Chapter 9

Patterns of Inheritance

Lecture by Dr. Prince

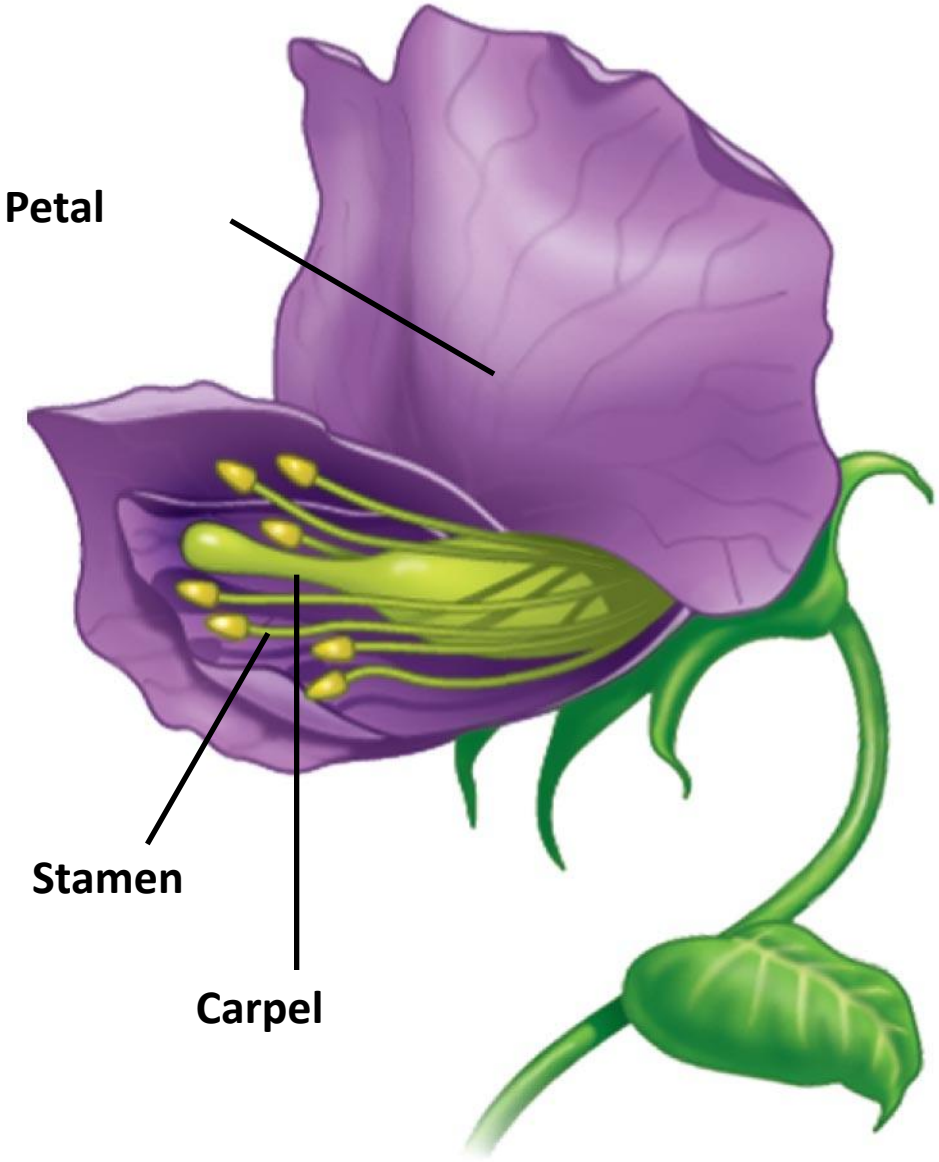
MENDEL'S LAWS

9.1 The science of genetics has ancient roots

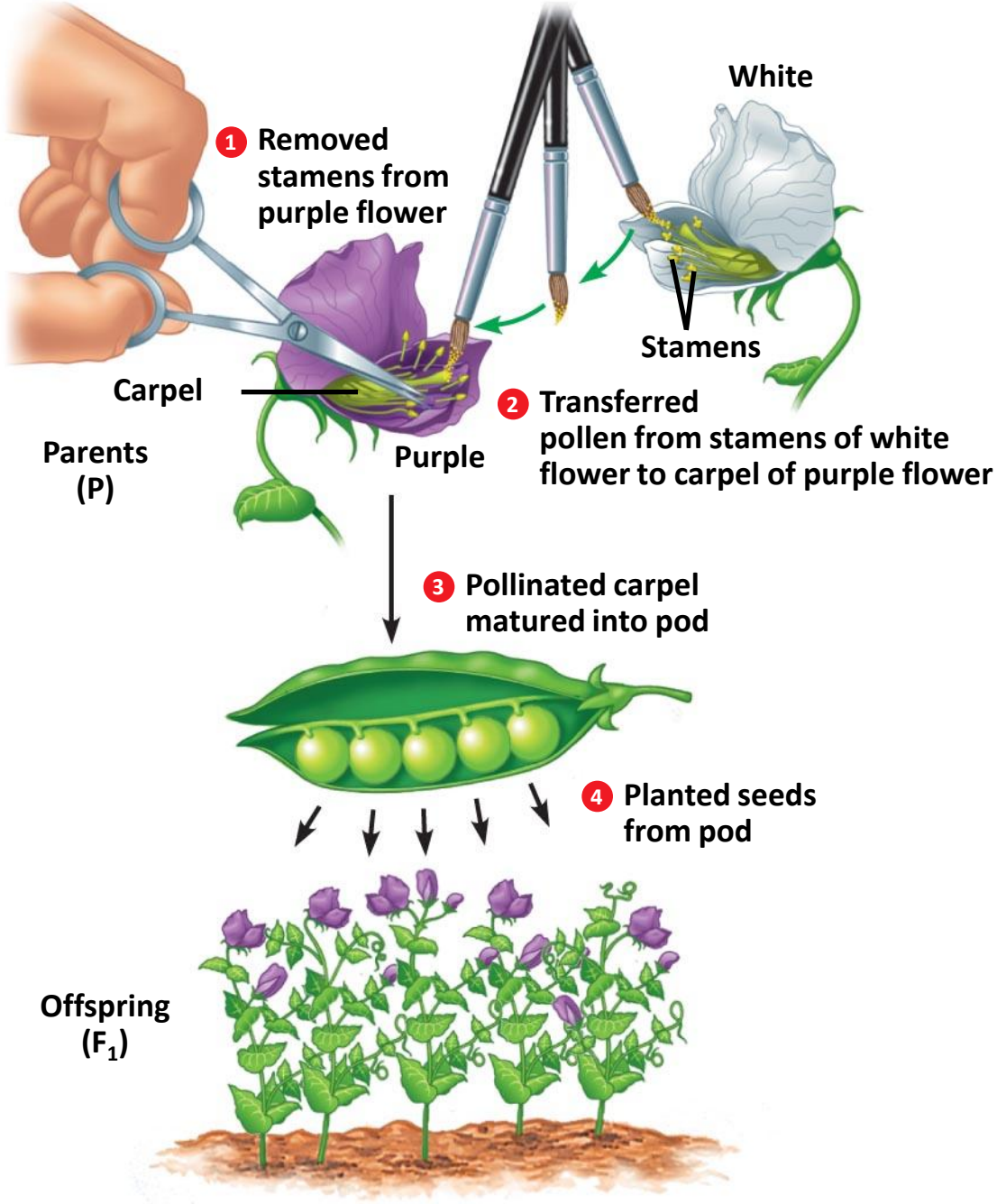
- Pangenesis was an early explanation for inheritance
  - It was proposed by Hippocrates
  - Particles called pangenes came from all parts of the organism to be incorporated into eggs or sperm
  - Characteristics acquired during the parents' lifetime could be transferred to the offspring
  - Aristotle rejected pangenesis and argued that instead of particles, the potential to produce the traits was inherited
- Blending was another idea, based on plant breeding
  - Hereditary material from parents mixes together to form an intermediate trait, like mixing paint

9.2 Experimental genetics began in an abbey garden

- Gregor Mendel discovered principles of genetics in experiments with the garden pea
  - Mendel showed that parents pass heritable factors to offspring (heritable factors are now called genes)
  - Advantages of using pea plants
    - Controlled matings
    - Self-fertilization or cross-fertilization
    - Observable characteristics with two distinct forms
    - True-breeding strains



