Chapter 11: Observing Patterns in Inherited Traits

Earlobe Variation

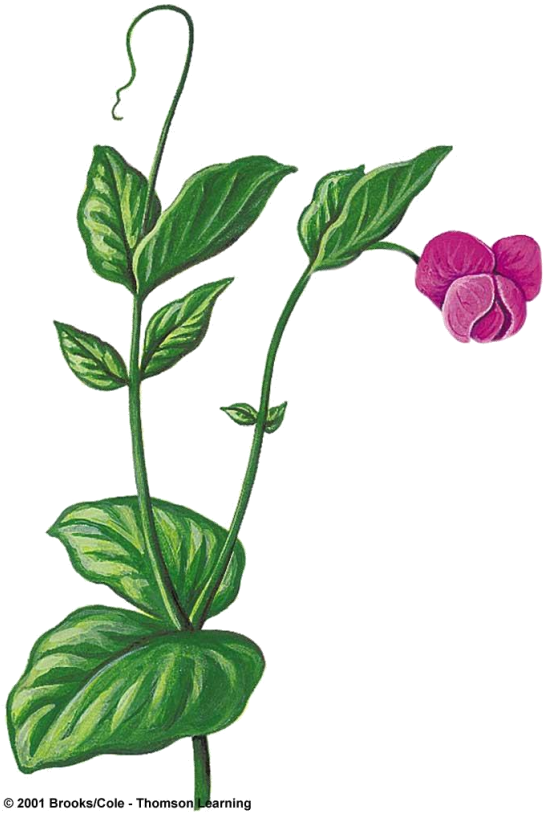
- Whether a person has attached or detached earlobes depends on a single gene
- Attached earlobes: two copies of the recessive allele for this gene
- Detached earlobes: either one or two copies of the dominant allele

Early Ideas about Heredity

- People knew that sperm and eggs transmitted information about traits
- Blending theory
- Problem:
  - Would expect variation to disappear
  - Variation in traits persists

Gregor Mendel

- Strong background in plant breeding and mathematics
- Using pea plants, found indirect but observable evidence of how parents transmit genes to offspring



Gregor Mendel

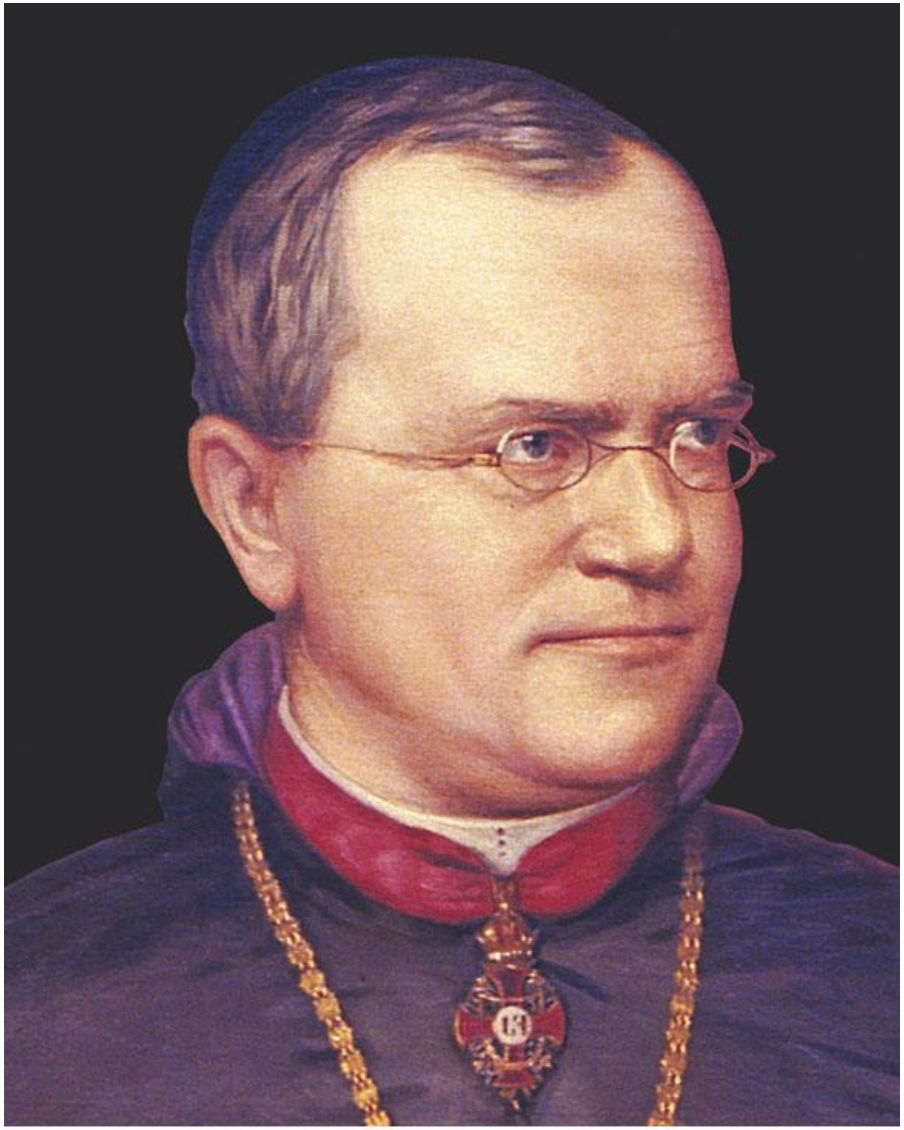


Fig. 11-2, p.170

Genes

- Units of information about specific traits
- Passed from parents to offspring
- Each has a specific location (locus) on a chromosome

Alleles

- Different molecular forms of a gene
- Arise by mutation
- Dominant allele masks a recessive allele that is paired with it

Allele Combinations

- Homozygous
  - having two identical alleles at a locus
  - *AA* or *aa*
- Heterozygous
  - having two different alleles at a locus
  - *Aa*

Genetic Terms

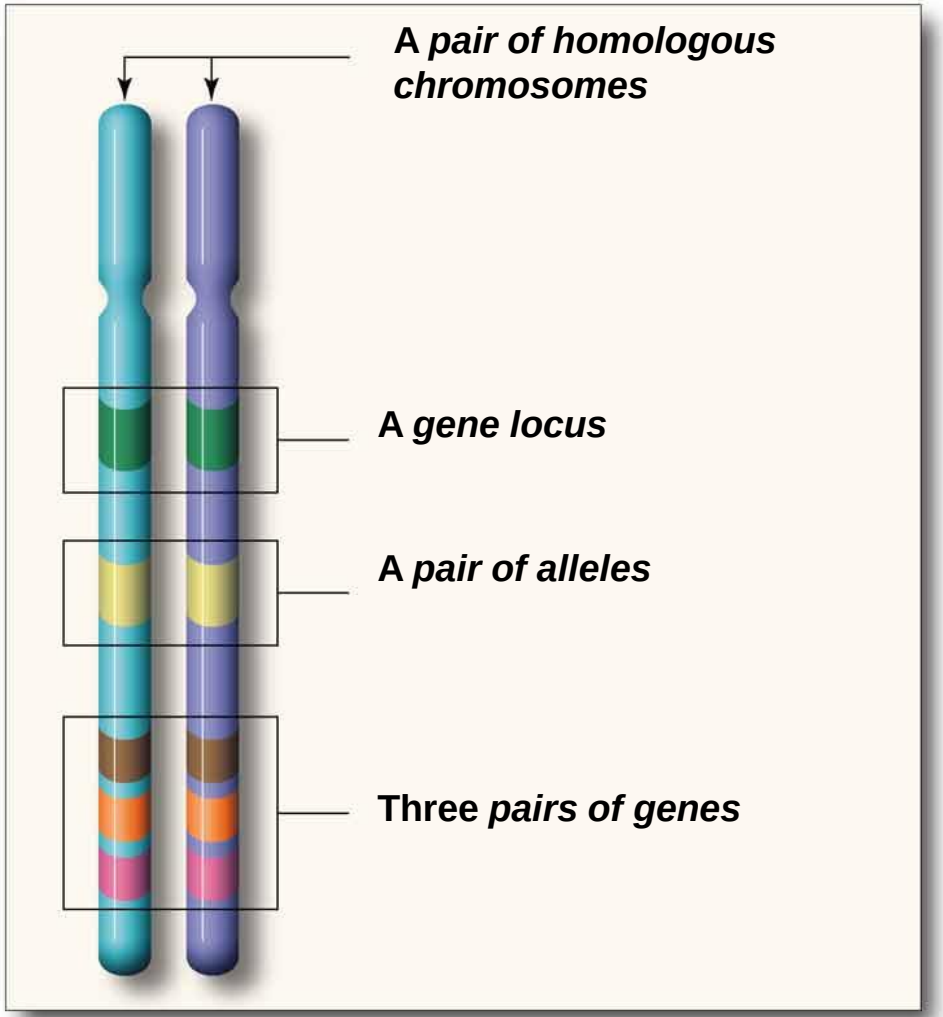


Figure 11.4

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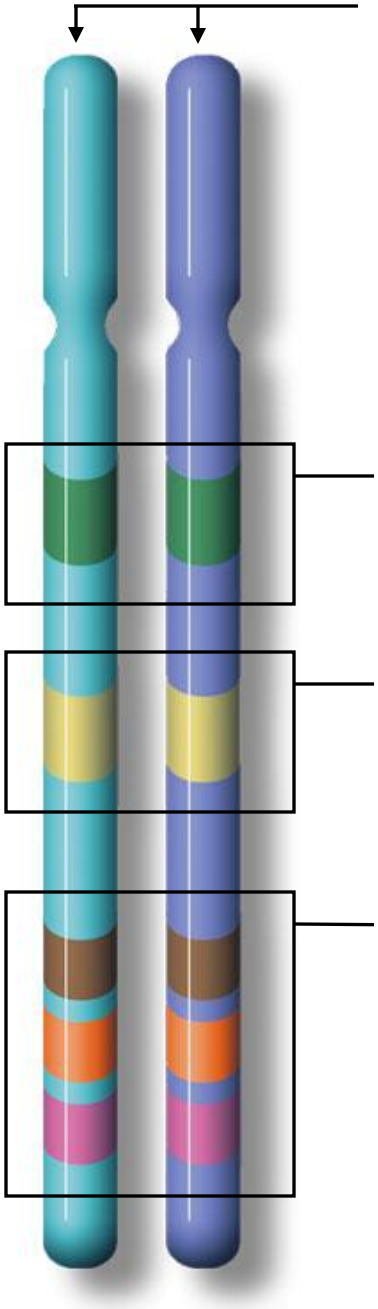

A ***pair of homologous chromosomes***, each in the unduplicated state (most often, one from a male parent and its partner from a female parent)

A ***gene locus*** (plural, loci), the location for a specific gene on a chromosome. Alleles are at corresponding loci on a pair of homologous chromosomes

A pair of alleles may be identical or nonidentical. They are represented in the text by letters such as D or d

Three ***pairs of genes*** (at three loci on this pair of homologous chromosomes); same thing as three pairs of alleles

Fig. 11-4, p.171

Genotype & Phenotype

- Genotype refers to particular genes an individual carries
- Phenotype refers to an individual's observable traits
- Cannot always determine genotype by observing phenotype

Tracking Generations

- Parental generation P

  mates to produce

- First-generation offspring F_1

  mate to produce

- Second-generation offspring F_2

Monohybrid Crosses

Experimental intercross between two F_1 heterozygotes

AA X *aa* ⟶ *Aa* (F_1 monohybrids)

Aa X *Aa* ⟶ ?

