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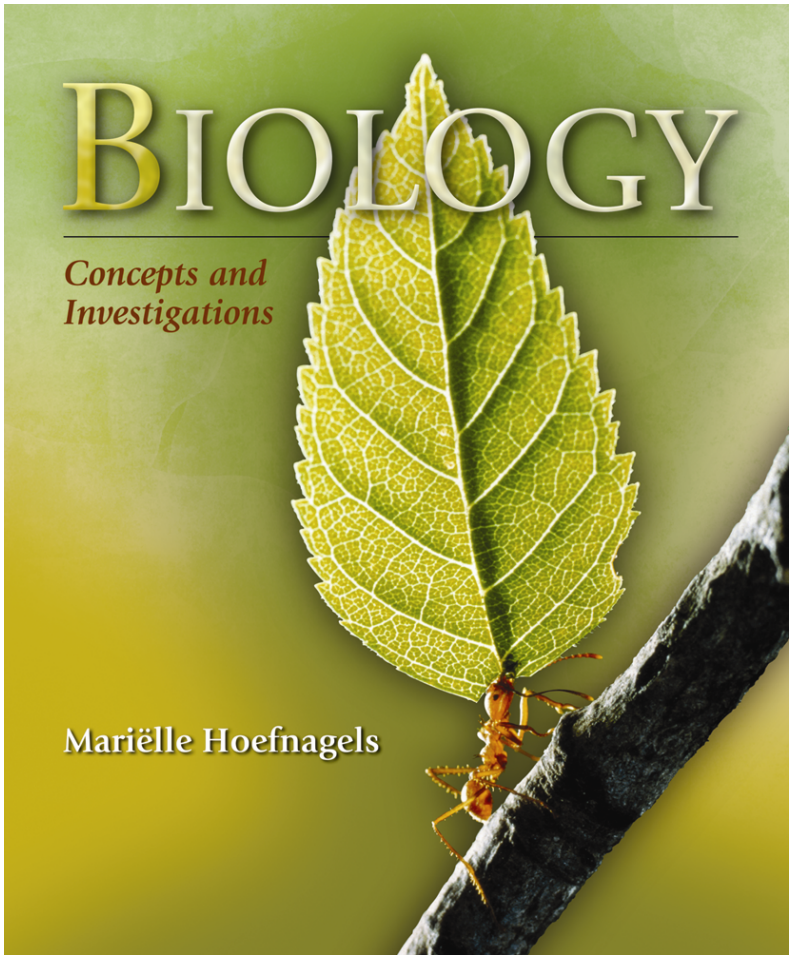
Chapter 10



10.1 Mendel and the Garden Pea

- The tendency for traits to be passed from parent to offspring is called heredity

Gregor Mendel (1822-1884)

- The first person to systematically study heredity
- Austrian monk who studied science and mathematics
- Worked with garden peas in his monastery

Mendel's Experimental System

- Mendel chose the garden pea for several reasons
  - 1. Many distinctive varieties were available
  - 2. Small and easy to grow
  - 3. Short generation time and lots of offspring
  - 4. Both male and female reproductive organs are enclosed within the pea flower

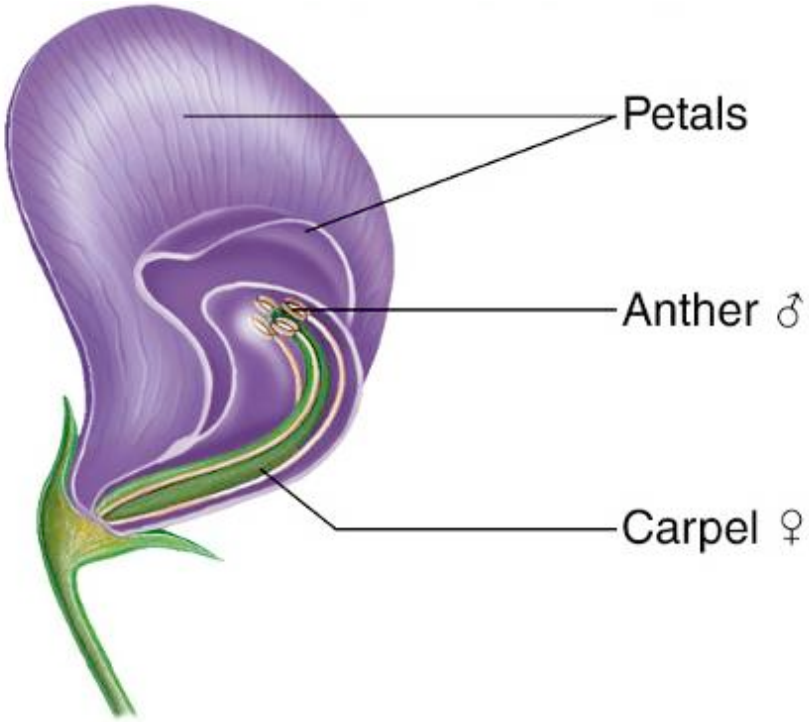
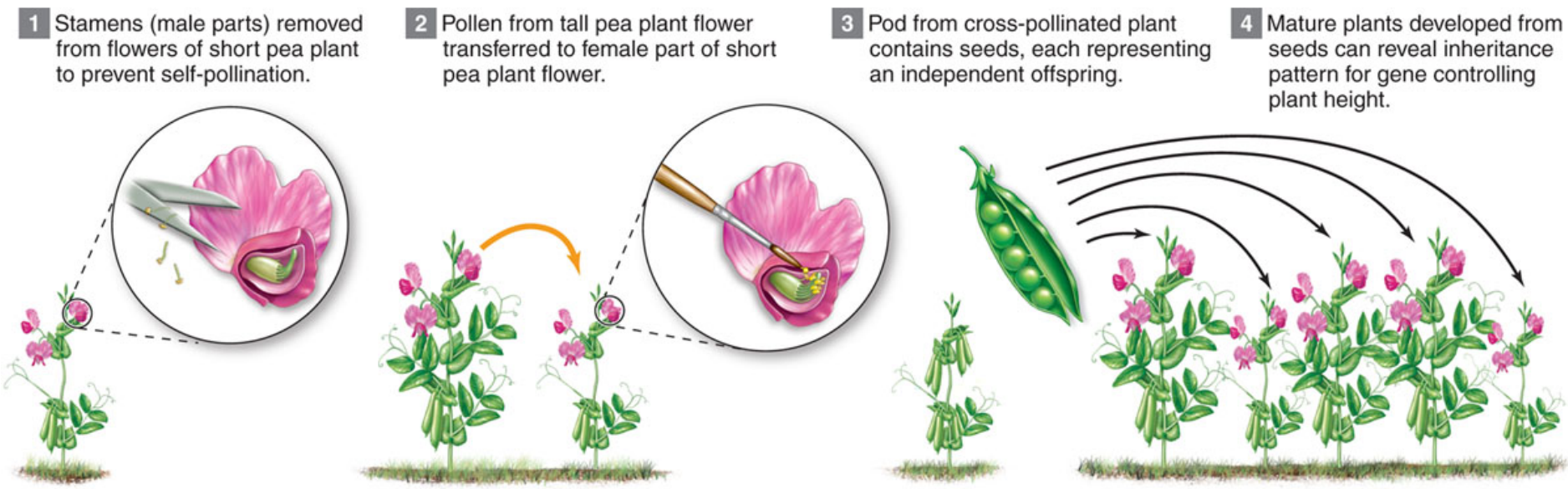


Fig. 10.2



Mendel's Experimental Design

- Mendel selected seven characteristics to study, each of which had two distinguishable traits
- He let each variety self-fertilize for many generations to ensure it was true-breeding
- He crossed individuals from two different varieties that differ in only one trait
  - P (parental) generation = Pure bred lines
  - F_1 (First filial) generation = Offspring of cross-fertilization of parentals
  - F_2 (Second filial) generation = Offspring of self-fertilization of F_1 plants

Mendel's Experimental Design

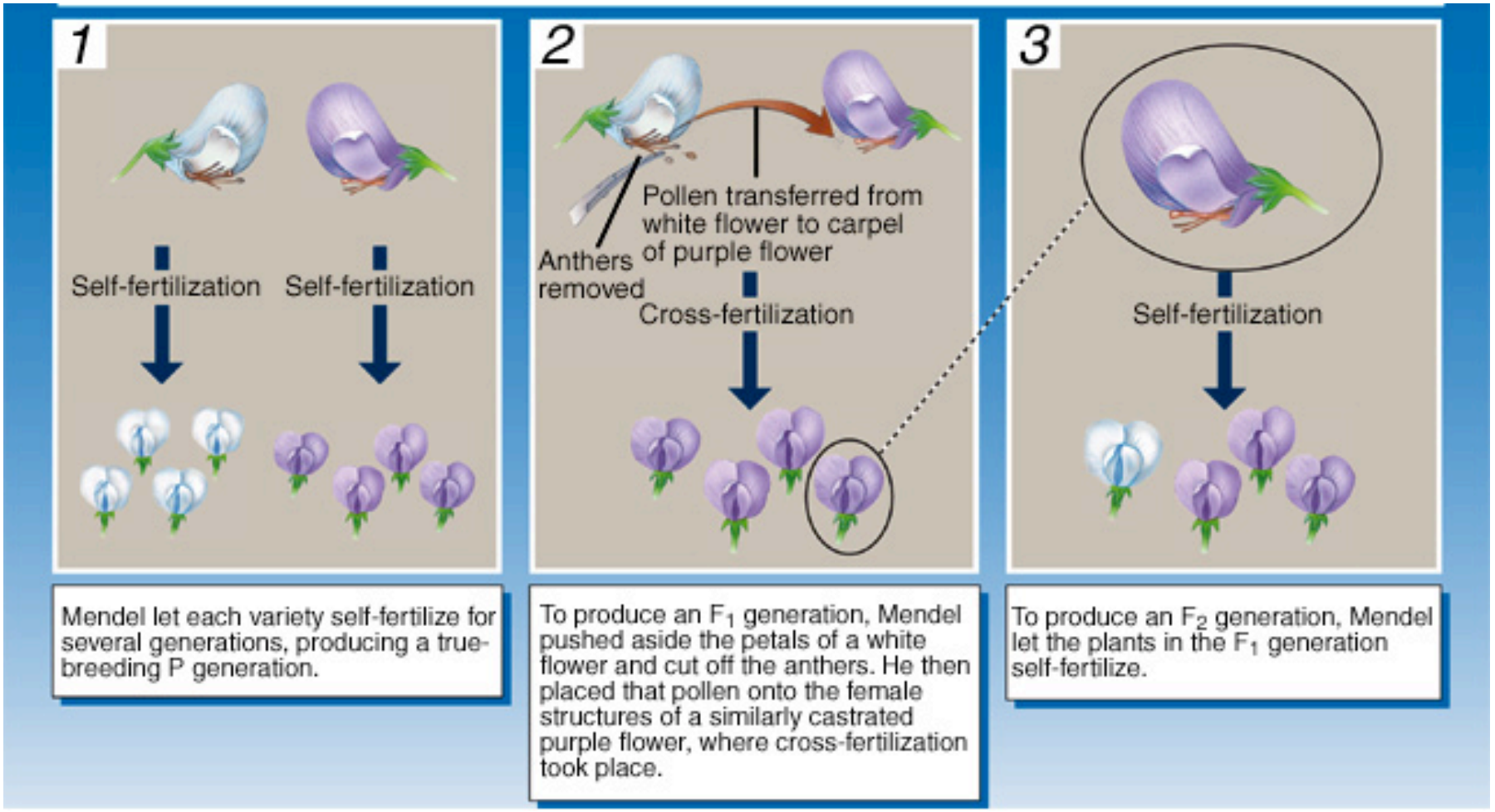


Fig. 10.3 How Mendel conducted his experiments

10.2 What Mendel Observed

- For all seven pairs of contrasting traits studied, Mendel observed the same results
  - 1. The F_1 generation showed only one of the two parental traits
    - He called it the dominant trait
    - The recessive trait was not expressed
  - 2. The F_2 generation showed an ~ 3:1 ratio of the dominant:recessive parental traits

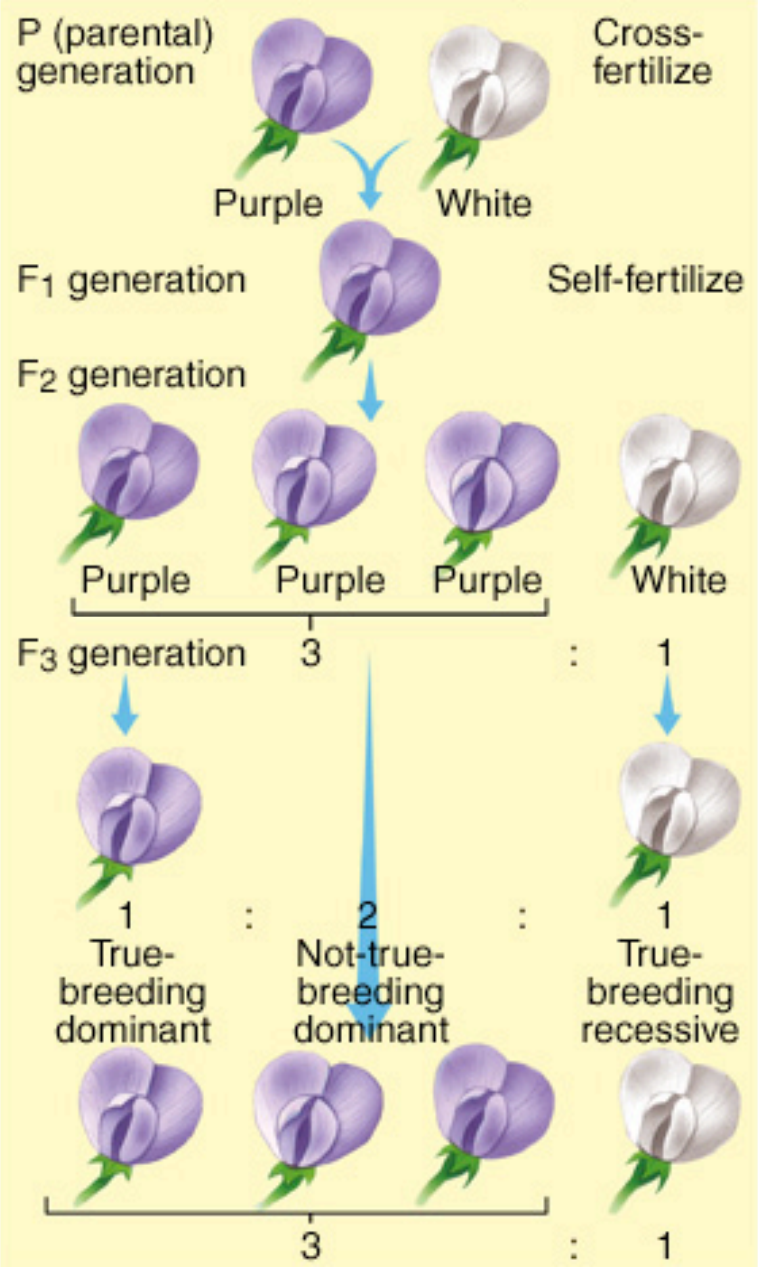
TABLE 8.1 SEVEN CHARACTERS MENDEL STUDIED IN HIS EXPERIMENTS

Character				F_2 Generation	
Dominant Form		×	Recessive Form	Dominant:Recessive	Ratio
	Purple flowers	×	White flowers 	705:224	3.15:1 (3/4:1/4)
	Yellow seeds	×	Green seeds 	6022:2001	3.01:1 (3/4:1/4)
	Round seeds	×	Wrinkled seeds 	5474:1850	2.96:1 (3/4:1/4)
	Green pods	×	Yellow pods 	428:152	2.82:1 (3/4:1/4)

TABLE 8.1 SEVEN CHARACTERS MENDEL STUDIED IN HIS EXPERIMENTS

Character				F_2 Generation	
Dominant Form	×	Recessive Form		Dominant:Recessive	Ratio
 Inflated pods	×	Constricted pods 		882:299	2.95:1 (3/4:1/4)
 Axial flowers	×	Terminal flowers 		651:207	3.14:1 (3/4:1/4)
 Tall plants	×	Dwarf plants 		787:277	2.84:1 (3/4:1/4)