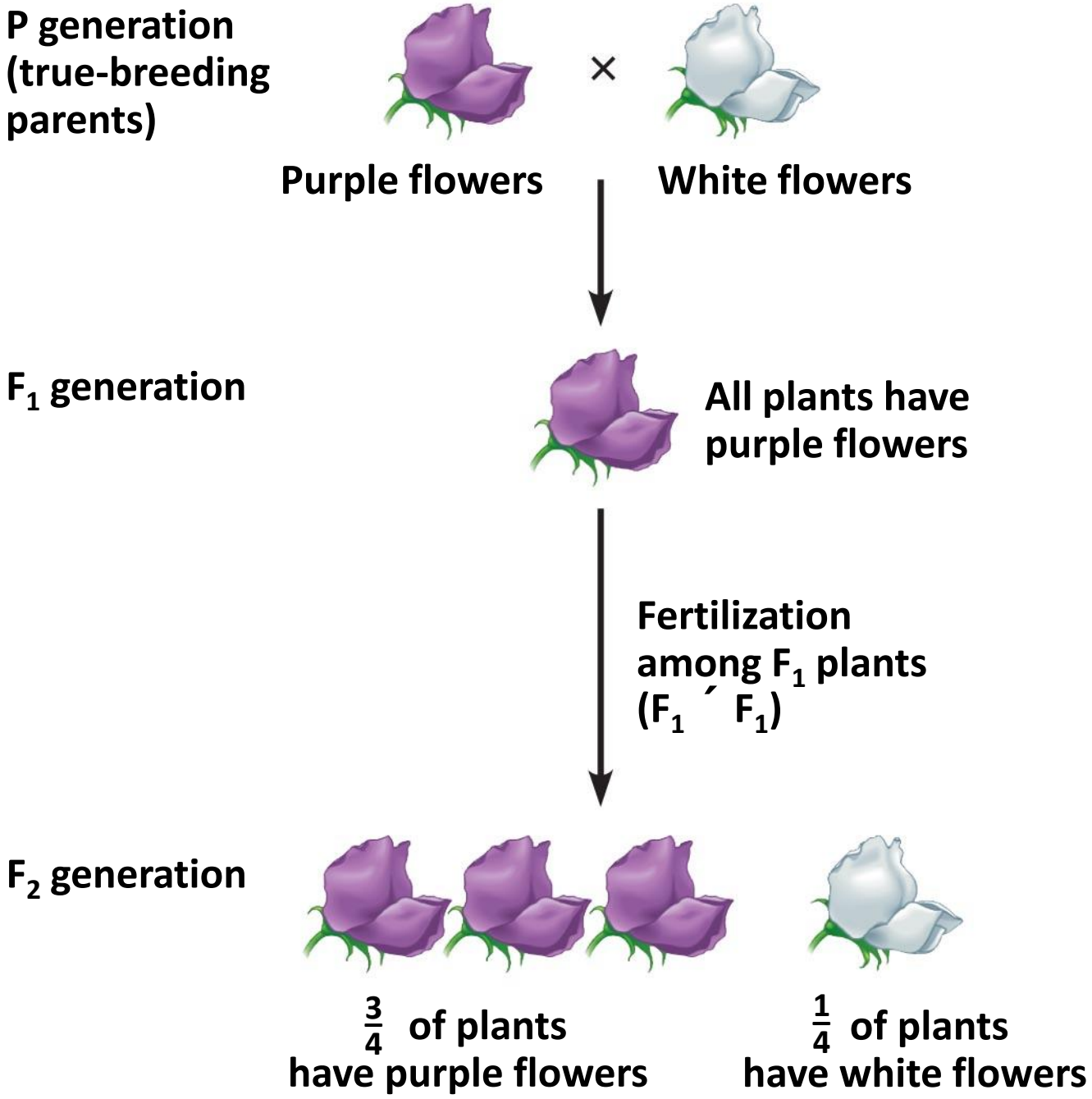
Flower color	 Purple	 White
Flower position	 Axial	 Terminal
Seed color	 Yellow	 Green
Seed shape	 Round	 Wrinkled
Pod shape	 Inflated	 Constricted
Pod color	 Green	 Yellow
Stem length	 Tall	 Dwarf



P generation (true-breeding parents)

Purple flowers **White flowers**

- Example of a **monohybrid cross**
- Parental generation: purple flowers × white flowers



