Chapter 15

The Chromosomal Basis of Inheritance

PowerPoint Lectures for

Biology, Seventh Edition

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Lectures by Chris Romero

- Overview: Locating Genes on Chromosomes
- Genes
  - Are located on chromosomes
  - Can be visualized using certain techniques

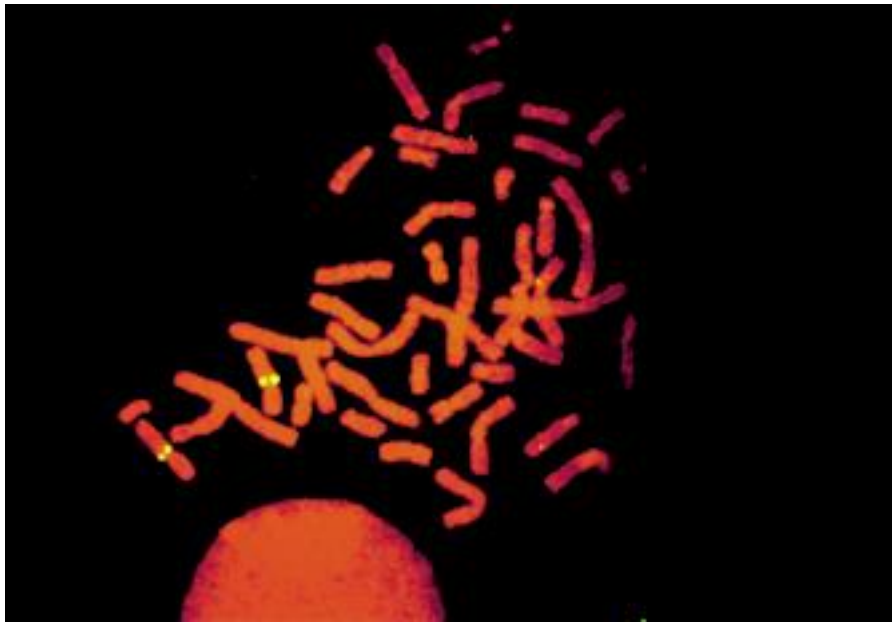


Figure 15.1

- Concept 15.1: Mendelian inheritance has its physical basis in the behavior of chromosomes
- Several researchers proposed in the early 1900s that genes are located on chromosomes
- The behavior of chromosomes during meiosis was said to account for Mendel's laws of segregation and independent assortment

- The chromosome theory of inheritance states that
  - Mendelian genes have specific loci on chromosomes
  - Chromosomes undergo segregation and independent assortment

- The chromosomal basis of Mendel's laws

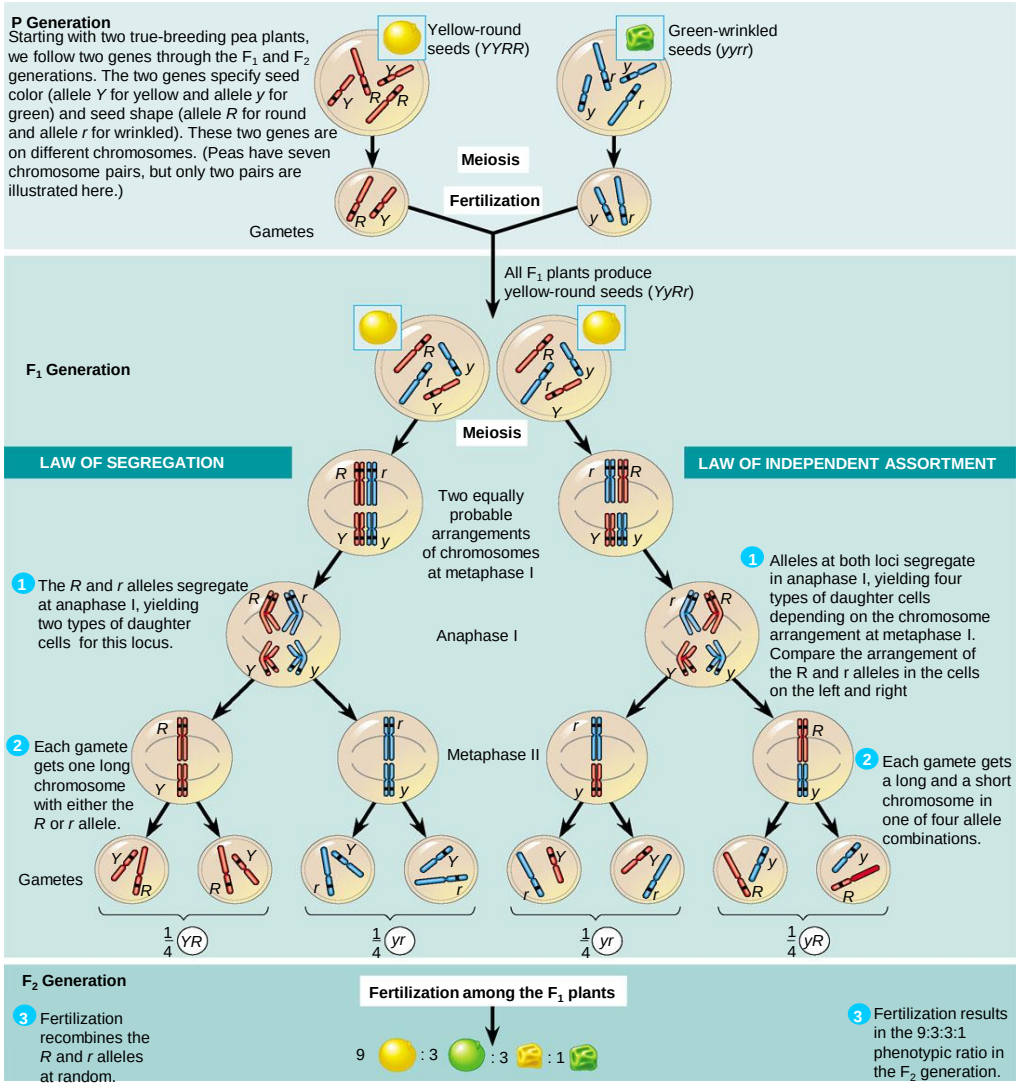


Figure 15.2

Morgan's Experimental Evidence: *Scientific Inquiry*

- Thomas Hunt Morgan
  - Provided convincing evidence that chromosomes are the location of Mendel's heritable factors

Morgan's Choice of Experimental Organism

- Morgan worked with fruit flies
  - Because they breed at a high rate
  - A new generation can be bred every two weeks
  - They have only four pairs of chromosomes

- Morgan first observed and noted
  - Wild type, or normal, phenotypes that were common in the fly populations
- Traits alternative to the wild type
  - Are called mutant phenotypes

