response under amino acid starvation.

stringent response A translational control mechanism in prokaryotes that represses transfer RNA and ribosomal RNA synthesis during amino acid starvation.

structural alleles Alleles whose alterations include some of the same base pairs.

submetacentric chromosome A chromosome whose centromere lies between its middle and its end, but closer to the middle.

subtelocentric chromosome

A chromosome whose centromere lies between its middle and its end, but closer to the end.

sum rule The rule that states that the probability that two mutually exclusive events will occur is the sum of the probabilities of the individual events.

supercoiling Negative or positive coiling of double-stranded DNA that differs from the relaxed state.

supergenes Several loci, which usually control related aspects of the phenotype, in close physical association.

suppressor gene A gene that, when mutated, apparently restores the wild-type phenotype to a mutant of another locus.

surveillance mechanism Used to describe mechanisms that oversee checkpoints in the cell cycle, points in which the process can be halted if certain conditions are not met.

survival of the fittest In evolutionary theory, survival of only those organisms best able to obtain and utilize resources (the fittest). This phenomenon is the cornerstone of Darwin's theory of evolution.

Svedberg unit A unit of sedimentation during centrifugation. Abbreviation is S, as in 50S.

swivelase See DNA gyrase.

sympatric speciation Speciation in which reproductive isolating mechanisms evolve within the range and habitat of the parent species. This type of speciation may be common in parasites.

synapsis The point-by-point pairing of homologous chromosomes during zygonema or in certain dipteran tissues that undergo endomitosis.

synaptonemal complex A proteinaceous complex that apparently mediates synapsis during the zygotene stage and then disintegrates.

syncytium A cell that has many nuclei not separated by cell membranes.

synexpression group A group of eukaryotic genes that are involved in the same function or pathway and are induced together.

synteny test A test that determines whether two loci belong to the same linkage group by observing concordance in hybrid cell lines.

synthetic medium A chemically defined substrate that microorganisms are grown upon.

TACTAAC box A consensus sequence surrounding the lariat branch point of eukaryotic messenger RNA introns.

TATA-binding protein A protein, part of TFIID, that binds the TATA consensus sequence in eukaryotic promoters.

TATA box An invariant DNA sequence at about -25 in the promoter region of eukaryotic genes; analogous to the Pribnow box in prokaryotes.

tautomeric shift Reversible shifts of proton positions in a molecule. The bases in nucleic acids shift between the keto and enol forms or between the amino and imino forms.

TBP-associated factors (TAFs) Proteins that bind with the TATA-binding protein to form TFIID. They aid in the selectivity of TFIID.

T-cell receptors Surface proteins of T cells that allow the T cells to recognize host cells that have been infected.

telocentric chromosome A chromosome whose centromere lies at one of its ends.

telomerase An enzyme that adds telomeric sequences to the ends of eukaryotic chromosomes.

telomeres The ends of linear chromosomes.

telophase The terminal stage of mitosis or meiosis in which chromosomes uncoil, the spindle breaks down, and cytokinesis usually occurs.

telson The posterior end of the arthropod embryo, where the end of the alimentary canal is located.

temperate phage A phage that can enter into lysogeny with its host.

temperature-sensitive mutant An organism with an allele that is normal at a permissive temperature but mutant at a restrictive temperature.

template A pattern serving as a mechanical guide. In DNA replication, each strand of the duplex acts as a template for the synthesis of a new double helix.

template strand See anticoding strand.

terminator sequence A sequence in DNA that signals the termination of transcription to RNA polymerase.

terminator stem A configuration of the leader transcript that signals transcriptional termination in attenuator-controlled amino acid operons.

tertiary structure The further folding of a protein, bringing α helices and β sheets into three-dimensional arrangements.

testcross The cross of an organism with a homozygous recessive organism.

testing of hypotheses The determination of whether to reject or fail to reject a proposed hypothesis based on the likelihood of the experimental results.

testis-determining factor (TDF) General term for the gene determining maleness in human beings (*Tdf* in mice).