9.3 Mendel's law of segregation describes the inheritance of a single character

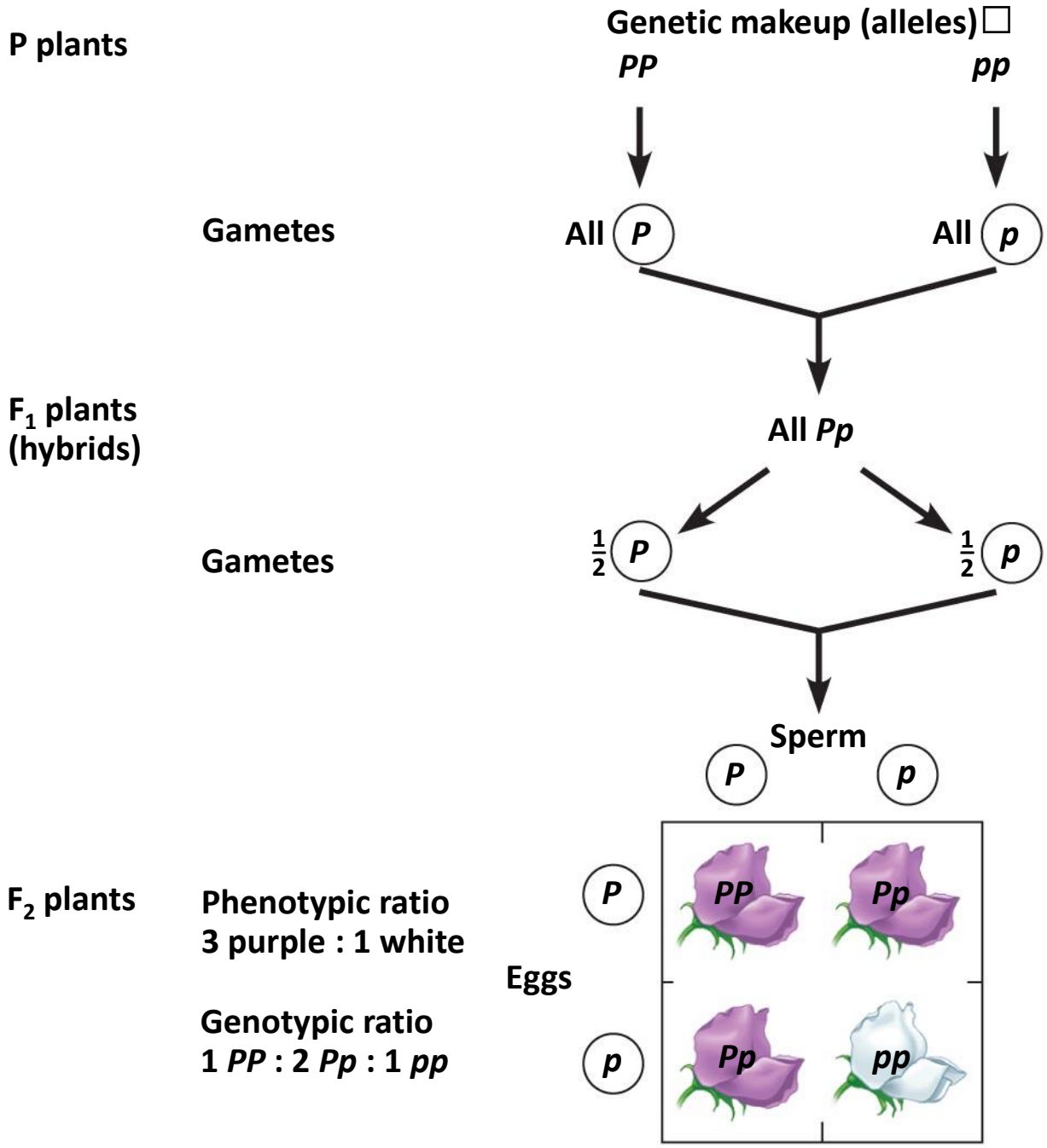
- Mendel needed to explain
- Why one trait seemed to disappear in the F_1 generation
- Why that trait reappeared in one quarter of the F_2 offspring

9.3 Mendel's law of segregation describes the inheritance of a single character

- Four Hypotheses
  1. Genes are found in alternative versions called **alleles**; a genotype is the listing of alleles an individual carries for a specific gene
  2. For each characteristic, an organism inherits two alleles, one from each parent; the alleles can be the same or different
     - A **homozygous** genotype has identical alleles
     - A **heterozygous** genotype has two different alleles