Mendel's Monohybrid Cross Results

F_2 plants showed dominant-to-recessive ratio that averaged 3:1

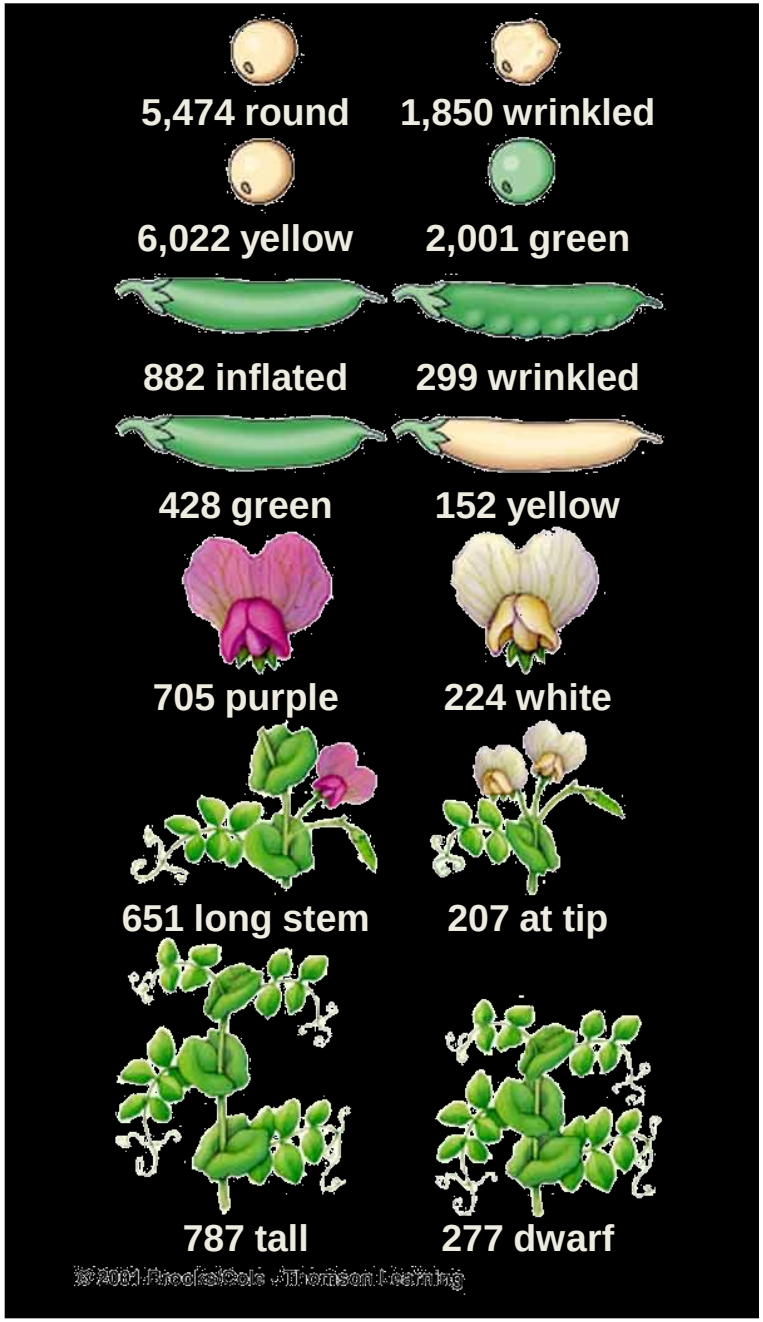

Fig. 11-6, p. 172

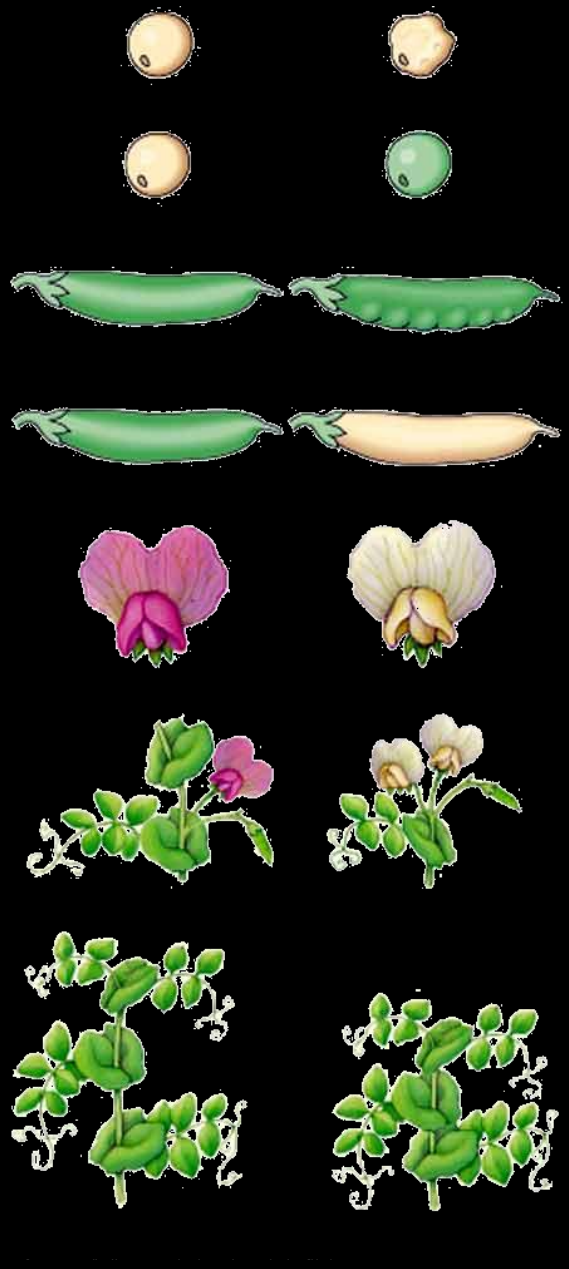

Fig. 11-6, p.172

Probability

The chance that each outcome of a given event will occur is proportional to the number of ways that event can be reached

Monohybrid Cross Illustrated

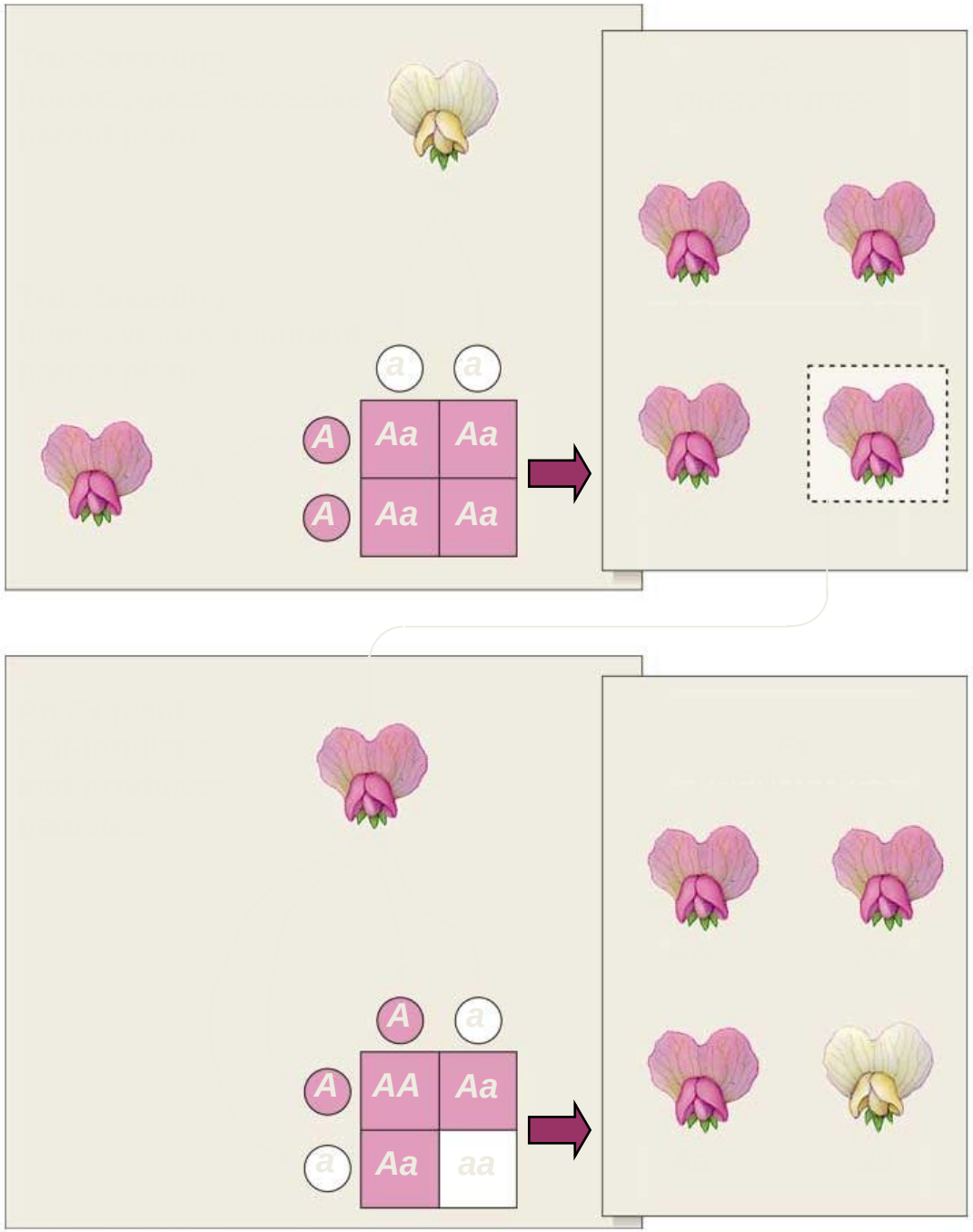

Figure 11.7

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Mendel's Theory of Segregation

- An individual inherits a unit of information (allele) about a trait from each parent

- During gamete formation, the alleles segregate from each other

Mendel's Theory of Segregation

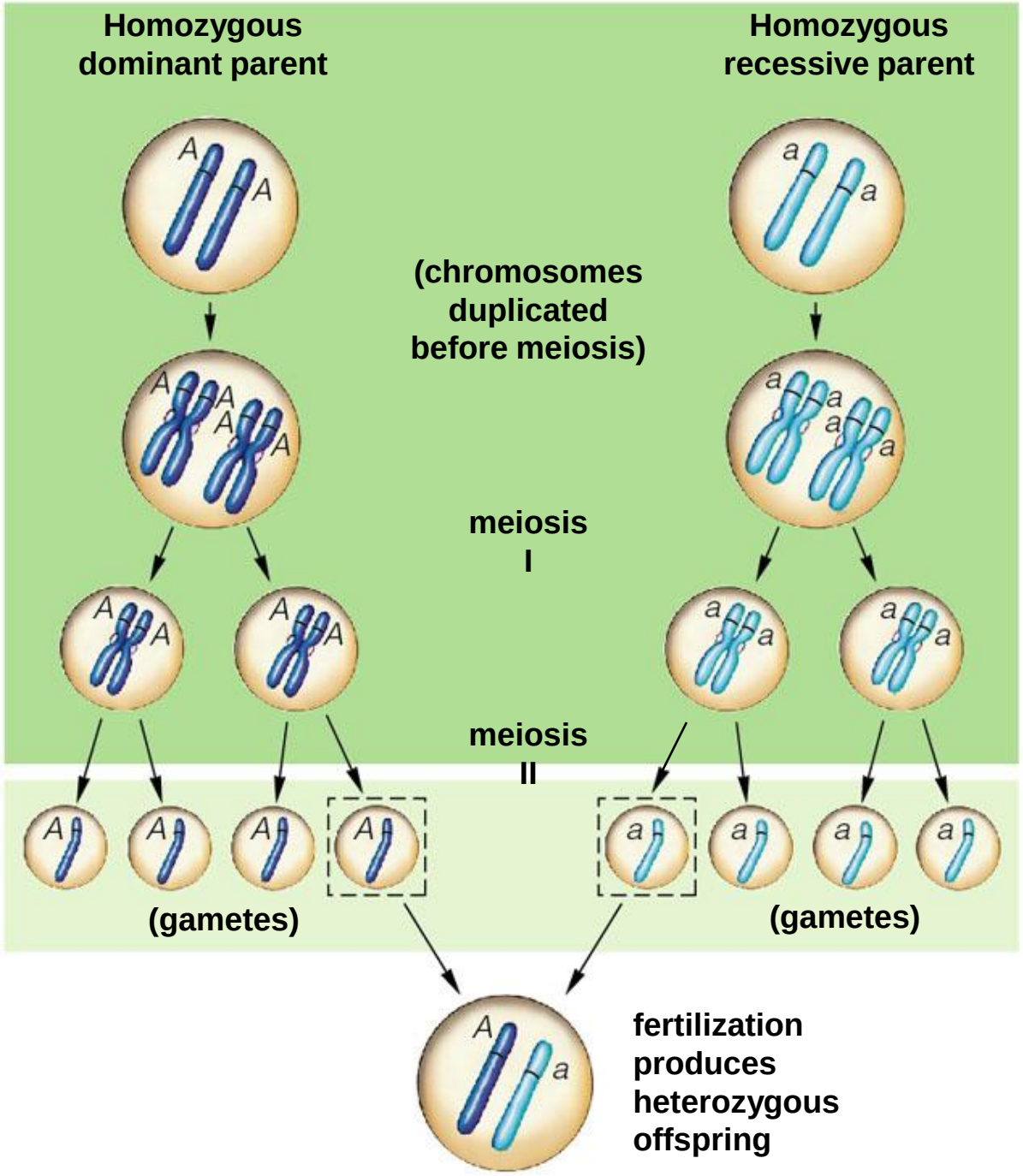


Fig. 11-5, p.172

Test Cross

- Individual that shows dominant phenotype is crossed with individual with recessive phenotype

- Examining offspring allows you to determine the genotype of the dominant individual

Punnett Squares of Test Crosses

Homozygous recessive

	a	*a*
A	*Aa*	*Aa*
a	*aa*	*aa*

Two phenotypes

Homozygous recessive

	a	*a*
A	*Aa*	*Aa*
A	*Aa*	*Aa*

All dominant phenotype

Punnett Squares of Test Crosses

POSSIBLE EVENT:	**PROBABLE OUTCOME:**
sperm *A* meets egg *A*	1/4 *AA* offspring
sperm *A* meets egg *a*	1/4 *Aa*
sperm *a* meets egg *A*	1/4 *Aa*
sperm *a* meets egg *a*	1/4 *aa*