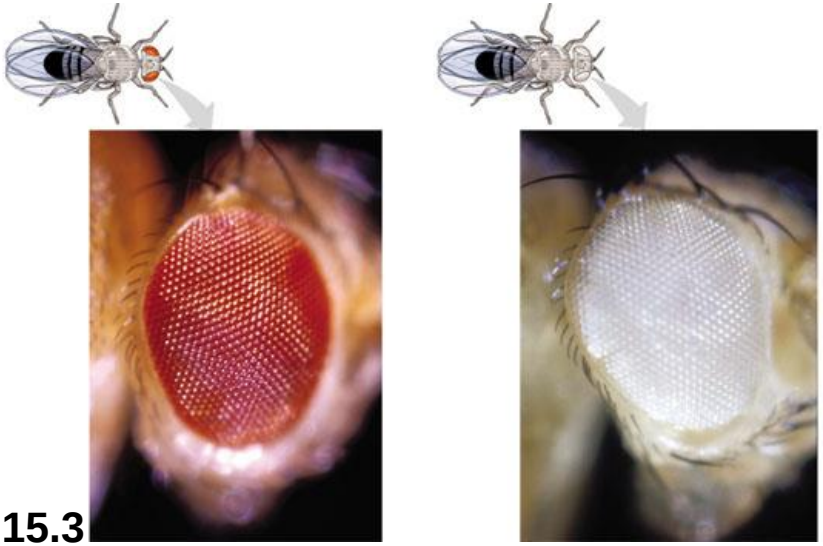


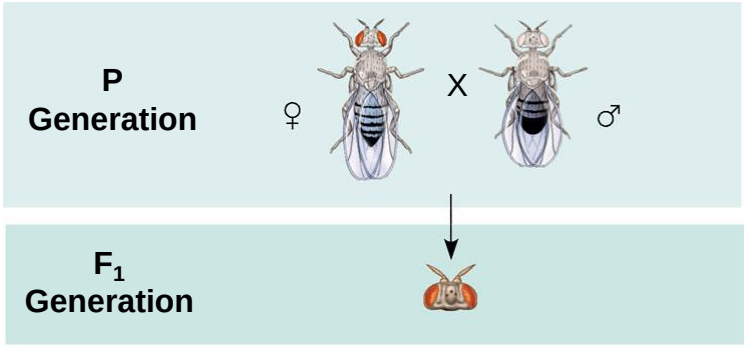
Figure 15.3

Correlating Behavior of a Gene's Alleles with Behavior of a Chromosome Pair

- In one experiment Morgan mated male flies with white eyes (mutant) with female flies with red eyes (wild type)
  - The F_1 generation all had red eyes
  - The F_2 generation showed the 3:1 red:white eye ratio, but only males had white eyes

- Morgan determined
  - That the white-eye mutant allele must be located on the X chromosome

EXPERIMENT Morgan mated a wild-type (red-eyed) female with a mutant white-eyed male. The F_1 offspring all had red eyes.



Morgan then bred an F_1 red-eyed female to an F_1 red-eyed male to produce the F_2 generation.

RESULTS The F_2 generation showed a typical Mendelian 3:1 ratio of red eyes to white eyes. However, no females displayed the white-eye trait; they all had red eyes. Half the males had white eyes, and half had red eyes.

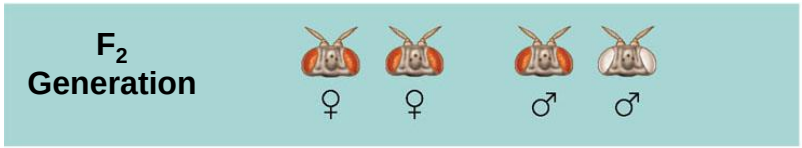
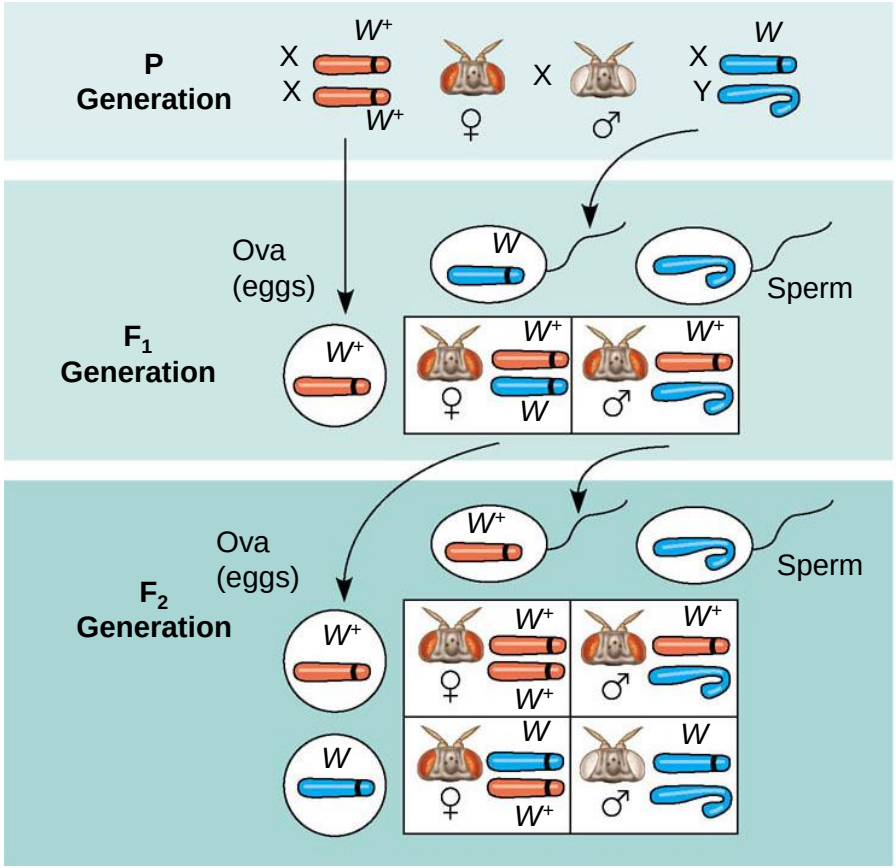


Figure 15.4

CONCLUSION Since all F_1 offspring had red eyes, the mutant white-eye trait (*w*) must be recessive to the wild-type red-eye trait (w^+). Since the recessive trait—white eyes—was expressed only in males in the F_2 generation, Morgan hypothesized that the eye-color gene is located on the X chromosome and that there is no corresponding locus on the Y chromosome, as diagrammed here.



- Morgan's discovery that transmission of the X chromosome in fruit flies correlates with inheritance of the eye-color trait
  - Was the first solid evidence indicating that a specific gene is associated with a specific chromosome

- Concept 15.2: Linked genes tend to be inherited together because they are located near each other on the same chromosome
- Each chromosome
  - Has hundreds or thousands of genes

How Linkage Affects Inheritance: *Scientific Inquiry*

- Morgan did other experiments with fruit flies
  - To see how linkage affects the inheritance of two different characters

- Morgan crossed flies
  - That differed in traits of two different characters

EXPERIMENT

Morgan first mated true-breeding wild-type flies with black, vestigial-winged flies to produce heterozygous F_1 dihybrids, all of which are wild-type in appearance. He then mated wild-type F_1 dihybrid females with black, vestigial-winged males, producing 2,300 F_2 offspring, which he "scored" (classified according to phenotype).

