Large-Scale Chromosomal Changes

WORKING WITH THE FIGURES

1. Based on Table 17-1, how would you categorize the following genomes? (Letters H through J stand for four different chromosomes.)

   \[
   \begin{align*}
   & HH \ II \ J \ KK \\
   & HH \ II \ JJ \ KKK \\
   & HHHH \ II \ III \ JJJJ \ KKKK
   \end{align*}
   \]

   Answer: Monosomic (\(2n-1\)) 7 chromosomes
   Trisomic (\(2n+1\)) 9 -II-
   Tetraploid (\(4n\)) 16 -II-

2. Based on Figure 17-4, how many chromatids are in a trivalent?

   Answer: There are 6 chromatids in a trivalent.

3. Based on Figure 17-5, if colchicine is used on a plant in which \(2n = 18\), how many chromosomes would be in the abnormal product?

   Answer: Colchicine prevents migration of chromatids, and the abnormal product of such treatment would keep all the chromatids (\(2n = 18\)) in one cell.

4. Basing your work on Figure 17-7, use colored pens to represent the chromosomes of the fertile amphidiploid.

   Answer: A fertile amphidiploids would be an organism produced from a hybrid with two different sets of chromosomes (\(n_1\) and \(n_2\)), which would be infertile until some tissue undergoes chromosomal doubling (\(2n_1 + 2n_2\)) and such chromosomal set would technically become a diploid (each chromosome has its pair; therefore they could undergo meiosis and produce gametes). This could be a new species.

   Picture example: 3 pairs of chromosomes/ different lengths/ red for \(n_1 = 3\)
5. If Emmer wheat (Figure 17-9) is crossed to another wild wheat CC (not shown), what would be the constitution of a sterile product of this cross? What amphidiploid could arise from the sterile product? Would the amphidiploid be fertile?

Answer: Emmer wheat was domesticated 10,000 years ago, as a tetraploid with two chromosome sets (2\(n_1 + 2 n_2\) or \(AA + BB\)). If \(AA BB\) tetraploid genome is combined with another wild wheat (\(n_3\) or \(C\) gamete), the product would be sterile. In such case, \(C\) chromosomes would not have homologous pairs in a hybrid parent wheat. Amphidiploid could occur, if chromosome doubling happens in a parental tissue (e.g., flower parts), and a fertile wheat species would be hexaploid (\(AA BB CC\)).

6. In Figure 17-12, what would be the constitution of an individual formed from the union of a monosomic from a first-division nondisjunction in a female and a disomic from a second-division nondisjunction in a male, assuming the gametes were functional?

Answer: A gamete from a first-division nondisjunction would be an egg without the chromosome in question (\(n - 1\)); while a gamete/sperm from a second-division non-disjunction would be a (\(n + 1\)). If both gametes are functional, they would result in a euploid (2\(n\)) zygote, with two copies of a father’s chromosome.

7. In Figure 17-14, what would be the expected percentage of each type of segregation?

Answer: These are three equally possible combinations/ segregation of chromosomes in meiosis, in a trisomic individual. Therefore each type is expected at 33.3%

8. In Figure 17-19, is there any difference between the inversion products formed from breakage and those formed from crossing over?

Answer: Inversion products look the same, having the sequence: 1-3-2-4, yet they could be genetically different. Breakage and rejoining (on the left) results in a rearrangement with possibly damaged gene sequence and yet the same length of the chromosome. Crossing over between repetitive DNA (blue) might
generate an inversion with less damage to the gene sequence, since the crossing over happens in homologous regions of a repetitive sequence.

9. Referring to Figure 17-19, draw a diagram showing the process whereby an inversion formed from crossing over could generate a normal sequence.

Answer: An inversion formed by a crossing over: _1 > 3 2 < 4_. Repetitive sequences are homologous (~< and ~> oriented) and they could pair and form crossing over in the next generation of gametes, producing a normal segment again: _1 < 2 3 > 4_.

10. In Figure 17-21, would the recessive _fa_ allele be expressed when paired with deletion 264-32? 258-11?

Answer: Deletion 264-32 overlaps with the segment carrying _fa_ allele (7) on polytene chromosome; this means that the _fa_ allele would be pseudodominant (expressed) in such combination of chromosomes. Deletion 258-11 is covering segments before the _fa_ allele and the gene would not be expressed. In this case a dominant allele (_fa+_) of the 258-11 chromosome will show in the phenotype.

11. Look at Figure 17-22 and state which bands are missing in the cri du chat deletion.

Answer: Cri du chat syndrome in humans is caused by the deletion of the tip of the p5 (bands 15.3 and 15.2).

12. In Figure 17-25, which species is most closely related to the ancestral yeast strain? Why are genes 3 and 13 referred to as duplicate?

Answer: _Kluyveromyces waitii_ seems to be most similar to the common ancestor. When genome of _Saccharomyces_ lineage doubled, genes such as 3 and 13 become duplicate in both species and in the same relative order. Some other genes were lost (2, 7, etc).

13. Referring to Figure 17-26, draw the product if breaks occurred within genes _A_ and _B_.

Answer: 5' _A_ X _p_ B_ X_ C D 3'

3' _A_ X _p_ _X_ C D 5'

Z Inversion _Z_
After the joining of the breaks, genes $A$ and $B$ become disrupted: $5'\text{“A” “BA” “B” C D 3’}$ and the final product may have the fused parts of genes $A$ and $B$: $5'\text{“A” B/A “B” C D 3’}$

14. In Figure 17-26, the bottom panel shows that genes $B$ and $C$ are oriented in a different direction (note the promoters). Do you think this difference in orientation would affect their functionality?

Answer: If promoter positions are changed, this could interfere with gene function because of the reading frame for RNA polymerase. An inversion of the gene order can alter normal expression of the genes by placing a gene in the new regulatory environment.

15. In Figure 17-28, what would be the consequence of a crossover between the centromere and locus $A$?

Answer: Inversions that include centromere are called pericentric. If the breakage happens between the centromere (o) and the locus $A$:

$\begin{align*}
o & \rightarrow \text{X} & A & B & C & D & E \\
o & \rightarrow \text{X} & A & D & C & B & E
\end{align*}$

Crossing over products might be both normal and inverted:

$E \ B \ C \ D \rightarrow o-- \ A$

16. Based on Figure 17-30, are normal genomes ever formed from the two types of segregation? Are normal genomes ever formed from an adjacent-1 segregation?

Answer: With the adjacent 1 segregation final meiotic products are often inviable due to a deletion of a significant gene segment. In plants such gametes do not function, while in animals gametes might be fine (but not the zygotes).

17. Referring to Figure 17-32, draw an inviable product from the same meiosis.

Answer: This figure shows a translocation heterozygote:

$$a \quad b \quad \text{Not linked} \quad b+ \quad a+ \quad \text{Pseudo-linked}$$

After meiosis some products would be inviable because they carry deletions of one or another gene segment, such as:

$$a \quad \text{or} \quad b \quad , \text{etc.}$$

The only viable progeny are the ones with parental genotypes and pseudo-linkage is an indication of reciprocal translocation.
18. Based on Figure 17-35, write a sentence stating how translocation can lead to cancer. Can you think of another genetic cause of cancer?

Answer: This figure illustrates how relocation of an oncogene (via translocation) might cause cancer in somatic cells, such as Burkitt’s lymphoma and chronic myelogenous leukemia. In general, any chromosomal rearrangement that changes the regulatory environment of a gene could lead to cancer.

Other causes would be based on a mutation in a gene which might control cell division or a gene that suppresses growth of tumors. Finally, mutations in genes responsible for repair mechanisms at the molecular or cellular level might also be genetic causes of different types of cancer.

19. Looking at Figure 17-36, why do you think the signal ratio is so much higher in the bottom panel?

Answer: Comparative genomic hybridization technique compares mutant to wild type ratios and the ratio of values of each cDNA is calculated. For example, duplication would have a ratio higher than 1 (more gene product), while a deletion will have less than 1 (less product) in a chromosomal microarray. The last graph indicated tandem amplification of a gene (multiple copies) which lead to a very high ratio (close to 10).

20. Using Figure 17-37, calculate what percentage of conceptions are triploid. The same figure shows XO in the spontaneous-abortion category; however, we know that many XO individuals are viable. In which of the viable categories would XO be grouped?

Answer: Figure 17-37 shows the proportion of chromosomal mutations in human conceptions (zygotes). Triploid zygotes (3n) were not found in the live births, but only in the spontaneous abortions (12,750 per 1,000,000 conceptions). Therefore only around 1.275 % of conceptions were triploid (or with 69 chromosomes).

Turner syndrome (45 X0) is an aneuploidy (2n-1). It is found in both live births (some of the 422 females with sex chromosome aneuploidy) and in spontaneous abortions (13,500 per 1,000,000 conceptions). Therefore viable XO (Turner syndrome females) belong to the category of sex chromosome aneuploids, under live births with chromosome abnormalities. Other than Turner syndrome, sex chromosome aneuploid females might also be 47 XXX, so the percentage of living XO is not specified in this figure (out of 422 births).
BASIC PROBLEMS

21. In keeping with the style of Table 17-1, what would you call organisms that are MM N OO; MM NN OO; MMM NN PP?

Answer: MM N OO would be classified as $2n - 1$ (monosomic); MM NN OO would be classified as $2n$ (euploid); and MMM NN PP would be classified as $2n + 1$ (trisomic).

22. A large plant arose in a natural population. Qualitatively, it looked just the same as the others, except much larger. Is it more likely to be an allopolyploid or an autopolyploid? How would you test that it was a polyploid and not just growing in rich soil?

Answer: It would more likely be an autopolyploid. To make sure it was polyploid, you would need to microscopically examine stained chromosomes from mitotically dividing cells and count the chromosome number.

23. Is a trisomic an aneuploid or a polyploid?

Answer: Aneuploid. Trisomic refers to three copies of one chromosome. Triploid refers to three copies of all chromosomes.

24. In a tetraploid $B/B/b/b$, how many quadrivalent possible pairings are there? Draw them (see Figure 17-5).

Answer: There would be one possible quadrivalent with 50 percent of recombinant products ($B/b$).

25. Someone tells you that cauliflower is an amphidiploid. Do you agree? Explain.

Answer: No. Amphidiploid means “doubled diploid ($2n_1 + 2n_2$).” Because cauliflower has $n = 9$ chromosome, it could not have arose in this fashion. It has, however, contributed to other amphidiploid species, such as rutabaga.

26. Why is Raphanobrassica fertile, whereas its progenitor wasn’t?

Answer: The progenitor had nine chromosomes from a cabbage parent and nine chromosomes from a radish parent. These chromosomes were different enough that pairs did not synapse and segregate normally at meiosis. By doubling the chromosomes in the progenitor ($2n = 36$), all chromosomes now had homologous partners and meiosis could proceed normally.
27. In the designation of wheat genomes, how many chromosomes are represented by the letter B?

Answer: In modern hexaploid wheat (*T. aestivum*) there are 2n or 6x chromosomes in its genome, with a total of 42. If each haploid set has the same number of chromosomes, B (or x) represents seven chromosomes.

28. How would you “re-create” hexaploid bread wheat from *Triticum tauschii* and Emmer?

Answer: Cross *T. tauschii* and Emmer to get ABD offspring. Treat the offspring with colchicine to double the chromosome number to AABBDD to get the hexaploid bread wheat.

29. How would you make a monoploid plantlet by starting with a diploid plant?

Answer: Cells destined to become pollen grains can be induced by cold treatment to grow into embryoids. These embryoids can then be grown on agar to form monoploid plantlets.

30. A disomic product of meiosis is obtained. What is its likely origin? What other genotypes would you expect among the products of that meiosis under your hypothesis?

Answer: The likely origin of a disomic (*n* + 1) gamete is nondisjunction during meiosis. Depending whether the nondisjunction took place during the first or second division, you would expect one nullosomic (*n* – 1), or two nullosomics and another disomic, respectively.

31. Can a trisomic A/A/a ever produce a gamete of genotype a?

Answer: Yes. You would expect that one-sixth of the gametes would be *a*. Also, two-sixths would be *A*, two-sixths would be *Aa*, and one-sixth would be *AA*.