Punnett Squares of Test Crosses

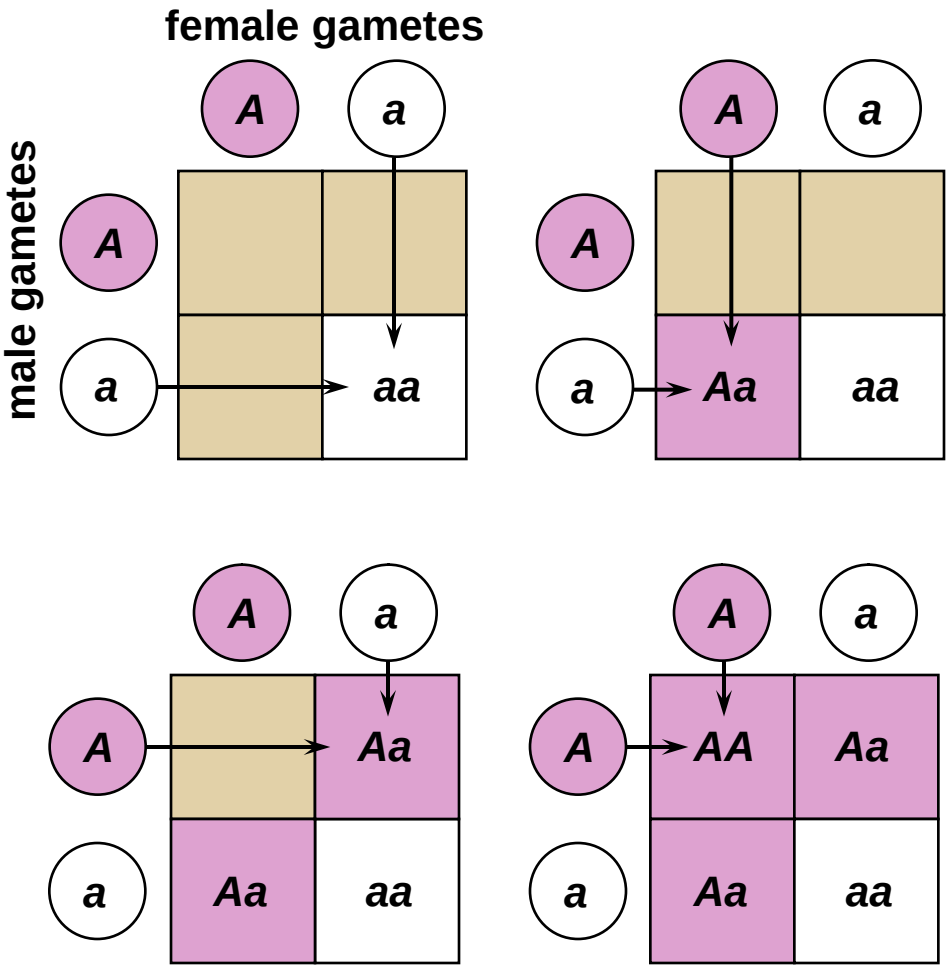
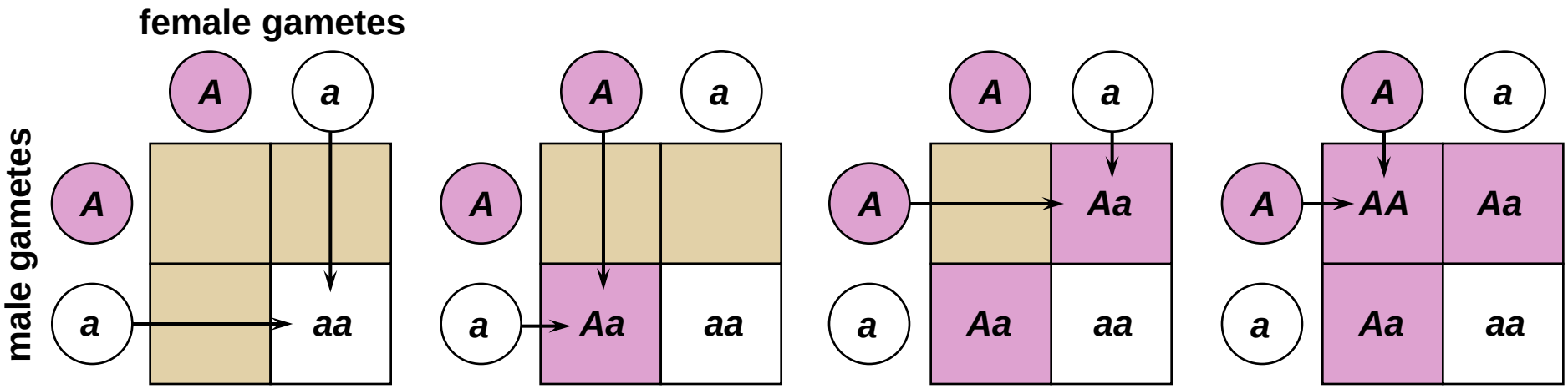


Fig. 11-7a, p.173

Punnett Squares of Test Crosses



Stepped Art

Fig. 11-7a, p.173

Punnett Squares of Test Crosses

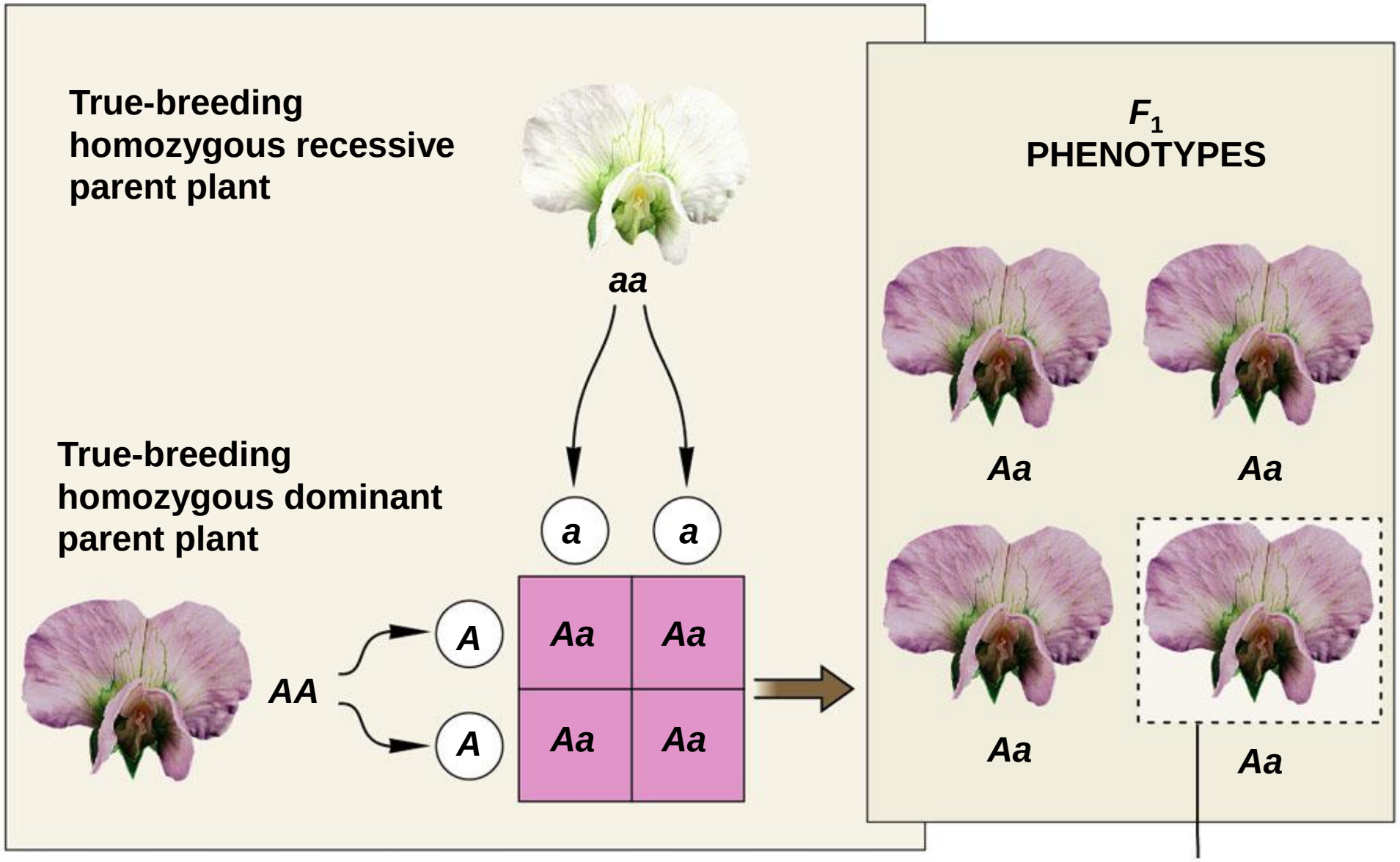


Fig. 11-7b1, p.173

Punnett Squares of Test Crosses

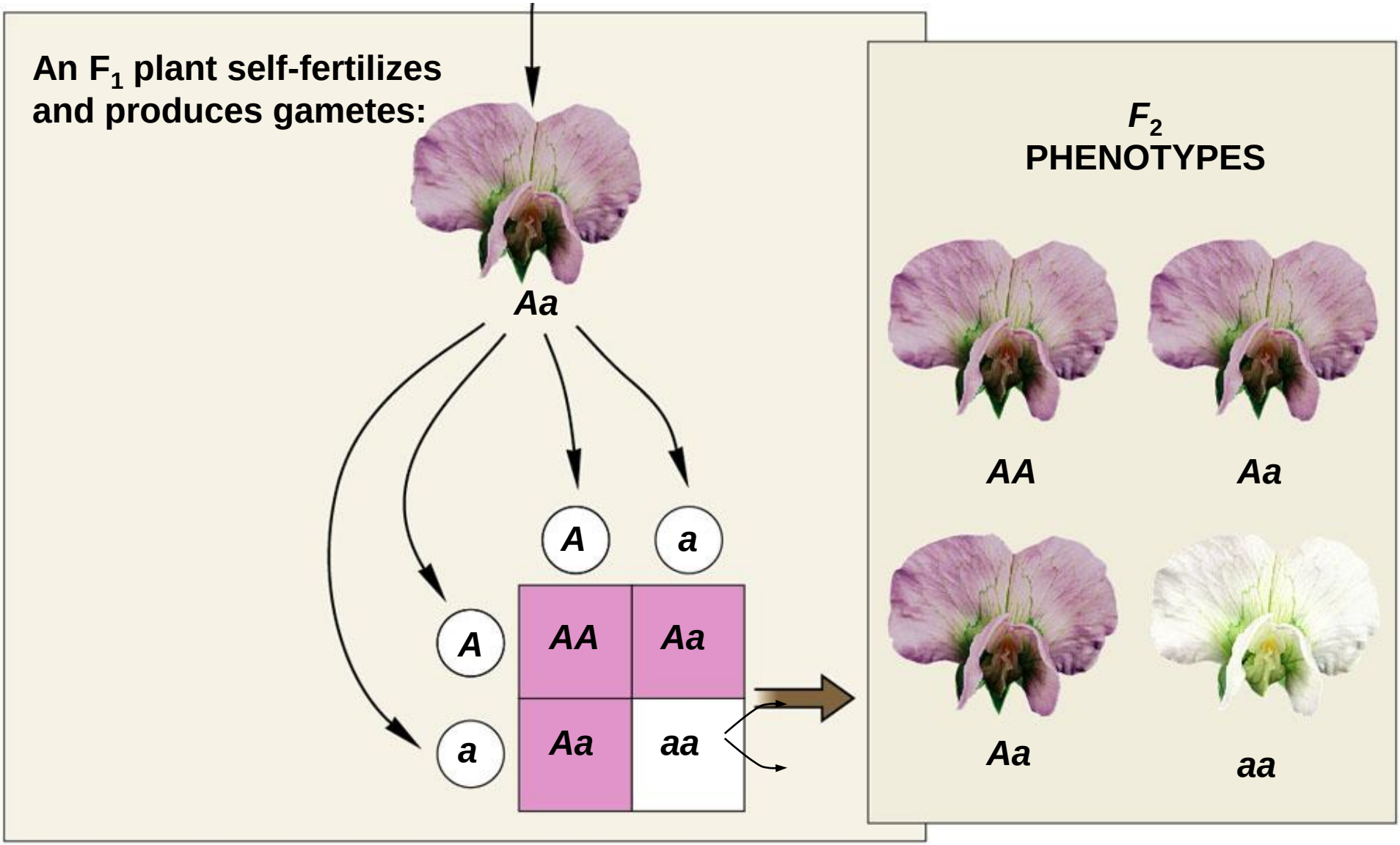
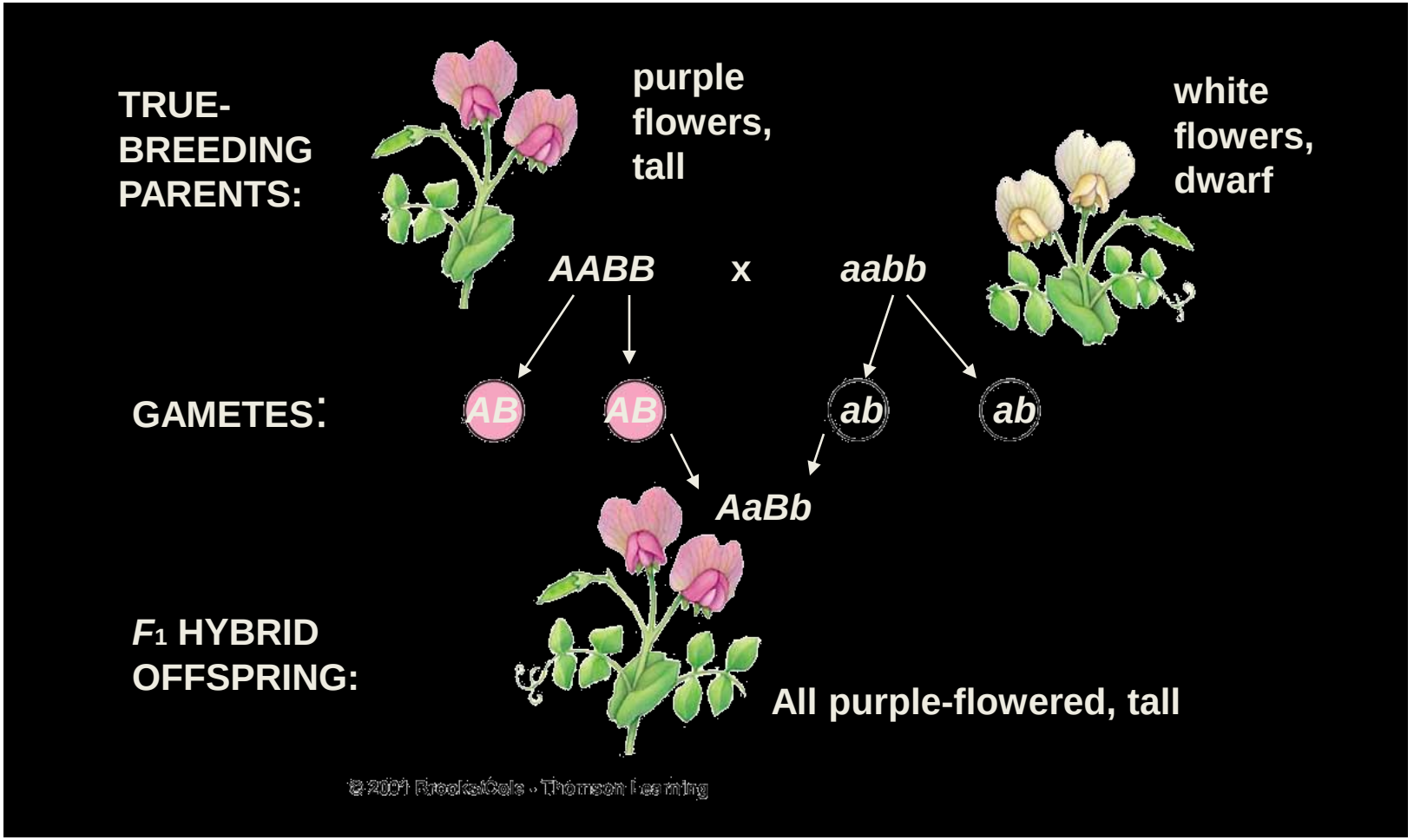


