C1. Duplications and deficiencies involve a change in the total amount of genetic material.

Duplication: a repeat of some genetic material
Deficiency: a shortage of some genetic material
Inversion: a segment of genetic material in the opposite orientation
Translocation: a segment of genetic material attached to the wrong chromosome

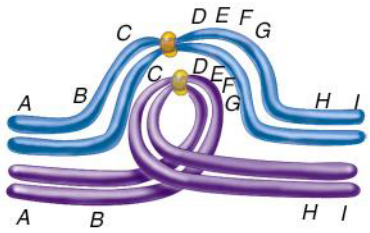
C2. Small deletions and duplications are less likely to affect phenotype simply because they usually involve fewer genes. If a small deletion did have a phenotypic effect, you would conclude that a gene or genes in this region are required in two functional copies in order to have a normal phenotype.

C3. It occurs when there is a misalignment during the crossing over of homologous chromosomes. One chromosome ends up with a deficiency and the other has a duplication.

C4. A gene family is a group of genes that are derived from the process of gene duplications. They have similar sequences, but the sequences have some differences due to the accumulation of mutations over many generations. The members of a gene family usually encode proteins with similar but specialized functions. The specialization may occur in different cells or at different stages of development.

C5. You would expect $a_1$ and $a_2$ to be more similar, because they have diverged more recently. Therefore, there has been less time for them to accumulate random mutations that would make their sequences different.

C6. It has a pericentric inversion.




C7. There are four products from meiosis. One would be a normal chromosome and one would contain the inversion shown in the answer to conceptual question C6. The other two chromosomes would have the following order of genes:

*A B C* centromere *D E F G H I J B A*
*M L K J I H G F E D* centromere *C K L M*

C8. There are four products from meiosis. One would be a normal chromosome and one would contain the inversion shown in the drawing to conceptual question C8. The other two chromosomes would be dicentric or acentric with the following order of genes:

centromere ↓ centromere ↓

*A B C D E F G H I J D C B A* Dicentric
*M L K J I H G F E K L M* Acentric

C9. Individuals who carry inversions and reciprocal translocations have the same amount of genetic material (i.e., the same number of genes) as do normal individuals. Therefore, they commonly have a normal phenotype. In some cases, however, the breakpoint in an inversion or translocation disrupts an important gene and thereby has a phenotypic consequence. In other cases, the chromosomal rearrangement may have a position effect that alters the expression of an important gene.

C10. In the absence of crossing over, alternate segregation would yield half the cells with two normal chromosomes and half with a balanced translocation. For adjacent-1 segregation, two cells would be

*A B C D E* + *A I J K L M*

And the other two cells would be:

*H B C D E* + *H I J K L M*

C11. A terminal piece of chromosome 11 broke off and attached to the short arm of chromosome 15. A crossover occurred between the long arm of chromosome 15 and the long arm of chromosome 18.

C12. One of the parents may carry a balanced translocation between chromosomes 5 and 7. The phenotypically abnormal offspring has inherited an imbalanced translocation due to the segregration of translocated chromosomes during meiosis.

C13. It is expected to be rare because the normal driving force for segregation is the segregation of centromeres. For example, the centromeres on chromosome 2 normally align during meiosis and segregate from each other whether or not the chromosome contains a translocation. On rare occasions, a misalignment of centromeres can lead to adjacent-2 segregation in which two centromeres from one chromosome travel to one pole and two centromeres from another chromosome travel to the opposite pole.

C14. A deficiency and an unbalanced translocation are more likely to have phenotypic effects because they create genetic imbalances. For a deficiency, there are too few copies of several genes, and for an unbalanced translocation, there are too many.

C15. It is because the homologous chromosomes are trying to synapse with each other. As shown in Figure 8.15, the formation of a translocation cross allows the homologous parts of the chromosomes to line up (i.e., synapse) with each other.

C16. You should draw out the inversion loop (as is done in Figure 8.12*a*). The crossover occurred between *P* and *U*.

C17. A. 16

B. 9

C. 7

D. 12

E. 17

C18. This person has a total of 46 chromosomes. However, this person would be considered aneuploid rather than euploid. This is because one of the sets is missing a sex chromosome and one set has an extra copy of chromosome 21.