32. Which, if any, of the following sex-chromosome aneuploids in humans are fertile: XXX, XXY, XYY, XO?

Answer: Both XYY (male) and XXX (female) would be fertile. XO (Turner syndrome) and XXY (Klinefelter syndrome) are known to be sterile.
33. Why are older expectant mothers routinely given amniocentesis or CVS?

Answer: Older mothers have an elevated risk of having a child with some chromosomal aberration, due to the age of their egg cells. Down syndrome and other aneuploidy due to the meiotic nondisjunction in mother’s gametogenesis are among the most common. However, age of the father also contributes to the increased risk of some chromosomal aberrations.

34. In an inversion, is a 5’ DNA end ever joined to another 5’ end? Explain.

Answer: No. The DNA backbone has strict 5’ to 3’ polarity, and 5’ ends can only be joined to 3’ ends.

35. If you observed a dicentric bridge at meiosis, what rearrangement would you predict had taken place?

Answer: A cross over within a paracentric inversion heterozygote results in a dicentric bridge (and an acentric fragment).

36. Why do acentric fragments get lost?

Answer: By definition, an acentric fragment has no centromere, so it cannot be aligned or moved during meiosis (or mitosis). Consequently, at the end of a cell division, it gets left in the cytoplasm where it is not replicated.

37. Diagram a translocation arising from repetitive DNA. Repeat for a deletion.

Answer:
Possible translocation:

```
  ------  ------  ------
            X
  ------  ------  ------
```

Possible deletion (and duplication):

```
  ------  ------  ------  ------
            X
  ------  ------  ------
```
38. From a large stock of *Neurospora* rearrangements available from the fungal genetics stock center, what type would you choose to synthesize a strain that had a duplication of the right arm of chromosome 3 and a deletion for the tip of chromosome 4?

Answer: You could cross a strain with the appropriate 3; 4 reciprocal translocation to a wild-type strain to generate heterozygotes. One of the meiotic products of adjacent-1 segregation in these heterozygotes will have a duplication of the translocated portion of chromosome 3 and a deletion for the translocated portion of chromosome 4.

39. You observe a very large pairing loop at meiosis. Is it more likely to be from a heterozygous inversion or heterozygous deletion? Explain.

Answer: Very large deletions tend to be lethal. This is likely due to genomic imbalance or the unmasking of recessive lethal genes. Therefore, the observed very large pairing loop is more likely to be from a heterozygous inversion.

40. A new recessive mutant allele doesn’t show pseudodominance with any of the deletions that span *Drosophila* chromosome 2. What might be the explanation?

Answer: Because the new mutant does not show pseudodominance with any of the deletions that span chromosome 2, it is likely that the mutation does not map to this chromosome, or it is located in a different region of the chromosome 2 in *Drosophila*.

41. Compare and contrast the origins of Turner syndrome, Williams syndrome, cri du chat syndrome, and Down syndrome. (Why are they called syndromes?)

Answer: Turner syndrome is a monosomy in X chromosomes (45 XO) due to a meiotic nondisjunction. Down syndrome (47 or trisomy 21) results from meiotic nondisjunction or from a Robertsonian translocation (with 46 chromosomes but a translocation between 21 and 14). Williams syndrome is the result of a deletion of the 7q11.23 region of chromosome 7. Cri du chat syndrome is the result of a deletion of a significant portion of the short arm of chromosome 5 (specifically bands 5p15.2 and 5p15.3). The term *syndrome* is used to describe a set of phenotypic changes (often complex and varied) that generally occur
together with a specific human chromosomal aberration. All four of these syndromes often include mental retardation and unique body and facial features. In addition, fatality rates are low and such children in most cases reach adulthood.

42. List the diagnostic features (genetic or cytological) that are used to identify these chromosomal alterations:

a. Deletions
b. Duplications
c. Inversions
d. Reciprocal translocations

Answer:

a. Cytologically, deletions lead to shorter chromosomes with missing bands (if banded) and an unpaired loop during meiotic pairing when heterozygous. Genetically, deletions are usually lethal when homozygous, do not revert, and when heterozygous, lower recombinational frequencies and can result in "pseudodominance" (the expression of recessive alleles on one homolog that are deleted on the other). Occasionally, heterozygous deletions express an abnormal (mutant) phenotype.

b. Cytologically, duplications lead to longer chromosomes and, depending on the type, unique pairing structures during meiosis when heterozygous. These may be simple unpaired loops or more complicated twisted loop structures. Genetically, duplications can lead to asymmetric pairing and unequal crossing-over events during meiosis, and duplications of some regions can produce specific mutant phenotypes. As in deletions, we could detect duplications using their hybridization signals.

c. Cytologically, inversions can be detected by banding, and when heterozygous, they show the typical twisted "inversion" loop during homologous pairing. Pericentric inversions can result in a change in the \( p:q \) ratio (the position of the centromere). Genetically, no viable crossover products are seen from recombination within the inversion when heterozygous, and as a result, flanking genes show a decrease in RF.

d. Cytologically, reciprocal translocations may be detected by banding, or they may drastically change the size of the involved chromosomes as well as the positions of their centromeres. Genetically, they establish new linkage relationships. When heterozygous, they show the typical cross structure during meiotic pairing and cause a diagnostic 50 percent reduction of viable gamete production, leading to semisterility.
43. The normal sequence of nine genes on a certain *Drosophila* chromosome is 123·456789, where the dot represents the centromere. Some fruit flies were found to have aberrant chromosomes with the following structures:

a. 123 · 476589  
b. 123 · 46789  
c. 1654 · 32789  
d. 123 · 4566789

Name each type of chromosomal rearrangement, and draw diagrams to show how each would synapse with the normal chromosome.

**Answer:**  
a. Paracentric inversion


![Paracentric inversion diagram](attachment:paracentric_inversion.png)  
b. Deletion


![Deletion diagram](attachment:deletion.png)  
c. Pericentric inversion


![Pericentric inversion diagram](attachment:pericentric_inversion.png)  
d. Duplication


![Duplication diagram](attachment:duplication.png)

44. The two loci *P* and *Bz* are normally 36 m.u. apart on the same arm of a certain plant chromosome. A paracentric inversion spans about one-fourth of this region but does not include either of the loci. What approximate recombinant frequency between *P* and *Bz* would you predict in plants that are

a. heterozygous for the paracentric inversion?  
b. homozygous for the paracentric inversion?

**Answer:**  
a. The products of crossing-over within the inversion will be inviable when the inversion is heterozygous. This paracentric inversion spans 25 percent of the region between the two loci and therefore will reduce the observed
recombination between these genes by a similar percentage (i.e., 9 percent.) The observed RF will be 27 percent.

b. When the inversion is homozygous, the products of crossing-over within the inversion will be viable, so the observed RF will be 36 percent.

45. As stated in Solved Problem 2, certain mice called waltzers have a recessive mutation that causes them to execute bizarre steps. W. H. Gates crossed waltzers with homozygous normals and found, among several hundred normal progeny, a single waltzing female mouse. When mated with a waltzing male, she produced all waltzing offspring. When mated with a homozygous normal male, she produced all normal progeny. Some males and females of this normal progeny were intercrossed, and there were no waltzing offspring among their progeny. T. S. Painter examined the chromosomes of waltzing mice that were derived from some of Gates’s crosses and that showed a breeding behavior similar to that of the original, unusual waltzing female. He found that these mice had 40 chromosomes, just as in normal mice or the usual waltzing mice. In the unusual waltzers, however, one member of a chromosome pair was abnormally short. Interpret these observations as completely as possible, both genetically and cytologically.

(Problem 45 is from A. M. Srb, R. D. Owen, and R. S. Edgar, General Genetics, 2nd ed. Copyright 1965, W. H. Freeman and Company.)

Answer: The following represents the crosses that are described in this problem:

Waltzers  \times

1 Waltzing female (among 100s of progeny)

Normal male ×  \times Waltzing male

All

Some of these are intercrossed

No

The single waltzing female that arose from a cross between waltzers and normals is expressing a recessive gene. It is possible that this represents a new “waltzer” mutation that was inherited from one of the “normal” mice, but given
the cytological evidence (the presence of a shortened chromosome), it is more likely that this exceptional female inherited a deletion of the wild-type allele, which allowed expression of the mutant recessive phenotype.

When this exceptional female was mated to a waltzing male, all the progeny were waltzers; when mated to a normal male, all the progeny were normal. When some of these normal offspring were intercrossed, there were no progeny that were waltzers. If a “new” recessive waltzer allele had been inherited, all these “normal” progeny would have been \( w^+/w \). Any intercross should have therefore produced 25 percent waltzers. On the other hand, if a deletion had occurred, half the progeny would be \( w^+/w \) and half would be \( w^+/w^{\text{deletion}} \). If \( w^+/w^{\text{deletion}} \) are intercrossed, 25 percent of the progeny would not develop (the homozygous deletion would likely be lethal), and no waltzers would be observed. This is consistent with the data.

46. Six bands in a salivary-gland chromosome of *Drosophila* are shown in the following illustration, along with the extent of five deletions (Del 1 to Del 5):

![Diagram of salivary-gland chromosome with deletions]

Recessive alleles \( a, b, c, d, e, \) and \( f \) are known to be in the region, but their order is unknown. When the deletions are combined with each allele, the following results are obtained:

<table>
<thead>
<tr>
<th>Deletion</th>
<th>( a )</th>
<th>( b )</th>
<th>( c )</th>
<th>( d )</th>
<th>( e )</th>
<th>( f )</th>
</tr>
</thead>
<tbody>
<tr>
<td>Del 1</td>
<td>–</td>
<td>–</td>
<td>–</td>
<td>+</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>Del 2</td>
<td>–</td>
<td>+</td>
<td>–</td>
<td>+</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>Del 3</td>
<td>–</td>
<td>+</td>
<td>–</td>
<td>+</td>
<td>–</td>
<td>+</td>
</tr>
<tr>
<td>Del 4</td>
<td>+</td>
<td>+</td>
<td>–</td>
<td>–</td>
<td>–</td>
<td>+</td>
</tr>
<tr>
<td>Del 5</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>–</td>
<td>–</td>
<td>–</td>
</tr>
</tbody>
</table>

In this table, a minus sign means that the deletion is missing the corresponding wild-type allele (the deletion uncovers the recessive), and a plus sign means that the corresponding wild-type allele is still present. Use these data to infer which salivary band contains each gene. (Problem 46 is from D. L. Hartl, D. Friefelder, and L. A. Snyder, *Basic Genetics*, Jones and Bartlett, 1988.)
Answer: This problem uses a known set of overlapping deletions to order a set of mutants. This is called deletion mapping and is based on the expression of the recessive mutant phenotype when heterozygous with a deletion of the corresponding allele on the other homolog. For example, mutants $a$, $b$, and $c$ are all expressed when heterozygous with Del1. Thus it can be assumed that these genes are deleted in Del1. When these results are compared with the crosses with Del2 and it is discovered that these progeny are $b^+$, the location of gene $b$ is mapped to the region deleted in Del1 that is not deleted in Del2. This logic can be applied in the following way:

Compare deletions 1 and 2: this places allele $b$ more to the left than alleles $a$ and $c$. The order is $b\ (a,\ c)$, where the parentheses indicate that the order is unknown.

Compare deletions 2 and 3: this places allele $e$ more to the right than $(a,\ c)$. The order is $b\ (a,\ c)\ e$.

Compare deletions 3 and 4: allele $a$ is more to the left than $c$ and $e$, and $d$ is more to the right than $e$. The order is $b\ a\ c\ e\ d$.

Compare deletions 4 and 5: allele $f$ is more to the right than $d$. The order is $b\ a\ c\ e\ d\ f$.

<table>
<thead>
<tr>
<th>Allele</th>
<th>Band</th>
</tr>
</thead>
<tbody>
<tr>
<td>$b$</td>
<td>1</td>
</tr>
<tr>
<td>$a$</td>
<td>2</td>
</tr>
<tr>
<td>$c$</td>
<td>3</td>
</tr>
<tr>
<td>$e$</td>
<td>4</td>
</tr>
<tr>
<td>$d$</td>
<td>5</td>
</tr>
<tr>
<td>$f$</td>
<td>6</td>
</tr>
</tbody>
</table>

47. A fruit fly was found to be heterozygous for a paracentric inversion. However, obtaining flies that were homozygous for the inversion was impossible even after many attempts. What is the most likely explanation for this inability to produce a homozygous inversion?