CONCLUSION

If these two genes were on different chromosomes, the alleles from the F_1 dihybrid would sort into gametes independently, and we would expect to see equal numbers of the four types of offspring. If these two genes were on the same chromosome, we would expect each allele combination, B^+ vg^+ and b vg, to stay together as gametes formed. In this case, only offspring with parental phenotypes would be produced. Since most offspring had a parental phenotype, Morgan concluded that the genes for body color and wing size are located on the same chromosome. However, the production of a small number of offspring with nonparental phenotypes indicated that some mechanism occasionally breaks the linkage between genes on the same chromosome.

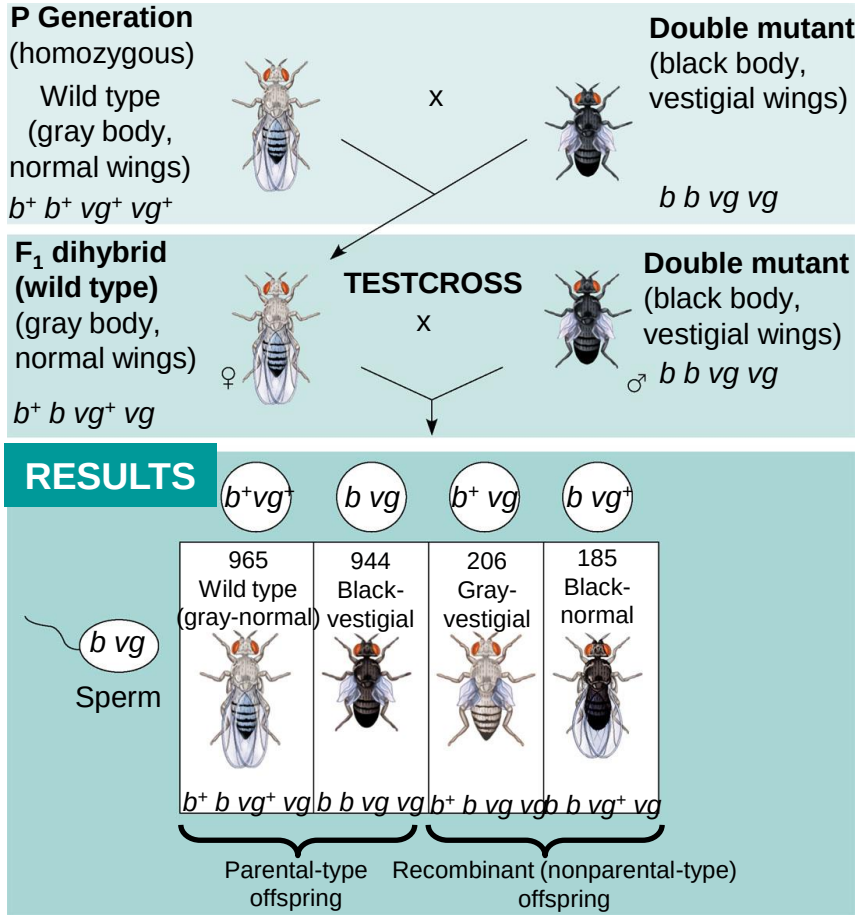
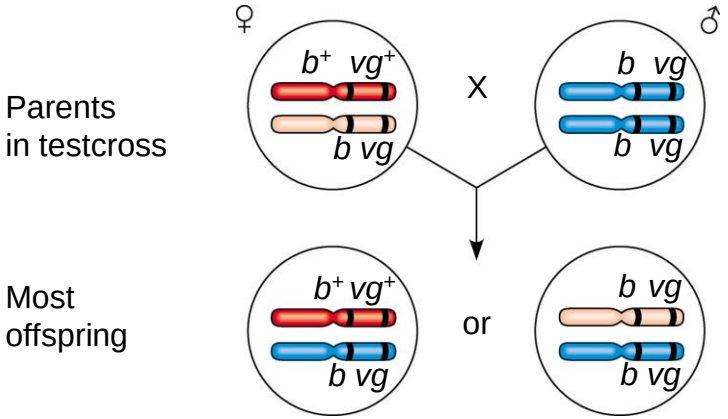


Figure 15.5

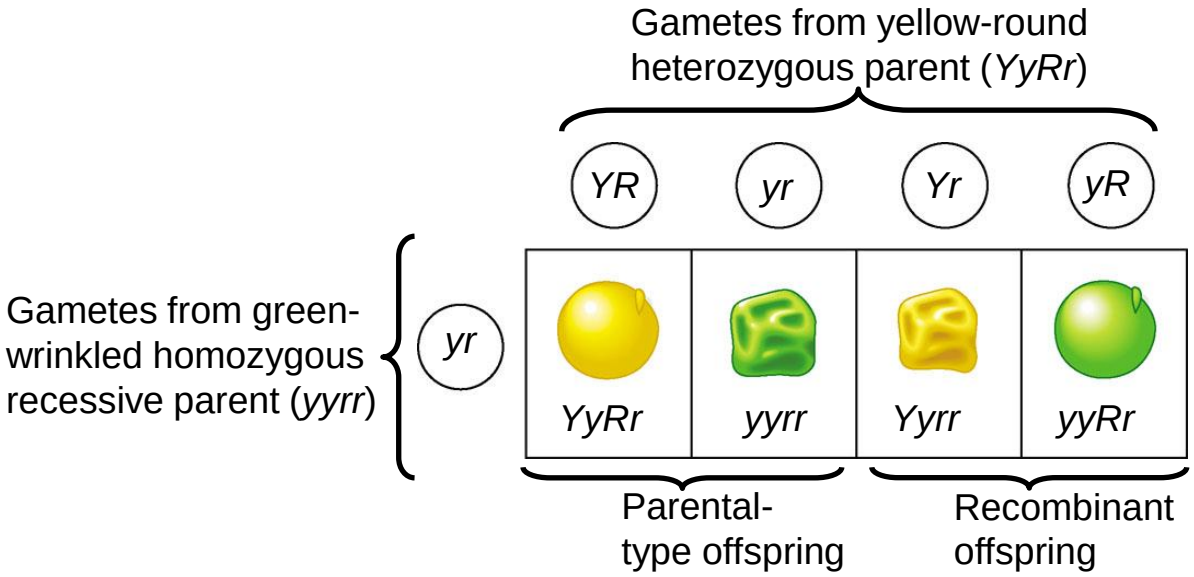
- Morgan determined that
  - Genes that are close together on the same chromosome are linked and do not assort independently
  - Unlinked genes are either on separate chromosomes of are far apart on the same chromosome and assort independently



Genetic Recombination and Linkage

Recombination of Unlinked Genes: Independent Assortment of Chromosomes

- When Mendel followed the inheritance of two characters
  - He observed that some offspring have combinations of traits that do not match either parent in the P generation



- Recombinant offspring
  - Are those that show new combinations of the parental traits
- When 50% of all offspring are recombinants
  - Geneticists say that there is a 50% frequency of recombination