C19. One parent is probably normal while the other parent has one normal copy of chromosomes 14 and 21 and one chromosome 14, which has most of chromosome 21 attached to it.

C20. It may be related to genetic balance. In aneuploidy, there is an imbalance in gene expression between the chromosomes found in their normal copy number versus those that are either too many or too few. In polyploidy, the balance in gene expression is still maintained.

C21. Imbalances in aneuploidy, deletions, and duplications are related to the copy number of genes. For many genes, the level of gene expression is directly related to the number of genes per cell. If there are too many copies, as in trisomy, or too few, as in monosomy, the level of gene expression will be too high or too low, respectively. It is difficult to say why deletions and monosomies are more detrimental although one could speculate that having too little of a gene product causes more cellular problems than having too much of a gene product.

C22. The male offspring is the result of nondisjunction during oogenesis. The female produced an egg without any sex chromosomes. The male parent transmitted a single X chromosome carrying the red allele. This produces an X0 male offspring with red eyes.

C23. A. The $F_1$ offspring would probably be phenotypically normal since they would carry the correct number of genes.

B. The $F_1$ offspring would have lowered fertility because they are inversion heterozygotes. Since this is a large inversion, crossing over is fairly likely in the inverted region. When this occurs, it will produce deletions and duplications that will probably be lethal in the resulting $F_2$ offspring.

C24. Trisomies 13, 18, and 21 survive because the chromosomes are small and probably contain fewer genes compared to the larger chromosomes. Individuals with abnormal numbers of X chromosomes can survive because the extra copies are converted to transcriptionally inactive Barr bodies. The other aneuploidies are lethal because they cause a great amount of imbalance between the level of gene expression on the normal diploid chromosomes relative to the chromosomes that are trisomic or monosomic.

C25. Maybe one is diploid and the other is a closely related tetraploid species. Their offspring would be triploid, which would explain the sterility. Another possibility is that one may carry a large inversion (see answer to conceptual question C23, part B).

C26. Endopolyploidy means that a particular somatic tissue is polyploid even though the rest of the organism is not. The biological significance is not entirely understood although it has been speculated that an increase in ploidy may enable the cell to make more gene products that the cell needs.

C27. A genetic mosaic is an individual having patches of tissue that are genetically different from each other. With regard to mosaics in chromosome number, it can be due to abnormal events during mitosis, such as the movement of two homologues to the same pole, or the loss of a chromosome.

C28. In certain types of cells, such as salivary cells, the homologous chromosomes pair with each other and then replicate about nine times to produce a polytene chromosome. The centromeres from each type of chromosome associate with each other at the chromocenter. This structure has six arms that arise from one arm of two telomeric chromosomes (the X and 4) and two arms each from chromosomes 2 and 3.

C29. Polyploid plants are often more robust than their diploid counterparts. With regard to agriculture, they may produce a greater yield of fruits and vegetables. In the field, they tend to be more resistant to harsh environmental conditions. When polyploid plants have an odd number of sets, they are typically seedless. This can be a desirable trait for certain fruit-producing crops such as bananas.

C30. The turtles are two distinct species that appear phenotypically identical. The turtles with 48 chromosomes are polyploid relatives (i.e., tetraploids) of the species with 24 chromosomes. In animals, it is somewhat hard to imagine how this could occur because animals cannot self-fertilize, so there had to be two animals (i.e., one male and one female) that became tetraploids. It is easy to imagine how one animal could become a tetraploid; complete nondisjunction could occur during the first cell division of a fertilized egg, thereby creating a tetraploid cell that continued to develop into a tetraploid animal. This would have to happen independently (i.e., in two individuals of opposite sex) to create a tetraploid species. If you mated a tetraploid turtle with a diploid turtle, the offspring would be triploid and probably phenotypically normal. However, the triploid offspring would be sterile because they would make highly aneuploid gametes.

C31. Aneuploid should not be used.

C32. Polyploid, triploid, and euploid should not be used.