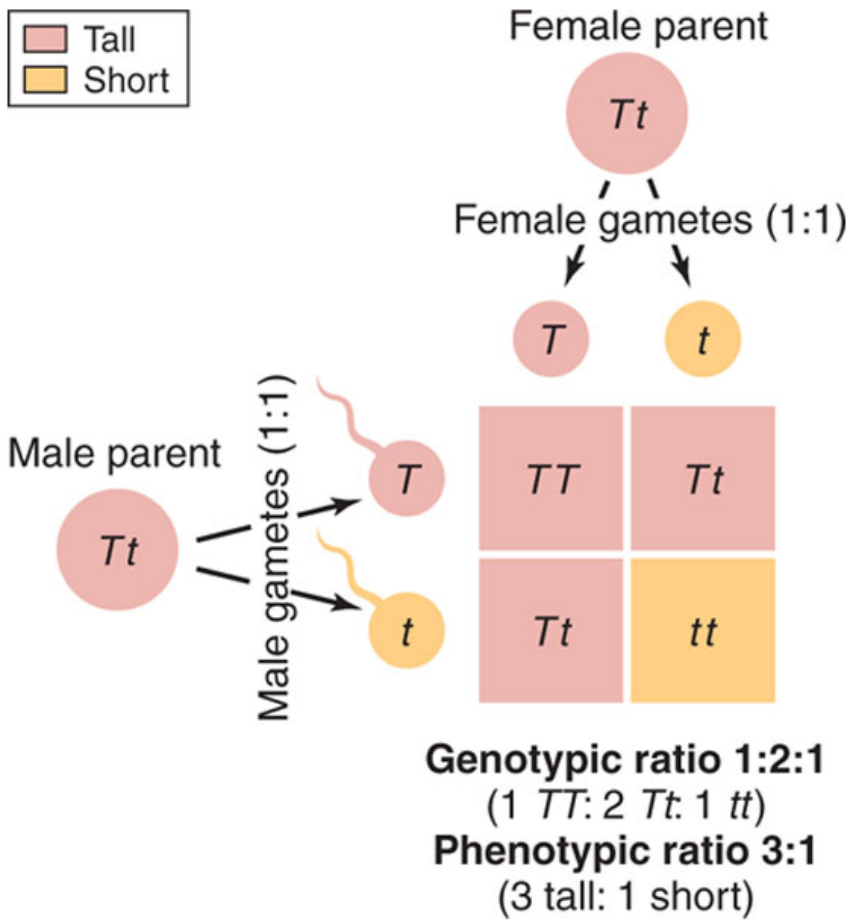
Fig. 10.5

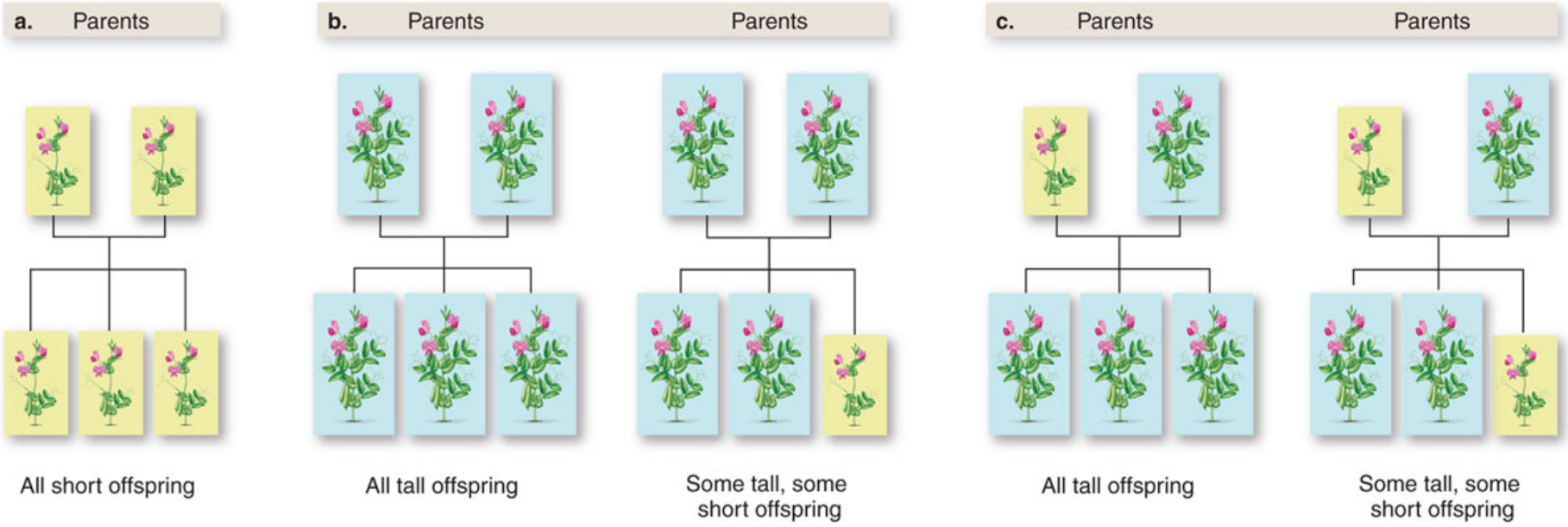


- Mendel let the F_2 plants self-fertilize for another generation
- He concluded from the results that the 3:1 ratio is a disguised 1:2:1 ratio

Two alleles end up in different gametes

- Monohybrid cross
  - Mating between 2 heterozygotes for one gene
  - F_2 has phenotypic ration of 3:1
  - Similar results in 7 pea plant traits
  - Punnet square reveals allele combinations





10.3 Mendel Proposes a Theory

- Mendel proposed a simple set of hypotheses
  - 1. Parents do not transmit traits directly to their offspring
    - They do so via factors (now termed genes)
  - 2. Each parent contains two copies of the factor governing each trait
    - If the two copies are the same, the individual is called homozygous
    - If the two copies are different, the individual is called heterozygous

10.3 Mendel Proposes a Theory

- Mendel proposed a simple set of hypotheses
  - 3. Alternative forms of a factor lead to different traits
    - Alternative forms are called alleles
    - The appearance of an individual is its phenotype
    - The genetic composition of an individual is its genotype
  - 4. The two alleles that an individual possesses do not affect each other
  - 5. The presence of an allele does not ensure that its trait will be expressed in the individual

Review the Concepts

- List the three expressions of genetic traits.
- When two true-breeding parental genotypes are crossed, the offspring are referred to as the _____ generation?

Analyzing Mendel's Results

- Each trait is determined by the inheritance of two alleles: one maternal and one paternal
  - These alleles, present on chromosomes, are distributed to gametes during meiosis

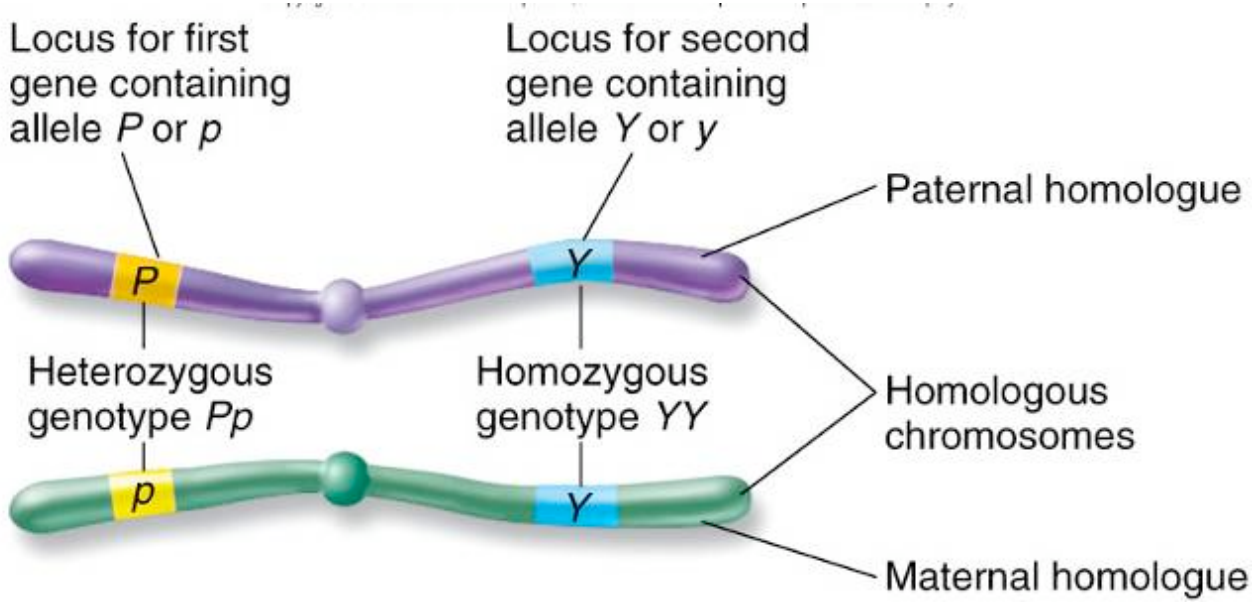


Fig. 10.6 Genes on homologous chromosomes

Analyzing Mendel's Results

- Consider Mendel's cross of purple-flowered with white-flowered pea plants
  - *P* (dominant) allele → Purple flowers
  - *p* (recessive) allele → White flowers
  - Using these conventions, the above cross can be symbolized as
    - *PP* X *pp*

Punnett Squares

- A Punnett square is a grid structure that enables the calculation of the results of simple genetic crosses
- Possible gametes are listed along two opposite sides
- Genotypes of potential offspring are represented by the cells in the square
- The frequency of these genotypes in the offspring is expressed by a probability

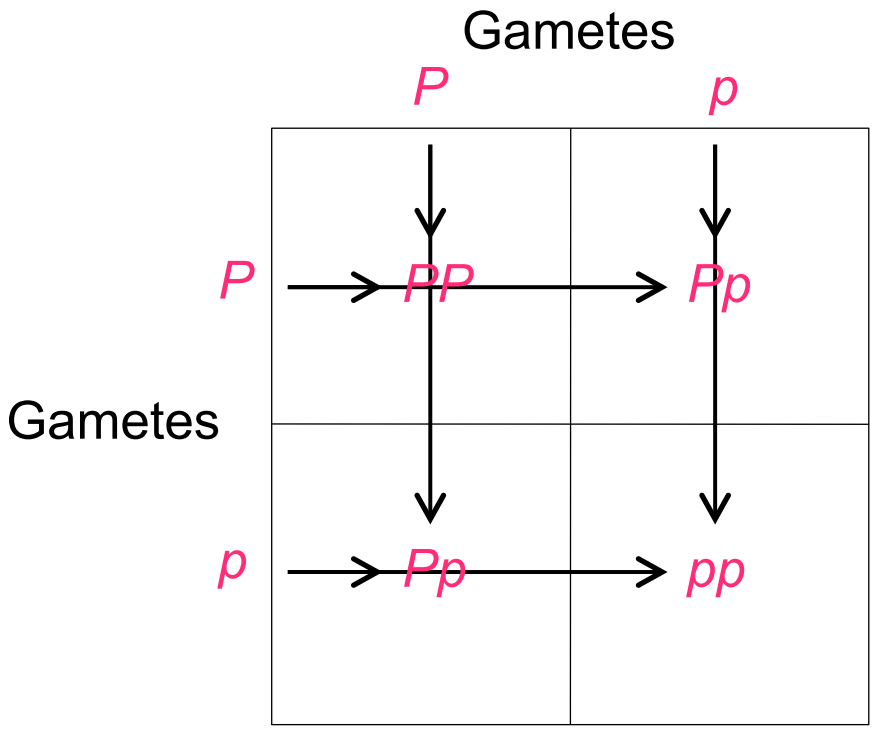
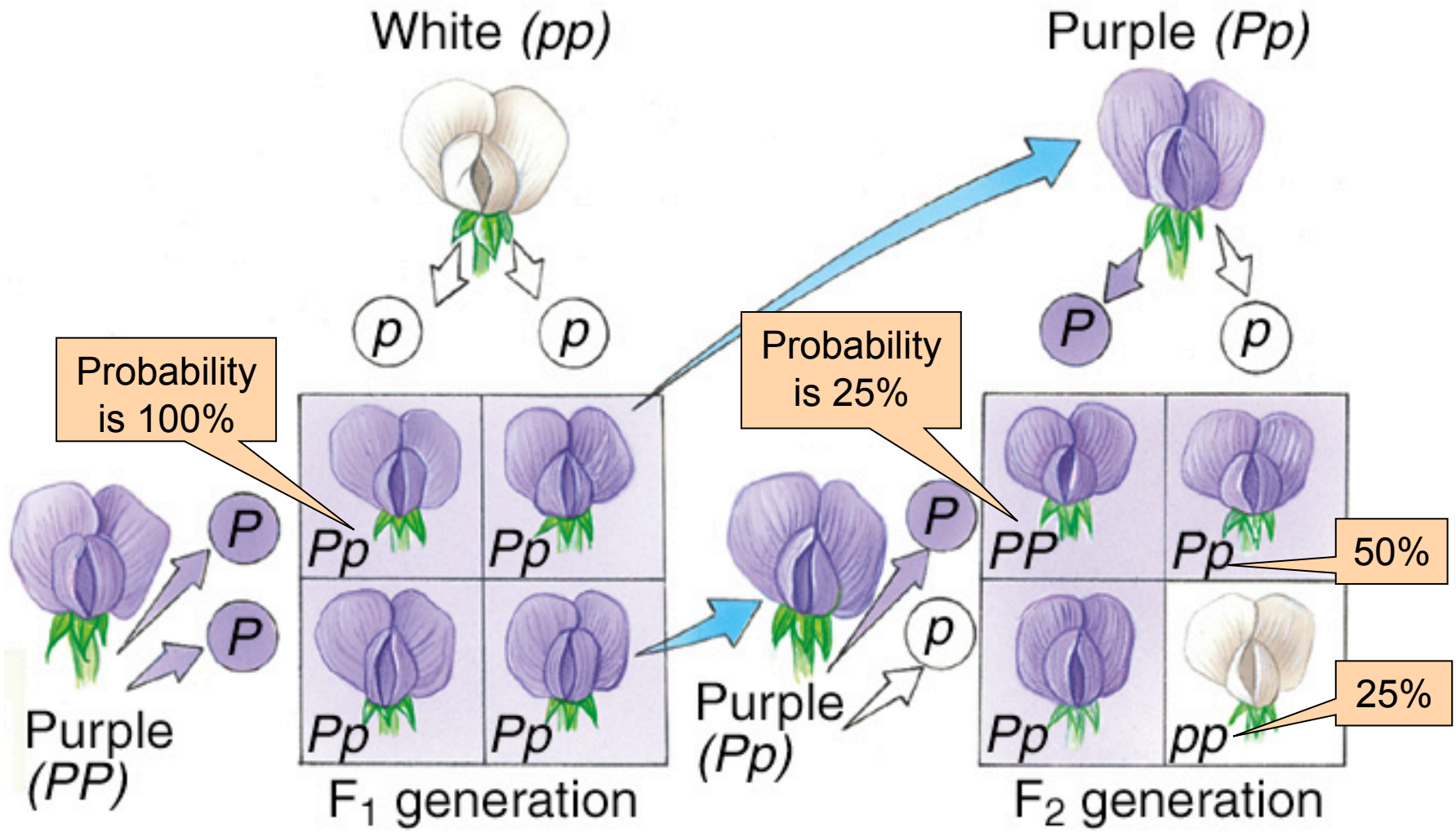


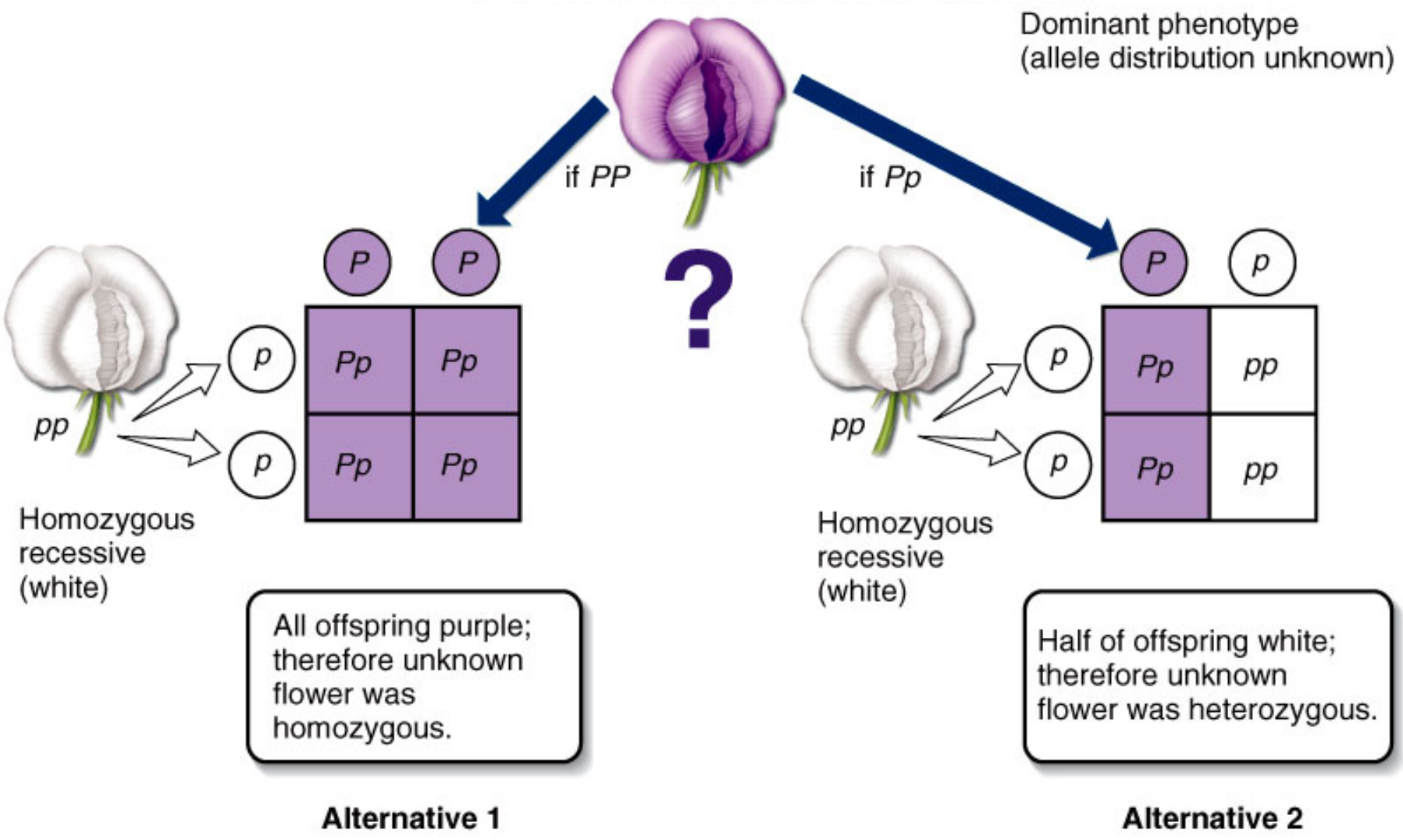
Fig. 10.10 How Mendel analyzed flower color