Answer: The data suggest that one or both breakpoints of the inversion are located within an essential gene, causing a recessive lethal mutation.

48. Orangutans are an endangered species in their natural environment (the islands of Borneo and Sumatra), and so a captive-breeding program has been established using orangutans currently held in zoos throughout the world. One component of this program is research into orangutan cytogenetics. This research has shown that all orangutans from Borneo carry one form of chromosome 2, as shown in the accompanying diagram, and all orangutans from
Sumatra carry the other form. Before this cytogenetic difference became known, some matings were carried out between animals from different islands, and 14 hybrid progeny are now being raised in captivity.

a. What term or terms describe the differences between these chromosomes?

b. Draw the chromosomes 2, paired in the first meiotic prophase, of such a hybrid orangutan. Be sure to show all the landmarks indicated in the accompanying diagram, and label all parts of your drawing.

c. In 30 percent of meioses, there will be a crossover somewhere in the region between bands p1.1 and q1.2. Draw the gamete chromosomes 2 that would result from a meiosis in which a single crossover occurred within band q1.1.

d. What fraction of the gametes produced by a hybrid orangutan will give rise to viable progeny, if these chromosomes are the only ones that differ between the parents? (Problem 48 is from Rosemary Redfield.)

Answer:
a. The Sumatra chromosome contains a pericentric inversion when compared with the Borneo chromosome.

b. 

c. 

d. 

Answer:
a. The Sumatra chromosome contains a pericentric inversion when compared with the Borneo chromosome.
d. Recall that all single crossovers within the inverted region will lead to four meiotic products: two that will be viable, nonrecombinant (parental) types and two that will be extremely unbalanced, (most likely nonviable), recombinant types. In other words, if 30 percent of the meioses have a crossover in this region, 15 percent of the gametes will not lead to viable progeny. That means that 85 percent of the gametes should produce viable progeny.

49. In corn, the genes for tassel length (alleles $T$ and $t$) and rust resistance (alleles $R$ and $r$) are known to be on separate chromosomes. In the course of making routine crosses, a breeder noticed that one $T/t; R/r$ plant gave unusual results in a testcross with the double-recessive pollen parent $t/t; r/r$. The results were

<table>
<thead>
<tr>
<th>Progeny</th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>$T/t; R/r$</td>
<td>98</td>
<td></td>
</tr>
<tr>
<td>$t/t; r/r$</td>
<td>104</td>
<td></td>
</tr>
<tr>
<td>$T/t; r/r$</td>
<td>3</td>
<td></td>
</tr>
<tr>
<td>$t/t; R/r$</td>
<td>5</td>
<td></td>
</tr>
</tbody>
</table>

Corncobs: Only about half as many seeds as usual

a. What key features of the data are different from the expected results?
b. State a concise hypothesis that explains the results.
c. Show genotypes of parents and progeny.
d. Draw a diagram showing the arrangement of alleles on the chromosomes.
e. Explain the origin of the two classes of progeny having three and five members.

**Unpacking Problem 49**

1. What do a “gene for tassel length” and a “gene for rust resistance” mean?

Answer: A “gene for tassel length” means that there is a gene with at least two alleles ($T$ and $t$) that controls the length of the tassel. A “gene for rust resistance” means that there is a gene that determines whether the corn plant is resistant to a rust infection or not ($R$ and $r$).
2. Does it matter that the precise meaning of the allelic symbols $T$, $t$, $R$, and $r$ is not given? Why or why not?

Answer: The precise meaning of the allelic symbols for the two genes is irrelevant to solving the problem because what is being investigated is the distance between the two genes.

3. How do the terms gene and allele, as used here, relate to the concepts of locus and gene pair?

Answer: A locus is the specific position occupied by a gene on a chromosome. It is implied that gene loci are the same on both homologous chromosomes. The gene pair can consist of identical or different alleles.

4. What prior experimental evidence would give the corn geneticist the idea that the two genes are on separate chromosomes?

Answer: Evidence that the two genes are normally on separate chromosomes would have come from previous experiments showing that the two genes independently assort during meiosis.

5. What do you imagine “routine crosses” are to a corn breeder?

Answer: Routine crosses could consist of $F_1$ crosses, $F_2$ crosses, backcrosses, and testcrosses.

6. What term is used to describe genotypes of the type $T/t; R/r$?

Answer: The genotype $T/t; R/r$ is a double heterozygote, or dihybrid, or $F_1$ genotype.

7. What is a “pollen parent”?

Answer: The pollen parent is the “male” parent that contributes to the pollen tube nucleus, the endosperm nucleus, and the progeny.

8. What are testcrosses, and why do geneticists find them so useful?
Answer: Testcrosses are crosses that involve a genotypically unknown and a homozygous recessive organism. They are used to reveal the complete genotype of the unknown organism and to study recombination during meiosis.

9. What progeny types and frequencies might the breeder have been expecting from the testcross?

Answer: The breeder was expecting to observe 1 \( T/t \); \( R/r \):1 \( T/t \); \( r/r \):1 \( t/t \); \( R/r \):1 \( t/t \); \( r/r \).

10. Describe how the observed progeny differ from expectations.

Answer: Instead of a 1:1:1:1 ratio indicating independent assortment, the testcross indicated that the two genes were linked, with a genetic distance of \( 100\% (3 + 5)/210 = 3.8 \) map units.

11. What does the approximate equality of the first two progeny classes tell you?

Answer: The equality and predominance of the first two classes indicate that the parentals were \( T R/t r \).

12. What does the approximate equality of the second two progeny classes tell you?

Answer: The equality and lack of predominance of the second two classes indicate that they represent recombinants.

13. What were the gametes from the unusual plant, and what were their proportions?

Answer: The gametes leading to this observation were:

- 46.7% \( T R \)
- 1.4% \( T r \)
- 49.5% \( t r \)
- 2.4% \( t R \)

14. Which gametes were in the majority?

Answer:

- 46.7% \( T R \)
- 49.5% \( t r \)

15. Which gametes were in the minority?

Answer:
16. Which of the progeny types seem to be recombinant?

Answer: $Tr$ and $tR$

17. Which allelic combinations appear to be linked in some way?

Answer: $T$ and $R$ are linked, as are $t$ and $r$.

18. How can there be linkage of genes supposedly on separate chromosomes?

Answer: Two genes on separate chromosomes can become linked through a translocation.

19. What do these majority and minority classes tell us about the genotypes of the parents of the unusual plant?

Answer: One parent of the hybrid plant contained a translocation that linked the $T$ and $R$ alleles and the $t$ and $r$ alleles.

20. What is a corncob?

Answer: A corncob is a structure that holds on its surface the seeds that will become the next generation of corn.

21. What does a normal corncob look like? (Sketch one and label it.)

Answer:

22. What do the corncobs from this cross look like? (Sketch one.)
23. What exactly is a kernel?

Answer: A kernel is one progeny on a corn cob.

24. What effect could lead to the absence of half the kernels?

Answer: Absence of half the kernels, or 50 percent aborted progeny (semisterility), could result from the random segregation of one normal with one translocated chromosome (T1 + N2 and T2 + N1) during meioses in a parent that is heterozygous for a reciprocal translocation.

25. Did half the kernels die? If so, was the female or the male parent the reason for the deaths?

Answer: Approximately 50 percent of the progeny died. It was the “female” that was heterozygous for the translocation.

*Now try to solve the problem.*

**Solution to the Problem**

Answer:

a. The progeny are not in the 1:1:1:1 ratio expected for independent assortment; instead, the data indicate close linkage. Also, half the progeny did not develop, indicating semisterility.

b. These observations are best explained by a translocation of material between the two chromosomes.

c. Parents: \[ T R/t \times t/t; r/r \]

Progeny:  
- 98 \[ T R/t; r \]
- 104 \[ t r/t; r \]
- 3 \[ T r/t; r \]
- 5 \[ t R/t; r \]
d. Assume a translocation heterozygote in coupling. If pairing is as diagrammed below, then you would observe the following:

No Crossover

\[
\begin{array}{c}
\text{T}_1 \quad R \\
\text{T} \\
\text{t} \\
\text{N}_1 \\
\text{t} \\
\text{T}_2
\end{array}
\times
\begin{array}{c}
\text{t} \\
\text{r} \\
\text{r}
\end{array}
\rightarrow
\begin{array}{c}
\text{T} \\
\text{T}_1, \text{T}_2
\end{array}
\text{and}
\begin{array}{c}
\text{r} \\
\text{r}
\end{array}
\begin{array}{c}
\text{t} \\
\text{N}_1, \text{N}_2
\end{array}
\]

Crossover between \(T\) and \(R\)

\[
\begin{array}{c}
\text{T}_1 \quad R \\
\text{T} \\
\text{t} \\
\text{N}_1 \\
\text{t} \\
\text{T}_2
\end{array}
\times
\begin{array}{c}
\text{t} \\
\text{r} \\
\text{r}
\end{array}
\rightarrow
\begin{array}{c}
\text{T} \\
\text{T}_1, \text{T}_2
\end{array}
\text{and}
\begin{array}{c}
\text{r} \\
\text{r}
\end{array}
\begin{array}{c}
\text{t} \\
\text{N}_1, \text{N}_2
\end{array}
\]

The locations of the genes on the chromosomes is not critical, as long as the cumulative distance between the genes and the breakpoints allows approximately 3.8 percent recombination.

e. The two recombinant classes result from a recombination event followed by proper segregation of chromosomes, as diagrammed above.

50. A yellow body in \textit{Drosophila} is caused by a mutant allele \(y\) of a gene located at the tip of the X chromosome (the wild-type allele causes a gray body). In a radiation experiment, a wild-type male was irradiated with X rays and then crossed with a yellow-bodied female. Most of the male progeny were yellow, as expected, but the scanning of thousands of flies revealed two gray-bodied (phenotypically wild-type) males. These gray-bodied males were crossed with yellow-bodied females, with the following results:

Progeny

<table>
<thead>
<tr>
<th></th>
<th>females all yellow</th>
<th>males all gray</th>
</tr>
</thead>
<tbody>
<tr>
<td>gray male 1 × yellow female</td>
<td></td>
<td></td>
</tr>
<tr>
<td>gray male 2 × yellow female</td>
<td>1/2 females yellow</td>
<td>1/2 males gray</td>
</tr>
<tr>
<td></td>
<td>1/2 females gray</td>
<td></td>
</tr>
<tr>
<td></td>
<td>1/2 males yellow</td>
<td></td>
</tr>
<tr>
<td></td>
<td>1/2 males gray</td>
<td></td>
</tr>
</tbody>
</table>

a. Explain the origin and crossing behavior of gray male 1.

b. Explain the origin and crossing behavior of gray male 2.
Answer: The cross was

\[
\begin{align*}
P & \quad X^{e+}/Y \text{ (irradiated)} \times X^e/X^e \\
F_1 & \quad \text{Most } X^e/Y \text{ yellow males} \\
& \quad \text{Two } ? \text{ gray males}
\end{align*}
\]

a. Gray male 1 was crossed with a yellow female, yielding yellow females and gray males, which is reversed sex linkage. If the \( e^+ \) allele was translocated to the Y chromosome, the gray male would be \( X^e/Y^{e+} \) or gray. When crossed with yellow females, the results would be

\[
\begin{align*}
X^e/Y^{e+} & \quad \text{gray males} \\
X^e/X^e & \quad \text{yellow females}
\end{align*}
\]

b. Gray male 2 was crossed with a yellow female, yielding gray and yellow males and females in equal proportions. If the \( e^+ \) allele was translocated to an autosome, the progeny would be as below, where “A” indicates autosome:

\[
\begin{align*}
P & \quad A^{e+}/A ; X^e/Y \times A/A \; X^e/X^e \\
F_1 & \quad \begin{align*}
A^{e+}/A & ; X^e/X^e \quad \text{gray female} \\
A^{e+}/A & ; X^e/Y \quad \text{gray male} \\
A/A & ; X^e/X^e \quad \text{yellow female} \\
A/A & ; X^e/Y \quad \text{yellow male}
\end{align*}
\]

Unpacking the Problem

51. In corn, the allele \( Pr \) stands for green stems, \( pr \) for purple stems. A corn plant of genotype \( pr/pr \) that has standard chromosomes is crossed with a \( Pr/Pr \) plant that is homozygous for a reciprocal translocation between chromosomes 2 and 5. The \( F_1 \) is semisterile and phenotypically \( Pr \). A backcross with the parent with standard chromosomes gives 764 semi-sterile \( Pr \), 145 semisterile \( pr \), 186 normal \( Pr \), and 727 normal \( pr \). What is the map distance between the \( Pr \) locus and the translocation point?