Recombination of Linked Genes: Crossing Over

- Morgan discovered that genes can be linked
  - But due to the appearance of recombinant phenotypes, the linkage appeared incomplete

- Morgan proposed that
  - Some process must occasionally break the physical connection between genes on the same chromosome
  - Crossing over of homologous chromosomes was the mechanism

- Linked genes
  - Exhibit recombination frequencies less than 50%

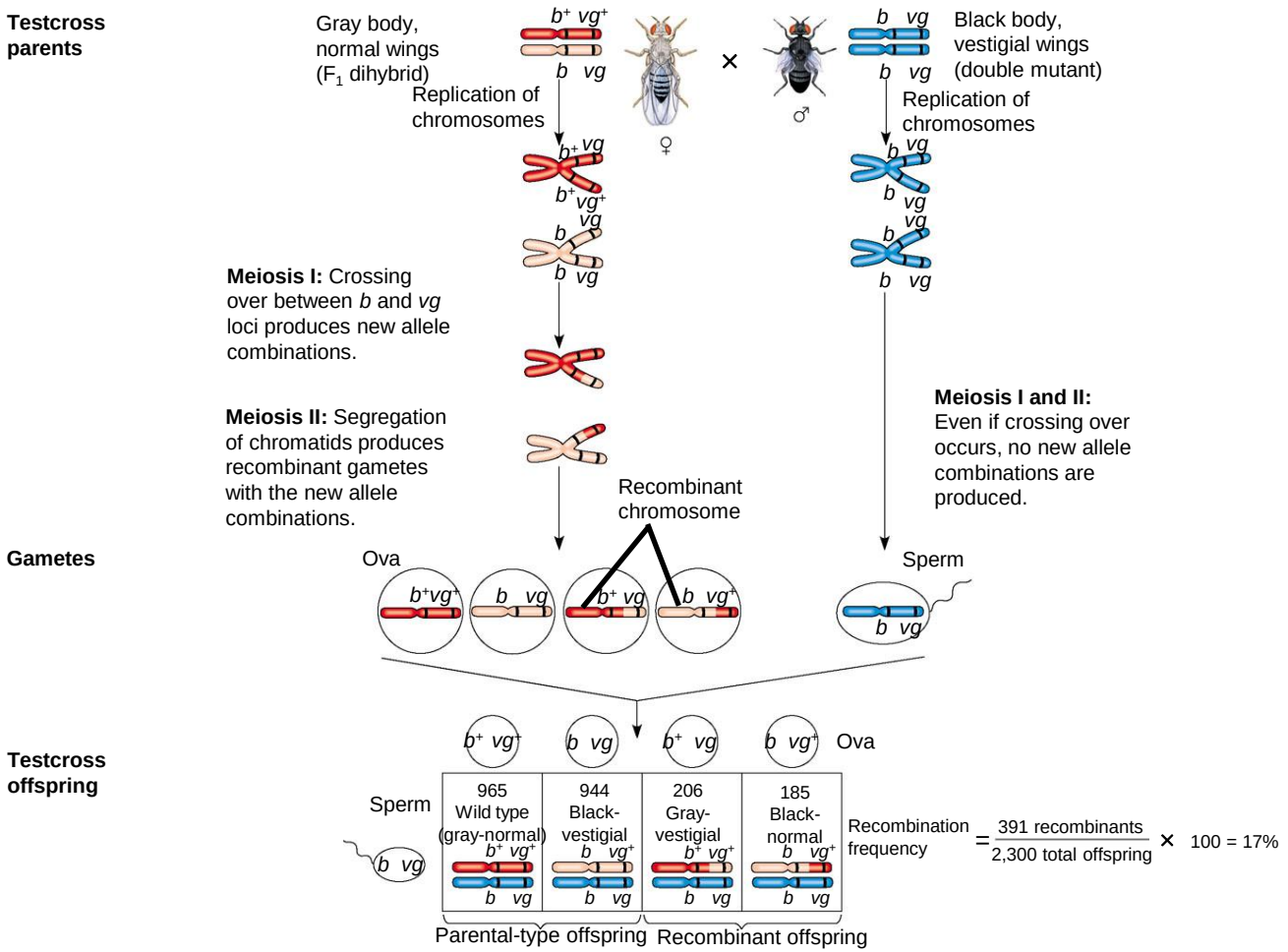


Figure 15.6

Linkage Mapping: Using Recombination Data: *Scientific Inquiry*

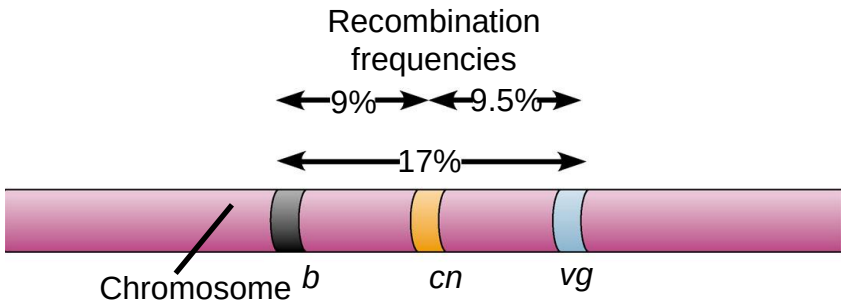
- A genetic map
  - Is an ordered list of the genetic loci along a particular chromosome
  - Can be developed using recombination frequencies

- A linkage map
  - Is the actual map of a chromosome based on recombination frequencies

APPLICATION A linkage map shows the relative locations of genes along a chromosome.

TECHNIQUE A linkage map is based on the assumption that the probability of a crossover between two genetic loci is proportional to the distance separating the loci. The recombination frequencies used to construct a linkage map for a particular chromosome are obtained from experimental crosses, such as the cross depicted in Figure 15.6. The distances between genes are expressed as map units (centimorgans), with one map unit equivalent to a 1% recombination frequency. Genes are arranged on the chromosome in the order that best fits the data.

RESULTS In this example, the observed recombination frequencies between three *Drosophila* gene pairs (*b*–*cn* 9%, *cn*–*vg* 9.5%, and *b*–*vg* 17%) best fit a linear order in which *cn* is positioned about halfway between the other two genes:



The *b*–*vg* recombination frequency is slightly less than the sum of the b–cn and *cn*–*vg* frequencies because double crossovers are fairly likely to occur between *b* and *vg* in matings tracking these two genes. A second crossover would "cancel out" the first and thus reduce the observed *b*–*vg* recombination frequency.