Fig. 11-7b2, p.173

Dihybrid Cross

Experimental cross between individuals that are homozygous for different versions of two traits

Dihybrid Cross: F_1 Results



Dihybrid Cross: F_2 Results

AaBb X ***AaBb***

	1/4 *AB*	1/4 *Ab*	1/4 *aB*	1/4 *ab*
1/4 *AB*	1/16 *AABB*	1/16 *AABb*	1/16 *AaBB*	1/16 *AaBb*
1/4 *Ab*	1/16 *AABb*	1/16 *AAbb*	1/16 *AaBb*	1/16 *Aabb*
1/4 *aB*	1/16 *AaBB*	1/16 *AaBb*	1/16 *aaBB*	1/16 *aaBb*
1/4 *ab*	1/16 *AaBb*	1/16 *Aabb*	1/16 *aaBb*	1/16 ***aabb***

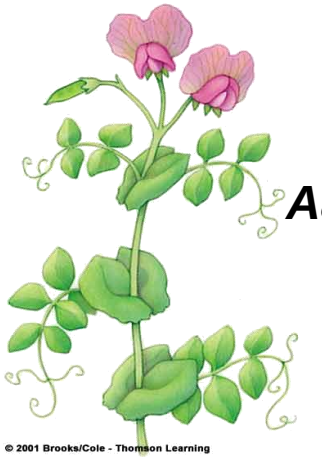
- 9/16 purple-flowered, tall
- 3/16 purple-flowered, dwarf
- 3/16 white-flowered, tall
- 1/16 white-flowered, dwarf

AABB
purple-flowered tall parent (homozygous dominant)

AB **X** *ab*

aabb
white-flowered dwarf parent (homozygous recessive)

F_1 OUTCOME:
All of the F_1 plants are *AaBb* heterozygotes (purple flowers, tall stems).



A*a****B****b***

Fig. 11-9a, p.175

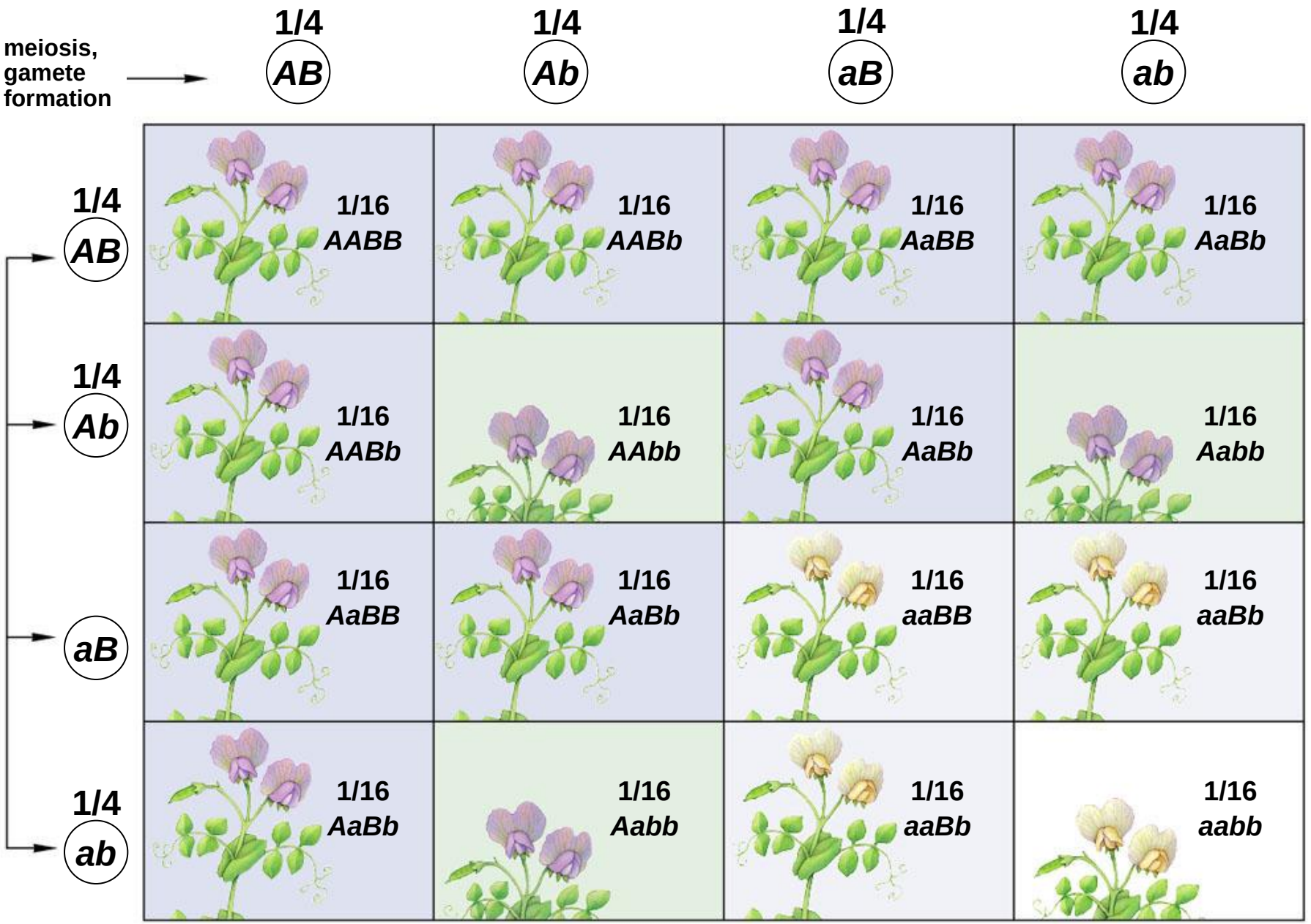


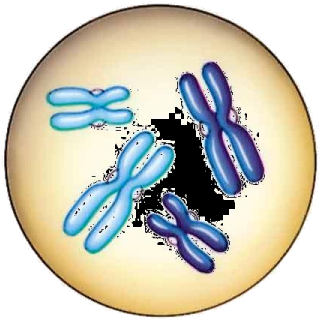
Fig. 11-9b, p.175

Independent Assortment

- Mendel concluded that the two “units” for the first trait were to be assorted into gametes independently of the two “units” for the other trait

- Members of each pair of homologous chromosomes are sorted into gametes at random during meiosis

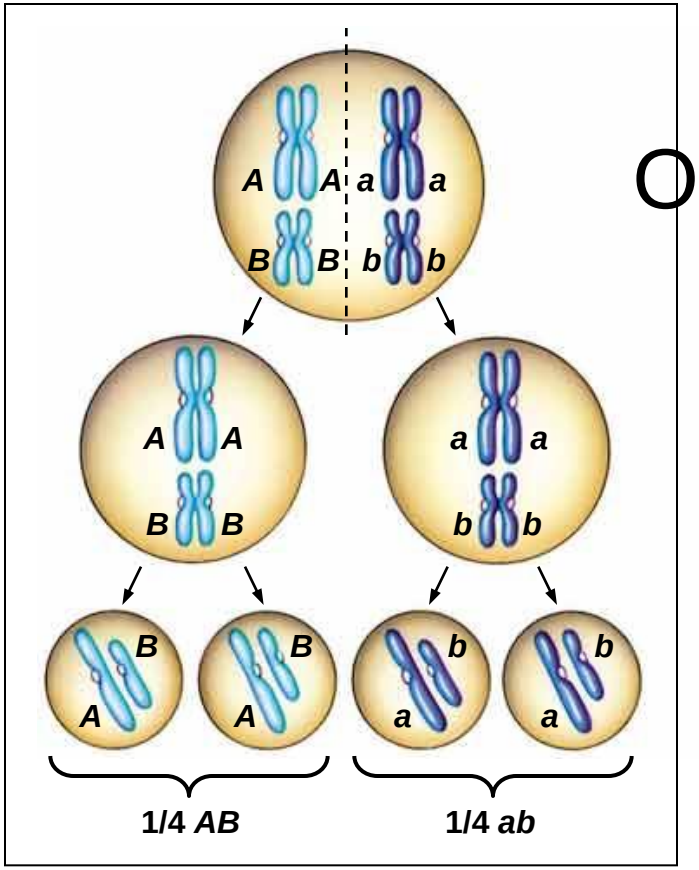
Independent Assortment



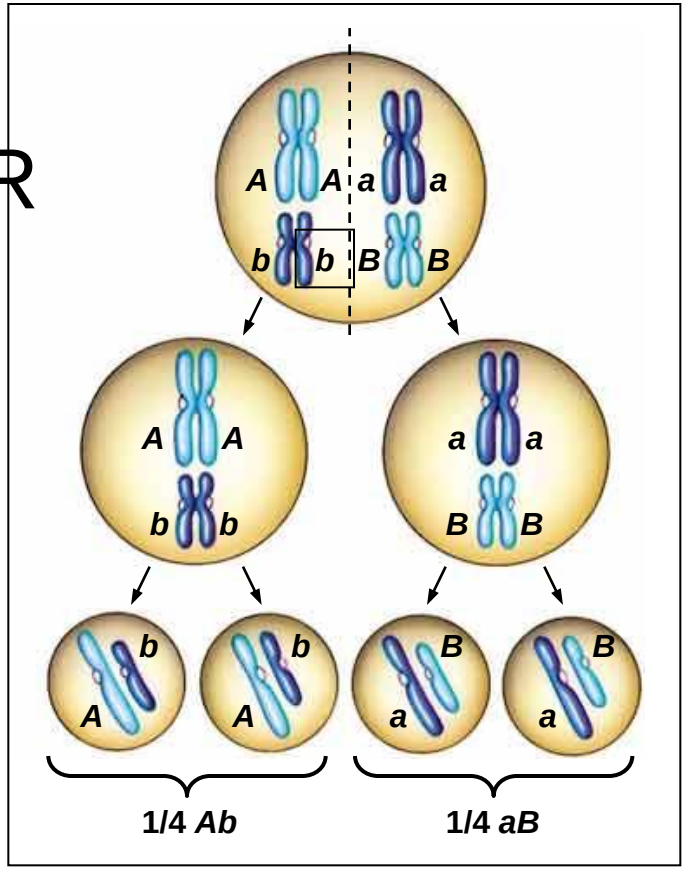
Metaphase I:

Metaphase II:

Gametes:



OR



Independent Assortment

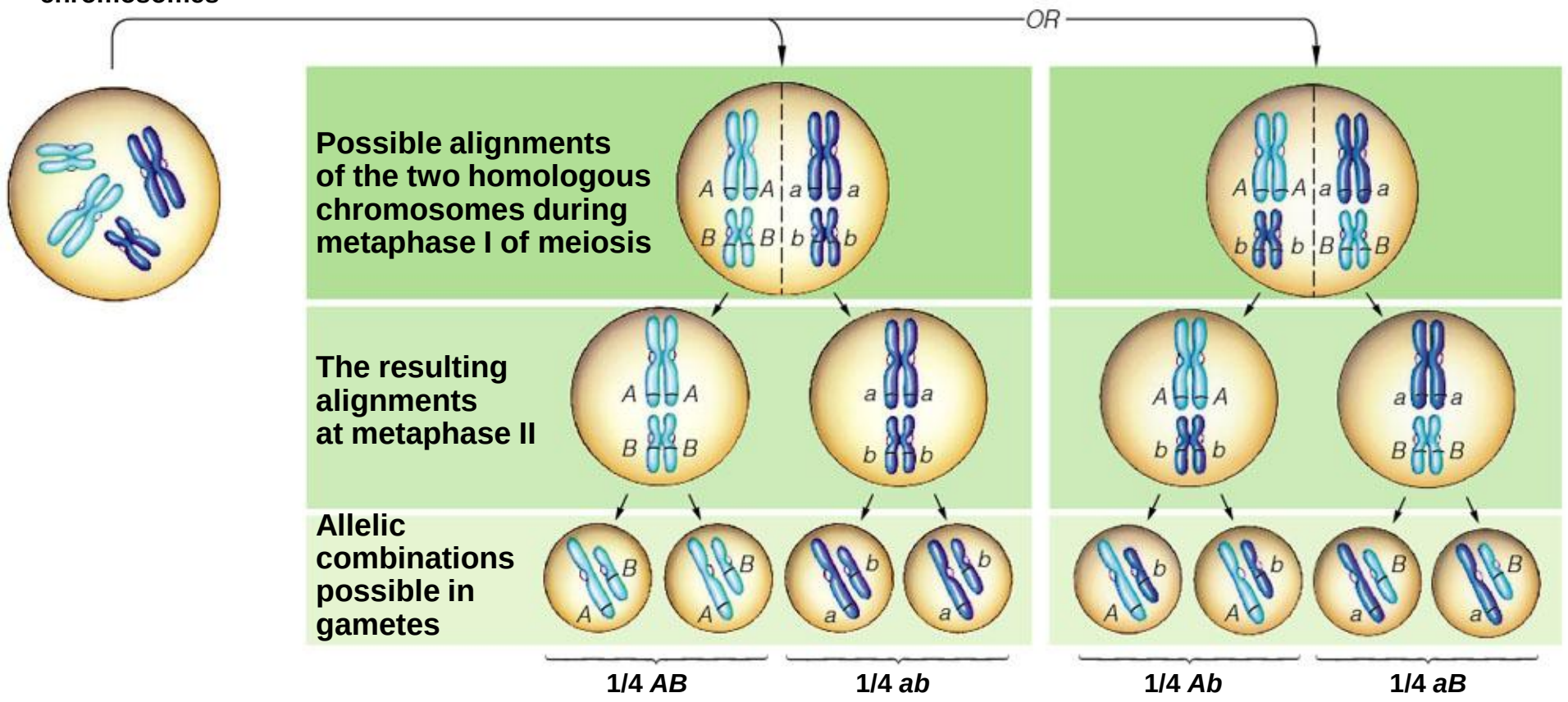


Fig. 11-8, p.174

Tremendous Variation

Number of genotypes possible in offspring as a result of independent assortment and hybrid crossing is

3^n

(*n* is the number of gene loci at which the parents differ)

Impact of Mendel's Work

- Mendel presented his results in 1865
- Paper received little notice
- Mendel discontinued his experiments in 1871
- Paper rediscovered in 1900

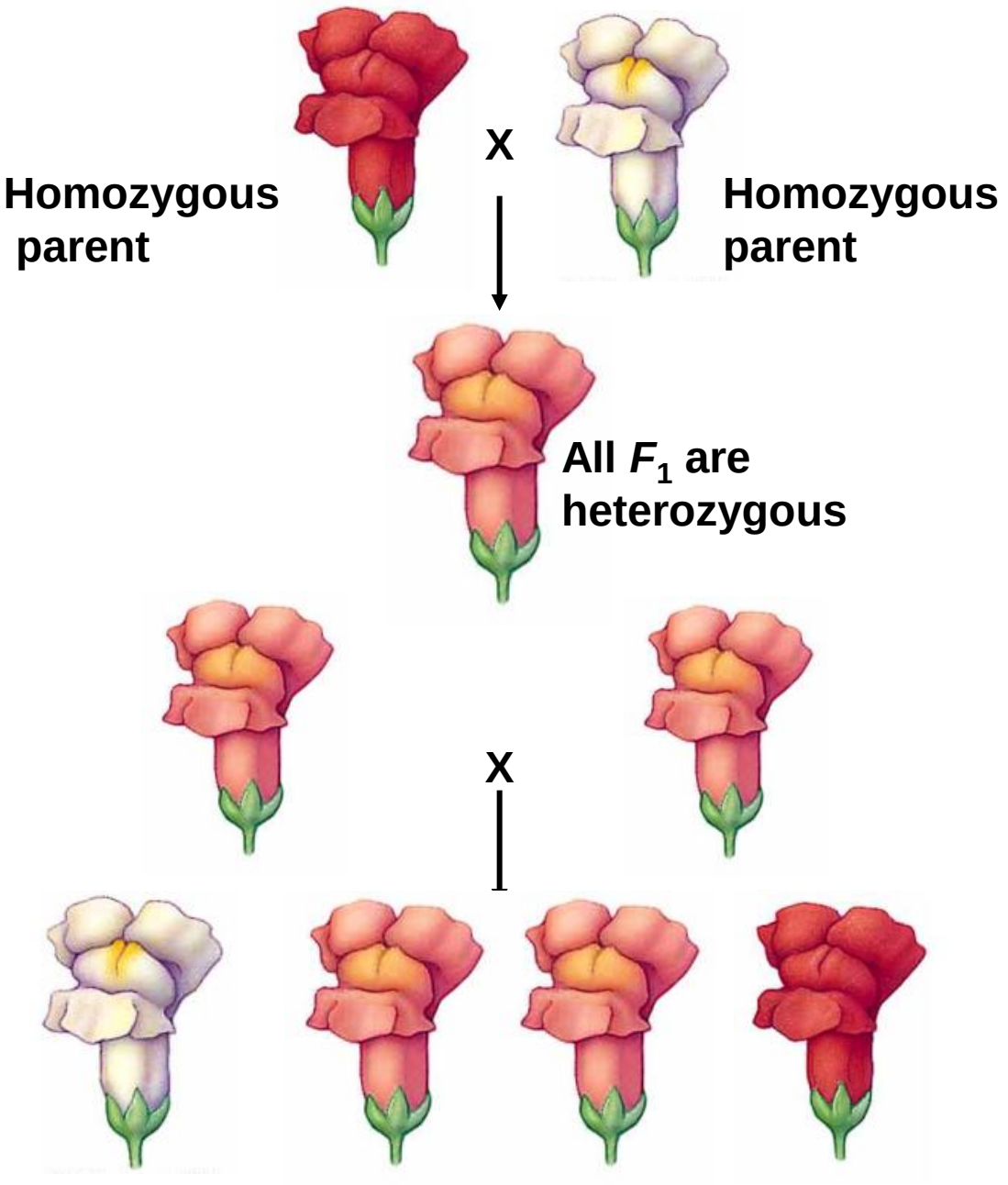
Dominance Relations

Complete dominance

Incomplete dominance

Codominance

Incomplete Dominance



F_2 shows three phenotypes in 1:2:1 ratio

Incomplete Dominance

homozygous parent X homozygous parent

All F_1 offspring heterozygous for flower color:

Cross two of the F_1 plants and the F_2 offspring will show three phenotypes in a 1:2:1 ratio:

Fig. 11-11, p.176

Codominance: ABO Blood Types

- Gene that controls ABO type codes for enzyme that dictates structure of a glycolipid on blood cells

- Two alleles (I^A and I^B) are codominant when paired

- Third allele (*i*) is recessive to others

ABO Blood Type

Range of genotypes:

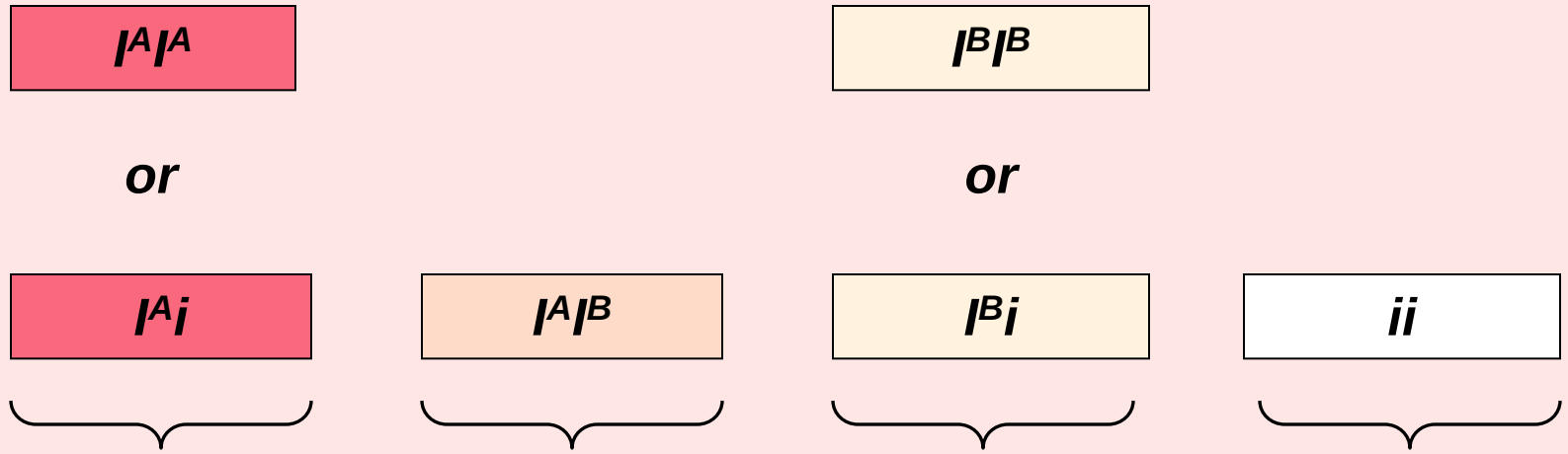


Fig. 11-10a, p.176

ABO Blood Type

Fig. 11-10b, p.176

ABO and Transfusions

- Recipient's immune system will attack blood cells that have an unfamiliar glycolipid on surface
- Type O is universal donor because it has neither type A nor type B glycolipid

Pleiotropy

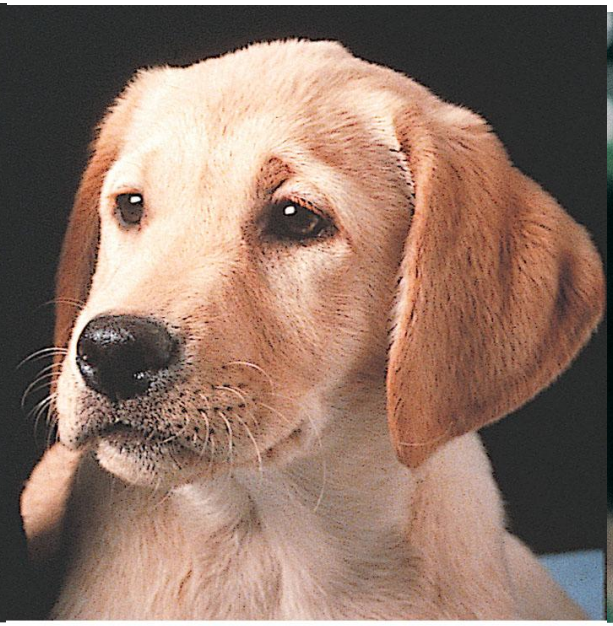
- Alleles at a single locus may have effects on two or more traits

- Marfan syndrome - Mutation in gene for fibrillin affects skeleton, cardiovascular system, lungs, eyes, and skin

Epistasis

- Interaction between the products of gene pairs
- Common among genes for hair color in mammals