tetrads The meiotic configuration of four chromatids first seen in pachynema. There is one tetrad (bivalent) per homologous pair of chromosomes.

tetranucleotide hypothesis

The hypothesis, based on incorrect information, that DNA could not be the genetic material because its structure was too simple—that is, that repeating subunits contain one copy each of the four DNA nucleotides.

tetraploids Organisms with four whole sets of chromosomes.

tetratype (TT) A spore arrangement in Ascomycete fungi that consists of the two parental and two recombinant spores.

theta structure An intermediate structure formed during the replication of a circular DNA molecule.

three-point cross A cross involving three loci.

thymine See pyrimidines.

t-loop A loop that forms at the end of mammalian telomeres by the interdigitation of the 3' free end into the DNA double helix.

topoisomerase An enzyme that can relieve (or create) supercoiling in DNA by creating transitory breaks in one (type I) or both (type II) strands of the helical backbone.

topoisomers Forms of DNA with the same sequence but differing in their linkage number (coiling).

totipotent The state of a cell that can give rise to any and all adult cell types, as compared with a differentiated cell whose fate is determined.

trailer The length of messenger RNA from the nonsense codon to the 3' end or, in polycistronic messenger RNAs, from a nonsense codon to the next gene's leader.

trans Meaning "across" and referring usually to the geometric configuration of mutant alleles across from each other on a homologous pair of chromosomes.

trans-acting Referring to mutations of, for example, a repressor gene, that act through a diffusible protein product; the normal mode of action of most recessive mutations.

transcription The process whereby RNA is synthesized from a DNA template.

transcription factors Eukaryotic proteins that aid RNA polymerase in recognizing promoters. See general transcription factors and specific transcription factors.

transducing particle A defective phage, carrying part of the host's genome.

transduction A process whereby a cell can gain access to and incorporate foreign DNA brought in by a viral particle.

transfection The introduction of foreign DNA into eukaryotic cells.

transfer operon (*tra*) Sequence of loci that impart the male (F-pili-producing) phenotype on a bacterium. The male cell can transfer the F plasmid to an F⁻ cell.

transfer RNA (tRNA) Small RNA molecules that carry amino acids to the ribosome for polymerization.

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Glossary

transformation A process whereby prokaryotes take up DNA from the environment and incorporate it into their genomes, or the conversion of a eukaryotic cell into a cancerous one.

transgenic Eukaryotic organisms that have taken up foreign DNA.

transition mutation A mutation in which a purine-pyrimidine base pair replaces a base pair in the same purine-pyrimidine relationship.

translation The process of protein synthesis wherein the nucleotide sequence in messenger RNA determines the primary structure of the protein.

translocase (EF-G) Elongation factor in prokaryotes necessary for proper translocation at the ribosome during the translation process. Replaced by eEF2 in eukaryotes.

translocation A chromosomal configuration in which part of a chromosome becomes attached to a different chromosome. Also a part of the translation process in which the messenger RNA is shifted one codon in relation to the ribosome.

translocation channel (translocon) A protein-lined pore or channel in a membrane through which nascent proteins are transported during translation.

transposable genetic element A region of the genome, flanked by inverted repeats, a copy of which can be inserted at another place; also called a transposon or a jumping gene.

transposon See transposable genetic element.

transversion mutation A mutation in which a purine replaces a pyrimidine, or vice versa.

trihybrid An organism heterozygous at three loci.

triploids Organisms with three whole sets of chromosomes.

trisomic A diploid cell with an extra chromosome.

tRNA See transfer RNA (tRNA).

trp RNA-binding attenuation protein (TRAP) Protein that can bind to the attenuation region of the messenger RNA of the tryptophan operon in *Bacillus subtilis*, causing a terminator stem to form and halting further transcription.

true heritability See heritability.

tumor Abnormal growth of tissue.

tumor-suppressor genes Genes that normally prevent unlimited cellular growth. When both copies of the gene are mutated, cellular transformation follows. Examples are the *p53* gene and the genes for retinoblastoma and Wilm's tumor.