Answer: The break point can be treated as a gene with two “alleles,” one for normal fertility and one for semisterility. The problem thus becomes a two-point cross.

\[
\begin{align*}
\text{Parental} & \quad 764 \quad \text{Semisterile } Pr \\
& \quad 727 \quad \text{Normal } pr \\
\text{Recombinant} & \quad 145 \quad \text{Semisterile } pr \\
& \quad 186 \quad \text{Normal } Pr \\
& \quad 1822
\end{align*}
\]
52. Distinguish among Klinefelter, Down, and Turner syndromes. Which syndromes are found in both sexes?

Answer:
- Klinefelter syndrome: XXY male
- Down syndrome: Trisomy 21
- Turner syndrome: XO female

53. Show how you could make an allotetraploid between two related diploid plant species, both of which are 2n = 28.

Answer: Create a hybrid by crossing the two plants and then double the chromosomes with a treatment that disrupts mitosis, such as colchicine treatment. Alternatively, diploid somatic cells from the two plants could be fused and then grown into plants through various culture techniques.

54. In *Drosophila*, trisomics and monosomics for the tiny chromosome 4 are viable, but nullisomics and tetrasomics are not. The *b* locus is on this chromosome. Deduce the phenotypic proportions in the progeny of the following crosses of trisomics.

a. \( b^+/b/b \times b/b \)
b. \( b^+/b^+/b \times b/b \)
c. \( b^+/b^+/b \times b^+/b \)

Answer:

a. \( b^+/b/ \times b/b \)

\[ \begin{align*}
\text{Gametes:} & \quad \frac{1}{6} b^+ b \\
& \quad \frac{1}{3} b \\
& \quad \frac{1}{3} b^+/b \\
& \quad \frac{1}{6} b/b
\end{align*} \]

Among the progeny of this cross, the phenotypic ratio will be 1 wild-type \((b^+) : 1 b.\)

b. \( b^+/b^+/b \times b/b \)

\[ \begin{align*}
\text{Gametes:} & \quad \frac{1}{6} b \\
& \quad \frac{1}{3} b^+ \\
& \quad \frac{1}{3} b^+/b \\
& \quad \frac{1}{6} b^+/b^+
\end{align*} \]
Among the progeny of this cross, the phenotypic ratio will be 5 wild-type \((b^+)\) : 1 \(b\).

c. \[ b^+/b^+ \times b^+/b \]

\[ \begin{array}{c}
\text{Gametes:} \\
1/6 \ b \\
1/3 \ b^+ \\
1/3 \ b^+/b \\
1/6 \ b^+/b^+
\end{array} \]

Among the progeny of this cross, the phenotypic ratio will be 11 wild-type \((b^+)\) : 1 \(b\).

55. A woman with Turner syndrome is found to be color-blind (an X-linked recessive phenotype). Both her mother and her father have normal vision.

a. Explain the simultaneous origin of Turner syndrome and color blindness by the abnormal behavior of chromosomes at meiosis.

b. Can your explanation distinguish whether the abnormal chromosome behavior occurred in the father or the mother?

c. Can your explanation distinguish whether the abnormal chromosome behavior occurred at the first or second division of meiosis?

d. Now assume that a color-blind Klinefelter man has parents with normal vision, and answer parts a, b, and c.

Answer:

a., b., and c. One of the parents of the woman with Turner syndrome (XO) must have been a carrier for color blindness, an X-linked recessive disorder. Because her father has normal vision, she could not have obtained her only X from him. Therefore, nondisjunction occurred in her father. A sperm lacking a sex chromosome fertilized an egg with the X chromosome carrying the color blindness allele. The nondisjunctive event could have occurred during either meiotic division.

d. If the color-blind patient had Klinefelter syndrome (XXY), then both X’s must carry the allele for color blindness. Therefore, nondisjunction had to occur in the mother. Remember that during meiosis I, given no crossover between the gene and the centromere, allelic alternatives separate from each other. During meiosis II, identical alleles on sister chromatids separate. Therefore, assuming there have been no crossovers between the color-blind allele and the centromere, the nondisjunctive event had to occur during meiosis II because both alleles are identical. If the gene is far from the
centromere, it would be difficult to determine if nondisjunction happened at M_I or M_{II} without molecular studies on haplotypes near the centromere.

56. a. How would you synthesize a pentaploid?

b. How would you synthesize a triploid of genotype A/a/a?

c. You have just obtained a rare recessive mutation a* in a diploid plant, which Mendelian analysis tells you is A/a*. From this plant, how would you synthesize a tetraploid (4n) of genotype A/A/a*/a*?

d. How would you synthesize a tetraploid of genotype A/a/a/a?

Answer:

a. If a 6x were crossed with a 4x, the result would be 5x.

b. Cross A/A with a/a/a/a to obtain A/a/a.

c. The easiest way is to expose the A/a* plant cells to colchicine for one cell division. This will result in a doubling of chromosomes to yield A/A/a*/a*.

d. Cross 6x (a/a/a/a/a/a) with 2x (A/A) to obtain A/a/a/a.

57. Suppose you have a line of mice that has cytologically distinct forms of chromosome 4. The tip of the chromosome can have a knob (called 4^K) or a satellite (4^S) or neither (4). Here are sketches of the three types:

![4^K](image)

![4^S](image)

![4](image)

You cross a 4^K/4^S female with a 4/4 male and find that most of the progeny are 4^K/4 or 4^S/4, as expected. However, you occasionally find some rare types as follows (all other chromosomes are normal):

a. 4^K/4^K/4
b. 4^K/4^S/4
c. 4^K
Explain the rare types that you have found. Give, as precisely as possible, the stages at which they originate, and state whether they originate in the male parent, the female parent, or the zygote. (Give reasons briefly.)

Answer: The following answers make the simplifying assumption that there are no crossovers. Without this assumption, it would be hard to tell which stage of meiosis led to the nondisjunction.

Type a: the extra chromosome must be from the mother. Because the chromosomes are identical, nondisjunction had to have occurred at MII.

Type b: the extra chromosome must be from the mother. Because the chromosomes are not identical, nondisjunction had to have occurred at MI.

Type c: the mother correctly contributed one chromosome, but the father did not contribute any chromosome 4. Therefore, nondisjunction occurred in the male during either meiotic division.

58. A cross is made in tomatoes between a female plant that is trisomic for chromosome 6 and a normal diploid male plant that is homozygous for the recessive allele for potato leaf (p/p). A trisomic F1 plant is backcrossed to the potato-leaved male.

a. What is the ratio of normal-leaved plants to potato-leaved plants when you assume that p is located on chromosome 6?

b. What is the ratio of normal-leaved to potato-leaved plants when you assume that p is not located on chromosome 6?

Answer:

a. The cross is P/P/p × p/p.

The gametes from the trisomic parent will occur in the following proportions:

<table>
<thead>
<tr>
<th>p</th>
<th>P</th>
<th>P/P</th>
<th>P/p</th>
</tr>
</thead>
<tbody>
<tr>
<td>1/6</td>
<td>2/6</td>
<td>1/6</td>
<td>2/6</td>
</tr>
</tbody>
</table>

Only gametes that are p can give rise to potato leaves, because potato is recessive. Therefore, the ratio of normal to potato will be 5:1.

b. If the gene is not on chromosome 6, there should be a 1:1 ratio of normal to potato.
59. A tomato geneticist attempts to assign five recessive mutations to specific chromosomes by using trisomics. She crosses each homozygous mutant \((2n)\) with each of three trisomics, in which chromosomes 1, 7, and 10 take part. From these crosses, the geneticist selects trisomic progeny (which are less vigorous) and backcrosses them to the appropriate homozygous recessive. The diploid progeny from these crosses are examined. Her results, in which the ratios are wild type:mutant, are as follows:

<table>
<thead>
<tr>
<th>Trisomic chromosome</th>
<th>d</th>
<th>y</th>
<th>c</th>
<th>h</th>
<th>cot</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>48:55</td>
<td>72:29</td>
<td>56:50</td>
<td>53:54</td>
<td>32:28</td>
</tr>
<tr>
<td>7</td>
<td>52:56</td>
<td>52:48</td>
<td>52:51</td>
<td>58:56</td>
<td>81:40</td>
</tr>
<tr>
<td>10</td>
<td>45:42</td>
<td>36:33</td>
<td>28:32</td>
<td>96:50</td>
<td>20:17</td>
</tr>
</tbody>
</table>

Which of the mutations can the geneticist assign to which chromosomes? (Explain your answer fully.)

Answer: The generalized cross is \(A/A/A \times a/a\), from which \(A/A/a\) progeny were selected. These progeny were crossed with \(a/a\) individuals, yielding the results given. Assume for a moment that each allele can be distinguished from the other, and let 1 = \(A\), 2 = \(A\) and 3 = \(a\). The gametic combinations possible are

1-2 \((A/A)\) and 3 \((a)\)
1-3 \((A/a)\) and 2 \((A)\)
2-3 \((A/a)\) and 1 \((A)\)

Because only diploid progeny were examined in the cross with \(a/a\), the progeny ratio should be 2 wild type:1 mutant if the gene is on the trisomic chromosome. With this in mind, the table indicates that \(y\) is on chromosome 1, \(cot\) is on chromosome 7, and \(h\) is on chromosome 10. Genes \(d\) and \(c\) do not map to any of these chromosomes.

60. A petunia is heterozygous for the following autosomal homologs:

\[
\begin{array}{cccccccc}
A & B & C & D & E & F & G & H & I \\
\end{array}
\begin{array}{cccccccc}
a & b & c & d & h & g & f & e & i \\
\end{array}
\]

a. Draw the pairing configuration that you would see at metaphase I, and identify all parts of your diagram. Number the chromatids sequentially from top to bottom of the page.

b. A three-strand double crossover occurs, with one crossover between the \(C\) and \(D\) loci on chromatids 1 and 3, and the second crossover between the \(G\) and \(H\) loci on chromatids 2 and 3. Diagram the results of these
recombination events as you would see them at anaphase I, and identify all parts of your diagram.

c. Draw the chromosome pattern that you would see at anaphase II after the crossovers described in part b.

d. Give the genotypes of the gametes from this meiosis that will lead to the formation of viable progeny. Assume that all gametes are fertilized by pollen that has the gene order A B C D E F G H I.

Answer:
a.

![Diagram](image1)

b.