Epistasis



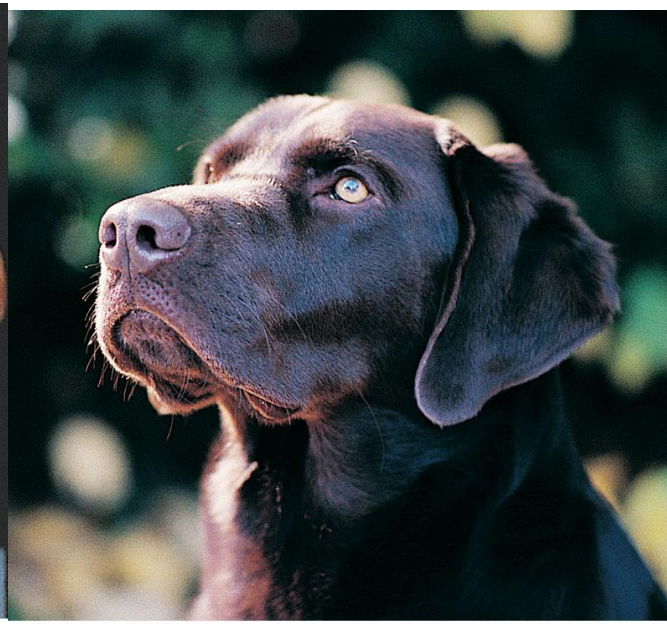


Fig. 11-13, p.177

Coat Color in Retrievers

BBEE **X**

bbee

F_1 puppies are all *BbEe*

F_2 puppies

	BE	***Be***	***bE***	***be***
BE	***BBEE***	***BBEe***	***BbEE***	***BbEe***
Be	***BBEe***	***BBee***	***BbEe***	***Bbee***
bE	***BbEE***	***BbEe***	***bbEE***	***bbEe***
be	***BbEe***	***Bbee***	***bbEe***	***bbee***

black

brown

yellow

Comb Shape in Poultry

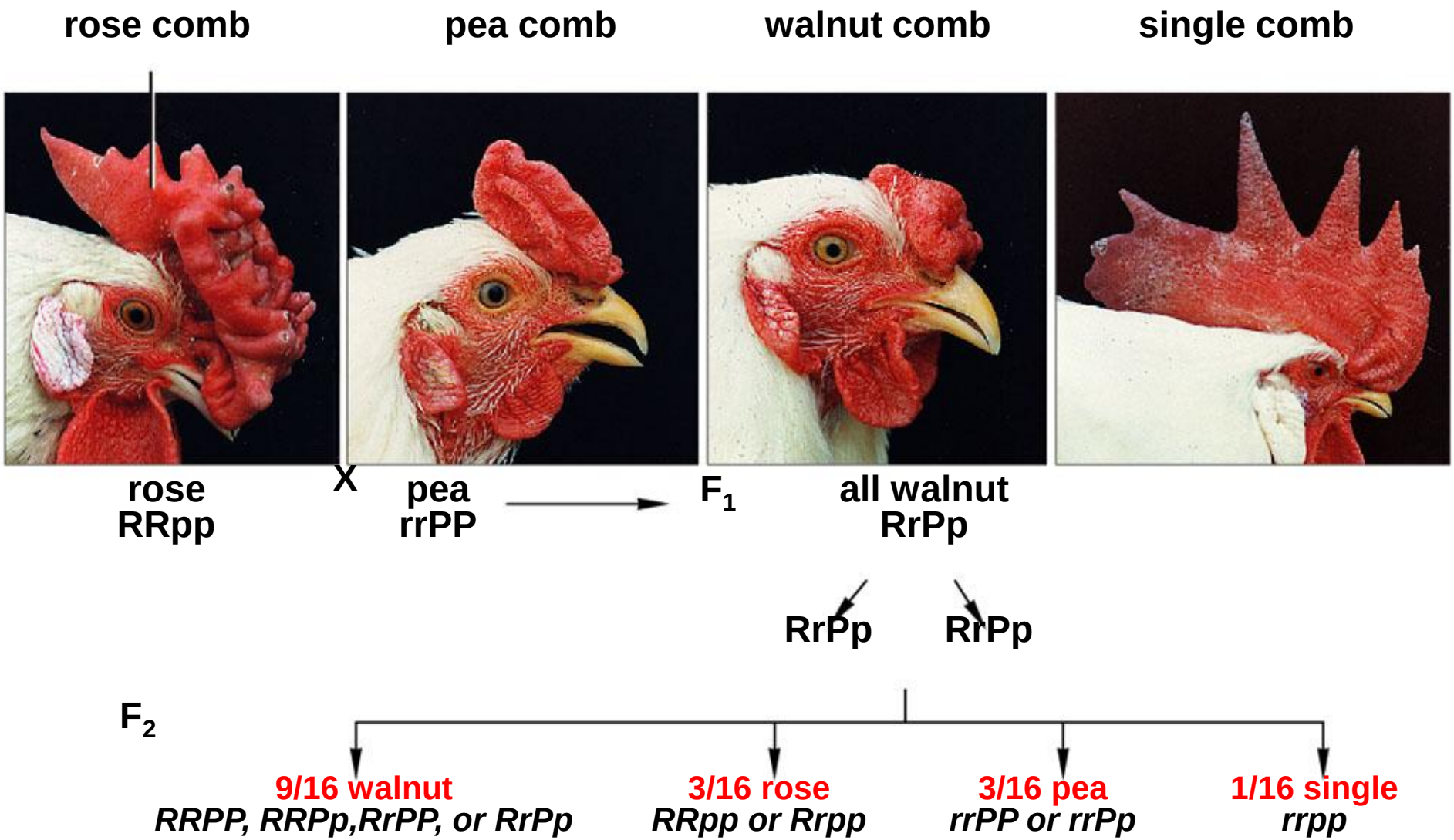
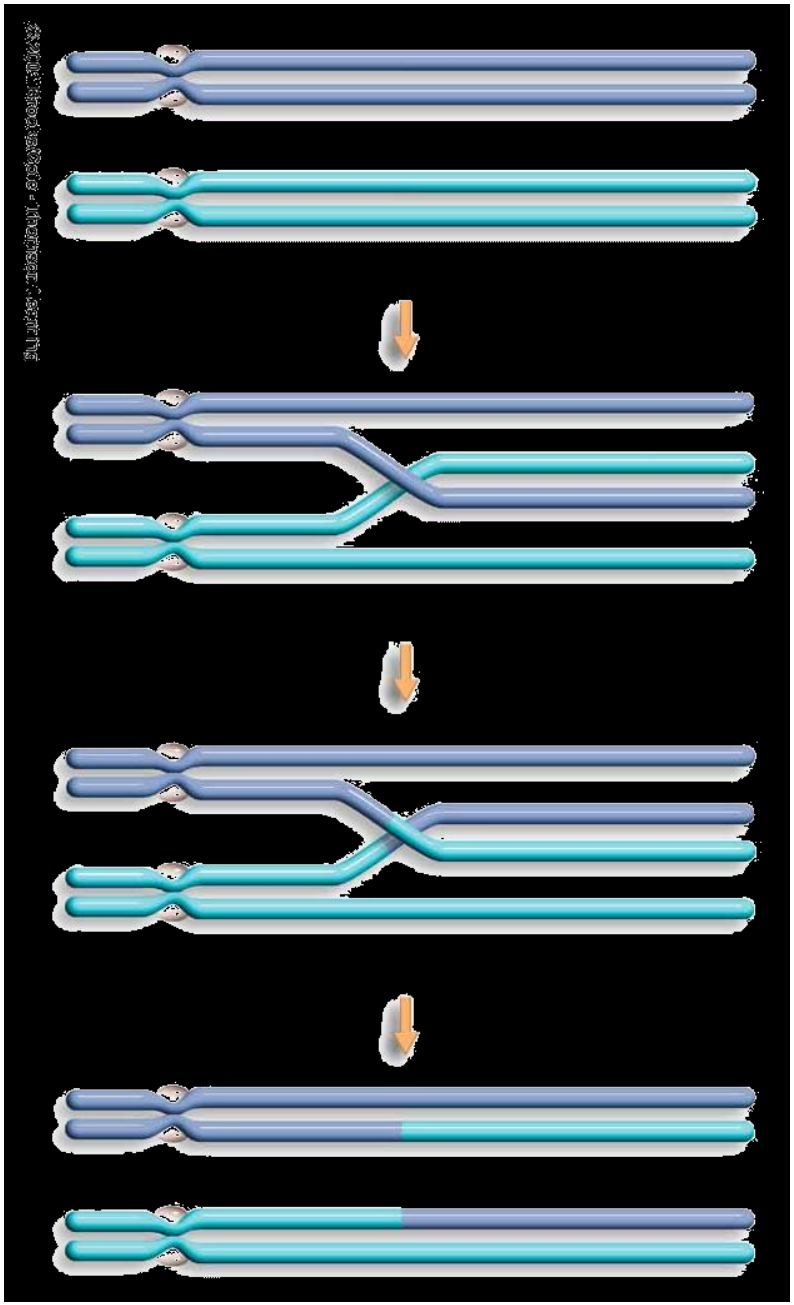


Fig. 11-12, p.177

Crossing Over

- Each chromosome becomes zippered to its homologue
- All four chromatids are closely aligned
- Nonsister chromosomes exchange segments



Effect of Crossing Over

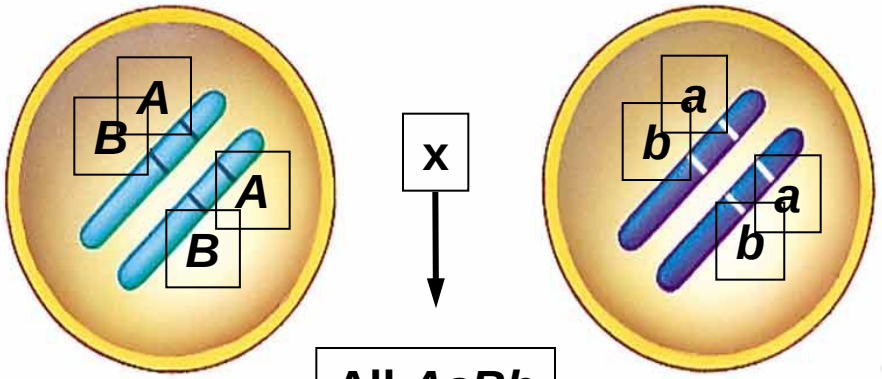
- After crossing over, each chromosome contains both maternal and paternal segments
- Creates new allele combinations in offspring

Linkage Groups

- Genes on one type of chromosome
- Fruit flies
  - 4 homologous chromosomes
  - 4 linkage groups
- Not all genes on chromosome are tightly linked

Full Linkage

Parents:

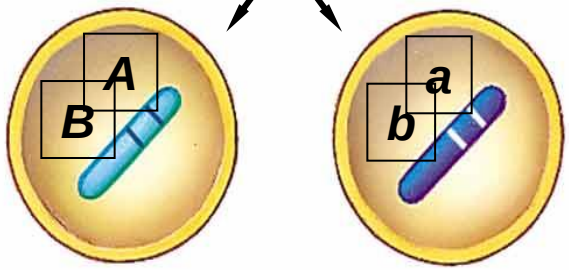
AB 

ab

F_1 offspring: **All *AaBb***

meiosis, gamete formation

Equal ratios of two types of gametes:



50% *AB* **50% *ab***

Incomplete Linkage

Parents:

***F_1* offspring:**

Unequal ratios of four types of gametes:

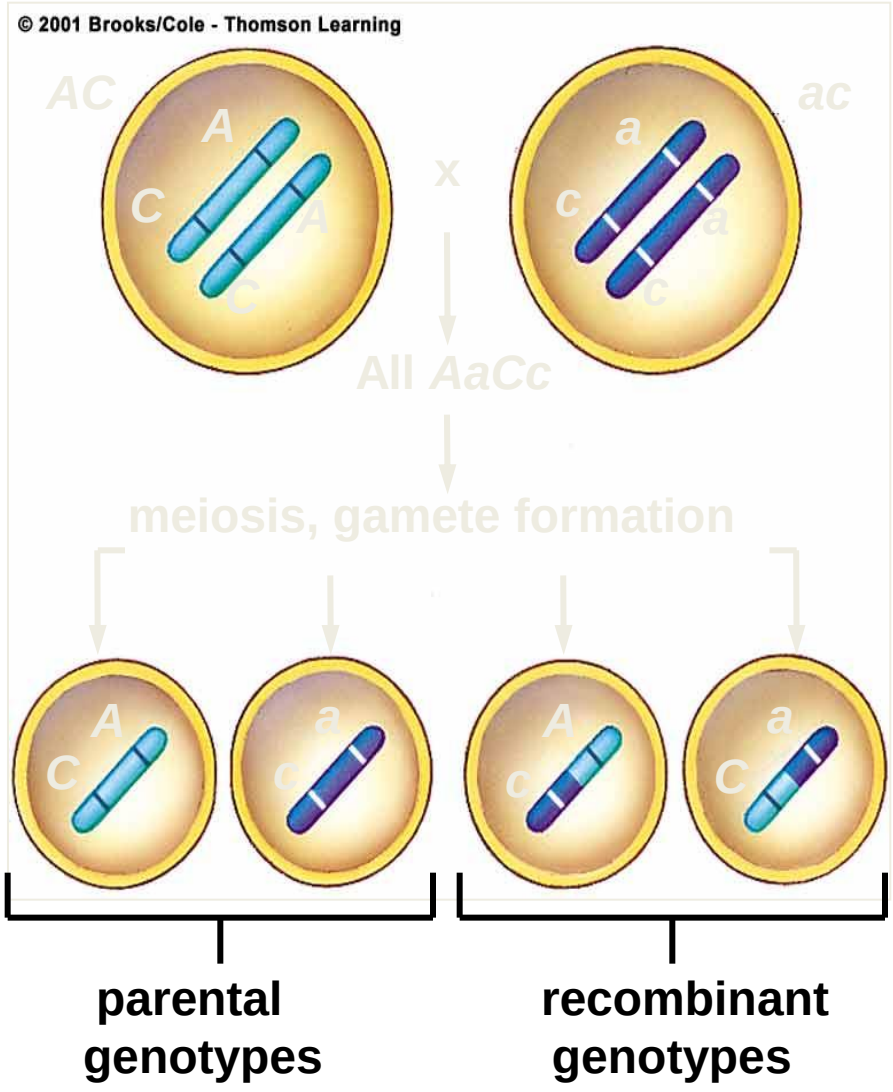


Figure 11.15

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Crossover Frequency

Proportional to the distance that separates genes

Crossing over will disrupt linkage between *A* and *B* more often than *C* and *D*