The Testcross

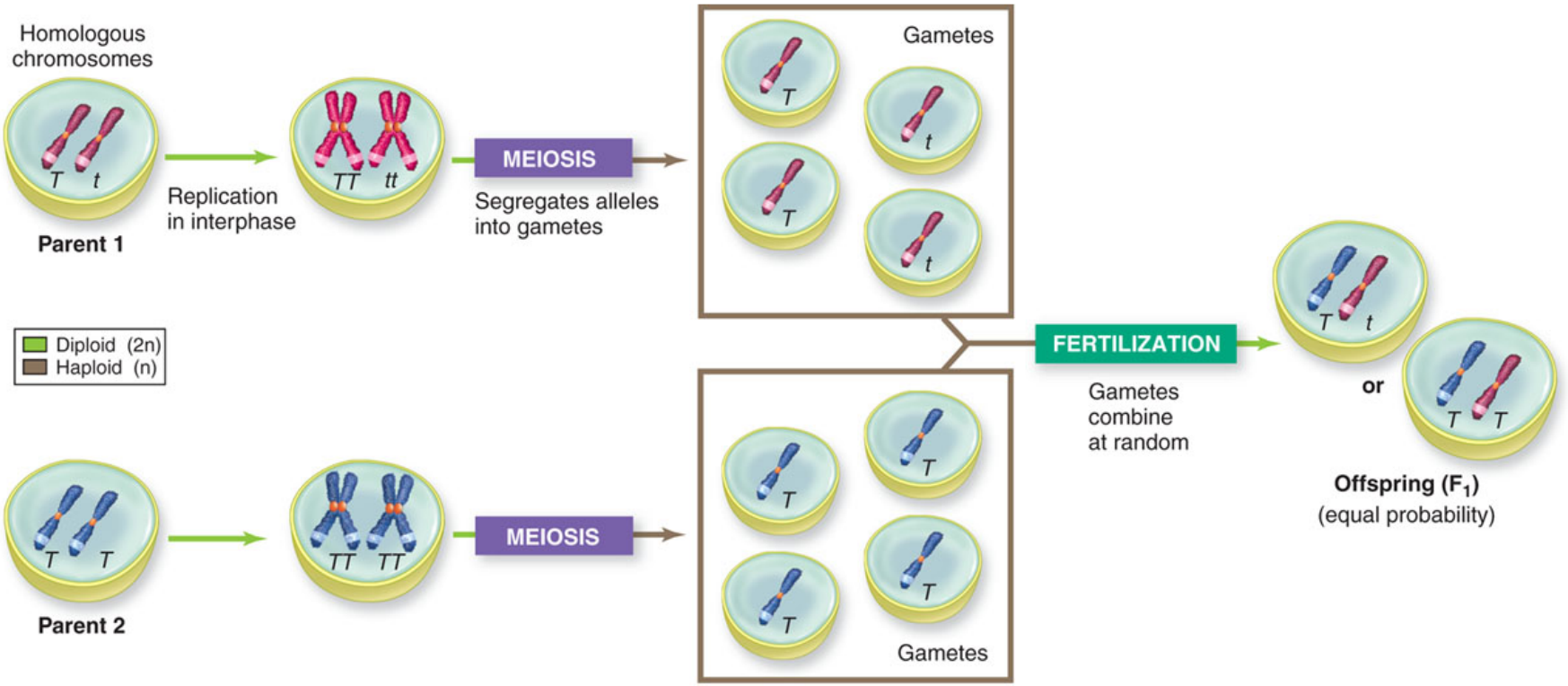
- A genetic procedure devised by Mendel to determine an individual's actual genetic composition
- A purple-flowered plant can be homozygous (*PP*) or heterozygous (*Pp*)
  - One cannot tell by simply looking at the phenotype
  - One *can* tell from the results of a cross between the test plant and a homozygous recessive plant

Fig. 10.9 How Mendel used the testcross to detect heterozygotes



10.4 Mendel's Laws

- Mendel's theory of heredity is one of the most important theories in the history of science
  - It has been so well supported by experimental results that his major proposals are considered "laws"
- Mendel's first law, or law of segregation
  - The two alleles of a gene separate when forming gametes, and gametes combine randomly in forming offspring
- Mendel's second law, or law of independent assortment
  - Alleles of genes located on different chromosomes are inherited independently of one another



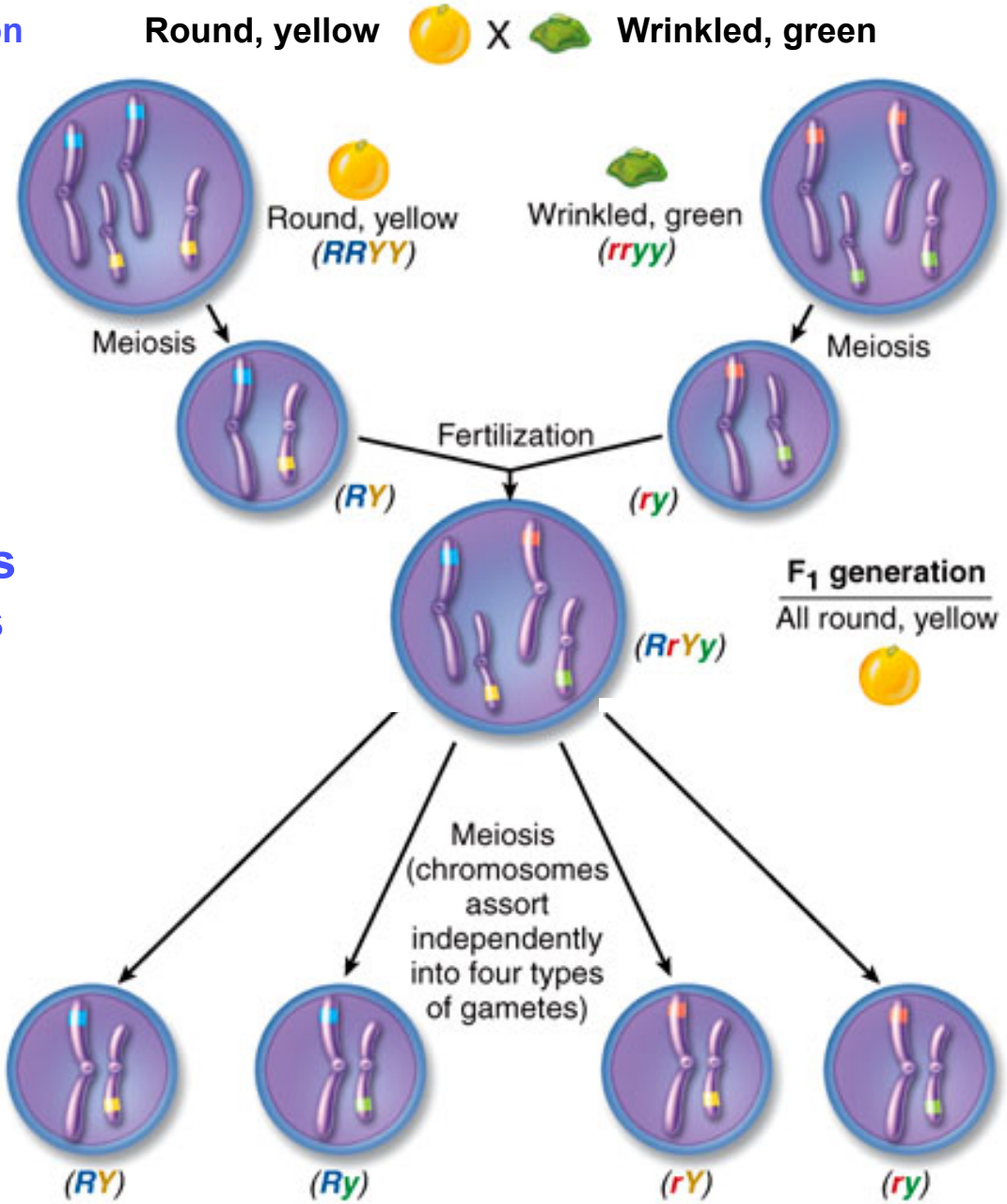
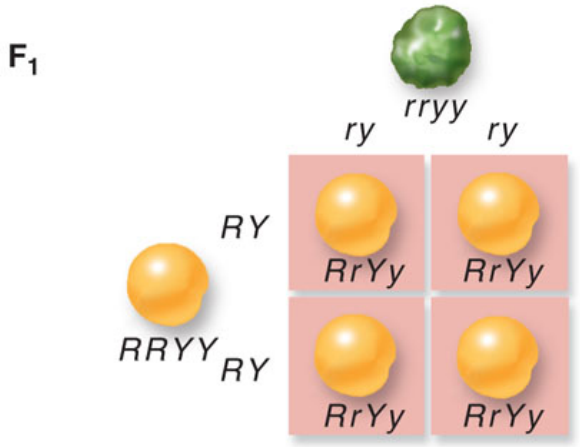
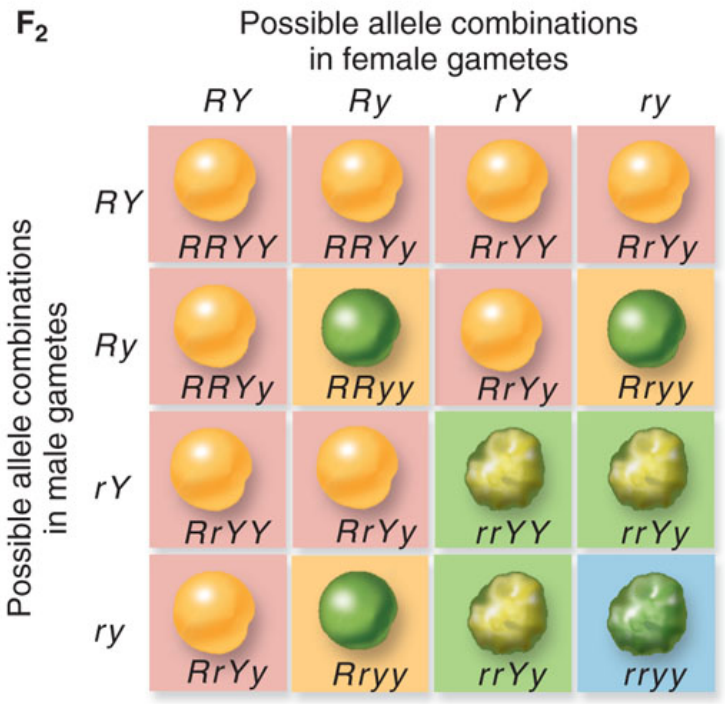


Fig. 10.10 Analysis of a dihybrid cross

a.



b.



10.5 How Genes Influence Traits

- Genes specify the amino acid sequence of proteins
  - The amino acid sequence determines the shape and activity of proteins
    - Proteins determine in large measure what the body looks like and how it functions
- Mutations in a gene result in alleles
  - This ultimately leads to a change in the amino acid sequence and, hence, activity of the protein
- Natural selection may favor one allele over another

Review the Concepts

- Tall is dominant to dwarf in pea plants. If two dwarf pea plants were crossed, their offspring will have which traits?
- Two true-breeding pea plants are crossed, one with purple flowers and the other with white. Their offspring will have which traits?
- Mendel tested for heterozygotes by using which kind of cross?