![Diagram](image2)

d. The chromosomes numbered 3 and 4 will give rise to viable progeny. The genotypes of those progeny will be A B C D E F G H I/a b c D E F G H I and A B C D E F G H I/a b c d h g f e i.
61. Two groups of geneticists, in California and in Chile, begin work to develop a linkage map of the medfly. They both independently find that the loci for body color \((B = \text{black}, \ b = \text{gray})\) and eye shape \((R = \text{round}, \ r = \text{star})\) are linked 28 m.u. apart. They send strains to each other and perform crosses; a summary of all their findings is shown here:

<table>
<thead>
<tr>
<th>Cross</th>
<th>F(_1)</th>
<th>Progeny of F(_1) × any b r/b r</th>
</tr>
</thead>
<tbody>
<tr>
<td>(B/R/B) (Calif.) × b r/b r (Calif.)</td>
<td>(B/B) b</td>
<td>(B/B) b</td>
</tr>
<tr>
<td></td>
<td></td>
<td>(36%)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>b r/b r</td>
</tr>
<tr>
<td></td>
<td></td>
<td>36</td>
</tr>
<tr>
<td></td>
<td></td>
<td>B r/b r</td>
</tr>
<tr>
<td></td>
<td></td>
<td>14</td>
</tr>
<tr>
<td></td>
<td></td>
<td>b R/b r</td>
</tr>
<tr>
<td></td>
<td></td>
<td>14</td>
</tr>
<tr>
<td>(B/R/B) (Chile) × b r/b r (Chile)</td>
<td>(B/B) b</td>
<td>(B/B) b</td>
</tr>
<tr>
<td></td>
<td></td>
<td>(36%)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>b r/b r</td>
</tr>
<tr>
<td></td>
<td></td>
<td>36</td>
</tr>
<tr>
<td></td>
<td></td>
<td>B r/b r</td>
</tr>
<tr>
<td></td>
<td></td>
<td>14</td>
</tr>
<tr>
<td></td>
<td></td>
<td>b R/b r</td>
</tr>
<tr>
<td></td>
<td></td>
<td>14</td>
</tr>
<tr>
<td>(B/R/B) (Calif.) × b r/b r (Calif.) or b r/b r (Calif.) × B/R/B (Chile)</td>
<td>(B/B) b</td>
<td>(B/B) b</td>
</tr>
<tr>
<td></td>
<td></td>
<td>(48%)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>b r/b r</td>
</tr>
<tr>
<td></td>
<td></td>
<td>48</td>
</tr>
<tr>
<td></td>
<td></td>
<td>B r/b r</td>
</tr>
<tr>
<td></td>
<td></td>
<td>2</td>
</tr>
<tr>
<td></td>
<td></td>
<td>b R/b r</td>
</tr>
<tr>
<td></td>
<td></td>
<td>2</td>
</tr>
</tbody>
</table>

a. Provide a genetic hypothesis that explains the three sets of testcross results.

b. Draw the key chromosomal features of meiosis in the F\(_1\) from a cross of the Californian and Chilean lines.

Answer:

a. Two crosses show 28 map units between the loci for body color and eye shape in a testcross of the F\(_1\): California × California and Chile × Chile. The third type of cross, California × Chile, leads to only four map units between the two genes when the hybrid is testcrossed. This indicates that the genetic distance has decreased by 24 map units, or 100% \((24/28) = 85.7\%\). A deletion cannot be used to explain this finding, nor can a translocation. Most likely the two lines are inverted with respect to each other for 85.7 percent of the distance between the two genes.

b.
A single crossover in either region would result in 4 percent crossing-over between \( B \) and \( R \). The products are

\[
\begin{array}{c|c}
\hline
& B & R \\
\hline
b & & r \\
\hline
& B & r \\
\hline
& b & R \\
\hline
\end{array}
\]

62. An aberrant corn plant gives the following RF values when testcrossed:

<table>
<thead>
<tr>
<th>Interval</th>
<th>( d-f )</th>
<th>( f-b )</th>
<th>( b-x )</th>
<th>( x-y )</th>
<th>( y-p )</th>
</tr>
</thead>
<tbody>
<tr>
<td>Control</td>
<td>5</td>
<td>18</td>
<td>23</td>
<td>12</td>
<td>6</td>
</tr>
<tr>
<td>Aberrant plant</td>
<td>5</td>
<td>2</td>
<td>2</td>
<td>0</td>
<td>6</td>
</tr>
</tbody>
</table>

(The locus order is centromere-\( d-f-b-x-y-p \).) The aberrant plant is a healthy plant, but it produces far fewer normal ovules and pollen than does the control plant.

a. Propose a hypothesis to account for the abnormal recombination values and the reduced fertility in the aberrant plant.

b. Use diagrams to explain the origin of the recombinants according to your hypothesis.

Answer:

a. The aberrant plant is semisterile, which suggests an inversion. Because the \( d-f \) and \( y-p \) frequencies of recombination in the aberrant plant are normal, the inversion must involve \( b \) through \( x \).

b. To obtain recombinant progeny when an inversion is involved, either a double crossover occurred within the inverted region or single crossovers occurred between \( f \) and the inversion, which occurred someplace between \( f \) and \( b \).

63. The following corn loci are on one arm of chromosome 9 in the order indicated (the distances between them are shown in map units):

\[
c-bz-wx-sh-d-centromere \\
12 8 10 20 10
\]

\( C \) gives colored aleurone; \( c \), white aleurone.
\( Bz \) gives green leaves; \( bz \), bronze leaves.
\( Wx \) gives starchy seeds; \( wx \), waxy seeds.
A plant from a standard stock that is homozygous for all five recessive alleles is crossed with a wild-type plant from Mexico that is homozygous for all five dominant alleles. The F₁ plants express all the dominant alleles and, when backcrossed to the recessive parent, give the following progeny phenotypes:

- Colored, green, starchy, smooth, tall: 360
- White, bronze, waxy, shrunk, dwarf: 355
- Colored, bronze, waxy, shrunk, dwarf: 40
- White, green, starchy, smooth, tall: 46
- Colored, green, starchy, smooth, tall: 85
- White, bronze, waxy, shrunk, tall: 84
- Colored, bronze, waxy, shrunk, tall: 8
- White, green, starchy, smooth, dwarf: 9
- Colored, green, waxy, smooth, tall: 7
- White, bronze, starchy, shrunk, dwarf: 6

Propose a hypothesis to explain these results. Include:

- a. a general statement of your hypothesis, with diagrams if necessary;
- b. why there are 10 classes;
- c. an account of the origin of each class, including its frequency; and
- d. at least one test of your hypothesis.

Answer: The cross is

\[ P \quad c \text{ bz wx sh d/c bz wx sh d} \times \quad C \text{ Bz Wx Sh D/C Bz Wx Sh D} \]

\[ F_1 \quad C \text{ Bz Wx Sh D/c bz wx sh d} \]

Backcross \[ C \text{ Bz Wx Sh D/c bz wx sh d} \times \quad c \text{ bz wx sh d/c bz wx sh d} \]

a. The total number of progeny is 1000. Classify the progeny as to where a crossover occurred for each type. Then, total the number of crossovers between each pair of genes. Calculate the observed map units.

<table>
<thead>
<tr>
<th>Region</th>
<th>#COs</th>
<th>M.U. observed</th>
<th>M.U. expected</th>
</tr>
</thead>
<tbody>
<tr>
<td>C–Bz</td>
<td>103</td>
<td>10.3</td>
<td>12</td>
</tr>
<tr>
<td>Bz–Wx</td>
<td>13</td>
<td>1.3</td>
<td>8</td>
</tr>
<tr>
<td>Wx–Sh</td>
<td>13</td>
<td>1.3</td>
<td>10</td>
</tr>
<tr>
<td>A–D</td>
<td>186</td>
<td>18.6</td>
<td>20</td>
</tr>
</tbody>
</table>
Notice that a reduction of map units, or crossing-over, is seen in two intervals. Results like this are suggestive of an inversion. The inversion most likely involves the $Bz$, $Wx$, and $Sh$ genes.

Further, notice that all those instances in which crossing-over occurred in the proposed inverted region involved a double crossover. This is the expected pattern.

b. A number of possible classes are missing: four single-crossover classes resulting from crossing-over in the inverted region, eight double-crossover classes involving the inverted region and the noninverted region, and triple crossovers and higher. The 10 classes detected were the only classes that were viable. They involved a single crossover outside the inverted region or a double crossover within the inverted region.

c. Class 1: parental; increased due to nonviability of some crossovers

Class 2: parental; increased due to nonviability of some crossovers

Class 3: crossing-over between $C$ and $Bz$; approximately expected frequency

Class 4: crossing-over between $C$ and $Bz$, approximately expected frequency

Class 5: crossing-over between $Sh$ and $D$; approximately expected frequency

Class 6: crossing-over between $Sh$ and $D$; approximately expected frequency

Class 7: double crossover between $C$ and $Bz$ and between $Sh$ and $D$; approximately expected frequency

Class 8: double crossover between $C$ and $Bz$ and between $A$ and $D$; approximately expected frequency

Class 9: double crossover between $Bz$ and $Wx$ and between $Wx$ and $Sh$; approximately expected frequency

Class 10: double crossover between $Bz$ and $Wx$ and between $Wx$ and $Sh$; approximately expected frequency

d. Cytological verification could be obtained by looking at chromosomes during meiotic pairing. Genetic verification could be achieved by mapping these genes in the wild-type strain and observing their altered relationships.
64. Chromosomally normal corn plants have a \( p \) locus on chromosome 1 and an \( s \) locus on chromosome 5.

\( P \) gives dark green leaves; \( p \), pale green leaves.
\( S \) gives large ears; \( s \), shrunken ears.

An original plant of genotype \( P/p; S/s \) has the expected phenotype (dark green, large ears) but gives unexpected results in crosses as follows:

- On selfing, fertility is normal, but the frequency of \( p/p; s/s \) types is 1/4 (not 1/16 as expected).
- When crossed with a normal tester of genotype \( p/p; s/s \), the \( F_1 \) progeny are 1/2; \( P/p; S/s \) and 1/2; \( p/p; s/s \); fertility is normal.
- When an \( F_1 \) \( P/p; S/s \) plant is crossed with a normal \( p/p; s/s \) tester, it proves to be semisterile, but, again, the progeny are 1/2; \( P/p; S/s \) and 1/2; \( p/p; s/s \).

Explain these results, showing the full genotypes of the original plant, the tester, and the \( F_1 \) plants. How would you test your hypothesis?

Answer: The original plant was homozygous for a translocation between chromosomes 1 and 5, with breakpoints very close to genes \( P \) and \( S \). Because of the close linkage, a ratio suggesting a monohybrid cross, instead of a dihybrid cross, was observed, both with selfing and with a testcross. All gametes are fertile because of homozygosity.

original plant: \( P S/p s \)
tester: \( p s/p s \)

\( F_1 \) progeny: heterozygous for the translocation:

![Diagram showing the translocation](image)

This figure is an example of one configuration that fits the data. One way to test for the presence of a translocation is to look at the chromosomes of heterozygotes during meiosis I.

65. A male rat that is phenotypically normal shows reproductive anomalies when compared with normal male rats, as shown in the following table. Propose a genetic explanation of these unusual results, and indicate how your idea could be tested.
Embryos (mean number)

<table>
<thead>
<tr>
<th>Mating</th>
<th>Implanted in the uterine wall</th>
<th>Degeneration after implantation</th>
<th>Normal</th>
<th>Degeneration (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>exceptional ♂ × normal ♀</td>
<td>8.7</td>
<td>5.0</td>
<td>3.7</td>
<td>37.5</td>
</tr>
<tr>
<td>normal ♂ × normal ♀</td>
<td>9.5</td>
<td>0.6</td>
<td>8.9</td>
<td>6.5</td>
</tr>
</tbody>
</table>

Answer: The percent degeneration seen in the progeny of the exceptional rat is roughly 50 percent larger than that seen in the progeny from the normal male. Fifty percent emisterility is an important diagnostic for translocation heterozygotes. This could be verified by cytological observation of the meiotic cells from the exceptional male.

66. A tomato geneticist working on $Fr$, a dominant mutant allele that causes rapid fruit ripening, decides to find out which chromosome contains this gene by using a set of lines of which each is trisomic for one chromosome. To do so, she crosses a homozygous diploid mutant with each of the wild-type trisomic lines.

a. A trisomic $F_1$ plant is crossed with a diploid wild-type plant. What is the ratio of fast- to slow-ripening plants in the diploid progeny of this second cross if $Fr$ is on the trisomic chromosome? Use diagrams to explain.

b. What is the ratio of fast- to slow-ripening plants in the diploid progeny of this second cross if $Fr$ is not located on the trisomic chromosome? Use diagrams to explain.

c. Here are the results of the crosses. On which chromosome is $Fr$, and why?