In-text figure

Linkage Mapping in Humans

- Linkage maps based on pedigree analysis through generations

- Color blindness and hemophilia are very closely linked on X chromosome

Environmental Effects on Plant Phenotype

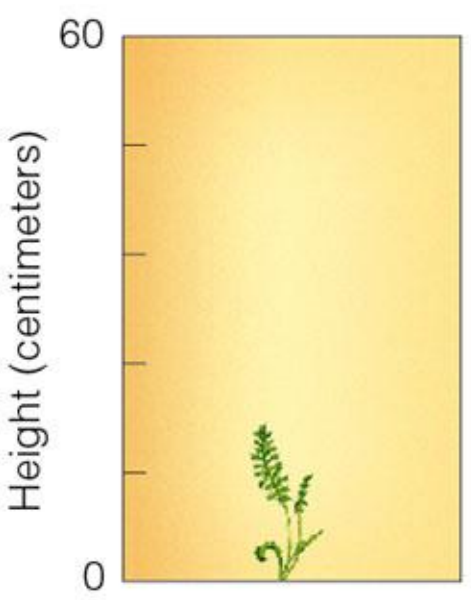
- *Hydrangea macrophylla*
- Action of gene responsible for floral color is influenced by soil acidity
- Flower color ranges from pink to blue

Environmental Effects on Plant Phenotype

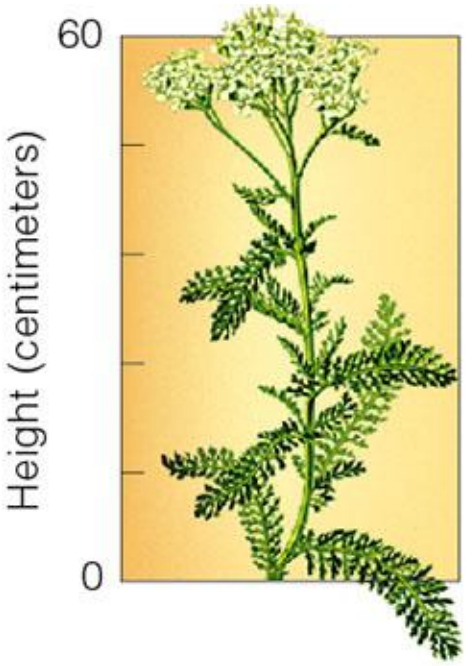
Fig. 11-17a, p.179

Environmental Effects on Plant Phenotype

a Mature cutting at high elevation (3,050 meters above sea level)



b Mature cutting at mid-elevation (1,400 meters above sea level)



c Mature cutting at low elevation (30 meters above sea level)

Fig. 11-17b, p.179

Temperature Effects on Phenotype

- Rabbit is homozygous for an allele that specifies a heat-sensitive version of an enzyme in melanin-producing pathway
- Melanin is produced in cooler areas of body

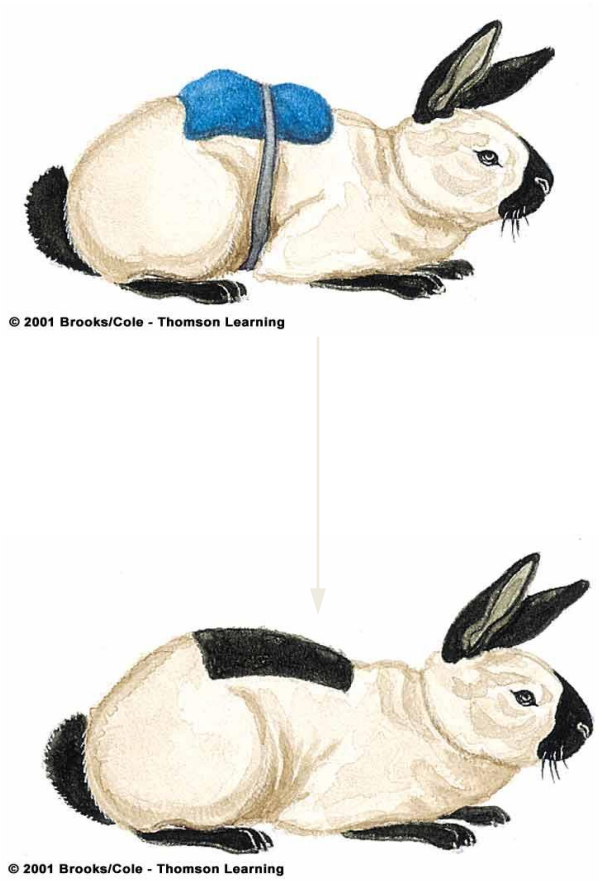


Figure 11.16

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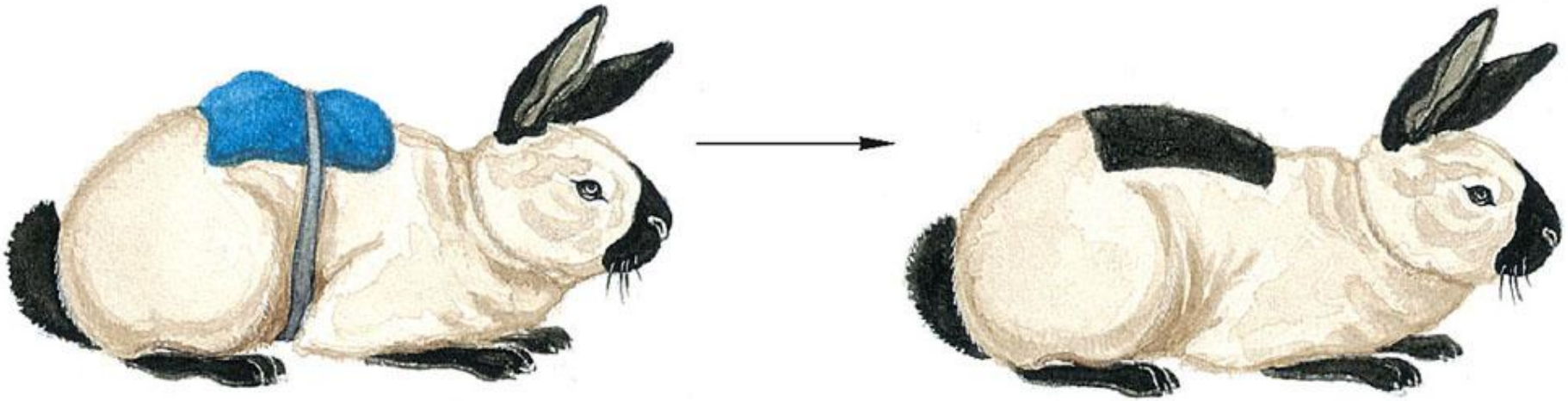

Fig. 11-16, p.179

Campodactyly: Unexpected Phenotypes

- Effect of allele varies:
  - Bent fingers on both hands
  - Bent fingers on one hand
  - No effect
- Many factors affect gene expression

Continuous Variation

- A more or less continuous range of small differences in a given trait among individuals
- The greater the number of genes and environmental factors that affect a trait, the more continuous the variation in versions of that trait

Human Variation

- Some human traits occur as a few discrete types
  - Attached or detached earlobes
  - Many genetic disorders
- Other traits show continuous variation
  - Height
  - Weight
  - Eye color

Continuous Variation

- Variation in human eye color

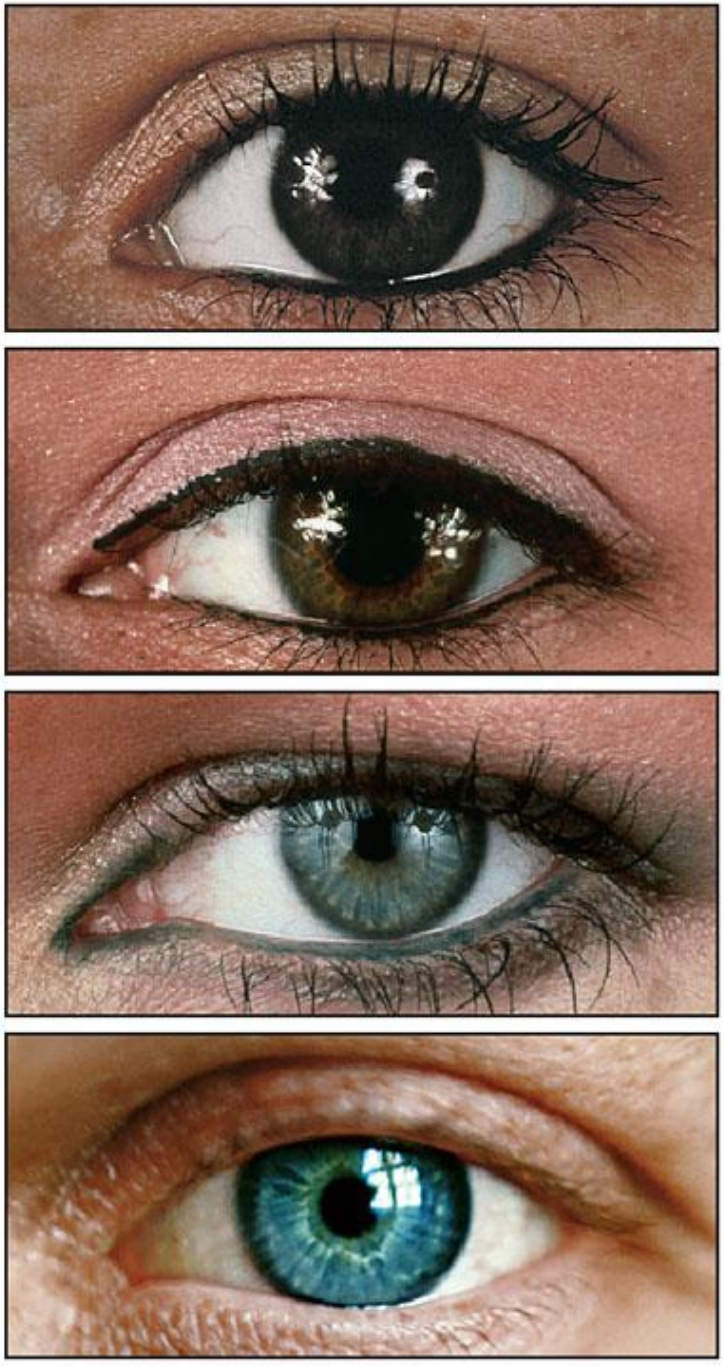

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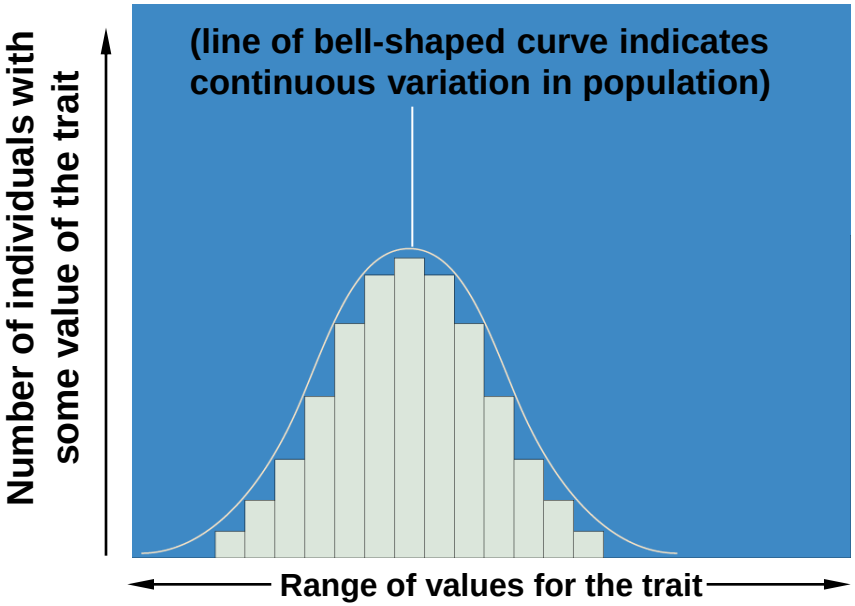
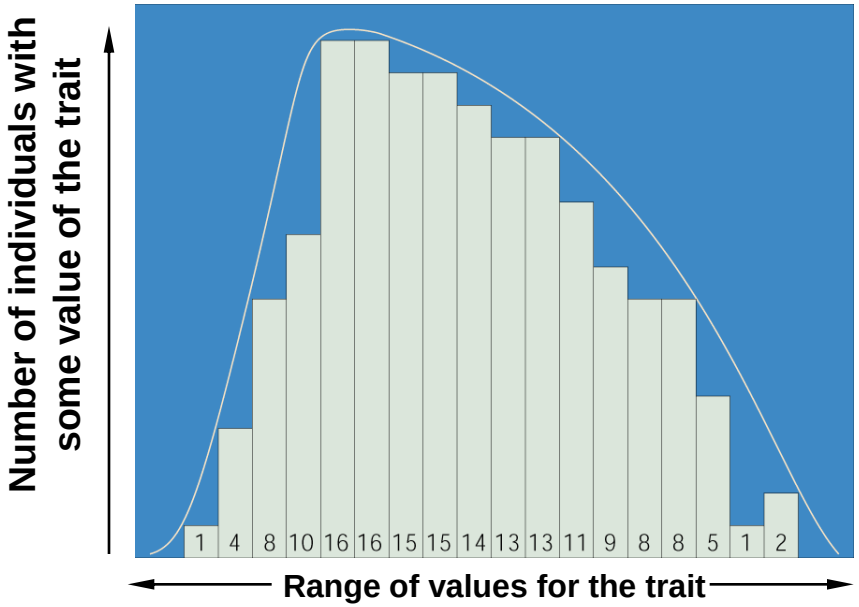
Fig. 11-18, p.180