Figure 15.7

- The farther apart genes are on a chromosome
  - The more likely they are to be separated during crossing over

- Many fruit fly genes
  - Were mapped initially using recombination frequencies

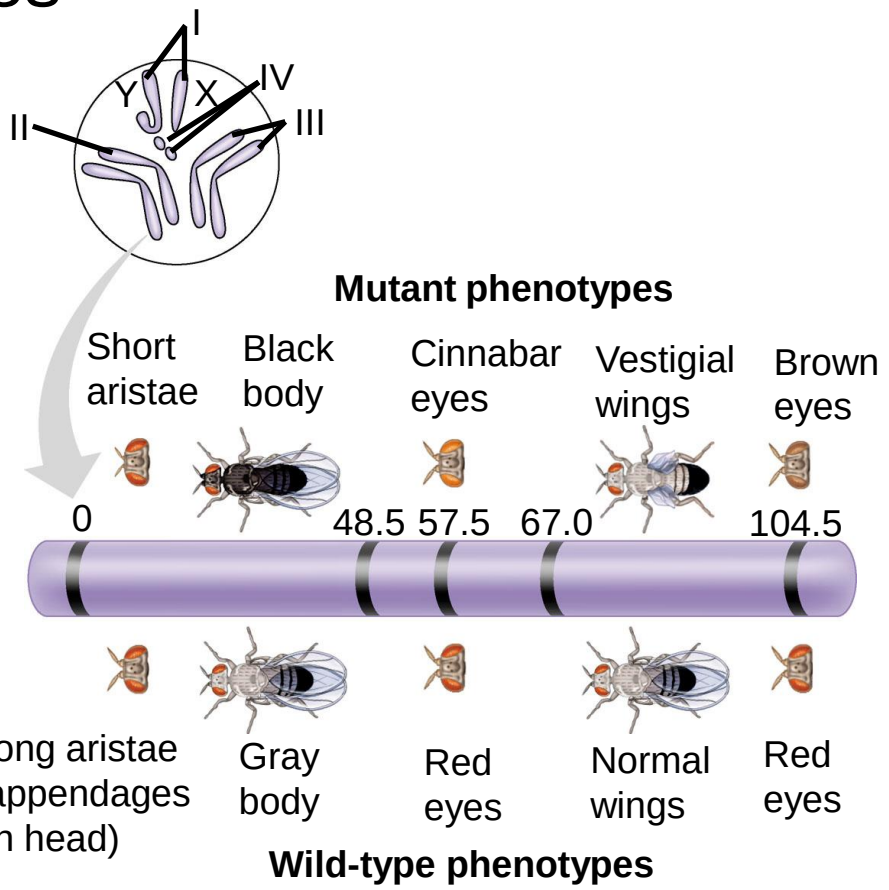


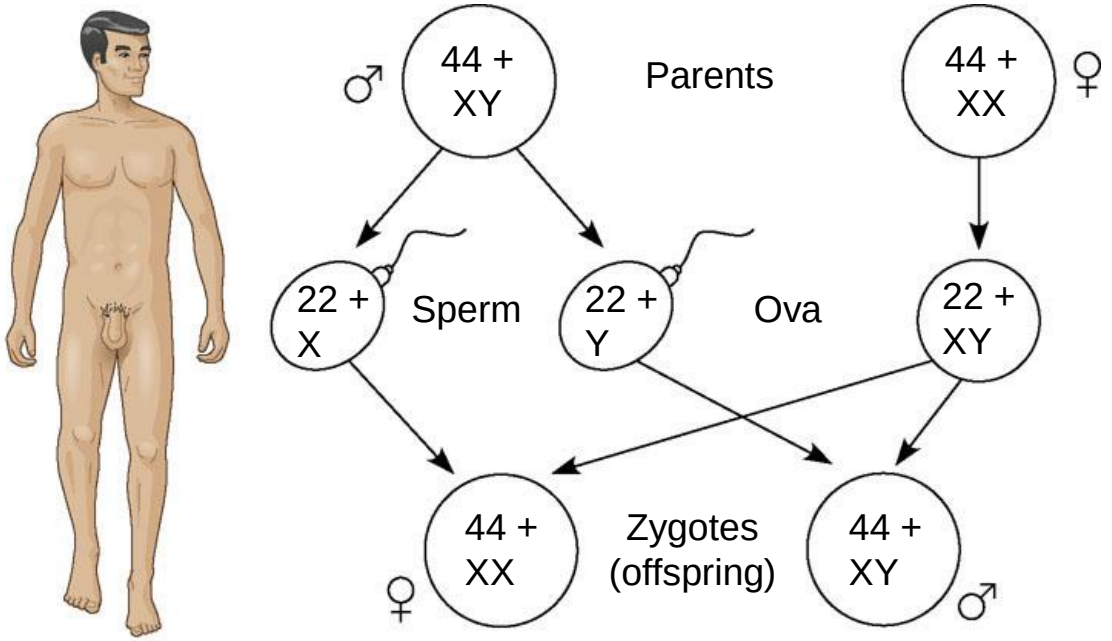
Figure 15.8

- Concept 15.3: Sex-linked genes exhibit unique patterns of inheritance

The Chromosomal Basis of Sex

- An organism's sex
  - Is an inherited phenotypic character determined by the presence or absence of certain chromosomes

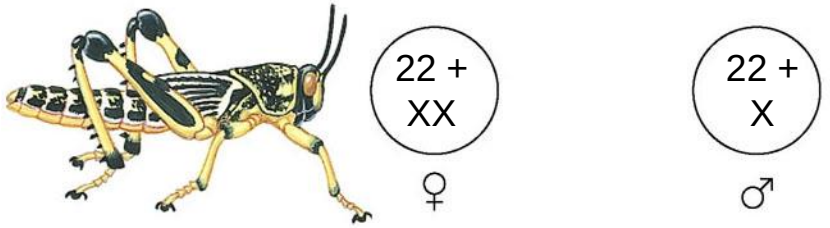
- In humans and other mammals
  - There are two varieties of sex chromosomes, X and Y



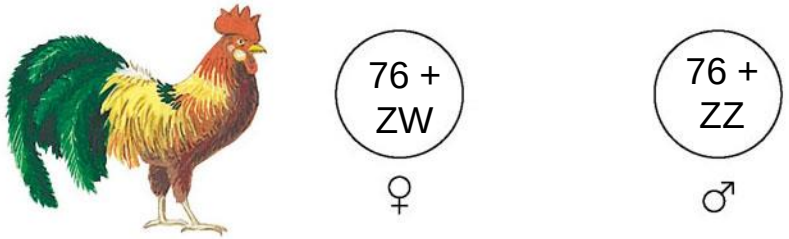
(a) The X-Y system

Figure 15.9a

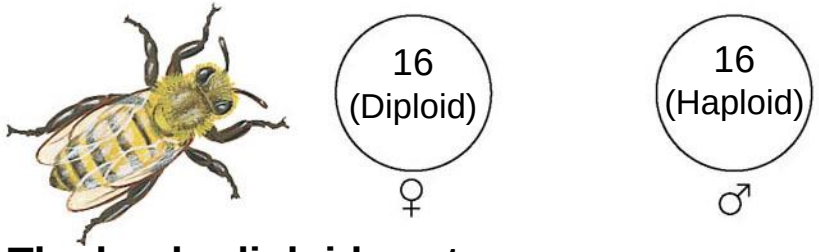
- Different systems of sex determination
  - Are found in other organisms



(b) The X–0 system



(c) The Z–W system



(d) The haplo-diploid system

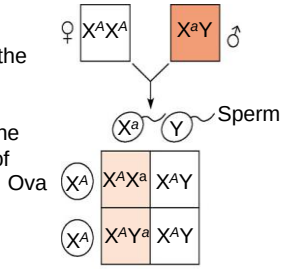
Figure 15.9b–d

Inheritance of Sex-Linked Genes

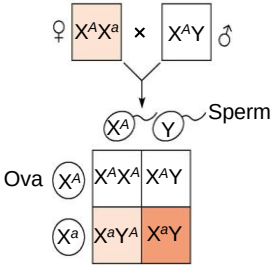
- The sex chromosomes
  - Have genes for many characters unrelated to sex
- A gene located on either sex chromosome
  - Is called a sex-linked gene

- Sex-linked genes
  - Follow specific patterns of inheritance

(a) A father with the disorder will transmit the mutant allele to all daughters but to no sons. When the mother is a dominant homozygote, the daughters will have the normal phenotype but will be carriers of the mutation.