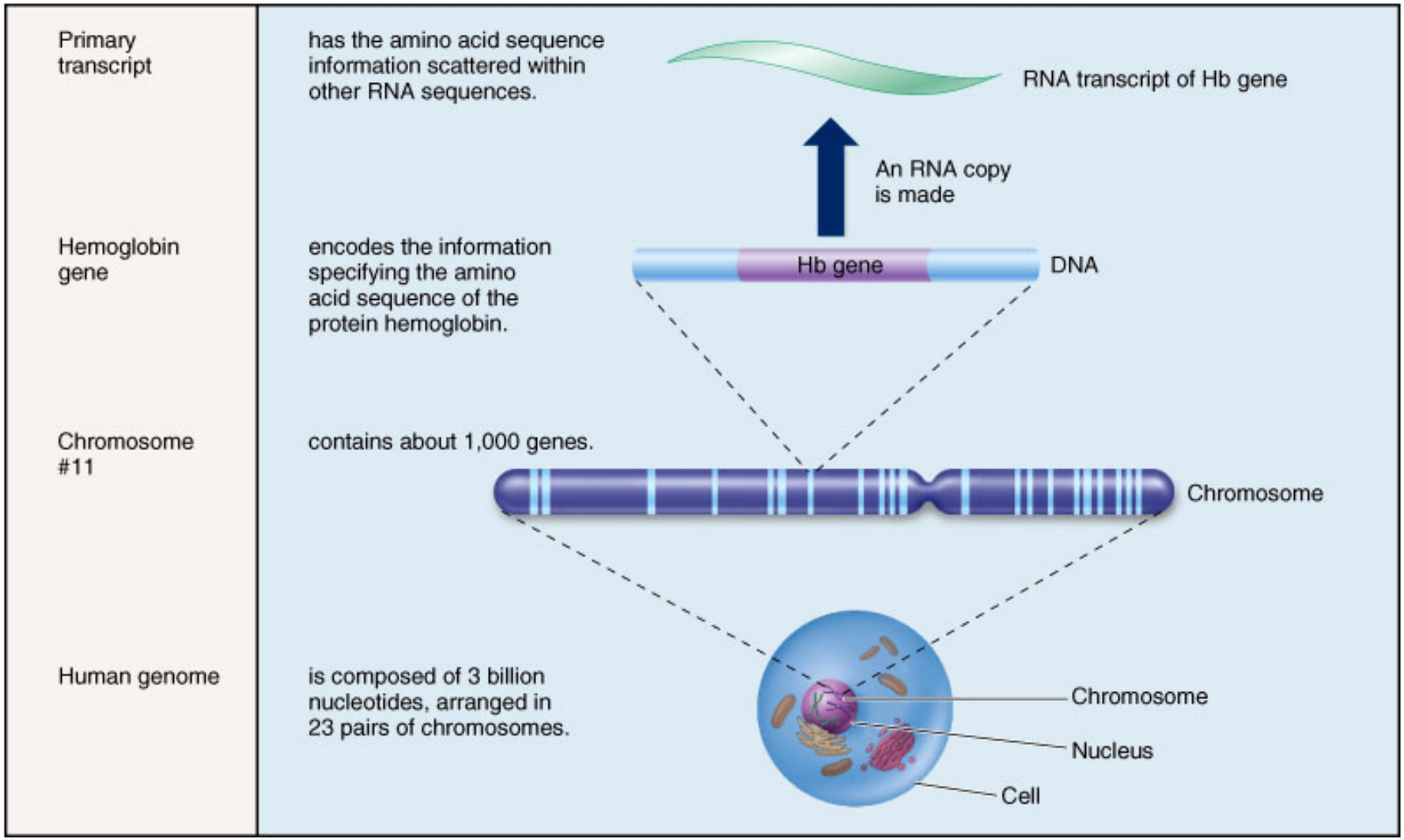


Fig. 10.11 The journey from DNA to phenotype

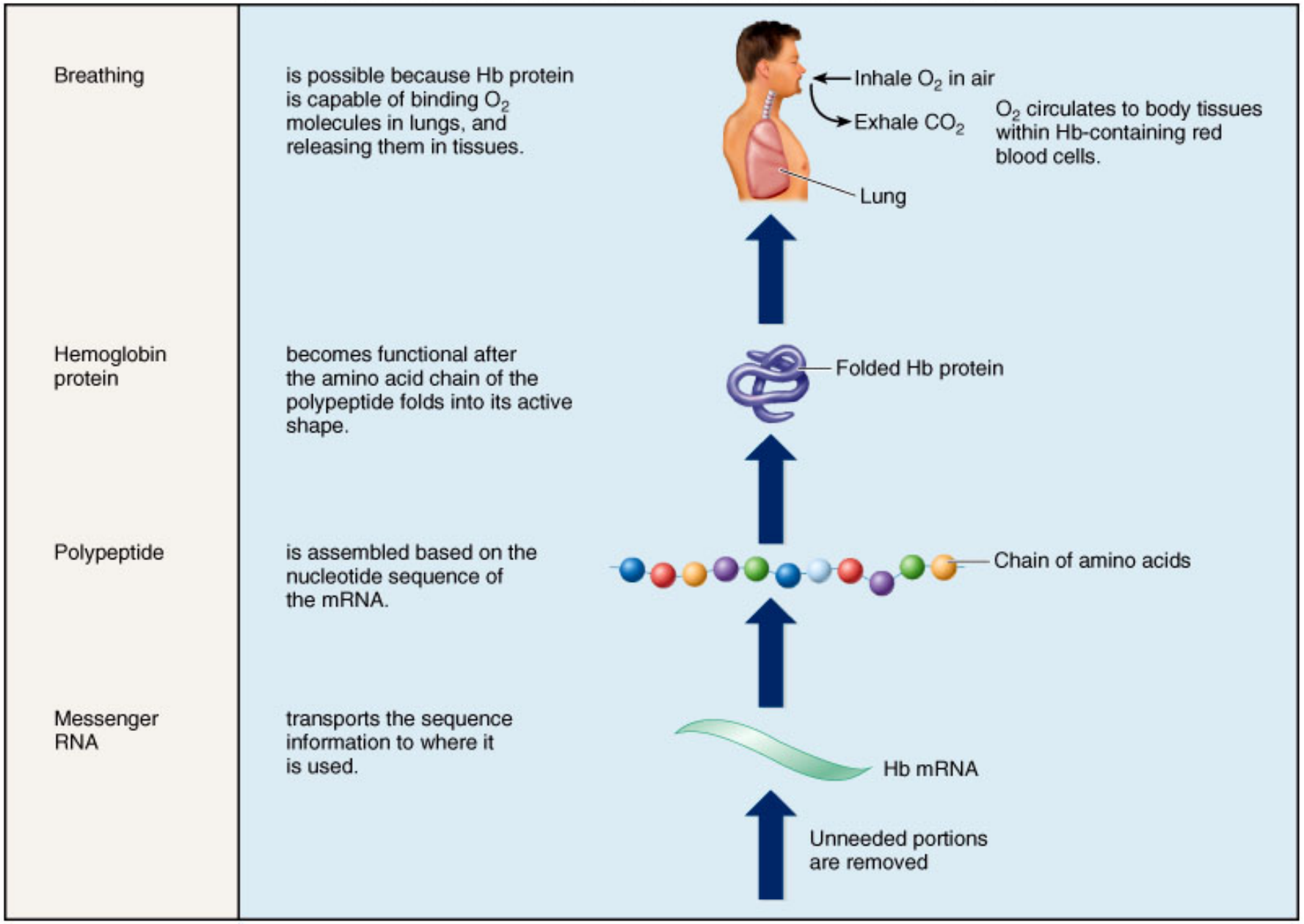


Fig. 10.11 The journey from DNA to phenotype

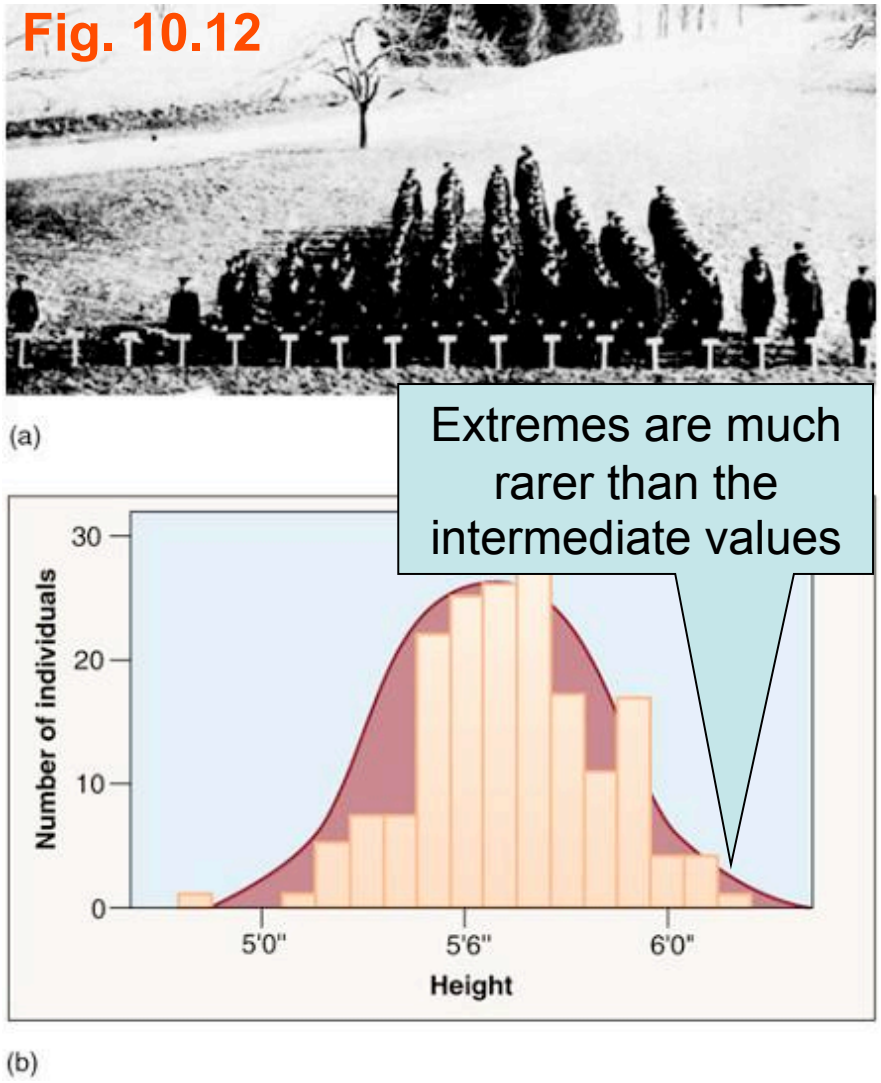
10.6 Why Some Traits Don't Show Mendelian Inheritance

- Mendelian segregation of alleles can be disguised by a variety of factors

1. Continuous variation
2. Pleiotropic effects
3. Incomplete dominance
4. Environmental effects
5. Epistasis
6. Codominance

Continuous Variation

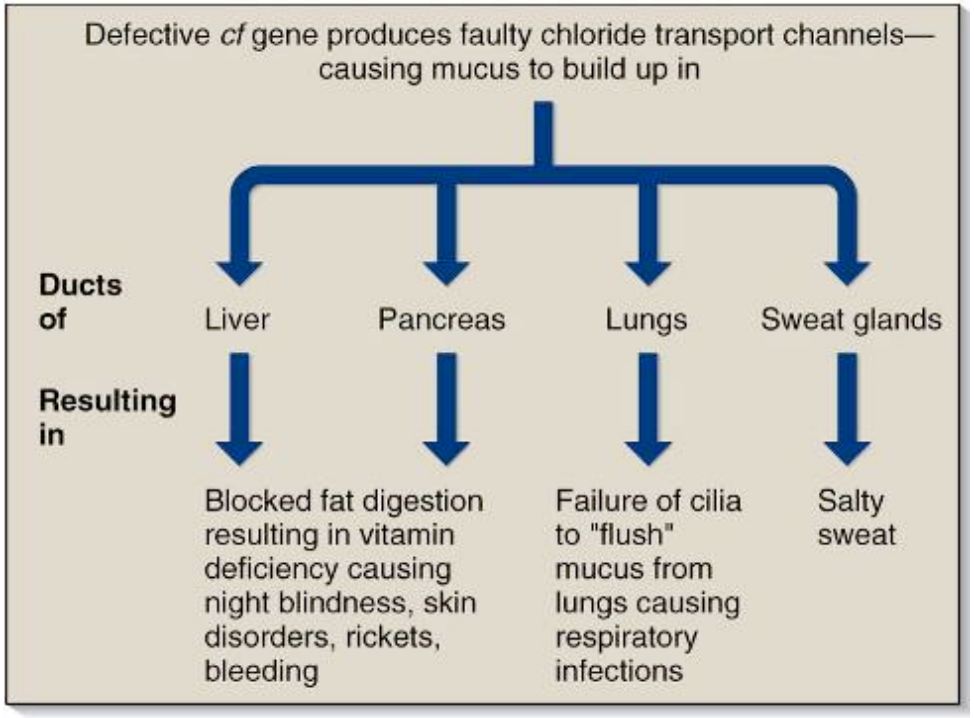
- Most traits are polygenic
  - They result from the action of more than one gene
- These genes contribute in a cumulative way to the phenotype
  - The result is a gradation in phenotypes or continuous variation



Pleiotropic Effects

- Alleles that have more than one phenotypic effect are said to be pleiotropic
- The effect may be dominant with respect to one phenotype, and recessive with respect to another
- Pleiotropic effects are characteristic of many inherited disorders
  - Cystic fibrosis

Fig. 10.13

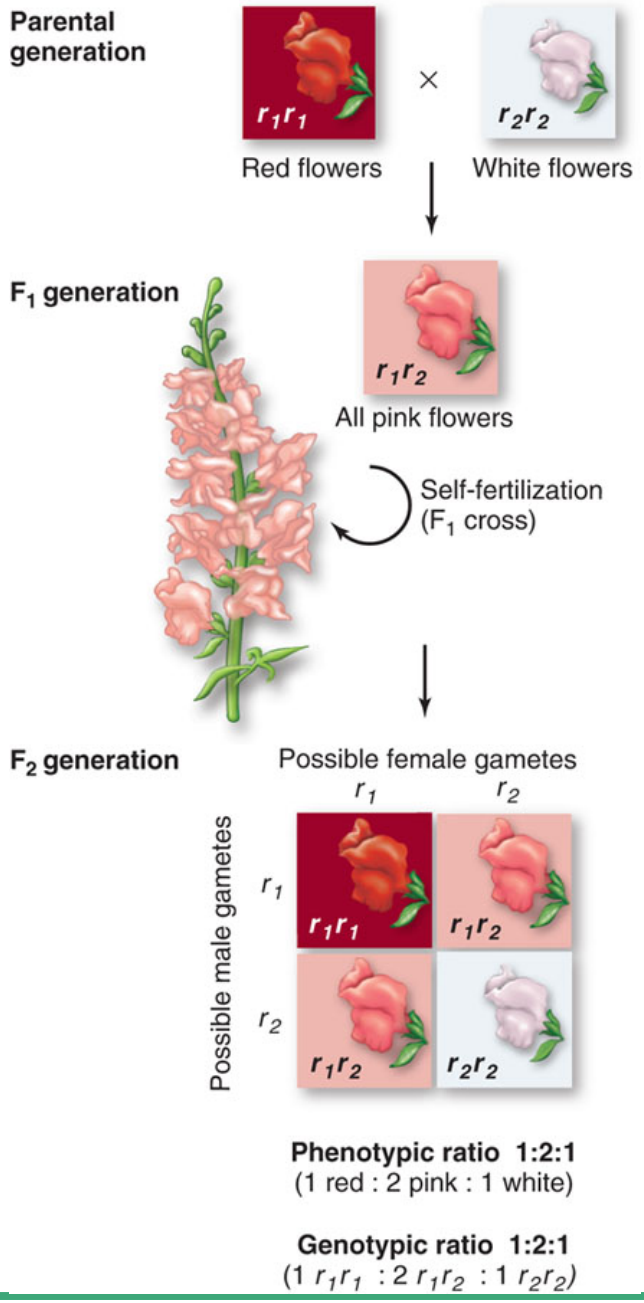


Incomplete Dominance

- Not all alternative alleles are fully dominant or fully recessive in heterozygotes

- Some pairs of alleles exhibit incomplete dominance
  - They produce a heterozygote phenotype that is *intermediate* between that of the homozygotes

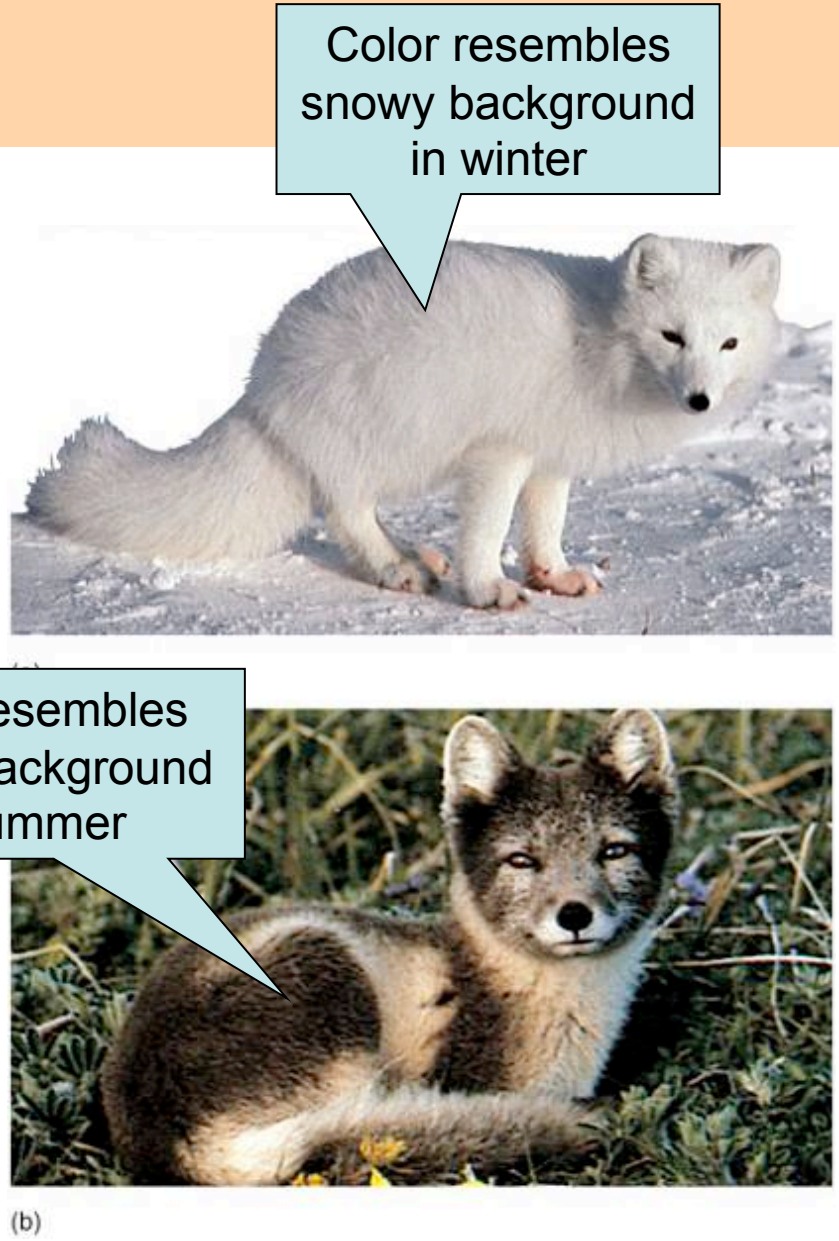
  - Example
    - Flower color in the Japanese four o'clock



Environmental Effects

- The expression of some genes is influenced by environmental factors, such as temperature

- Some alleles are heat-sensitive
  - Arctic foxes make fur pigment only when the weather is warm



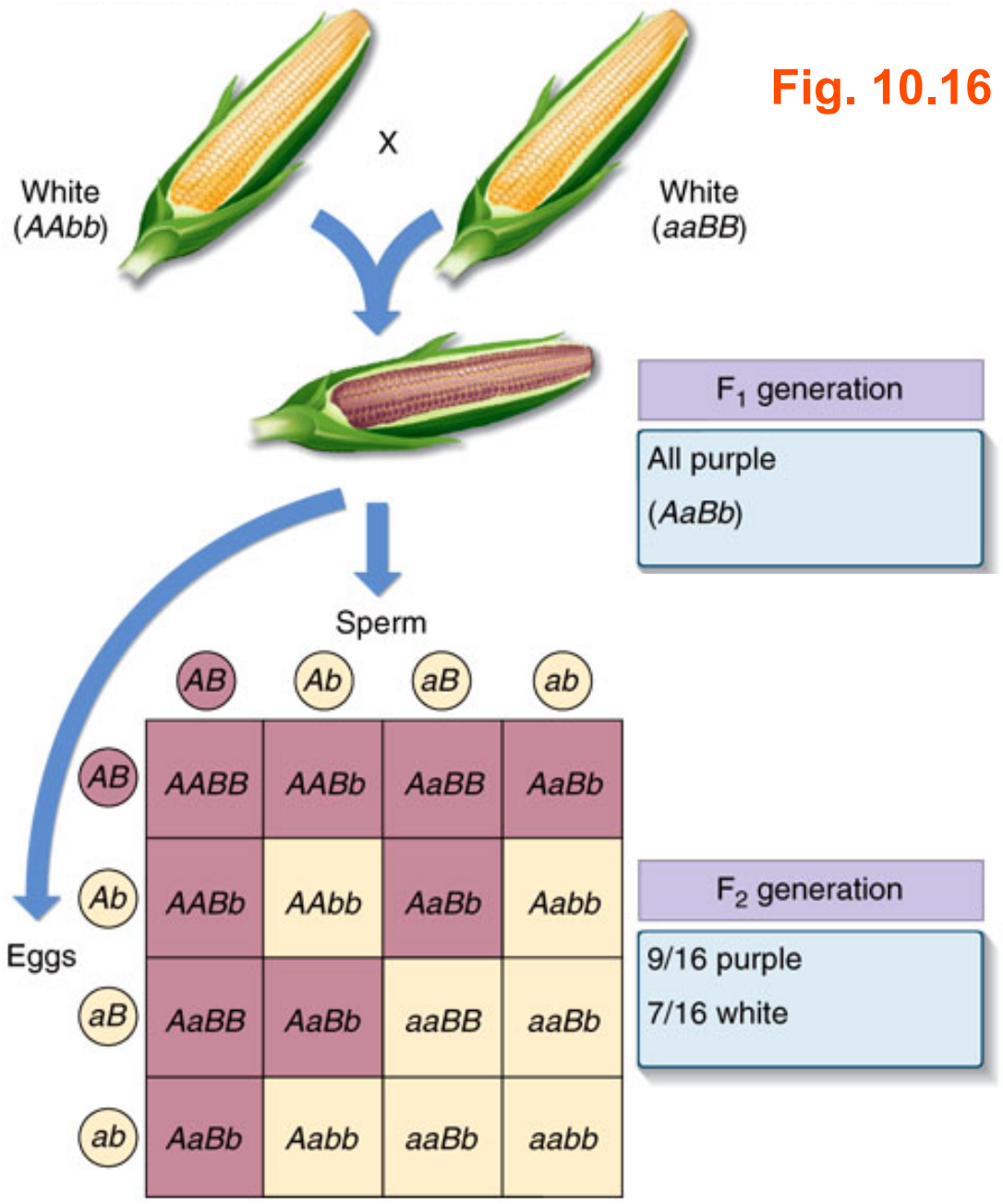
Epistasis

- Interaction between two genes where one of them modifies the phenotypic expression of the other

