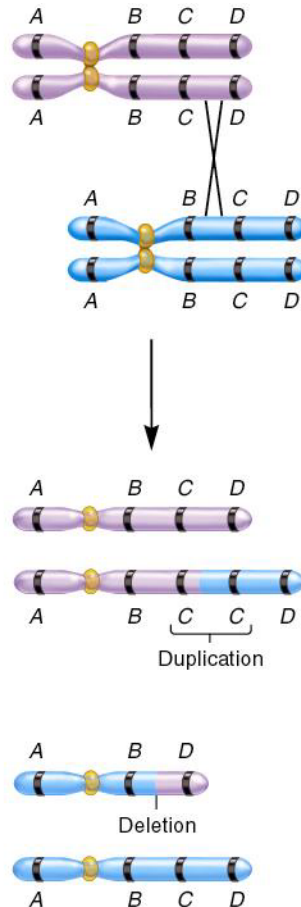


S1. Describe how a gene family is produced. Discuss the common and unique features of the family members in the globin gene family.

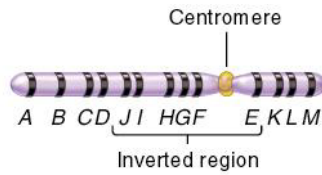
Answer: A gene family is produced when a single gene is copied one or more times by a gene duplication event. This duplication occurs by an abnormal (misaligned) crossover, which produces a chromosome with a deficiency and another chromosome with a gene duplication.



Over time, this type of duplication may occur several times to produce many copies of a particular gene. In addition, translocations may move the duplicated genes to other chromosomes, so that the members of the gene family may be dispersed among several different chromosomes. Eventually, each member of a gene family will accumulate mutations, which may subtly alter their function.

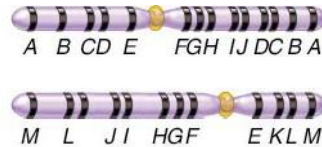
All the members of the globin gene family bind oxygen. Myoglobin tends to bind it more tightly; therefore, it is good at storing oxygen. Hemoglobin binds it more loosely, so it can transport oxygen throughout the body (via red blood cells) and release it to the tissues that need oxygen. The polypeptides that form hemoglobins are predominantly expressed in red blood cells, whereas myoglobin genes are expressed in many different cell types. The expression pattern of the globin genes changes during different stages of development. The ϵ - and ζ -globin genes are expressed in the early embryo, they are turned off near the end of the first trimester, and then the γ -globin genes exhibit their maximal expression during the second and third trimesters of gestation. Following birth, the γ -globin genes are silenced and the β -globin gene is expressed for the rest of a person's life. These differences in the expression of the globin genes reflect the differences in the oxygen transport needs of humans during the different stages of life. Overall, the evolution of gene families has resulted in gene products that are better suited to a particular tissue or stage of development. This has allowed a better "fine-tuning" of human traits.

S2. An inversion heterozygote has the following inverted chromosome:



What is the result if a crossover occurs between genes *F* and *G* on one inversion and one normal chromosome?

Answer: The resulting product is four chromosomes. One chromosome is normal, one is an inversion chromosome, and two chromosomes have duplications and deficiencies. The two duplicated/deficient chromosomes are shown here:



S3. In humans, the number of chromosomes per set equals 23. Even though the following conditions are lethal, what would be the total number of chromosomes for the following individuals?

- A. Trisomy 22
- B. Monosomy 11
- C. Triploid individual

Answer:

- A. 47 (the diploid number, 46, plus 1)
- B. 45 (the diploid number, 46, minus 1)
- C. 69 (3 times 23)

S4. A diploid species with 44 chromosomes (i.e., 22/set) is crossed to another diploid species with 38 chromosomes (i.e., 19/set). What would be the number of chromosomes in an allodiploid or allotetraploid produced from this cross? Would you expect the offspring to be sterile or fertile?