<table>
<thead>
<tr>
<th>Trisomic chromosome</th>
<th>Fast ripening:slow ripening in diploid progeny</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>45:47</td>
</tr>
<tr>
<td>2</td>
<td>33:34</td>
</tr>
<tr>
<td>3</td>
<td>55:52</td>
</tr>
<tr>
<td>4</td>
<td>26:30</td>
</tr>
<tr>
<td>5</td>
<td>31:32</td>
</tr>
<tr>
<td>6</td>
<td>37:41</td>
</tr>
<tr>
<td>7</td>
<td>44:79</td>
</tr>
<tr>
<td>8</td>
<td>49:53</td>
</tr>
<tr>
<td>9</td>
<td>34:34</td>
</tr>
<tr>
<td>10</td>
<td>37:39</td>
</tr>
</tbody>
</table>

(Problem 66 is from Tamara Western.)

Answer:
a. The cross is $Fr/Fr \times fr/fr/fr$. 
Trisomic progeny are then crossed to a diploid wild-type plant.

Fr/fr/fr × fr/fr

Because only diploid progeny of this cross are evaluated, the ratio of fast- to slow-ripening plants will be 1:2.

b. If Fr is not located on the trisomic chromosome, the crosses are

Fr/Fr × fr/fr

and

Fr/fr × fr/fr

Therefore, the ratio of fast- to slow-ripening plants will be 1:1.

c. The 1:2 ratio of fast- to slow ripening plants indicates that the Fr gene is on chromosome 7.

CHALLENGING PROBLEMS

67. The Neurospora un-3 locus is near the centromere on chromosome 1, and crossovers between un-3 and the centromere are very rare. The ad-3 locus is on the other side of the centromere of the same chromosome, and crossovers occur between ad-3 and the centromere in about 20 percent of meioses (no multiple crossovers occur).

a. What types of linear asci (see Chapter 4) do you predict, and in what frequencies, in a normal cross of un-3 ad-3 × wild type? (Specify genotypes of spores in the asci.)

b. Most of the time such crosses behave predictably, but, in one case, a standard un-3 ad-3 strain was crossed with a wild type isolated from a field of sugarcane in Hawaii. The results follow:
Explain these results, and state how you could test your idea. *(Note: In Neurospora, ascospores with extra chromosomal material survive and are the normal black color, whereas ascospores lacking any chromosome region are white and inviable.)*

**Answer:**

a. Single crossovers between a gene and its centromere lead to a tetratype (second-division segregation.) Thus, a total of 20 percent of the asci should show second division segregation, and 80 percent will show first-division segregation. The following are representative asci

\[
\begin{align*}
\text{un}^3+ \text{ad}^3+ & \quad \text{un}^3+ \text{ad}^3+ & \quad \text{un}^3+ \text{ad}^3+ & \quad \text{un}^3+ \text{ad}^3+ & \quad \text{un}^3+ \text{ad}^3+ \\
\text{un}^3+ \text{ad}^3+ & \quad \text{un}^3+ \text{ad}^3+ & \quad \text{un}^3+ \text{ad}^3+ & \quad \text{un}^3+ \text{ad}^3+ & \quad \text{un}^3+ \text{ad}^3+ \\
\text{un}^3+ \text{ad}^3+ & \quad \text{un}^3+ \text{ad}^3+ & \quad \text{un}^3+ \text{ad}^3+ & \quad \text{un}^3+ \text{ad}^3+ & \quad \text{un}^3+ \text{ad}^3+ \\
\text{un}^3+ \text{ad}^3+ & \quad \text{un}^3+ \text{ad}^3+ & \quad \text{un}^3+ \text{ad}^3+ & \quad \text{un}^3+ \text{ad}^3+ & \quad \text{un}^3+ \text{ad}^3+ \\
\text{un}^3+ \text{ad}^3+ & \quad \text{un}^3+ \text{ad}^3+ & \quad \text{un}^3+ \text{ad}^3+ & \quad \text{un}^3+ \text{ad}^3+ & \quad \text{un}^3+ \text{ad}^3+ \\
80\% & \quad 5\% & \quad 5\% & \quad 5\% & \quad 5\%
\end{align*}
\]

In all cases, the “upside-down” version would be equally likely.

b. The aborted spores could result from a crossing-over event within an inversion of the wild type, compared with the standard strain. Crossing-over within heterozygous inversions leads to unbalanced chromosomes and nonviable spores. This could be tested by using the wild type from Hawaii in mapping experiments of other markers on chromosome 1 in crosses with the standard strain and looking for altered map distances.

68. Two mutations in *Neurospora, ad-3* and *pan-2*, are located on chromosomes 1 and 6, respectively. An unusual *ad-3* line arises in the laboratory, giving the results shown in the table below. Explain all three results with the aid of clearly labeled diagrams. *(Note: In Neurospora, ascospores with extra chromosomal material survive and are the normal black color, whereas ascospores lacking any chromosome region are white and inviable.)*

<table>
<thead>
<tr>
<th>Ascospore appearance</th>
<th>RF between <em>ad-3</em> and <em>pan-2</em></th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Normal <em>ad-3</em> × normal <em>pan-2</em></td>
<td>All black</td>
</tr>
<tr>
<td>2. Abnormal <em>ad-3</em> × normal <em>pan-2</em></td>
<td>About 1/2 black and 1/2 white (inviable)</td>
</tr>
<tr>
<td>3. Of the black spores from cross 2, about half were completely normal and half repeated the same behavior as the original abnormal <em>ad-3</em> strain</td>
<td></td>
</tr>
</tbody>
</table>
Chapter Seventeen 419

Answer:
Cross 1: independent assortment of the 2 genes (expected for genes on separate chromosomes).

Cross 2: the 2 genes now appear to be linked (the observed RF is 1 percent); also, half of the progeny are inviable. These data suggest a reciprocal translocation occurred and both genes are very close to the breakpoints.

Cross 3: the viable spores are of 2 types: half contain the normal (nontranslocated chromosomes) and half contain the translocated chromosomes.

69. Deduce the phenotypic proportions in the progeny of the following crosses of autotetraploids in which the $a^+/a$ locus is very close to the centromere. (Assume that the four homologous chromosomes of any one type pair randomly two by two and that only one copy of the $a^+$ allele is necessary for the wild-type phenotype.)

a. $a^+/a^+/a/a \times a/a/a/a$
b. $a^+/a/a/a \times a/a/a/a$
c. $a^+/a/a/a \times a^+/a/a/a$
d. $a^+/a^+/a/a \times a^+/a/a/a$

Answer:

a. $a^+/a^+/a/a \times a/a/a/a$
   \[ \downarrow \quad \downarrow \]
   Gametes: 1/6 $a^+/a^+$, 2/3 $a^+/a$, 1/6 $a/a$

Among the progeny of this cross, the phenotypic ratio will be 5 wild-type $(a^+)$ : 1 $a$.

b. $a^+/a/a/a \times a/a/a/a$
   \[ \downarrow \quad \downarrow \]
   Gametes: 1/2 $a^+/a$, 1/2 $a/a$

Among the progeny of this cross, the phenotypic ratio will be 1 wild-type $(a^+)$ : 1 $a$.

c. $a^+/a/a/a \times a^+/a/a/a$
   \[ \downarrow \quad \downarrow \]
   Gametes: 1/2 $a^+/a$, 1/2 $a^+/a$, 1/2 $a/a$, 1/2 $a/a$

Among the progeny of this cross, the phenotypic ratio will be 1 wild-type $(a^+)$ : 1 $a$. 
Among the progeny of this cross, the phenotypic ratio will be 3 wild-type \((a^+)^2 : \) 1 a.

d. \(a^+/a^+/a^+/a^+ \times a^+/a+/a+/a\)

\[
\begin{array}{c|c|c}
\text{Gametes} & \text{1/6} & a^+/a^+ \\
& \text{1/2} & a^+/a \\
& \text{2/3} & a^+/a \\
& \text{1/6} & a/a \\
\end{array}
\]

Among the progeny of this cross, the phenotypic ratio will be 11 wild-type \((a^+)^2 : \) 1 a.

70. The New World cotton species *Gossypium hirsutum* has a \(2n\) chromosome number of 52. The Old World species *G. thurberi* and *G. herbaceum* each have a \(2n\) number of 26. Hybrids between these species show the following chromosome pairing arrangements at meiosis:

<table>
<thead>
<tr>
<th>Hybrid</th>
<th>Pairing arrangement</th>
</tr>
</thead>
<tbody>
<tr>
<td><em>G. hirsutum</em> × <em>G. thurberi</em></td>
<td>13 small bivalents + 13 large univalents</td>
</tr>
<tr>
<td><em>G. hirsutum</em> × <em>G. herbaceum</em></td>
<td>13 large bivalents + 13 small univalents</td>
</tr>
<tr>
<td><em>G. thurberi</em> × <em>G. herbaceum</em></td>
<td>13 large univalents + 13 small univalents</td>
</tr>
</tbody>
</table>

Draw diagrams to interpret these observations phylogenetically, clearly indicating the relationships between the species. How would you go about proving that your interpretation is correct? (Problem 70 is adapted from A. M. Srb, R. D. Owen, and R. S. Edgar, *General Genetics*, 2nd ed. W. H. Freeman and Company, 1965.)

Answer: Consider the following table, in which “L” and “S” stand for 13 large and 13 small chromosomes, respectively:

<table>
<thead>
<tr>
<th>Hybrid</th>
<th>Chromosomes</th>
</tr>
</thead>
<tbody>
<tr>
<td><em>G. hirsutum</em> × <em>G. thurberi</em></td>
<td>S, S, L</td>
</tr>
<tr>
<td><em>G. hirsutum</em> × <em>G. herbaceum</em></td>
<td>S, S, L</td>
</tr>
<tr>
<td><em>G. thurberi</em> × <em>G. herbaceum</em></td>
<td>S, L</td>
</tr>
</tbody>
</table>

Each parent in the cross must contribute half its chromosomes to the hybrid offspring. It is known that *G. hirsutum* has twice as many chromosomes as the other two species. Furthermore, because the *G. hirsutum* chromosomes form bivalent pairs in the hybrids, the *G. hirsutum* karyotype must consist of chromosomes donated by the other two species. Therefore, the genome of *G. hirsutum* must consist of one large and one small set of chromosomes. Once this is realized, the rest of the problem essentially solves itself. In the first hybrid, the genome of *G. thurberi* must consist of one set of small chromosomes. In the second hybrid, the genome of *G. herbaceum* must consist of one set of large
chromosomes. The third hybrid confirms the conclusions reached from the first two hybrids.

The original parents must have had the following chromosome constitution:

\[
\begin{align*}
G. \text{ hirsutum} & \quad 26 \text{ large, 26 small} \\
G. \text{ thurberi} & \quad 26 \text{ small} \\
G. \text{ herbaceum} & \quad 26 \text{ large}
\end{align*}
\]

\[G. \text{ hirsutum}\] is a polyploid derivative of a cross between the two Old World species. This could easily be checked by looking at the chromosomes.

71. There are six main species in the \textit{Brassica} genus: \textit{B. carinata}, \textit{B. campestris}, \textit{B. nigra}, \textit{B. oleracea}, \textit{B. juncea}, and \textit{B. napus}. You can deduce the interrelationships among these six species from the following table:

<table>
<thead>
<tr>
<th>Species or F(_1) hybrid</th>
<th>Chromosome number</th>
<th>Number of bivalents</th>
<th>Number of univalents</th>
</tr>
</thead>
<tbody>
<tr>
<td>\textit{B. juncea}</td>
<td>36</td>
<td>18</td>
<td>0</td>
</tr>
<tr>
<td>\textit{B. carinata}</td>
<td>34</td>
<td>17</td>
<td>0</td>
</tr>
<tr>
<td>\textit{B. napus}</td>
<td>38</td>
<td>19</td>
<td>0</td>
</tr>
<tr>
<td>\textit{B. juncea} \times \textit{B. nigra}</td>
<td>26</td>
<td>8</td>
<td>10</td>
</tr>
<tr>
<td>\textit{B. napus} \times \textit{B. campestris}</td>
<td>29</td>
<td>10</td>
<td>9</td>
</tr>
<tr>
<td>\textit{B. carinata} \times \textit{B. oleracea}</td>
<td>26</td>
<td>9</td>
<td>8</td>
</tr>
<tr>
<td>\textit{B. juncea} \times \textit{B. oleracea}</td>
<td>27</td>
<td>0</td>
<td>27</td>
</tr>
<tr>
<td>\textit{B. carinata} \times \textit{B. campestris}</td>
<td>27</td>
<td>0</td>
<td>27</td>
</tr>
<tr>
<td>\textit{B. napus} \times \textit{B. nigra}</td>
<td>27</td>
<td>0</td>
<td>27</td>
</tr>
</tbody>
</table>

\textbf{a.} Deduce the chromosome number of \textit{B. campestris}, \textit{B. nigra}, and \textit{B. oleracea}.