- In 19110, the geneticist R. A. Emerson crossed two true-breeding corn varieties with white kernels
  - To his surprise, all F_1 plants had purple kernels
  - The plants of the F_2 generation showed a ratio of 9 purple : 7 white

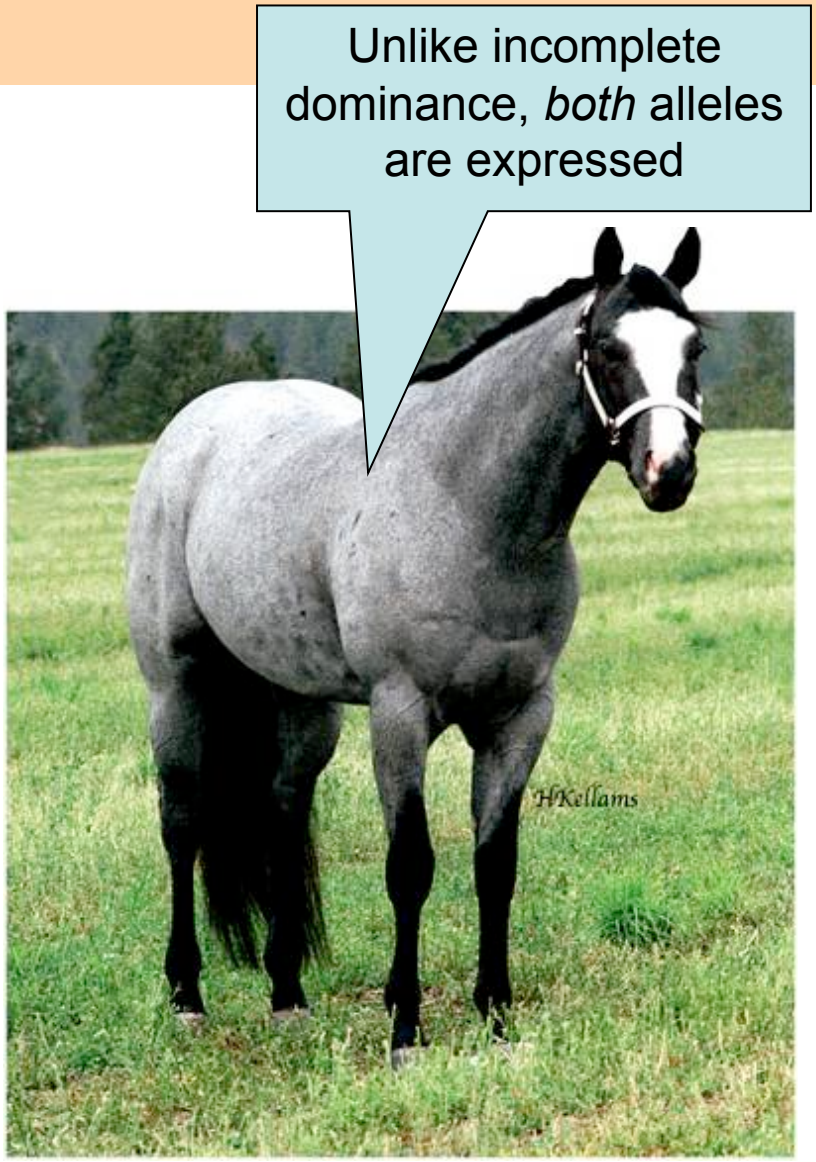
  - Mendelian genetics predicts a 9:3:3:1 ratio
  - So why is Emerson's ratio modified?

- There are two genes that contribute to kernel color
  - B → Production of pigment
  - A → Deposition of pigment
- Either gene can block the other's expression
  - To produce pigment a plant must possess at least one functional copy of each gene

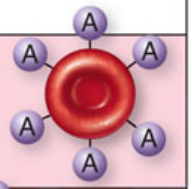
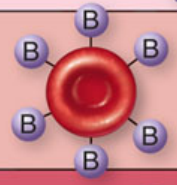
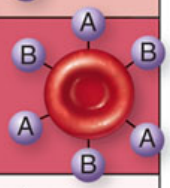


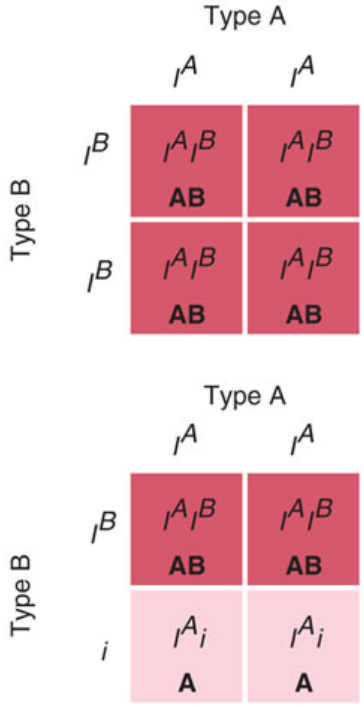
Codominance

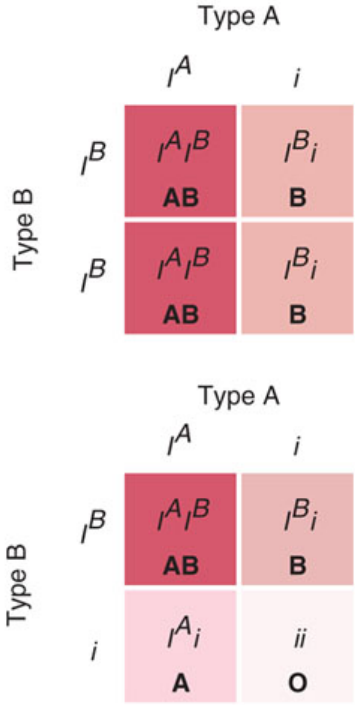
- Some pairs of alleles exhibit codominance
  - They produce a heterozygote phenotype that is a *combination* of that of the two homozygotes
  - Example
    - Roan color in horses



- The gene (termed *I*) that determines the ABO blood group in humans has more than one dominant allele
  - The encoded enzyme adds sugar molecules to lipids on the surface of red blood cells
    - I^A adds galactosamine
    - I^B adds galactose
    - *i* adds neither sugar
    - The I^A *and* I^B alleles are codominant
    - The *i* allele is recessive to both
    - The different combinations of the three alleles produces four different phenotypes

Genotypes	Phenotypes	
	Surface antigens	ABO blood type
$I^A I^A$ $I^A i$	Only A	Type A 
$I^B I^B$ $I^B i$	Only B	Type B 
$I^A I^B$	Both A and B	Type AB 
ii	None	Type O 





10.7 Chromosomes Are the Vehicles of Mendelian Inheritance

- The chromosomal theory of inheritance was first formulated by Walter Sutton in 1902
  - It basically states that Mendelian factors are found on chromosomes

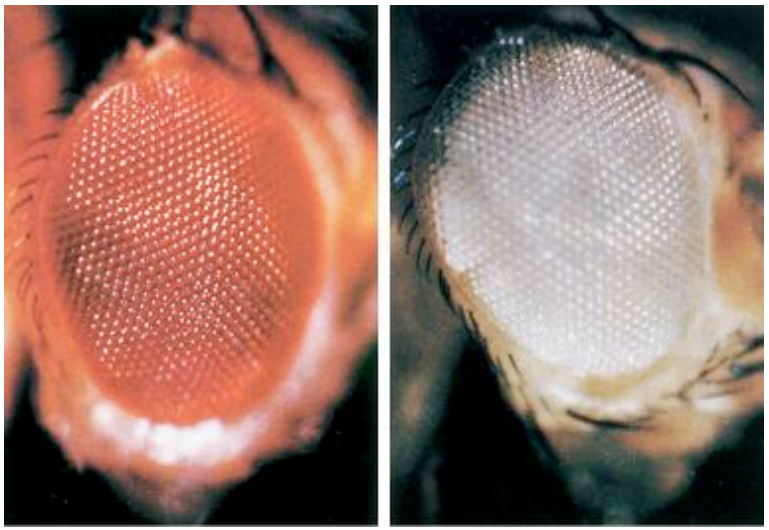
- Investigators soon pointed out a major problem with the theory
  - Number of independently assorting traits is far more than an organism's number of chromosome pairs

Review the Concepts

- In snapdragons, pink-flowered plants are produced when red-flowered plants are crossed with white-flowered plants. What type of inheritance can best be described as?
- What are called several traits that are affected by the same allele?
- In the human ABO blood grouping, there are four basic blood types, type A, type B, type AB, and type O. The blood proteins express themselves due to what trait?
- Foxes, cats, and rabbits have enzymes that are heat-sensitive. What causes the seasonal variation in coat color?

10.7 Chromosomes Are the Vehicles of Mendelian Inheritance

- Confirmation of Sutton's theory was provided by a single fruit fly, discovered by Thomas Hunt Morgan in 1910
  - The mutant was a white-eyed male
  - *Drosophila* wild-type flies are red-eyed



- Morgan immediately set out to determine whether this new trait is inherited in a Mendelian fashion

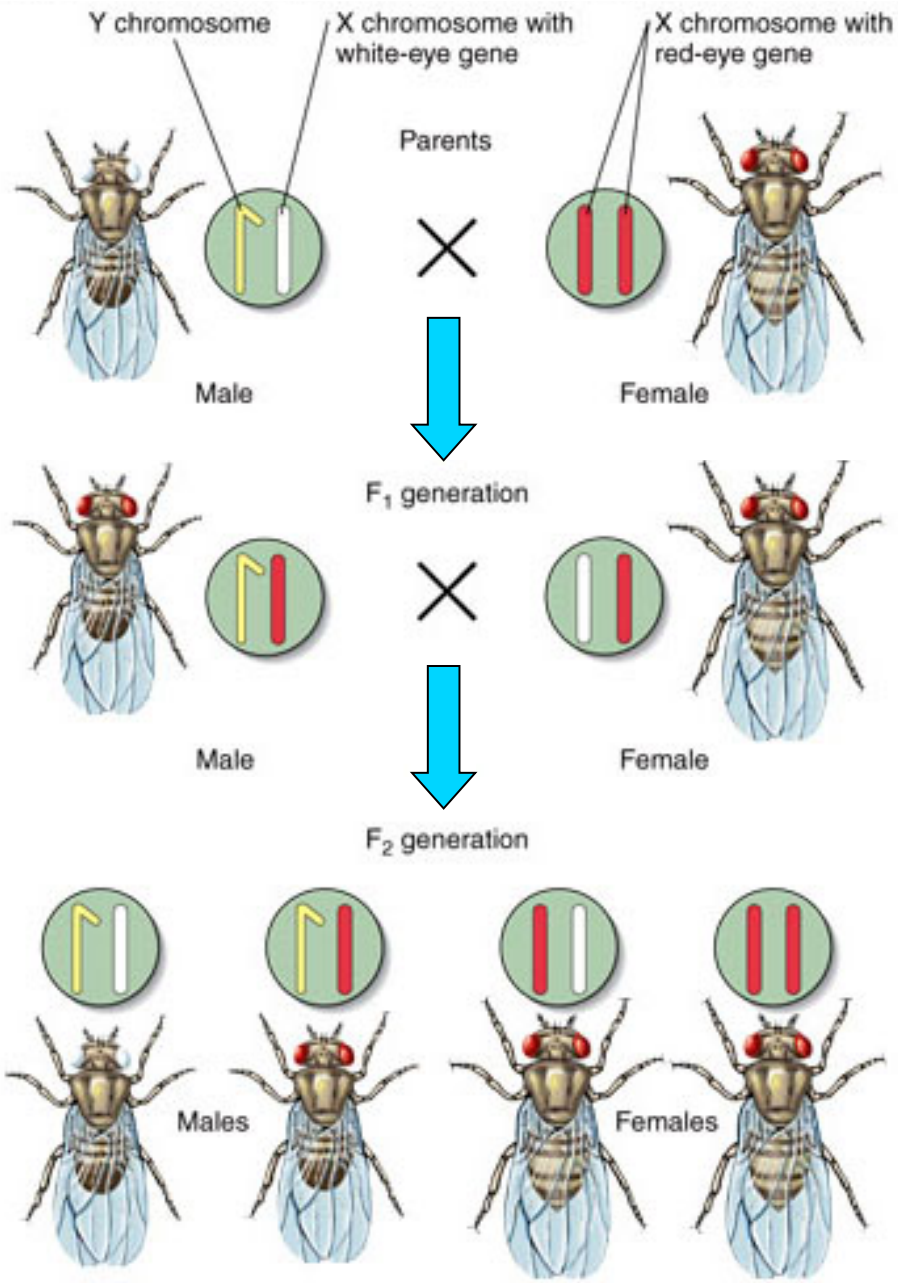
- P: white-eyed male X red-eyed female
  - All F_1 was red-eyed
    - *Thus, red eye color is dominant over white*

- The F_1 was then self-crossed
  - The F_2 was
    - ~ 1/2 red-eyed female
    - ~ 1/4 red-eyed male
    - ~ 1/4 white-eyed male
  - *Thus, the eye color segregates*
    - *But why is white-eyes only found in males?*

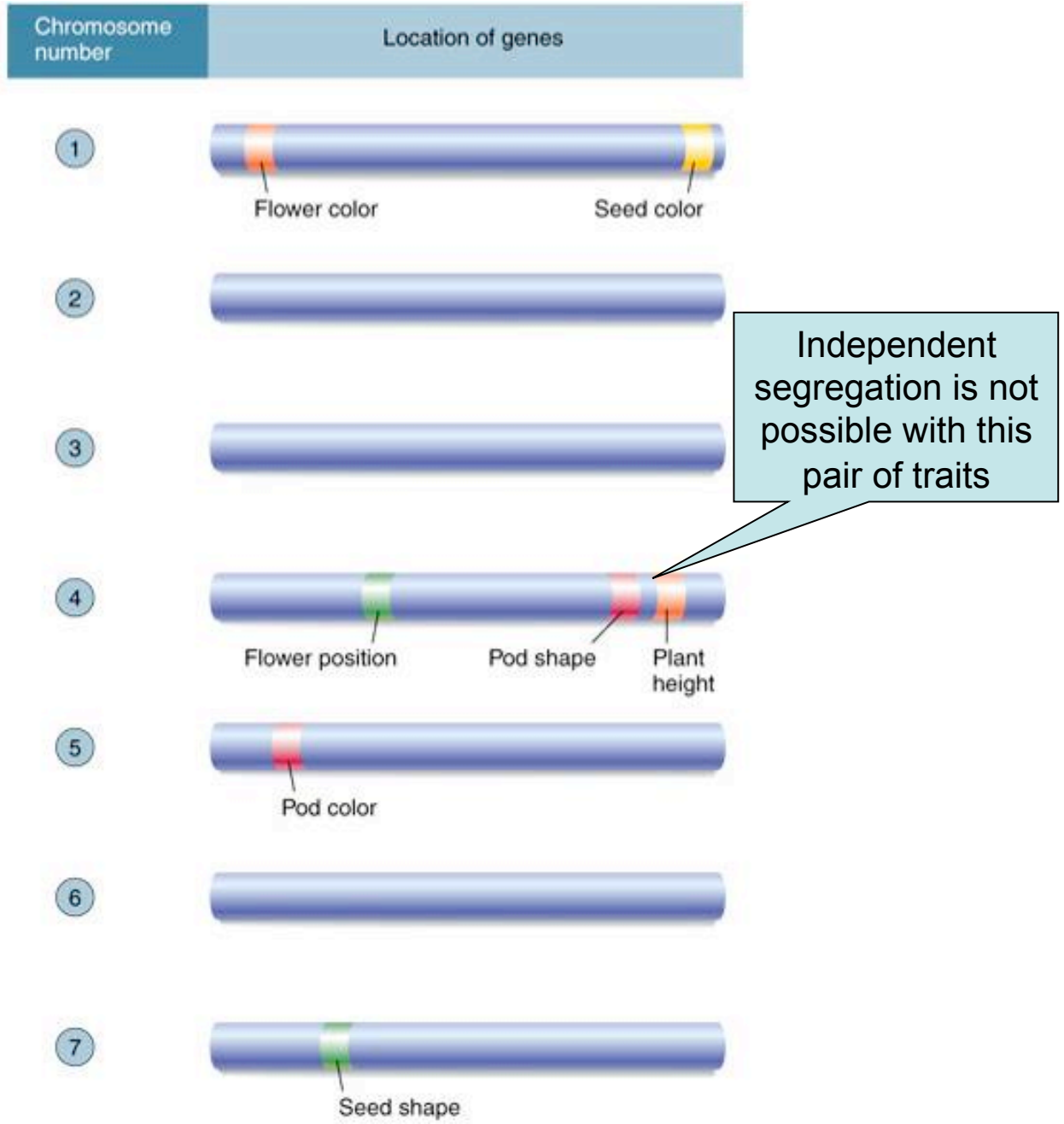
- In *Drosophila*, the sex of an individual is determined by the number of X chromosomes
  - Female flies have two X chromosomes
  - Male flies have only one