(b) If a carrier mates with a male of normal phenotype, there is a 50% chance that each daughter will be a carrier like her mother, and a 50% chance that each son will have the disorder.



(c) If a carrier mates with a male who has the disorder, there is a 50% chance that each child born to them will have the disorder, regardless of sex. Daughters who do not have the disorder will be carriers, where as males without the disorder will be completely free of the recessive allele.

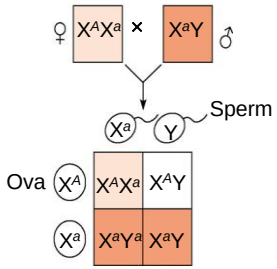


Figure 15.10a–c

- Some recessive alleles found on the X chromosome in humans cause certain types of disorders
  - Color blindness
  - Duchenne muscular dystrophy
  - Hemophilia

X inactivation in Female Mammals

- In mammalian females
  - One of the two X chromosomes in each cell is randomly inactivated during embryonic development

- If a female is heterozygous for a particular gene located on the X chromosome
  - She will be a mosaic for that character

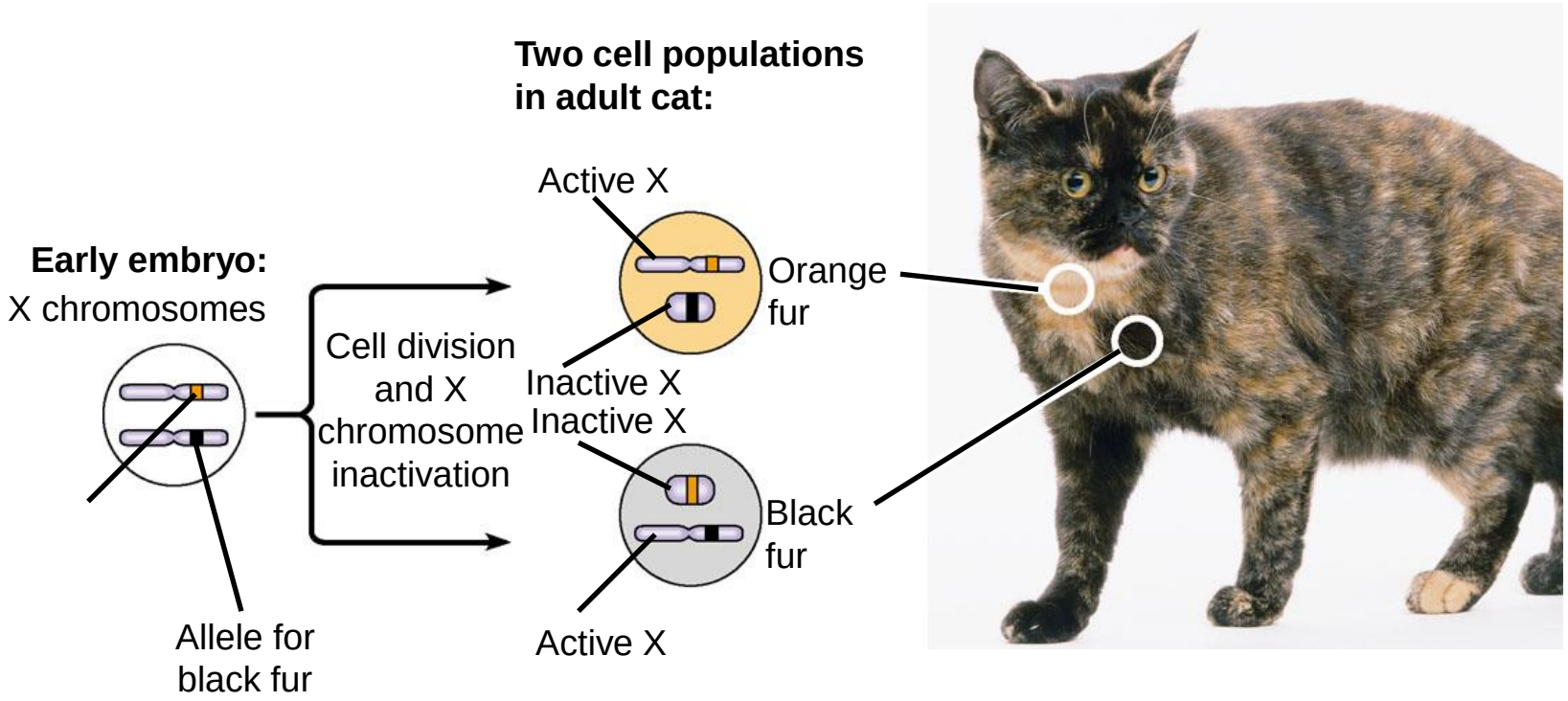


Figure 15.11

- Concept 15.4: Alterations of chromosome number or structure cause some genetic disorders
- Large-scale chromosomal alterations
  - Often lead to spontaneous abortions or cause a variety of developmental disorders

Abnormal Chromosome Number

- When nondisjunction occurs
  - Pairs of homologous chromosomes do not separate normally during meiosis
  - Gametes contain two copies or no copies of a particular chromosome

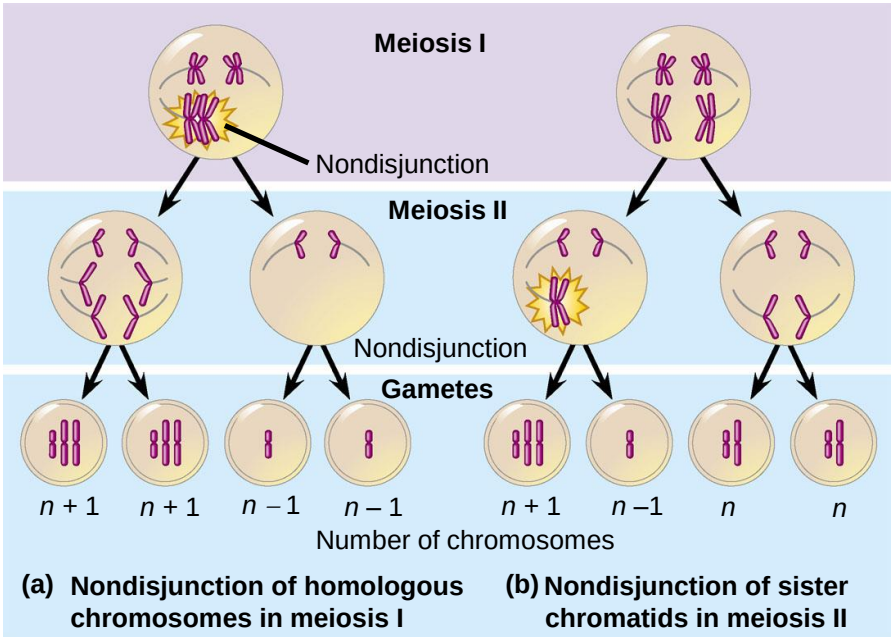


Figure 15.12a, b

- Aneuploidy
  - Results from the fertilization of gametes in which nondisjunction occurred
  - Is a condition in which offspring have an abnormal number of a particular chromosome