two-point cross A cross involving two loci.

type I error In statistics, the rejection of a true hypothesis.

type II error In statistics, the accepting of a false hypothesis.

typological thinking The concept that organisms of a species conform to a specific norm. In this view, variation is considered abnormal.

ubiquitin A peptide of twenty-six amino acid residues that enzymes attach to proteins that the proteasome will degrade.

unequal crossing over Nonreciprocal crossing over caused by the mismatching of homologous chromosomes. Usually occurs in regions of tandem repeats.

uninemic chromosome A chromosome consisting of one DNA double helix.

unique DNA A length of DNA with no repetitive nucleotide sequences.

unmixed families Groups of four codons sharing their first two bases and coding for the same amino acid.

unusual bases Other bases, in addition to adenine, cytosine, guanine, and uracil, found primarily in transfer RNAs.

UP element See upstream element.

upstream A convention on DNA related to the position and direction of transcription by RNA polymerase (5'→3'). Downstream (or 3' to) is in the direction of transcription whereas upstream (5' to) is in the direction from which the polymerase has come.

upstream element A sequence of about twenty AT-rich bases centered at -50 in promoters of prokaryotic genes that are expressed strongly.

uracil See pyrimidines.

variable-number-of-tandem-repeats (VNTR) loci Loci that are hypervariable because of tandem repeats. Presumably, variability is generated by unequal crossing over.

variance The average squared deviation about the mean of a set of data.

variegation Patchiness; a type of position effect that results when particular loci are contiguous with heterochromatin.

virion A virus particle.

viroids Bare RNA particles that are plant pathogens.

V(D)J joining The process of joining variable, diversity, and joining gene segments (V-J, V-D, or D-J joining) in the formation of a functioning immunoglobulin gene.

Wahlund effect The effect of subdivision on a population, causing it to contain fewer heterozygotes than predicted despite the fact that all subdivisions are in Hardy-Weinberg proportions.

western blotting A technique for probing for a particular protein using antibodies. See Southern blotting.

whole-genome shotgun method

A method of sequencing entire genomes by breaking up the genomes into small pieces, sequencing the pieces, and then using computers to establish order by overlapping the sequences.

wild-type The phenotype of a particular organism when it is first seen in nature.

Wilm's tumor A childhood kidney cancer caused by the inactivation of an anti-oncogene.

wobble Referring to the reduced constraint over the third base of an anticodon as compared with the other bases, thus allowing additional complementary base pairings.

xeroderma pigmentosum A disease in human beings caused by a defect in the UV mutation repair system.

X-inactivation center (XIC) Locus at which inactivation is initiated on the X chromosome in mammals.

X linked See sex-linked.

X-ray crystallography A technique, using X rays, to determine the atomic structure of molecules that have been crystallized.

yeast artificial chromosome (YAC) Originating from a bacterial plasmid, a YAC contains additionally a yeast centromeric region (CEN) and a yeast origin of DNA replication (ARS). YACs are capable of including large pieces of foreign DNA during cloning.

Y-junction The point of active DNA replication where the double helix opens up so that each strand can serve as a template.

Y linked Inheritance pattern of loci located on the Y chromosome only. Also refers to the loci themselves.

Z DNA A left-handed form of DNA found under physiological conditions in short GC segments that are methylated. It may be involved in regulating gene expression in eukaryotes.

ZFY gene Originally believed to be the human male sex-switch gene, located on the short arm of the Y chromosome. *ZFY* stands for zinc finger on the Y chromosome.

zinc finger Configuration of a DNA-binding protein that resembles a finger with a base, usually cysteines and histidines, binding a zinc ion. Discovered in a transcription factor in *Xenopus*.

zygonema (zygotene stage) The stage of prophase I of meiosis in which synapsis occurs.

zygotic induction The beginning of vegetative growth when a prophage is passed into an F⁺ cell during conjugation.

zygotic selection The forces acting to cause differential mortality of an organism at any stage (other than gametes) in its life cycle.