\textbf{b.} Show clearly any evolutionary relationships between the six species that you can deduce at the chromosomal level.

Answer:

\textbf{a.} \textit{B. campestris} was crossed with \textit{B. napus}, and the hybrid had 29 chromosomes consisting of 10 bivalents; and 9 univalents. \textit{B. napus} had to have contributed a total of 19 chromosomes to the hybrid. Therefore, \textit{B. campestris} had to have contributed 10 chromosomes. The 2\textit{n} number of \textit{B. campestris} is 20.

When \textit{B. nigra} was crossed with \textit{B. napus}, \textit{B. nigra} had to have contributed 8 chromosomes to the hybrid. The 2\textit{n} number of \textit{B. nigra} is 16.

\textit{B. oleracea} had to have contributed 9 chromosomes to the hybrid formed with \textit{B. juncea}. The 2\textit{n} number in \textit{B. oleracea} is 18.
b. First, list the haploid and diploid number for each species:

<table>
<thead>
<tr>
<th>Species</th>
<th>Haploid</th>
<th>Diploid</th>
</tr>
</thead>
<tbody>
<tr>
<td><em>B. nigra</em></td>
<td>8</td>
<td>16</td>
</tr>
<tr>
<td><em>B. oleracea</em></td>
<td>9</td>
<td>18</td>
</tr>
<tr>
<td><em>B. campestris</em></td>
<td>10</td>
<td>20</td>
</tr>
<tr>
<td><em>B. carinata</em></td>
<td>17</td>
<td>34</td>
</tr>
<tr>
<td><em>B. juncea</em></td>
<td>18</td>
<td>36</td>
</tr>
<tr>
<td><em>B. napus</em></td>
<td>19</td>
<td>38</td>
</tr>
</tbody>
</table>

Now, recall that a bivalent in a hybrid indicates that the chromosomes are essentially identical. Therefore, the more bivalents formed in a hybrid, the closer the two parent species. Three crosses result in no bivalents, suggesting that the parents of each set of hybrids are not closely related:

<table>
<thead>
<tr>
<th>Cross</th>
<th>Haploid number</th>
</tr>
</thead>
<tbody>
<tr>
<td><em>B. juncea</em> × <em>B. oleracea</em></td>
<td>18 vs. 9</td>
</tr>
<tr>
<td><em>B. carinata</em> × <em>B. campestris</em></td>
<td>17 vs. 10</td>
</tr>
<tr>
<td><em>B. napus</em> × <em>B. nigra</em></td>
<td>19 vs. 8</td>
</tr>
</tbody>
</table>

Three additional crosses resulted in bivalents, suggesting a closer relationship among the parents:

<table>
<thead>
<tr>
<th>Cross</th>
<th>Haploid #</th>
<th>Bivalents</th>
<th>Univalents</th>
</tr>
</thead>
<tbody>
<tr>
<td><em>B. juncea</em> × <em>B. nigra</em></td>
<td>18 vs. 8</td>
<td>8</td>
<td>10</td>
</tr>
<tr>
<td><em>B. napus</em> × <em>B. campestris</em></td>
<td>19 vs. 10</td>
<td>10</td>
<td>9</td>
</tr>
<tr>
<td><em>B. carinata</em> × <em>B. oleracea</em></td>
<td>17 vs. 9</td>
<td>9</td>
<td>8</td>
</tr>
</tbody>
</table>

Note that in each cross the number of bivalents is equal to the haploid number of one species. This suggests that the species with the larger haploid number is a hybrid composed of the second species and some other species. In each case, the haploid number of the unknown species is the number of univalents. Therefore, the following relationships can be deduced:

* *B. juncea* is an amphidiploid formed by the cross of *B. nigra* and *B. campestris*. *

* *B. napus* is an amphidiploid formed by the cross of *B. campestris* and *B. oleracea*. *

* *B. carinata* is an amphidiploid formed by the cross of *B. nigra* and *B. oleracea*. *

These conclusions are in accord with the three crosses that did not yield bivalents:
72. Several kinds of sexual mosaicism are well documented in humans. Suggest how each of the following examples may have arisen by nondisjunction at mitosis:

a. XX/XO (that is, there are two cell types in the body, XX and XO)
b. XX/XXYY
c. XO/XXX
d. XX/XY
e. XO/XX/XXX

Answer:

a. Loss of one X in the developing fetus after the two-cell stage

b. Nondisjunction leading to Klinefelter syndrome (XXY), followed by a nondisjunctive event in one cell for the Y chromosome after the two-cell stage, resulting in XX and XXYY

c. Nondisjunction of the X at the one-cell stage

d. Fused XX and XY zygotes (from the separate fertilizations either of two eggs or of an egg and a polar body by one X-bearing and one Y-bearing sperm)

e. Nondisjunction of the X at the two-cell stage or later

73. In *Drosophila*, a cross (cross 1) was made between two mutant flies, one homozygous for the recessive mutation bent wing (*b*) and the other homozygous for the recessive mutation eyeless (*e*). The mutations *e* and *b* are alleles of two different genes that are known to be very closely linked on the tiny autosomal chromosome 4. All the progeny had a wild-type phenotype. One of the female progeny was crossed with a male of genotype *b e/b e*; we will call this cross 2.
Most of the progeny of cross 2 were of the expected types, but there was also one rare female of wild-type phenotype.

a. Explain what the common progeny are expected to be from cross 2.

b. Could the rare wild-type female have arisen by (1) crossing over or (2) nondisjunction? Explain.

c. The rare wild-type female was testcrossed to a male of genotype \( b e/b e \) (cross 3). The progeny were

\[
\begin{align*}
1/6 & \text{ wild type} \\
1/6 & \text{ bent, eyeless} \\
1/3 & \text{ bent} \\
1/3 & \text{ eyeless}
\end{align*}
\]

Which of the explanations in part b is compatible with this result? Explain the genotypes and phenotypes of the progeny of cross 3 and their proportions.

Unpacking Problem 73

1. Define homozygous, mutation, allele, closely linked, recessive, wild type, crossing over, nondisjunction, testcross, phenotype, and genotype.

Answer:
*Homozygous* means that an organism has two identical alleles.
*A mutation* is any deviation from wild type.
*An allele* is one particular form of a gene.
*Closely linked* means two genes are almost always transmitted together through meiosis.
*Recessive* refers to a type of allele that is expressed only when it is the sole type of allele for that gene found in an individual.
*Wild type* is the most frequent type found in a laboratory population or in a population in the “wild.”
*Crossing over* refers to the physical exchange of alleles between homologous chromosomes.
*Nondisjunction* is the failure of separation of either homologous chromosomes or sister chromatids in the two meiotic divisions.
*A testcross* is a cross to a homozygous recessive organism for the trait or traits being studied.
*Phenotype* is the appearance of an organism.
*Genotype* is the genetic constitution of an organism.

2. Does this problem concern sex linkage? Explain.
3. How many chromosomes does *Drosophila* have?

Answer: The most common lab species, *Drosophila melanogaster*, has eight chromosomes.

4. Draw a clear pedigree summarizing the results of crosses 1, 2, and 3.

Answer:

<table>
<thead>
<tr>
<th>Pedigree</th>
<th>Genotype</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>P</td>
<td><em>b e</em>+/<em>b e</em> × <em>b</em>+/<em>b e</em></td>
<td>(cross 1)</td>
</tr>
<tr>
<td>F1</td>
<td><em>b e</em>+/<em>b e</em> × <em>b e</em>/<em>b e</em></td>
<td>(cross 2)</td>
</tr>
<tr>
<td>Progeny</td>
<td><em>b e</em>+/<em>b e</em></td>
<td>expected parental</td>
</tr>
<tr>
<td></td>
<td><em>b</em>+/<em>b e</em></td>
<td>expected parental</td>
</tr>
<tr>
<td></td>
<td><em>b</em>+/<em>b e</em></td>
<td>unexpected recombinant (&quot;very closely linked&quot; so rare)</td>
</tr>
<tr>
<td></td>
<td><em>b e</em>/<em>b e</em></td>
<td>unexpected recombinant (&quot;very closely linked&quot; so rare)</td>
</tr>
</tbody>
</table>

Progeny

1/6 wild type
1/6 bent, eyeless
1/3 bent
1/3 eyeless

5. Draw the gametes produced by both parents in cross 1.

Answer: *b e*+ and *b*+ e

6. Draw the chromosome 4 constitution of the progeny of cross 1.

Answer: *b e*+/*b*+ e

7. Is it surprising that the progeny of cross 1 are wild-type phenotype? What does this outcome tell you?

Answer: It is not at all surprising that the F$_1$ are wild type. This means that both mutations are recessive and complement (are in different genes).
8. Draw the chromosome 4 constitution of the male tester used in cross 2 and the
gametes that he can produce.

Answer: \( b/e/b/e \rightarrow \text{gametes: } b/e \)

9. With respect to chromosome 4, what gametes can the female parent in cross 2
produce in the absence of nondisjunction? Which would be common and which
rare?

Answer: The two common gametes are \( b/e^+ \) and \( b^+e \). The two rare gametes are
\( b^+e^+ \) and \( b/e \).

10. Draw first- and second-division meiotic nondisjunction in the female parent of
cross 2, as well as in the resulting gametes.

Answer:

Normal

\[ \begin{align*}
\text{b} & \quad \text{e}^+ \\
\text{b}^+ & \quad \text{e}
\end{align*} \]

\[ \begin{align*}
\text{M}_1 & \\
\text{b} & \quad \text{e}^+ \\
\text{b}^+ & \quad \text{e}
\end{align*} \]

\[ \begin{align*}
\text{M}_II & \\
b & \quad \text{e}^+ \\
b^+ & \quad \text{e}
\end{align*} \]

First-Division Nondisjunction: all gametes are aneuploid

\[ \begin{align*}
\text{b} & \quad \text{e}^+ \\
\text{b}^+ & \quad \text{e}
\end{align*} \]

\[ \begin{align*}
\text{M}_1 & \\
\text{b} & \quad \text{e}^+ \\
\text{b}^+ & \quad \text{e}
\end{align*} \]

\[ \begin{align*}
\text{M}_II & \\
b & \quad \text{e}^+ \\
b^+ & \quad \text{e}
\end{align*} \]

Second-Division Nondisjunction: half the gametes are aneuploid

\[ \begin{align*}
\text{b} & \quad \text{e}^+ \\
\text{b}^+ & \quad \text{e}
\end{align*} \]

\[ \begin{align*}
\text{M}_1 & \\
\text{b} & \quad \text{e}^+ \\
\text{b}^+ & \quad \text{e}
\end{align*} \]

\[ \begin{align*}
\text{M}_II & \\
b & \quad \text{e}^+ \\
b^+ & \quad \text{e}
\end{align*} \]

\[ \begin{align*}
\text{M}_II & \\
b & \quad \text{e}^+ \\
b^+ & \quad \text{e}
\end{align*} \]

OR

\[ \begin{align*}
\text{b} & \quad \text{e}^+ \\
\text{b}^+ & \quad \text{e}
\end{align*} \]

\[ \begin{align*}
\text{M}_1 & \\
\text{b} & \quad \text{e}^+ \\
\text{b}^+ & \quad \text{e}
\end{align*} \]

\[ \begin{align*}
\text{M}_II & \\
b & \quad \text{e}^+ \\
b^+ & \quad \text{e}
\end{align*} \]

\[ \begin{align*}
\text{M}_II & \\
b & \quad \text{e}^+ \\
b^+ & \quad \text{e}
\end{align*} \]
11. Are any of the gametes from part 10 aneuploid?

Answer: This is answered in 10, above.

12. Would you expect aneuploid gametes to give rise to viable progeny? Would these progeny be nullisomic, monosomic, disomic, or trisomic?