Answer: An allodiploid would have $22 + 19 = 41$ chromosomes. This individual would likely be sterile, because all the chromosomes would not have homeologous partners to pair with during meiosis. This yields aneuploidy, which usually causes sterility. An allotetraploid would have $44 + 38 = 82$ chromosomes. Since each chromosome would have a homologous partner, the allotetraploid would likely be fertile.

S5. Pseudodominance occurs when a single copy of a recessive allele is phenotypically expressed because the second copy of the gene has been deleted from the homologous chromosome; the individual is hemizygous for the recessive allele. As an example, we can consider the “notch” phenotype in *Drosophila*, which is an X-linked trait. Fruit flies with this condition have wings with a notched appearance at their edges. Female flies that are heterozygous for this mutation have notched wings; homozygous females and hemizygous males are unable to survive. The notched phenotype is due to a defect in a single gene called *notch* (*N*). When geneticists have studied fruit flies with this phenotype, some of the mutant flies are due to a small deletion that includes the *notch* gene as well as a few genes on either side of it. Other notch mutations are due to small mutations confined within the *notch* gene itself. A genetic analysis can distinguish between “notched” fruit flies carrying a deletion versus those that carry a single-gene mutation. This is possible because the *notch* gene happens to be located next to the red/white eye color gene on the X chromosome. How would you distinguish between a notched phenotype due to a deletion that included the *notch* gene and the adjacent eye color gene versus a notch phenotype due to a small mutation only within the *notch* gene itself?

Answer: To determine if the notch mutation is due to a deletion, red-eyed females with the notched phenotype can be crossed to white-eyed males. Only the daughters with notched wings need to be analyzed. If they have red eyes, this means that the notch mutation has not deleted the red-eye allele from the X chromosome. Alternatively, if they have white eyes, this indicates that the red-eye allele has been deleted from the X chromosome, which carries the notch mutation. In this case, the white-eye allele is expressed because it is present in a single copy in a female fly with two X chromosomes. This phenomenon is pseudodominance.

S6. Albert Blakeslee began using the Jimson weed (*Datura stramonium*) as an experimental organism to teach his students the laws of Mendelian inheritance. Although this plant has not gained widespread use in genetic studies, Blakeslee's work provided a convincing demonstration that changes in chromosome number have an impact on the phenotype of organisms (also see fig. 8.18). Blakeslee's assistant, B. T. Avery, identified a Jimson weed mutant, which he called "globe" because the capsule is more rounded than normal. In genetic crosses, he found that the globe mutant had a peculiar pattern of inheritance. The globe trait was passed to about 25% of the offspring when the globe plants were allowed to self-fertilize. Unexpectedly, about 25% of the offspring also had the globe phenotype when globe plants were pollinated by a normal plant. In contrast, when pollen from a globe plant was used to pollinate a normal plant, less than 2% of the offspring had the globe phenotype. This non-Mendelian pattern of inheritance caused Blakeslee and his colleagues to investigate the nature of this trait further. We now know that the globe phenotype is due to trisomy 11. Can you explain this unusual pattern of inheritance knowing that it is due to trisomy 11?

Answer: This unusual pattern of inheritance of these aneuploid strains can be explained by the viability of euploid versus aneuploid gametes and/or gametophytes. When an individual is trisomic, there is a 50% chance that an egg or sperm nucleus will inherit an extra chromosome and a 50% chance that a gamete will be normal. Blakeslee's results indicate that when a pollen grain inherited an extra copy of chromosome 11, it was almost always nonviable and unable to produce an aneuploid offspring. However, a significant percentage of aneuploid eggs were viable so that some (25%) of the offspring were aneuploid. Since an aneuploid plant should produce a 1:1 ratio between euploid and aneuploid eggs, the observation that only 25% of the offspring were aneuploid also indicates that about half of the female gametophytes or aneuploid eggs from such gametophytes were also nonviable. Overall, these results provide compelling evidence that imbalances in chromosome number can alter reproductive viability and also cause significant phenotypic consequences.