- The solution to Morgan's puzzle is that the gene for white eye color resides only on the X chromosome
  - A trait determined by genes on the sex chromosomes is said to be sex-linked

Morgan's experiment



The chromosomal location of Mendel's seven characters

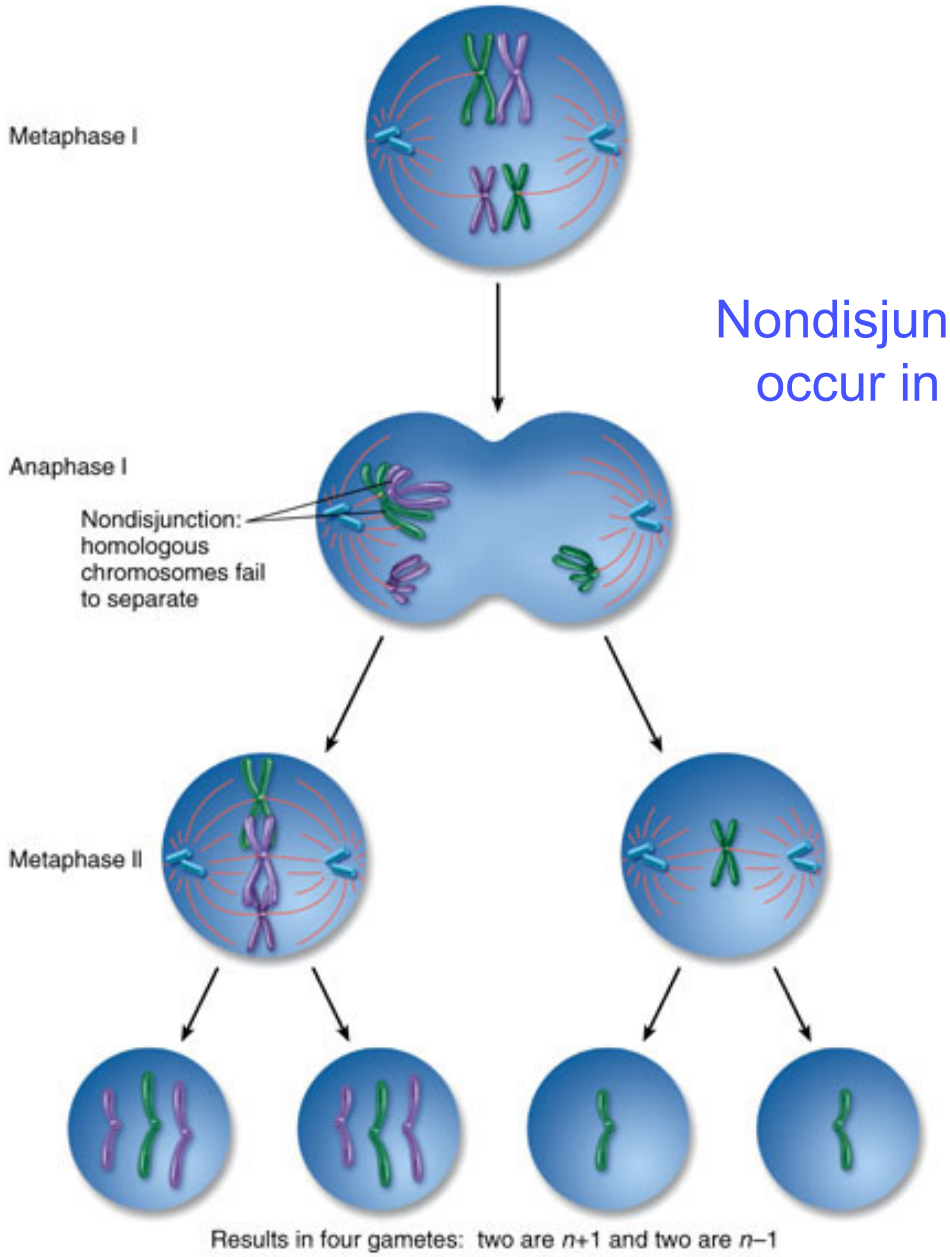


10.10 Human Chromosomes

- Human somatic cells have 23 pairs of chromosomes
  - 22 pairs of autosomes
  - 1 pair of sex chromosomes
    - XX in females
    - XY in males

- Failure of chromosomes to separate correctly in meiosis I or II is termed non-disjunction
  - This leads to an abnormal number of chromosomes, or aneuploidy

Nondisjunction in anaphase I



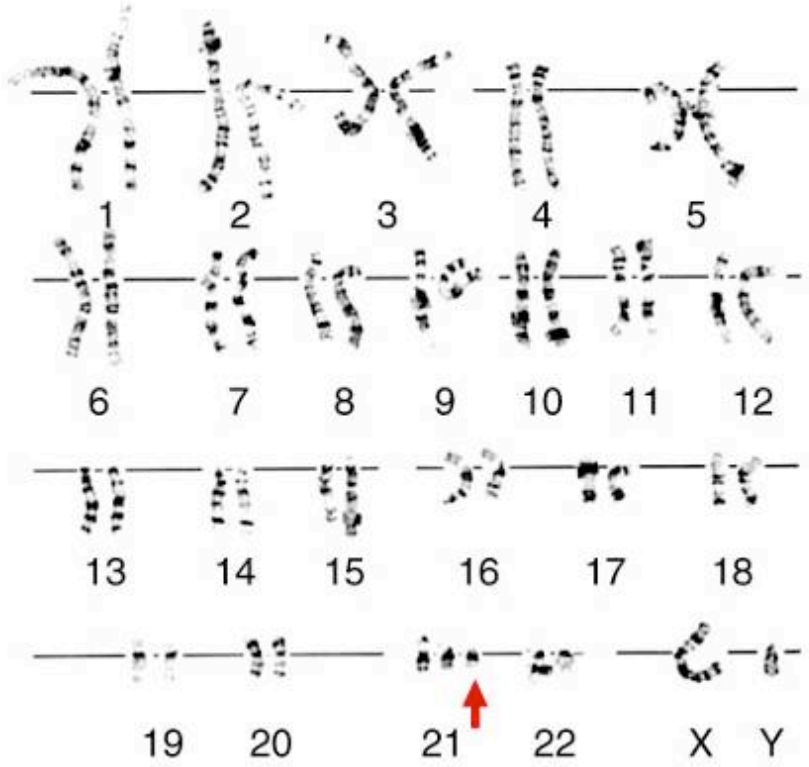
Nondisjunction can also occur in anaphase II

10.10 Human Chromosomes

- Humans with one less autosome are called monosomics
  - These do not survive development
- Humans with one extra autosome are called trisomics
  - The vast majority do not survive
  - Trisomy for only a few chromosomes is compatible with survival
    - However, there are severe developmental defects

Down Syndrome

- Caused by trisomy 21



(a)

(b)

Down Syndrome

- Frequency is about 1 in 750 children
  - However, it is much more common among children of older women

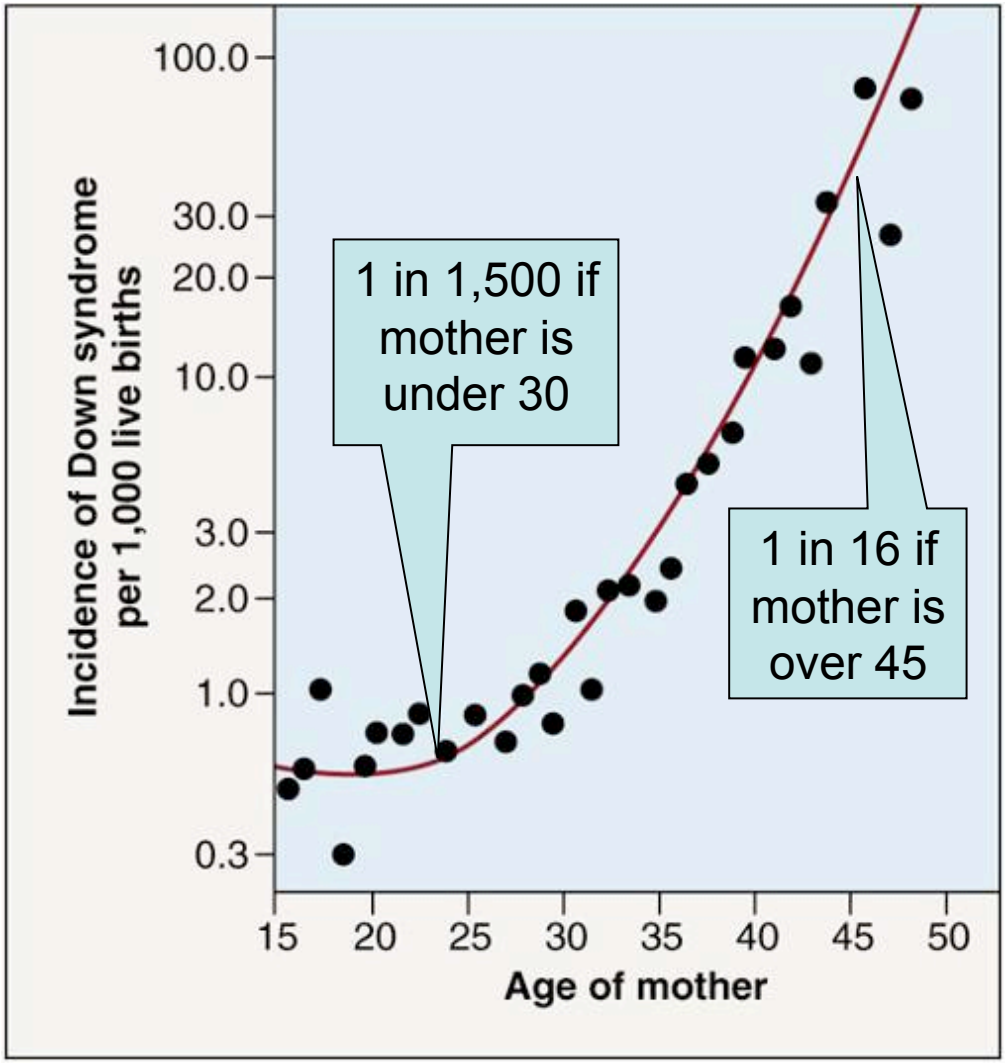


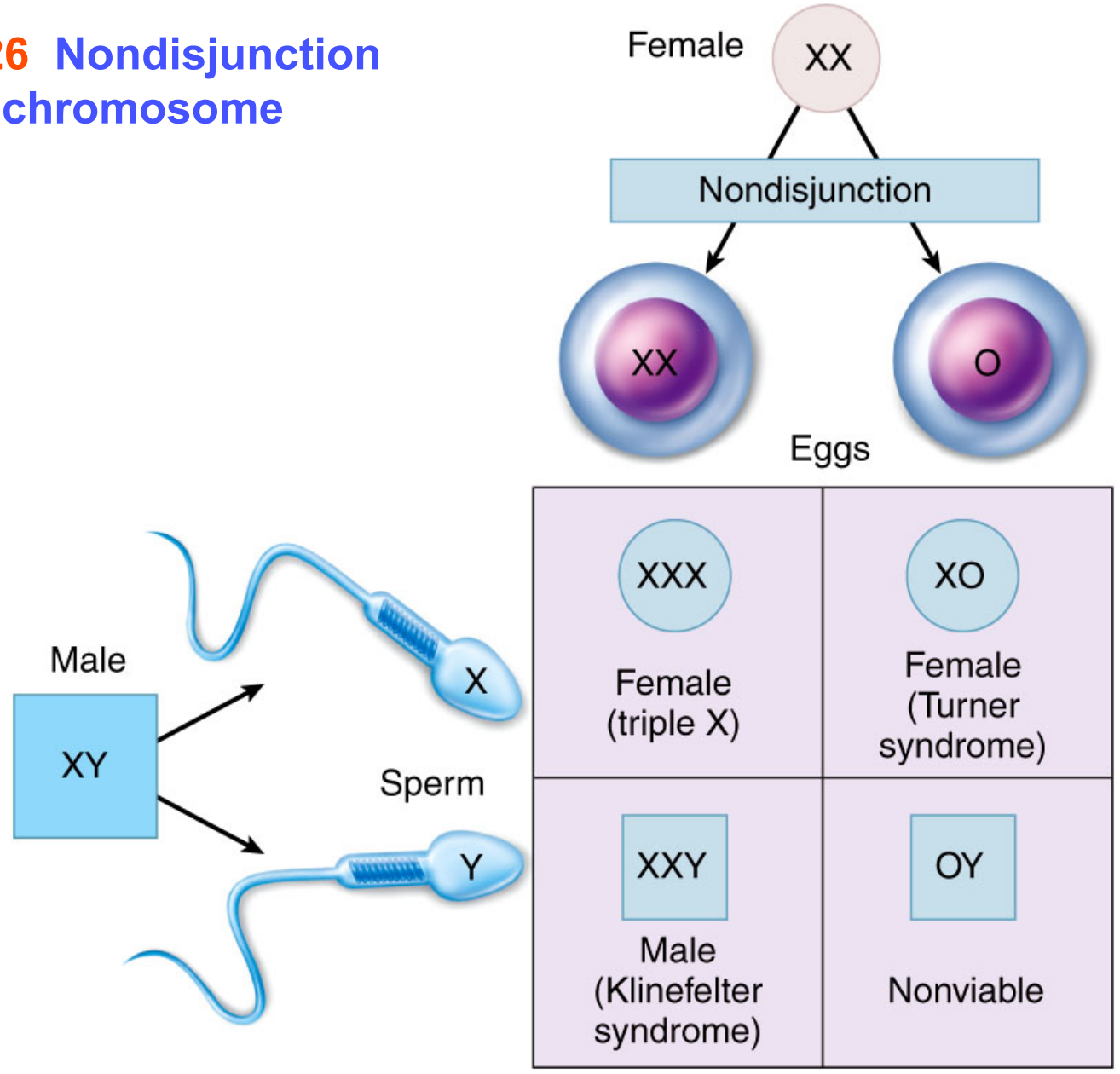
Fig. 10.25

Nondisjunction Involving Sex Chromosomes

- Aneuploidies of sex chromosomes have less serious consequences than those of autosomes
  - However, they can lead to sterility

- Nondisjunction of the Y chromosome
  - Yields YY gametes and ultimately XYY zygotes
  - Frequency of XYY is 1 in 1,000 males
    - In general, these are phenotypically normal

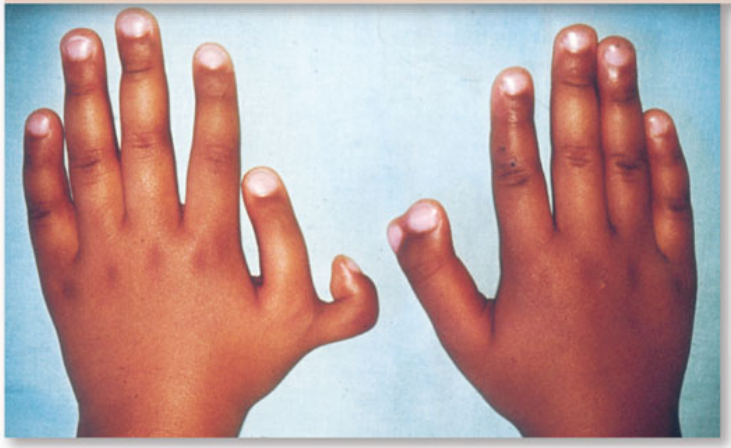
Fig. 10.26 Nondisjunction of the X chromosome