Answer: Viable progeny may be able to arise from aneuploid genotypes because chromosome 4 is very small and, percentage-wise, contributes little to the genome. The progeny would be monosomic and trisomic.

13. What progeny phenotypes would be produced by the various gametes considered in parts 9 and 10?

Answer: Listed below are the gametes from 9 and 10 above, the contribution of the male parent, and the phenotype of the progeny.

<table>
<thead>
<tr>
<th>Female gamete</th>
<th>Male gamete</th>
<th>Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>(b^+ e)</td>
<td>(b e)</td>
<td>eyeless</td>
</tr>
<tr>
<td>(b e^+)</td>
<td>(b e)</td>
<td>bent</td>
</tr>
<tr>
<td>(b^+ e^+)</td>
<td>(b e)</td>
<td>wild type</td>
</tr>
<tr>
<td>(b e)</td>
<td>(b e)</td>
<td>bent, eyeless</td>
</tr>
<tr>
<td>(b^+ e/b e^+)</td>
<td>(b e)</td>
<td>wild type</td>
</tr>
<tr>
<td>(--)</td>
<td>(b e)</td>
<td>bent, eyeless</td>
</tr>
<tr>
<td>(b e^+/b e^+)</td>
<td>(b e)</td>
<td>bent</td>
</tr>
<tr>
<td>(b^+ e/b^+ e)</td>
<td>(b e)</td>
<td>eyeless</td>
</tr>
</tbody>
</table>

14. Consider the phenotypic ratio in the progeny of cross 3. Many genetic ratios are based on halves and quarters, but this ratio is based on thirds and sixths. To what might this ratio point?

Answer: The ratio points to meiosis of a trisomic.

15. Could there be any significance to the fact that the crosses concern genes on a very small chromosome? When is chromosome size relevant in genetics?

Answer: Research with artificial chromosomes has indicated that extremely small chromosomes segregate improperly at higher rates than longer chromosomes. It is suspected that the chromatids from homologous chromosomes need to intertwine in order to remain together until the onset of anaphase. Very short chromosomes are thought to have some difficulty in doing this and therefore have a higher rate of nondisjunction. In this instance, which deals with natural chromosomes as opposed to artificial chromosomes, very
small chromosomes would be expected to have very little genetic material in them, and therefore their loss or gain may not be of too much importance during development.

16. Draw the progeny expected from cross 3 under the two hypotheses, and give some idea of relative proportions.

Answer:
rare wild type × e b/e b (cross 3)

If the rare wild type is from recombination, then the cross becomes

\[ b^+ e^+ / b e × b e/b e \]

Progeny

- \[ b^+ e^+ / b e \] parental: wild type
- \[ b e/b e \] parental: bent, eyeless
- \[ b^+ / e b e \] rare recombinant: eyeless
- \[ b e^+/ b e \] rare recombinant: bent

If the rare wild type is from nondisjunction, then the cross becomes

\[ b e^+/ b^+ e / b e \]

Progeny

- \[ b e^+/ b^+ e / b e \] wild type
- \[ b^+ e / b e \] bent
- \[ b^+ / b e \] eyeless
- \[ b e^+/ b e \] bent
- \[ b^+ / b e \] eyeless
- \[ b e / b e \] bent, eyeless

**Solution to the Problem**

**Cross 1:** P 
\[ b e^+/ b e^+ \times \]
\[ b^+ e/b e^+ \]
F\(_1\) 
\[ b^+ e / b e^+ \]

**Cross 2:** P  
\[ X/X ; \] \[ b^+ e/b e^+ \times X/Y ; \] \[ b e/b e \]
F\(_1\)  
expect 1 \[ b e^+/ b e \] : 1 \[ b^+ e/b e \], X/X and X/Y one rare observed X/X ; \[ b^+ e^+ \]

a. The common progeny are \[ b^+ e/b e \] and \[ b e^+/ b e \].

b. The rare female could have come from crossing-over, which would have resulted in a gamete that was \[ b^+ e^- \]. The rare female could also have come from nondisjunction that gave a gamete that was \[ b e^+/ b^+ e \]. Such a gamete might give rise to viable progeny.
c. If the female had been wild type \((b^+/e^+/b\, e)\) as a result of crossing-over, her progeny would have been as follows:

<table>
<thead>
<tr>
<th>Parental:</th>
<th>(b^+/e^+/b, e)</th>
<th>wild type (common)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>(b, e/b, e)</td>
<td>bent, eyeless (common)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Recombinant:</th>
<th>(b, e^+/b, e)</th>
<th>bent (rare)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>(b^+, e/b, e)</td>
<td>eyeless (rare)</td>
</tr>
</tbody>
</table>

These expected results are very far from what was observed, so the rare female was not the result of recombination.

If the female had been the product of nondisjunction \((b\, e^+/b^+/e^+/b\, e)\), her progeny when crossed to \(b\, e/b\, e\) would be as follows:

<table>
<thead>
<tr>
<th></th>
<th>(b^+/e)</th>
<th>eyeless</th>
</tr>
</thead>
<tbody>
<tr>
<td>(1/6)</td>
<td>(b, e)</td>
<td>bent</td>
</tr>
<tr>
<td>(1/6)</td>
<td>(b^+/e \times b, e)</td>
<td>bent</td>
</tr>
<tr>
<td>(1/6)</td>
<td>(b^+, e/b, e)</td>
<td>eyeless</td>
</tr>
<tr>
<td>(1/6)</td>
<td>(b, e^+/b, e)</td>
<td>bent</td>
</tr>
<tr>
<td>(1/6)</td>
<td>(b, e/b, e)</td>
<td>bent, eyeless</td>
</tr>
<tr>
<td>(1/6)</td>
<td>(b, e^+/b^+, e/b, e)</td>
<td>wild type</td>
</tr>
</tbody>
</table>

Overall, 2 bent:2 eyeless:1 bent eyeless:1 wild type

These results are in accord with the observed results, indicating that the female was a product of nondisjunction.

74. In the fungus *Ascobolus* (similar to *Neurospora*), ascospores are normally black. The mutation \(f\), producing fawn-colored ascospores, is in a gene just to the right of the centromere on chromosome 6, whereas mutation \(b\), producing beige ascospores, is in a gene just to the left of the same centromere. In a cross of fawn and beige parents \((+f \times b^+)\), most octads showed four fawn and four beige ascospores, but three rare exceptional octads were found, as shown in the accompanying illustration. In the sketch, black is the wild-type phenotype, a vertical line is fawn, a horizontal line is beige, and an empty circle represents an aborted (dead) ascospore.
Chapter Seventeen

**a.** Provide reasonable explanations for these three exceptional octads.

**b.** Diagram the meiosis that gave rise to octad 2.

**Answer:** Recall that ascospores are haploid. The normal genotype associated with the phenotype of each spore is given below.

<table>
<thead>
<tr>
<th></th>
<th>1</th>
<th>2</th>
<th>3</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>$b^+ f^+$</td>
<td>$b f^+$</td>
<td>$b^+ f$</td>
</tr>
<tr>
<td></td>
<td>$b^+ f^+$</td>
<td>$b f^+$</td>
<td>$b^+ f$</td>
</tr>
<tr>
<td></td>
<td>$b^+ f^+$</td>
<td>abort</td>
<td>$b^+ f^+$</td>
</tr>
<tr>
<td></td>
<td>astropt</td>
<td>$b^+ f^+$</td>
<td>abort</td>
</tr>
<tr>
<td></td>
<td>abort</td>
<td>$b^+ f$</td>
<td>$b f$</td>
</tr>
<tr>
<td></td>
<td>abort</td>
<td>$b^+ f$</td>
<td>$b f^+$</td>
</tr>
<tr>
<td></td>
<td>abort</td>
<td>$b^+ f$</td>
<td>$b f^+$</td>
</tr>
</tbody>
</table>

**a.** For the first ascus, the most reasonable explanation is that nondisjunction occurred at the first meiotic division. Second-division nondisjunction or chromosome loss are two explanations of the second ascus. Crossing-over best explains the third ascus.

**b.**

![Diagram of meiosis process](image)
The life cycle of the haploid fungus *Ascobolus* is similar to that of *Neurospora*. A mutational treatment produced two mutant strains, 1 and 2, both of which when crossed with wild type gave unordered tetrads, all of the following type (fawn is a light brown color; normally, crosses produce all black ascospores):

- spore pair 1 black
- spore pair 2 black
- spore pair 3 fawn
- spore pair 4 fawn

**a.** What does this result show? Explain.

The two mutant strains were crossed. Most of the unordered tetrads were of the following type:

- spore pair 1 fawn
- spore pair 2 fawn
- spore pair 3 fawn
- spore pair 4 fawn

**b.** What does this result suggest? Explain.

When large numbers of unordered tetrads were screened under the microscope, some rare ones that contained black spores were found. Four cases are shown here:

<table>
<thead>
<tr>
<th>Case A</th>
<th>Case B</th>
<th>Case C</th>
<th>Case D</th>
</tr>
</thead>
<tbody>
<tr>
<td>spore pair 1</td>
<td>black</td>
<td>black</td>
<td>black</td>
</tr>
<tr>
<td>spore pair 2</td>
<td>black</td>
<td>fawn</td>
<td>black</td>
</tr>
<tr>
<td>spore pair 3</td>
<td>fawn</td>
<td>fawn</td>
<td>abort</td>
</tr>
<tr>
<td>spore pair 4</td>
<td>fawn</td>
<td>abort</td>
<td>fawn</td>
</tr>
</tbody>
</table>

*Note:* Ascospores with extra genetic material survive, but those with less than a haploid genome abort.

**c.** Propose reasonable genetic explanations for each of these four rare cases.

**d.** Do you think the mutations in the two original mutant strains were in one single gene? Explain.
Answer:

a. Each mutant is crossed with wild type, or

\[ m \times m^+ \]

The resulting tetrads (octads) show 1:1 segregation, indicating that each mutant is the result of a mutation in a single gene.

b. The results from crossing the two mutant strains indicate either both strains are mutant for the same gene,

\[ m_1 \times m_2 \]

or, that they are mutant in different but closely linked genes

\[ m_1 m_2^+ \times m_1+m_2 \]

c. and d. Because phenotypically black offspring can result from nondisjunction (notice that in Case C and Case D, black appears in conjunction with aborted spores), it is likely that mutant 1 and mutant 2 are mutant in different but closely linked genes. The cross is therefore

\[ m_1 m_2^+ \times m_1+m_2 \]

Case A is an NPD tetrad and would be the result of a four-strand double crossover.

\[
\begin{array}{c|c}
&m_1^+ m_2^+ \text{ black} \\
&m_1^+ m_2^+ \text{ black} \\
&m_1 m_2 \text{ fawn} \\
&m_1 m_2 \text{ fawn} \\
\end{array}
\]

Case B is a tetratype and would be the result of a single crossover between one of the genes and the centromere.

\[
\begin{array}{c|c}
&m_1^+ m_2^+ \text{ black} \\
&m_1^+ m_2 \text{ fawn} \\
&m_1 m_2^+ \text{ fawn} \\
&m_1 m_2 \text{ fawn} \\
\end{array}
\]

Case C is the result of nondisjunction during meiosis I.

\[
\begin{array}{c|c}
&m_1^+ m_2^+ m_1 m_2^+ \text{ black} \\
&m_1^+ m_2^+ m_1 m_2^+ \text{ black} \\
&\text{no chromosome abort} \\
&\text{no chromosome abort} \\
\end{array}
\]
Case D is a the result of recombination between one of the genes and the centromere followed by nondisjunction during meiosis II. For example:

\[
\begin{align*}
\bullet \times \bullet \quad & \quad m_1^+ m_2^+ \\
\bullet \quad & \quad m_1^+ m_2 \\
\bullet \quad & \quad m_1^+ m_2
\end{align*}
\]

\[
\begin{align*}
\bullet \quad & \quad m_1 m_2^+ \\
\bullet \quad & \quad m_1^+ m_2 \\
\bullet \quad & \quad m_1 m_2^+
\end{align*}
\]

- \( m_1^+ m_2 \); \( m_1 m_2^+ \) black
- no chromosome abort
- \( m_1^+ m_2^+ \) fawn
- \( m_1^+ m_2 \) fawn