C33. There are 11 chromosomes per set, so there are 11 possible trisomics: trisomy 1, trisomy 2, trisomy 3, trisomy 4, trisomy 5, trisomy 6, trisomy 7, trisomy 8, trisomy 9, trisomy 10, and trisomy 11.

C34. The boy carries a translocation involving chromosome 21: probably a translocation in which nearly all of chromosome 21 is translocated to chromosome 14. He would have one normal copy of chromosome 14, one normal copy of chromosome 21, and the translocated chromosome that contains both chromosome 14 and chromosome 21. This boy is phenotypically normal because the total amount of genetic material is normal, although the total number of chromosomes is 45 (because chromosome 14 and chromosome 21 are fused into a single chromosome). His sister has Down syndrome because she has inherited the translocated chromosome, but she also must have one copy of chromosome 14 and two copies of chromosome 21. She has the equivalent of three copies of chromosome 21 (i.e., two normal copies and one copy fused with chromosome 14). This is why she has Down syndrome. One of the parents of these two children is probably normal with regard to karyotype (i.e., the parent has 46 normal chromosomes). The other parent would have a karyotype that would be like the phenotypically normal boy.

C35. The odds of producing a euploid gamete are $(1/2)^{n-1}$, which equals $(1/2)^5$ or a 1 in 32 chance. The chance of producing an aneuploid gamete is 31 out of 32 gametes will be aneuploid. We use the product rule to determine the chances of getting a euploid individual, since a euploid individual is produced from two euploid gametes: $1/32 \times 1/32 = 1/1{,}024$. In other words, if this plant self-fertilized, only 1 in 1,024 offspring would be euploid. The euploid offspring could be diploid, triploid, or tetraploid.

C36. Nondisjunction is a mechanism whereby the chromosomes do not segregate equally into the two daughter cells. This can occur during meiosis to produce cells with altered numbers of chromosomes, or it can occur during mitosis to produce a genetic mosaic individual. A third way to alter chromosome number is by interspecies crosses to produce an alloploid.

C37. It usually occurs during meiosis I when the homologues synapse to form bivalents. During meiosis II and mitosis, the homologues do not synapse. Instead, the chromosomes align randomly along the metaphase plate and then the centromeres separate.

C38. A mutation occurred during early embryonic development to create the blue patch of tissue. One possibility is a mitotic nondisjunction in which the two chromosomes carrying the *b* allele went to one cell and the two chromosomes carrying the *B* allele went to the other daughter cell. A second possibility is that the chromosome carrying the *B* allele could be lost. A third possibility is that the *B* allele could have been deleted. This would cause the recessive *b* allele to exhibit pseudodominance.

C39. An allodiploid is an organism having one set of chromosomes from two different species. Unless the two species are closely related evolutionarily, the chromosomes do not synapse during meiosis. Therefore, they do not segregate properly. This produces aneuploid gametes that are usually inviable. By comparison, allotetraploids that have two sets of chromosomes from each species are more likely to be fertile because each chromosome has a homologue to pair with during meiosis.

C40. Homeologous chromosomes are chromosomes from two species that are evolutionarily related to each other. For example, chromosome 1 in chimpanzees and gorillas is homeologous; it carries the same types of genes.

C41. For meiotic nondisjunction, the bivalents are not separating correctly during meiosis I. During mitotic nondisjunction, the sister chromatids are not separating properly.

C42. In general, Turner syndrome could be due to nondisjunction during oogenesis or spermatogenesis. However, the Turner individual with color blindness is due to nondisjunction during spermatogenesis. The sperm lacked a sex chromosome, due to nondisjunction, and the egg carried an X chromosome with the recessive color blindness allele. This X chromosome had to be inherited from the mother, because the father was not color-blind. The mother must be heterozygous for the recessive color blind allele, and the father is hemizygous for the normal allele. Therefore, the mother must have transmitted a single X chromosome carrying the color-blind allele to her offspring indicating that nondisjunction did not occur during oogenesis.

C43. Complete nondisjunction occurs during meiosis I so that one nucleus receives all the chromosomes and the other nucleus does not get any. The nucleus with all the chromosomes then proceeds through a normal meiosis II to produce two haploid sperm cells.