Describing Continuous Variation





Describing Continuous Variation

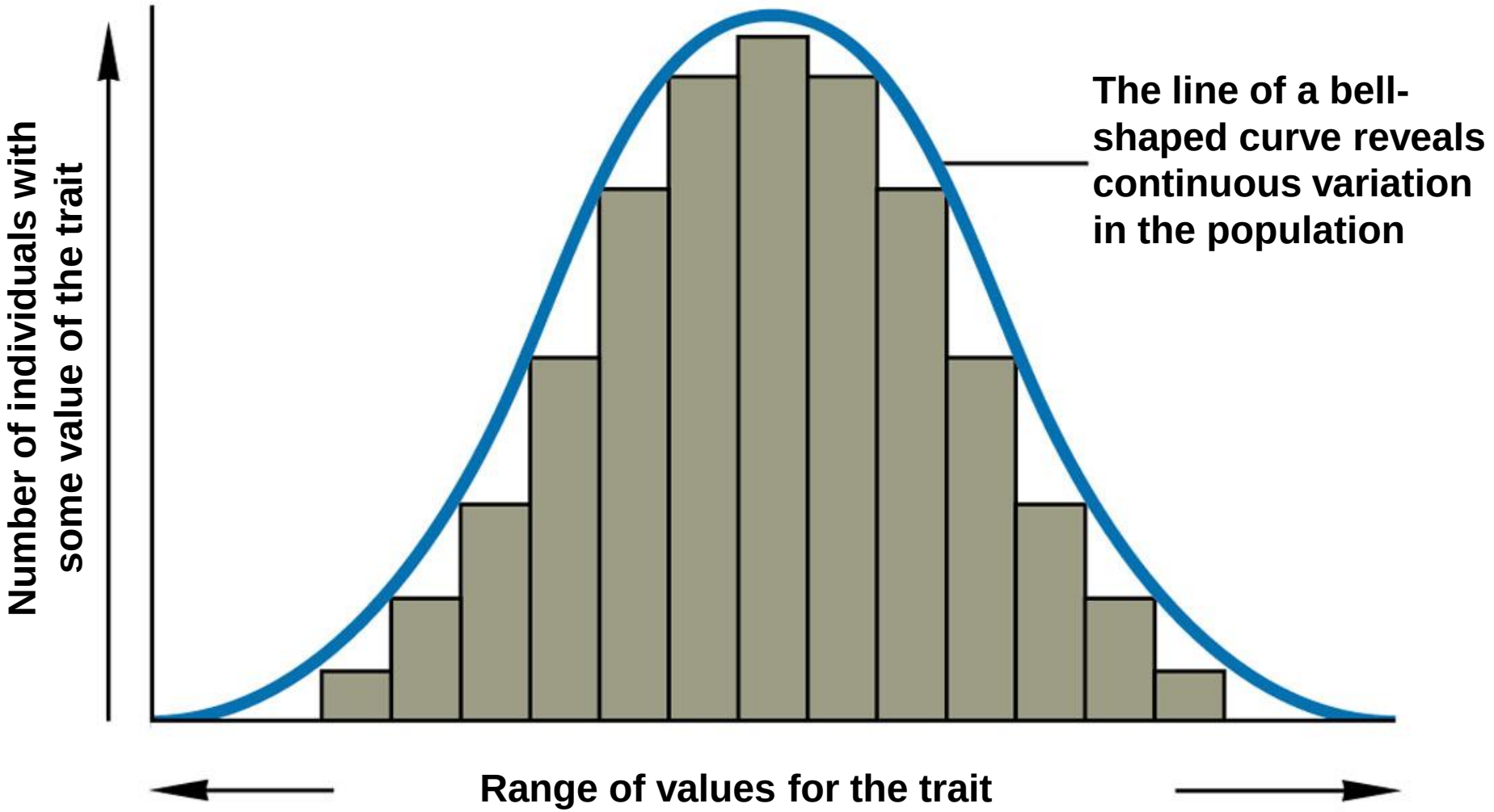


Fig. 11-19a, p.180

Describing Continuous Variation

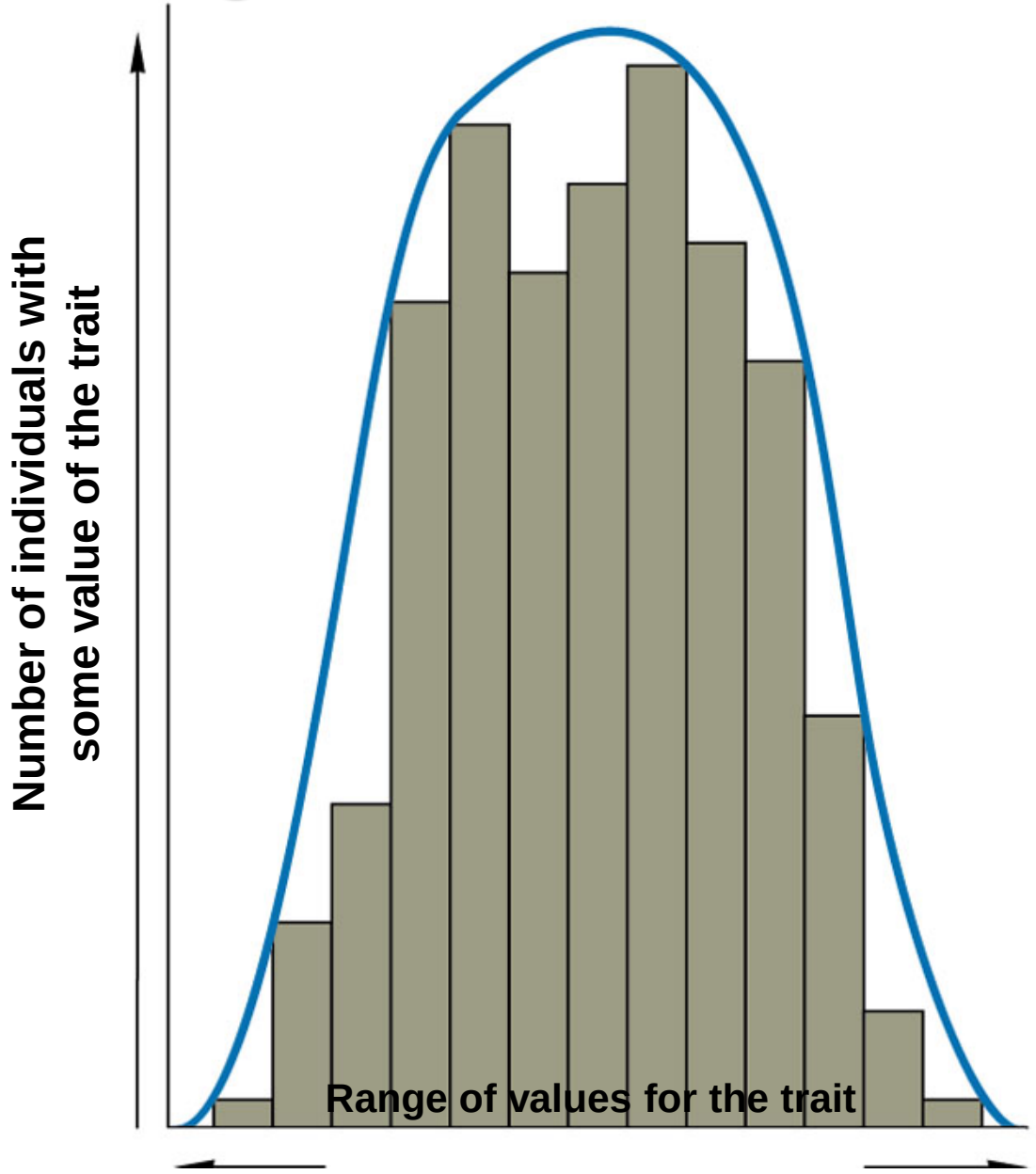
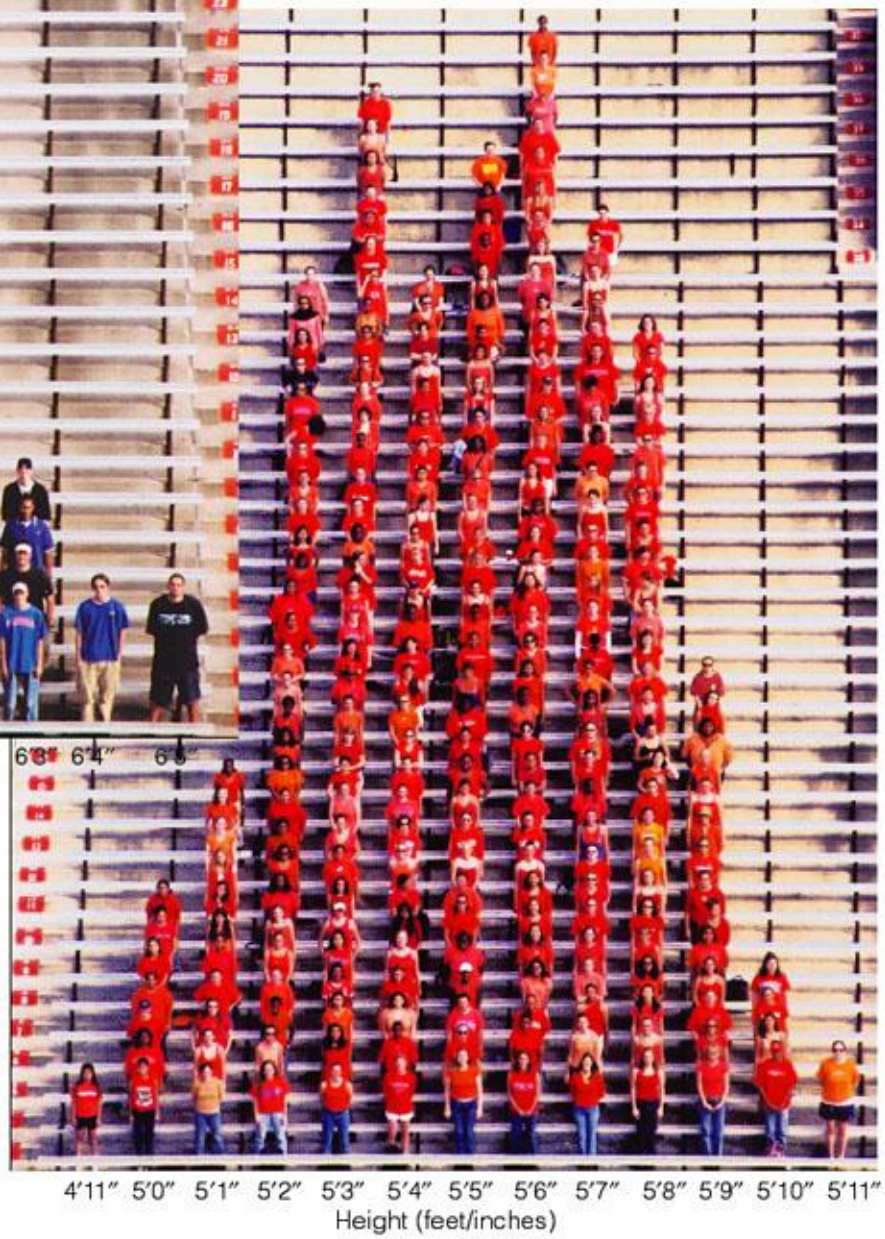


Fig. 11-19b, p.180



Describing Continuous Variation

Fig. 11-20, p.181

Fig. 11-21, p.183

dominant

recessive

Fig. 11-22, p.183

dominant

recessive

Fig. 11-22, p.183

Fig. 11-24, p.185