- If a zygote is trisomic
  - It has three copies of a particular chromosome
- If a zygote is monosomic
  - It has only one copy of a particular chromosome

- Polyploidy
  - Is a condition in which there are more than two complete sets of chromosomes in an organism

Figure 15.13

Alterations of Chromosome Structure

- Breakage of a chromosome can lead to four types of changes in chromosome structure
  - Deletion
  - Duplication
  - Inversion
  - Translocation

- # Alterations of chromosome structure

(a) A **deletion** removes a chromosomal segment.

(b) A **duplication** repeats a segment.

(c) An **inversion** reverses a segment within a chromosome.

(d) A **translocation** moves a segment from one chromosome to another, nonhomologous one. In a reciprocal translocation, the most common type, nonhomologous chromosomes exchange fragments. Nonreciprocal translocations also occur, in which a chromosome transfers a fragment without receiving a fragment in return.

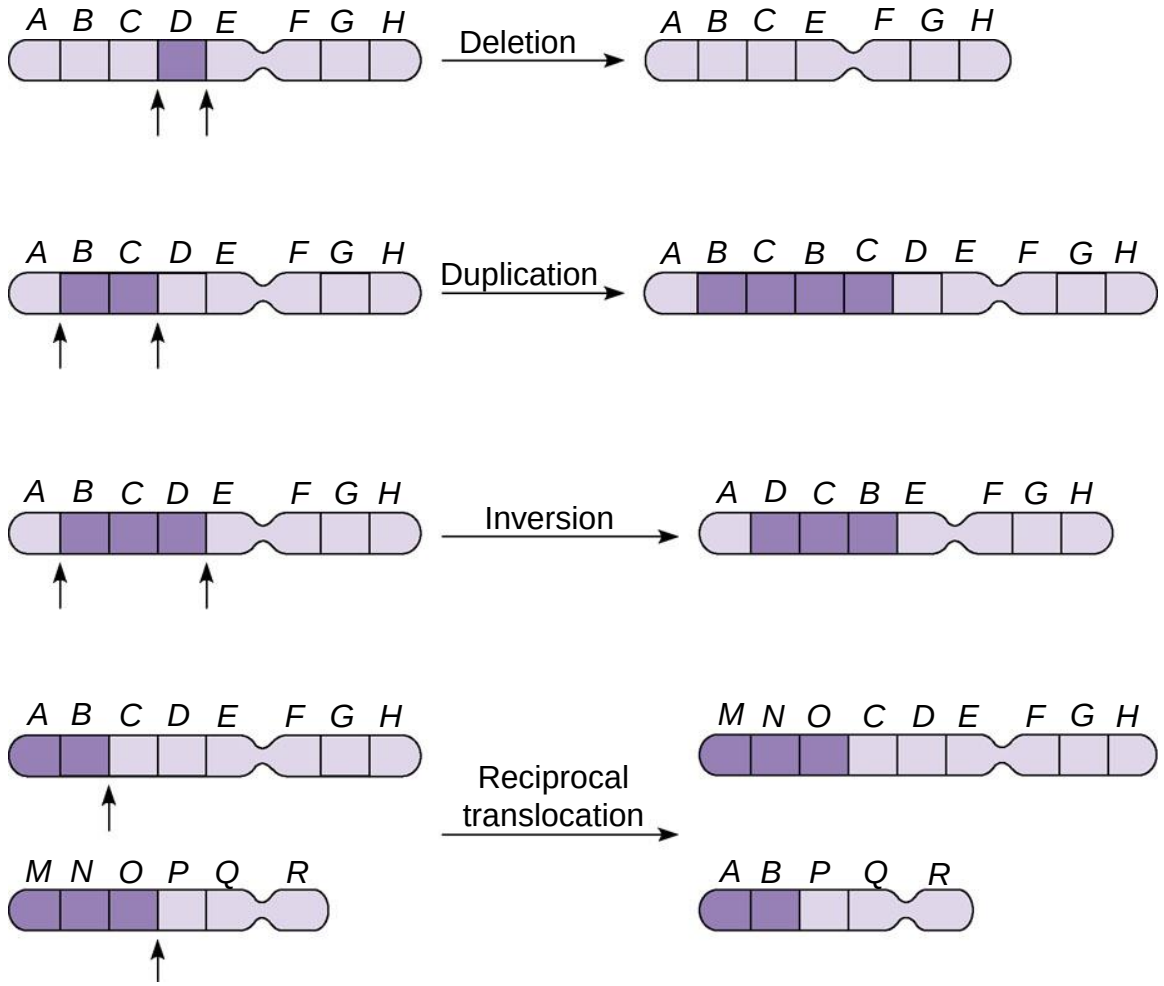


Figure 15.14a–d

Human Disorders Due to Chromosomal Alterations

- Alterations of chromosome number and structure
  - Are associated with a number of serious human disorders

Down Syndrome

- Down syndrome
  - Is usually the result of an extra chromosome 21, trisomy 21

Figure 15.15

Aneuploidy of Sex Chromosomes

- Nondisjunction of sex chromosomes
  - Produces a variety of aneuploid conditions

- Klinefelter syndrome
  - Is the result of an extra chromosome in a male, producing XXY individuals
- Turner syndrome
  - Is the result of monosomy X, producing an X0 karyotype