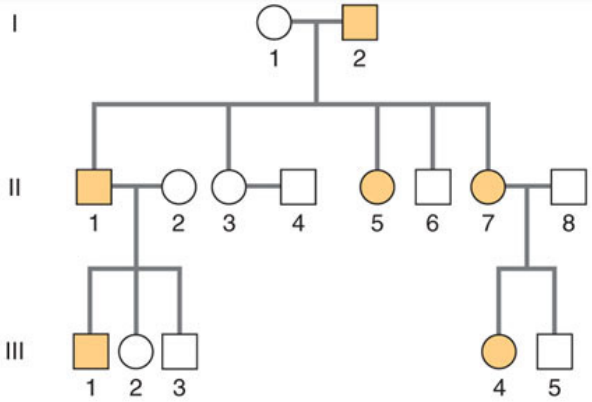


10.9 The Role of Mutations in Human Heredity

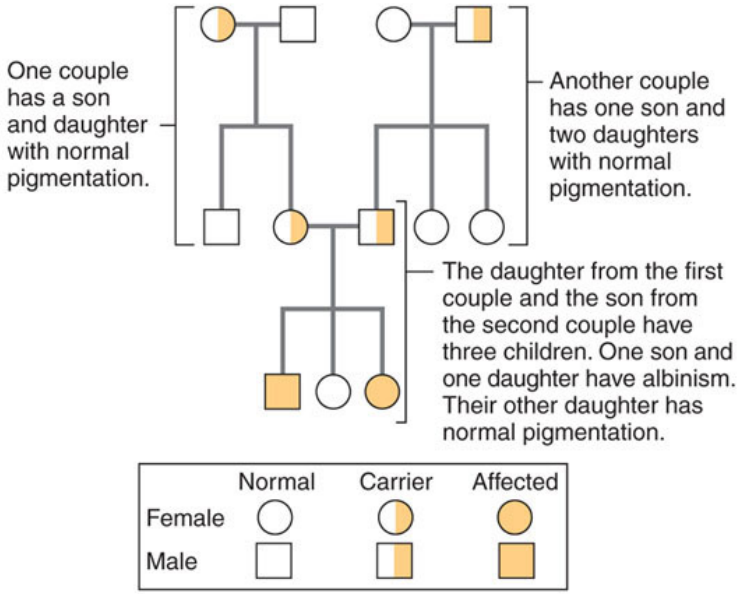
- Mutations are accidental changes in genes
  - They are rare, random and tend to produce recessive alleles
- Mutations cause genetic disorders
- The inheritance of these disorders, as well as harmless traits, is studied by looking at pedigrees
  - Family trees that identify individuals with the disease/trait

a. Polydactyly





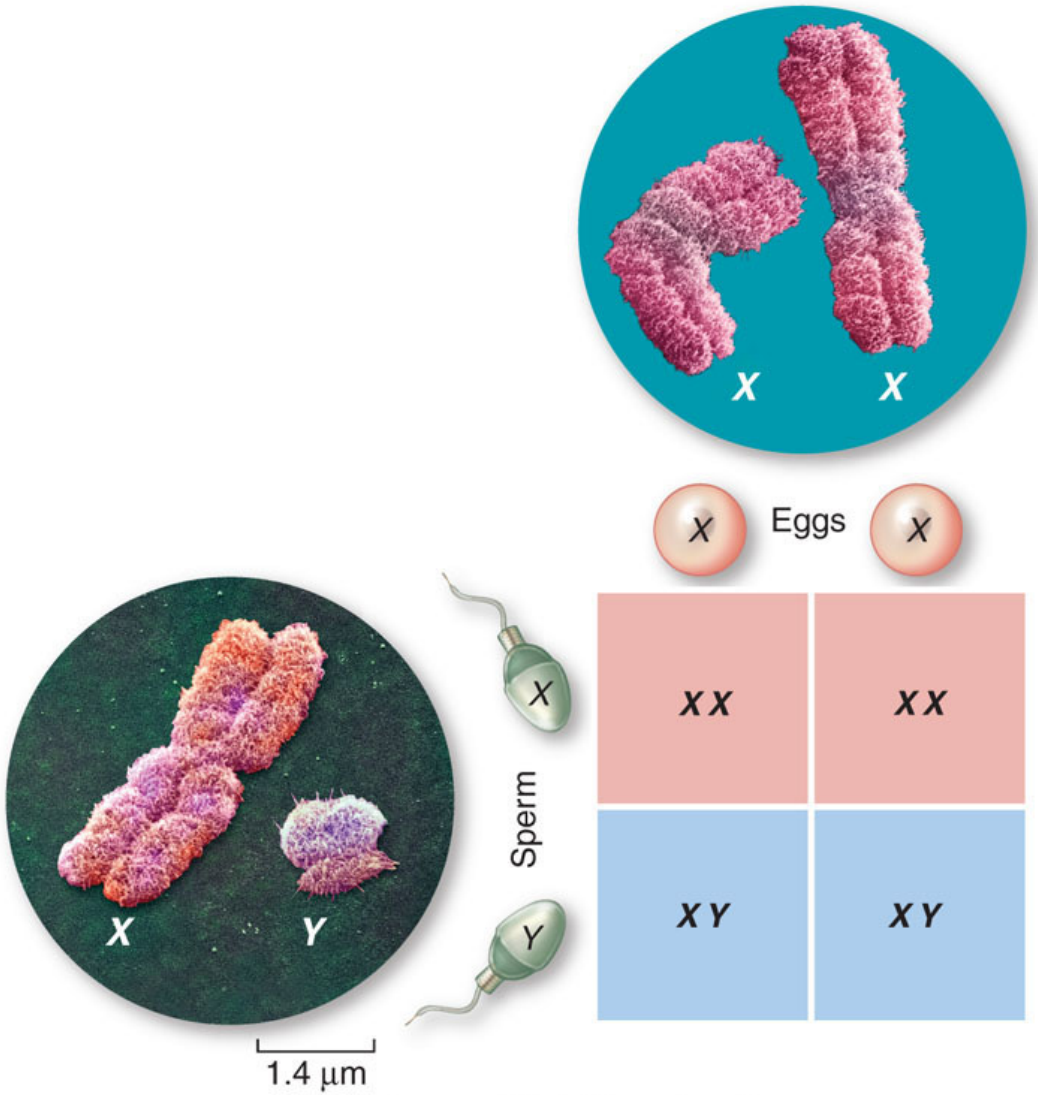
b. Albinism



a: © Lester Bergman; b: © Joe McDonald/Visuals Unlimited

Hemophilia: A Sex-Linked Trait

- Hemophilia is an inherited condition in which the blood clots slowly or not at all
- Two genes that encode blood-clotting proteins reside on the X chromosome
- Hemophilia is an X-linked recessive disorder
  - Males develop hemophilia if they inherit one mutant allele from their mother
  - For females to develop hemophilia, they have to inherit two mutant alleles, one from each parent



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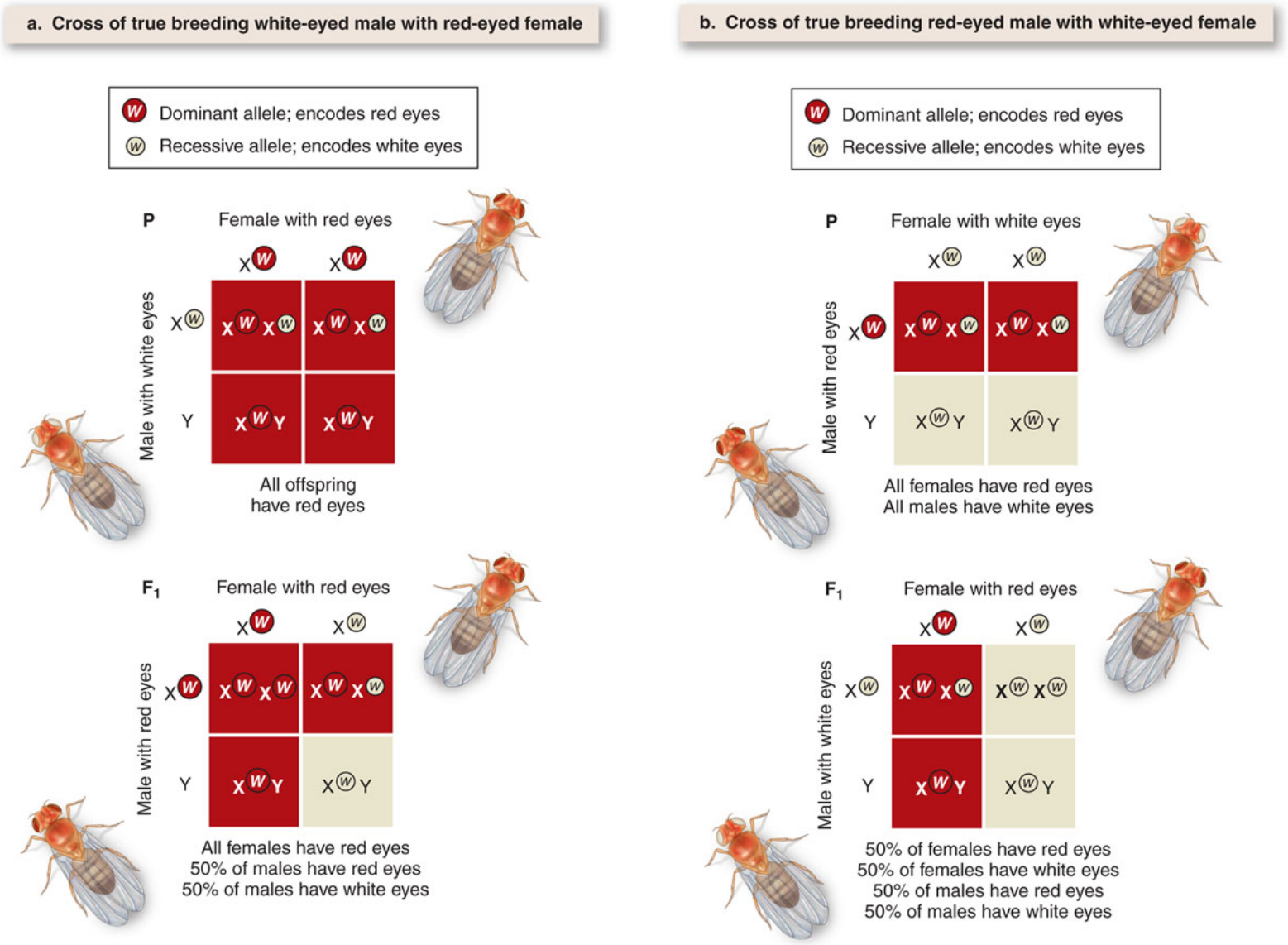
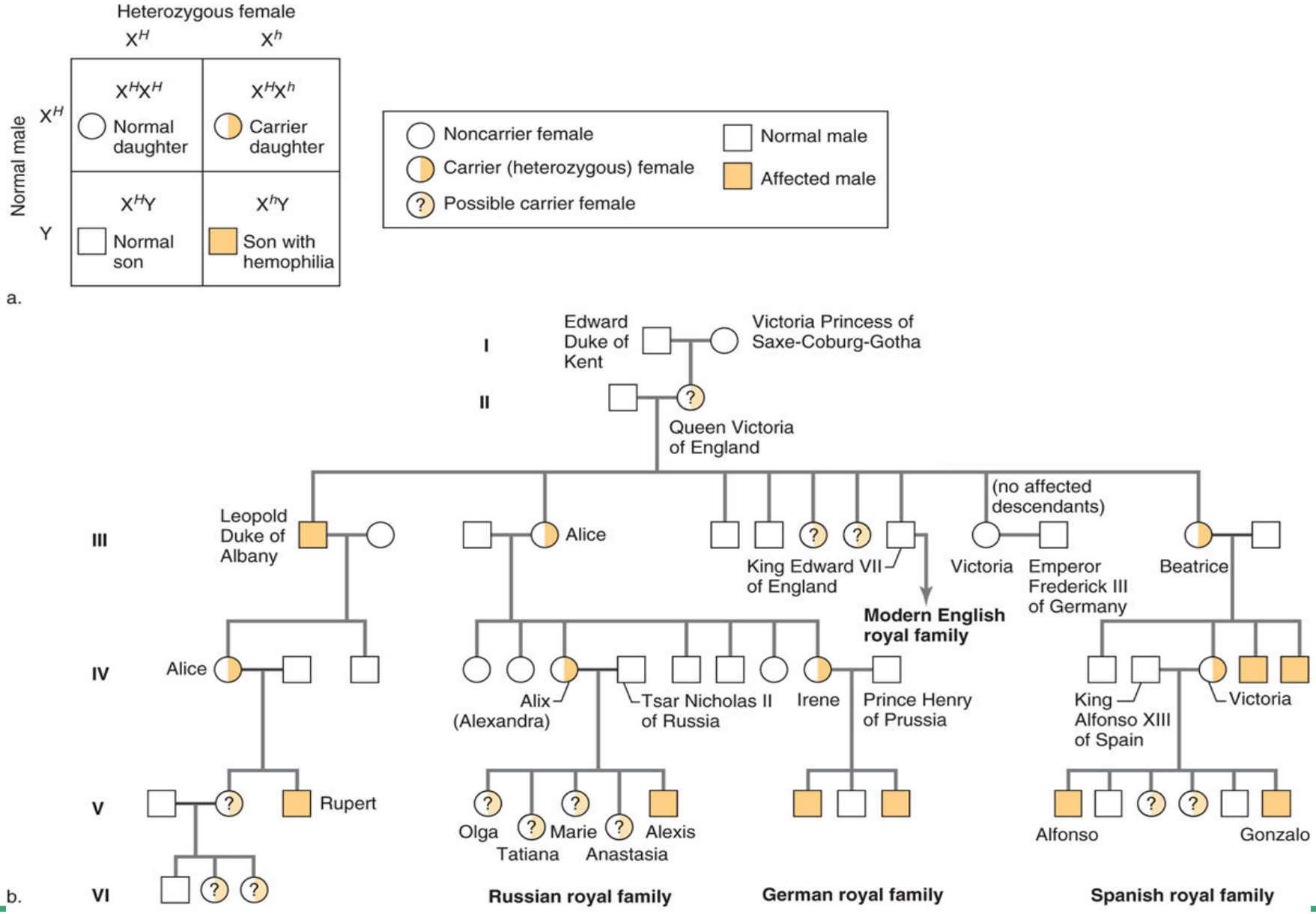


- Royal hemophilia
  - Started by a mutant allele in Queen Victoria of England
  - Three of her nine children received the defective allele
    - They transferred it by marriage to other royal families

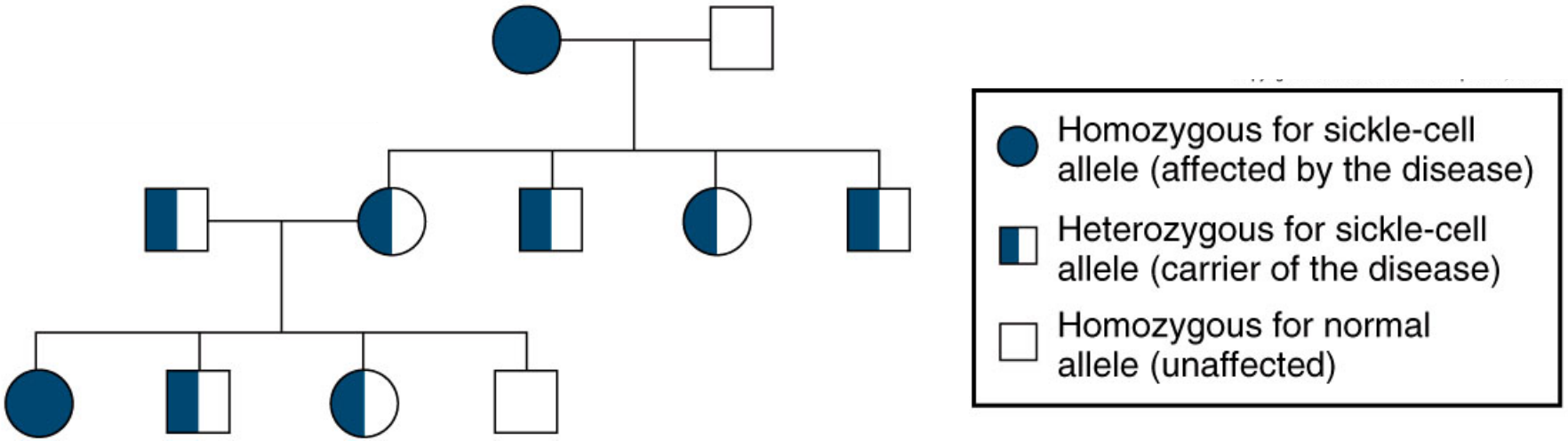
Fig. 10.210

- In all, 10 of Victoria's male descendants had hemophilia

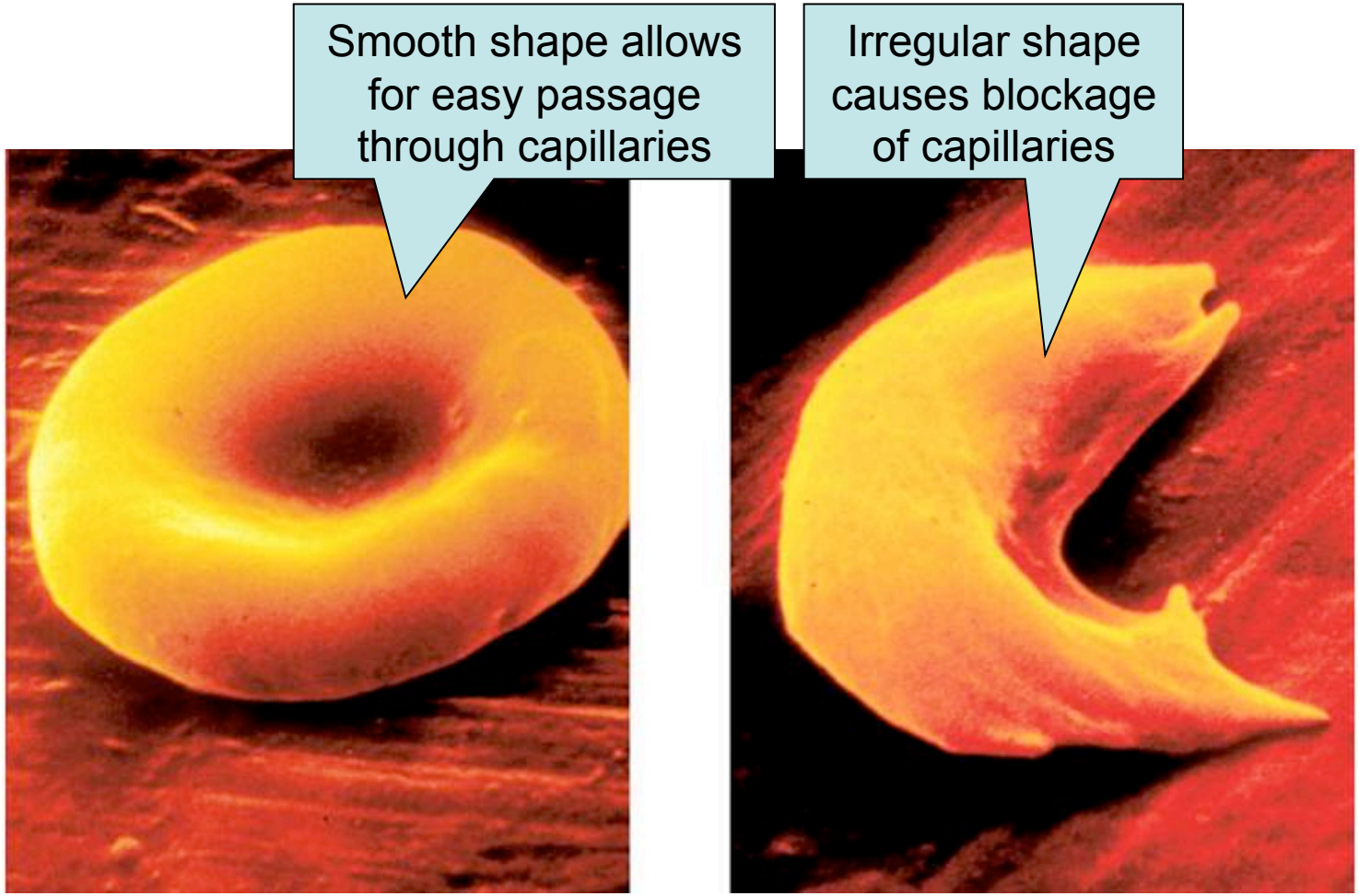


Sickle-Cell Anemia: Recessive Trait

- Sickle-cell anemia is an autosomal recessive trait in which the protein hemoglobin is defective
  - Affected individuals cannot properly transport oxygen to their tissues