Disorders Caused by Structurally Altered Chromosomes

- *Cri du chat*
  - Is a disorder caused by a deletion in a chromosome

- Certain cancers
  - Are caused by translocations of chromosomes

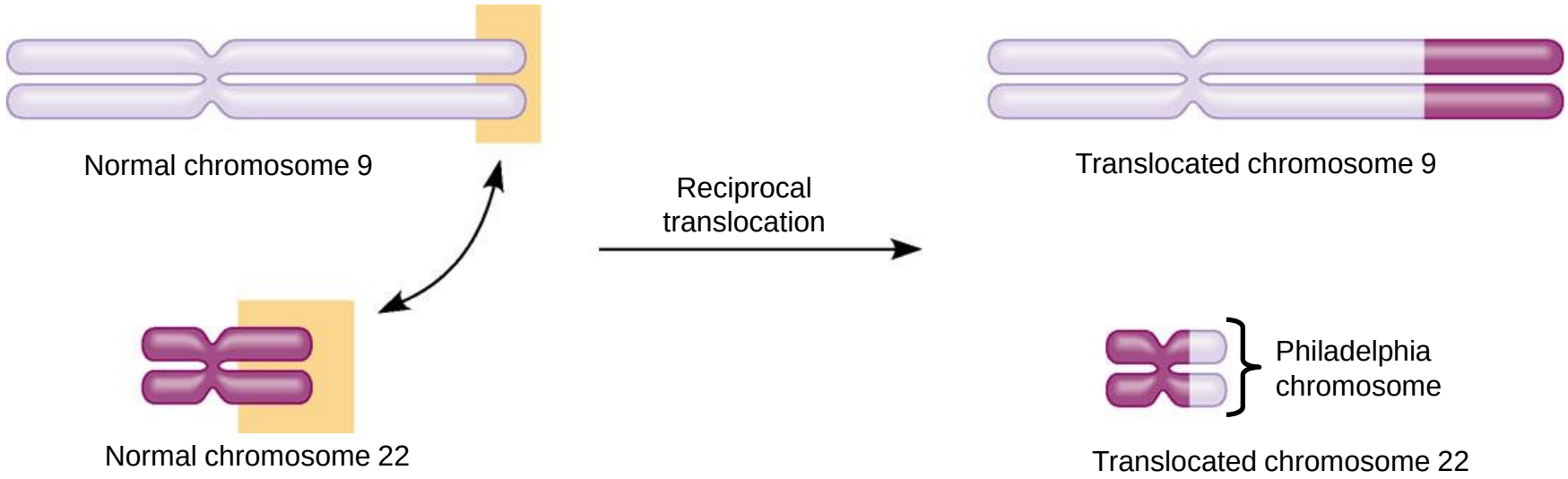


Figure 15.16

- Concept 15.5: Some inheritance patterns are exceptions to the standard chromosome theory
- Two normal exceptions to Mendelian genetics include
  - Genes located in the nucleus
  - Genes located outside the nucleus

Genomic Imprinting

- In mammals
  - The phenotypic effects of certain genes depend on which allele is inherited from the mother and which is inherited from the father

- Genomic imprinting
  - Involves the silencing of certain genes that are "stamped" with an imprint during gamete production

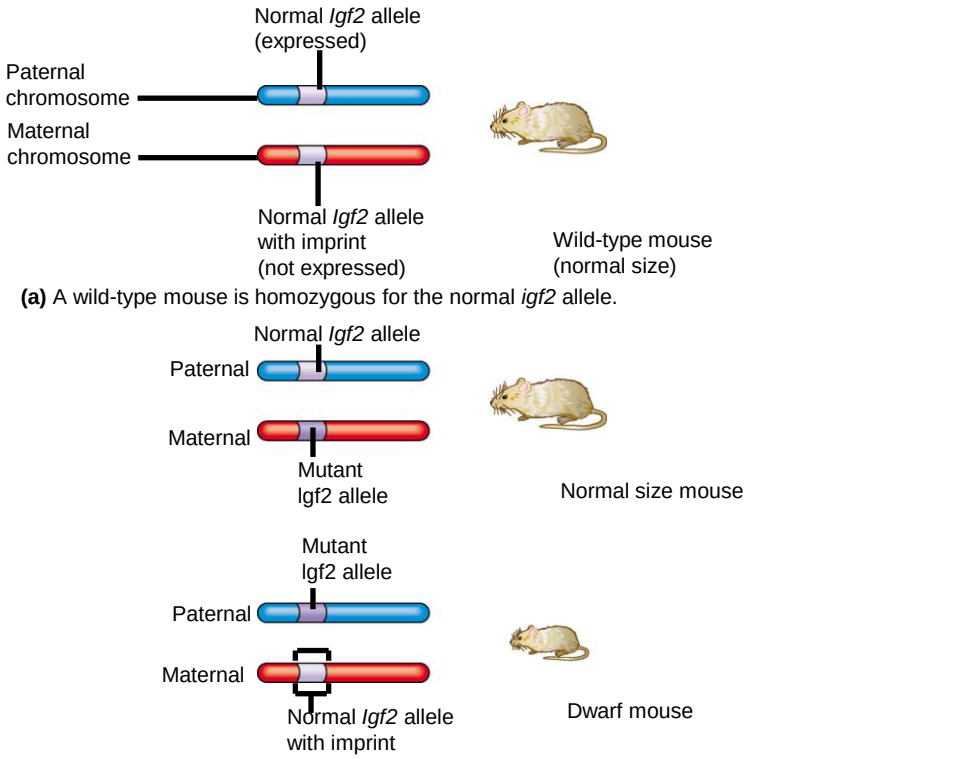


Figure 15.17a, b

Inheritance of Organelle Genes

- Extranuclear genes
  - Are genes found in organelles in the cytoplasm

- The inheritance of traits controlled by genes present in the chloroplasts or mitochondria
  - Depends solely on the maternal parent because the zygote's cytoplasm comes from the egg

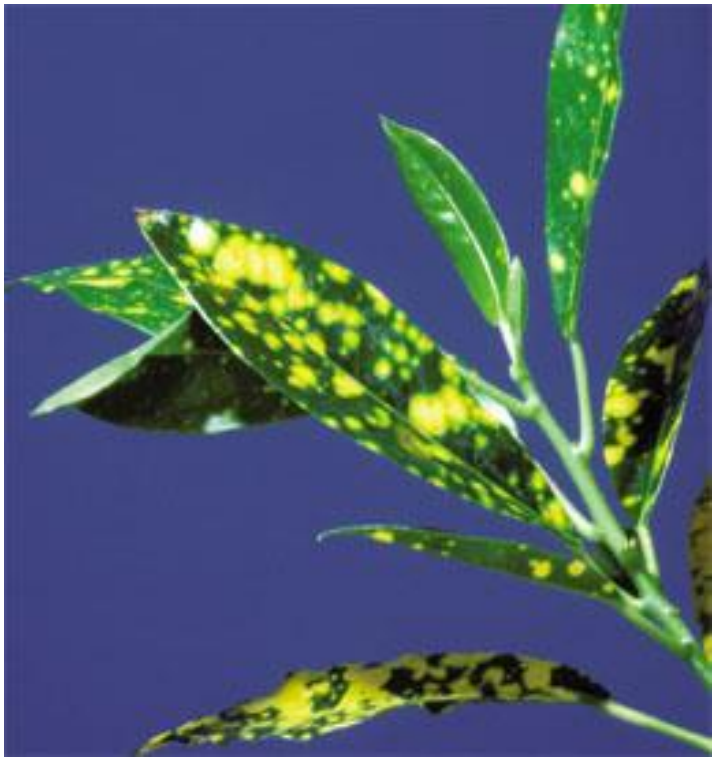


Figure 15.18

- Some diseases affecting the muscular and nervous systems
  - Are caused by defects in mitochondrial genes that prevent cells from making enough ATP