Sickle-Cell Anemia: Recessive Trait

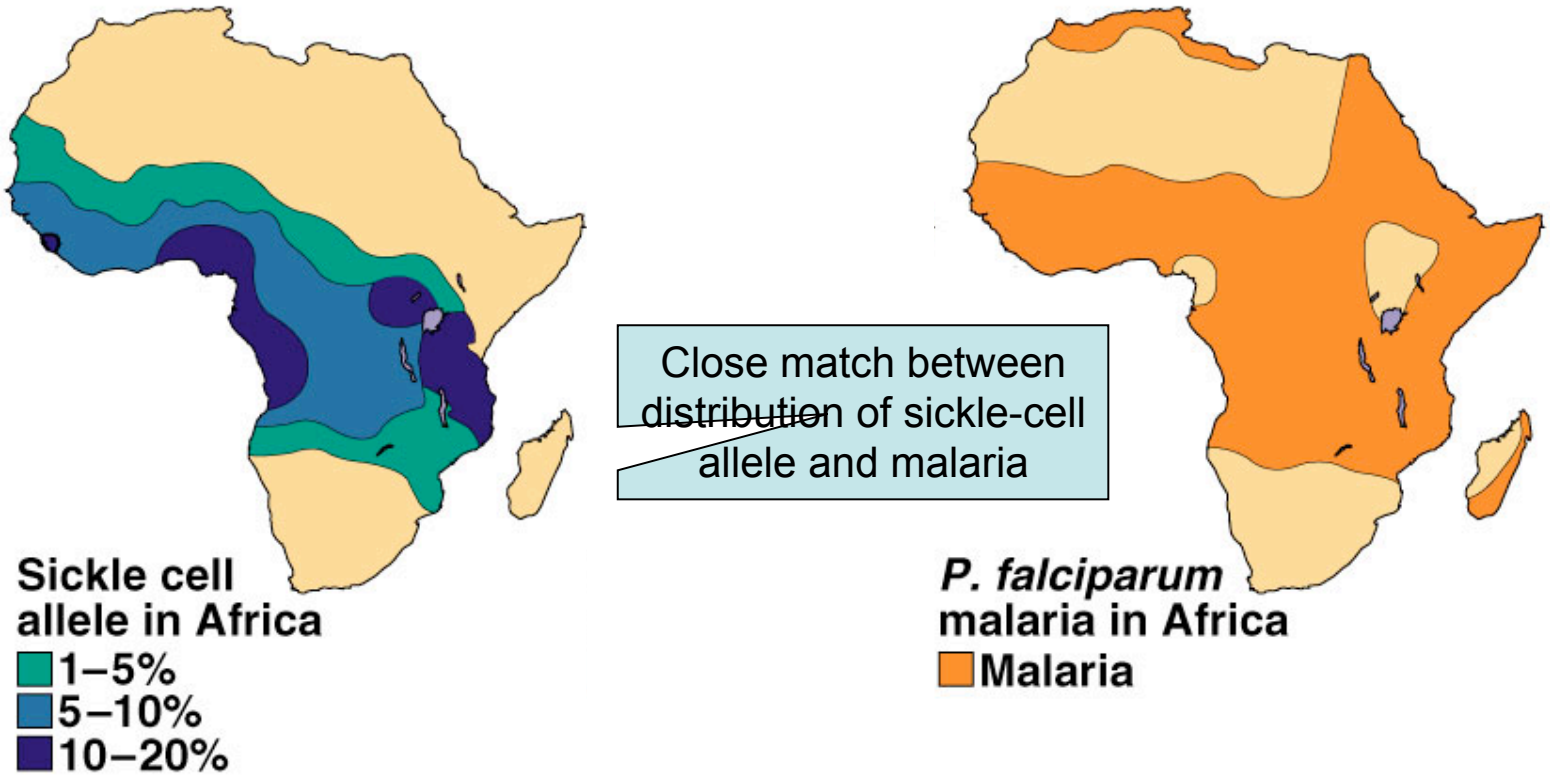


Normal red blood cell

Sickled red blood cell

Sickle-Cell Anemia: Recessive Trait

- The sickle-cell allele is particularly common among people of African descent
  - It increases resistance against malaria

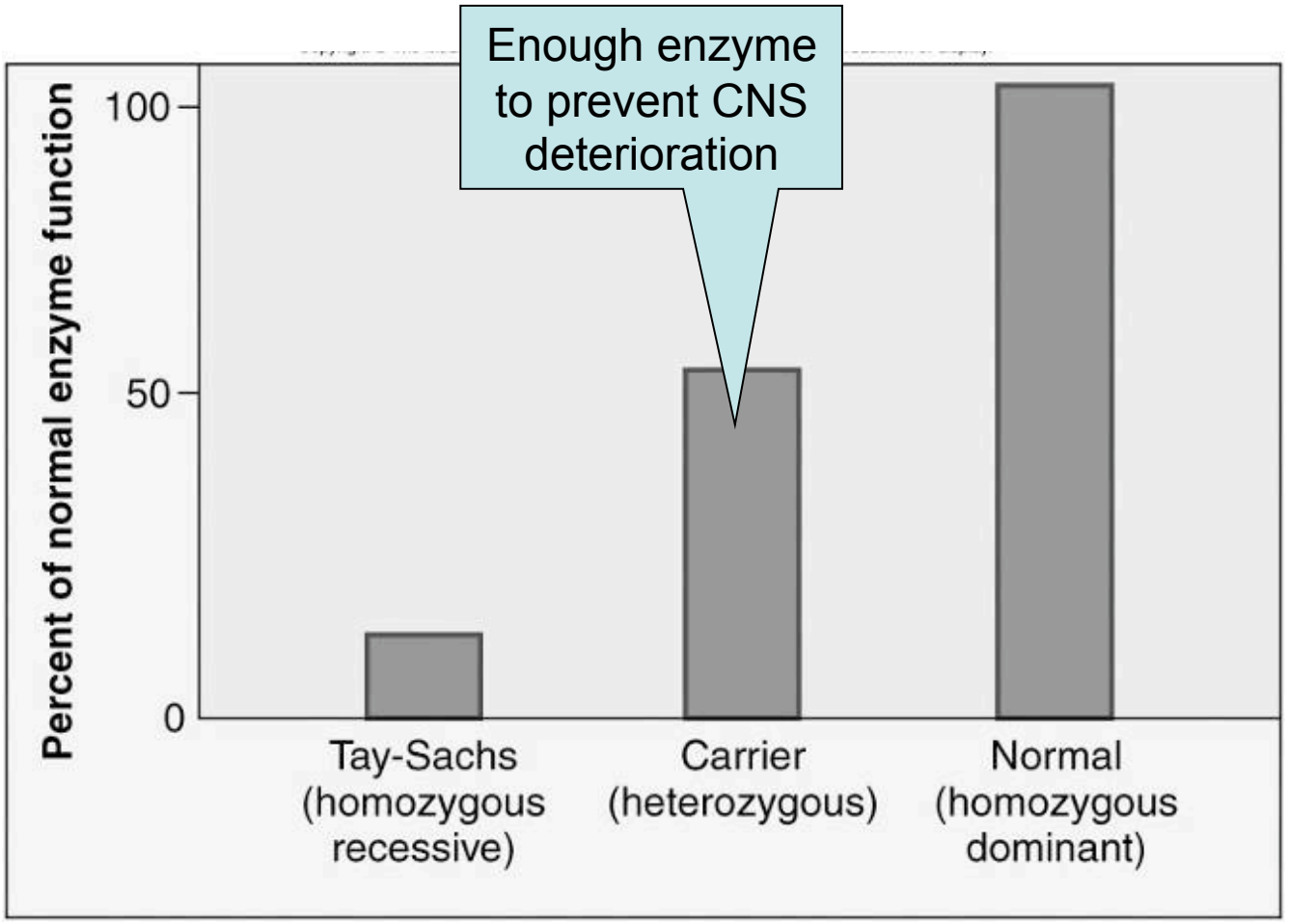


Tay-Sachs Disease: Recessive Trait

- Tay-Sachs disease is an autosomal recessive trait in which the enzyme hexosaminidase A is defective
  - Affected individuals cannot break down specific lipids
    - These lipids accumulate in brain cells
      - Children die by five years of age

- The disease is very rare in human populations
  - However, it has high incidence in Ashkenazi Jews

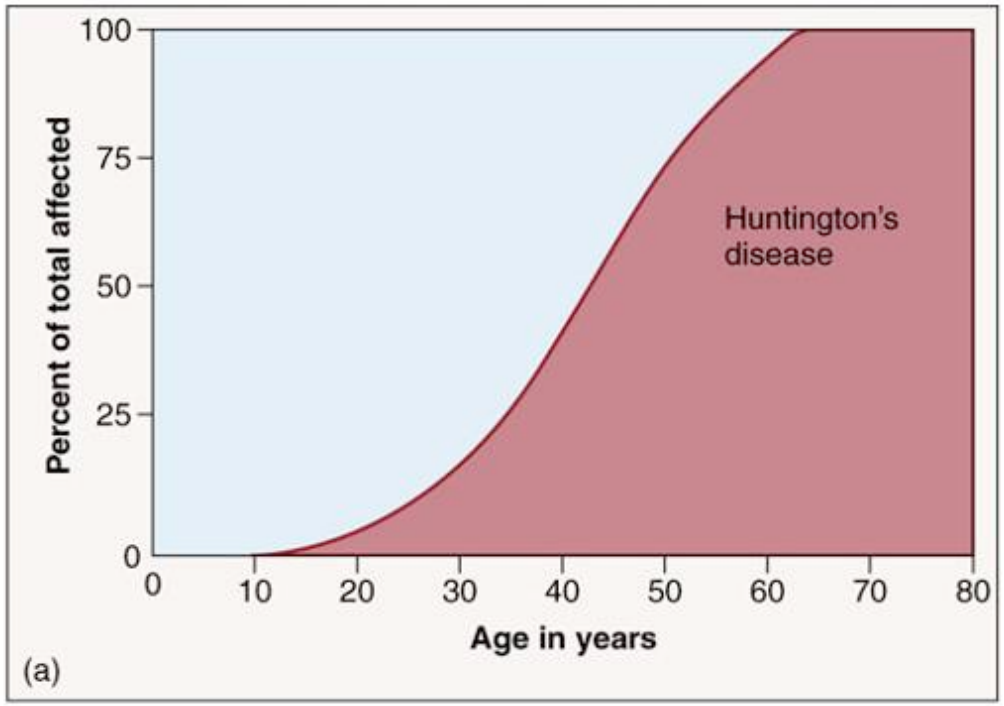
Tay-Sachs Disease: Recessive Trait



Huntington's Disease: Dominant Trait

- Huntington's disease is an autosomal dominant trait that causes progressive deterioration of brain cells

- It is a fatal disease
  - However, it persists in human populations because it has a late onset



Review the Concepts

- Down syndrome in humans is due to the alteration of which chromosome?
- Humans who have lost one copy of a chromosome are called _____ and generally do not survive development.
- Hemophilia is a genetic trait link to what chromosome?

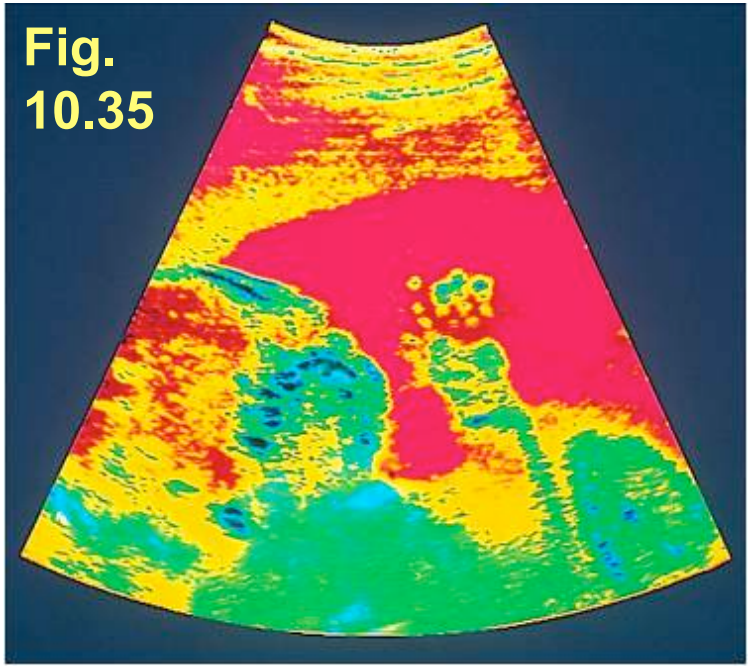
10.10 Genetic Counseling and Therapy

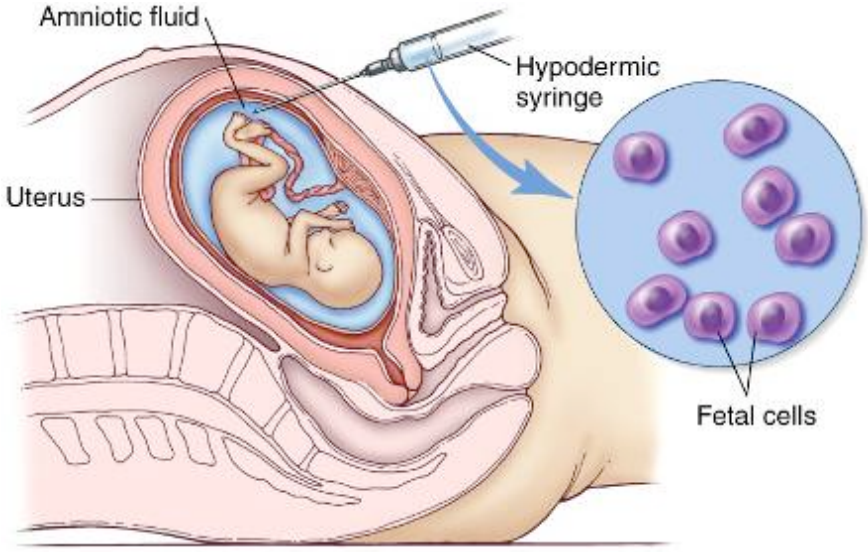
- Genetic counseling identifies parents at risk of producing children with genetic defects and assesses the state of early embryos
  - It also offers advise on medical treatments and options

- High-risk pregnancies
  - Parents with recessive alleles
  - Mothers older than 35

Genetic Screening

- Amniocentesis
  - Usually performed in the fourth month of pregnancy





- Ultrasound
  - Used to locate the fetus during amniocentesis
  - Used to examine the fetus for signs of major abnormalities

Genetic Screening

- Chorionic villus sampling
  - Cells are removed from a region of the placenta termed the chorion
  - This procedure offer advantages over amniocentesis
    - Used earlier in pregnancy
    - Yields results faster
  - However, it increases the risk of miscarriage
- The fetal cells obtained by these procedures are then grown in culture

Genetic Screening

- Genetic counselors examine fetal cell cultures for three main things
  - 1. Chromosomal karyotype
    - Aneuploidies or gross chromosomal alterations
  - 2. Enzyme activity
    - Lack of enzyme activity signals a disorder
  - 3. Genetic markers
    - The genes for certain genetic disorders are associated with other nearby mutations

DNA Screening

- Versions of the same gene in different individuals may differ in only one DNA nucleotide
  - These differences are termed single nucleotide polymorphisms (SNP)

- Screening of SNPs and comparing them to known SNP databases may detect certain genetic disorders
  - However, SNP profiling raises important ethical issues