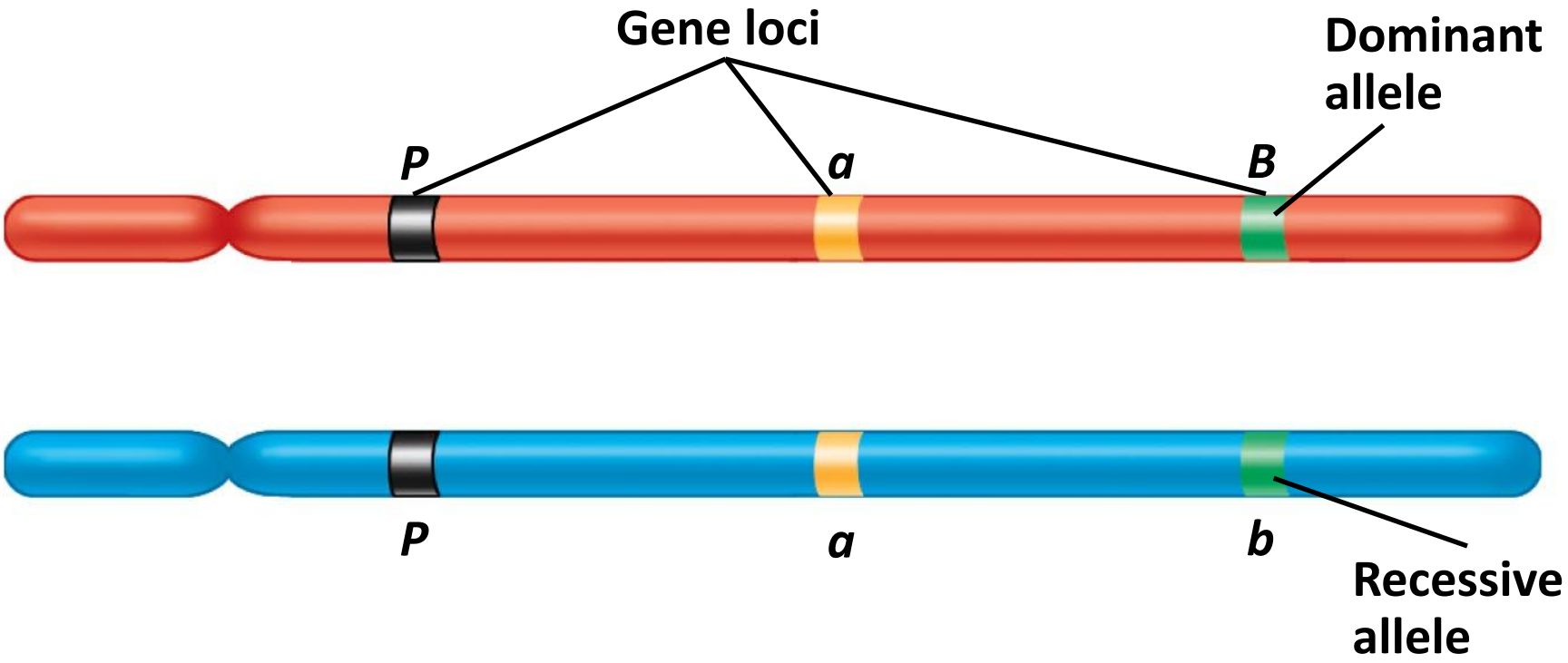
9.3 Mendel's law of segregation describes the inheritance of a single character

- Four Hypotheses
  3. If the alleles differ, the dominant allele determines the organism's appearance, and the recessive allele has no noticeable effect
     - The phenotype is the appearance or expression of a trait
     - The same phenotype may be determined by more than one genotype
  4. Law of segregation: Allele pairs separate (segregate) from each other during the production of gametes so that a sperm or egg carries only one allele for each gene



9.4 Homologous chromosomes bear the alleles for each character

- For a pair of homologous chromosomes, alleles of a gene reside at the same locus
  - Homozygous individuals have the same allele on both homologues
  - Heterozygous individuals have a different allele on each homologue



Genotype: ***PP*** ***aa*** ***Bb***

Homozygous for the dominant allele

Homozygous for the recessive allele

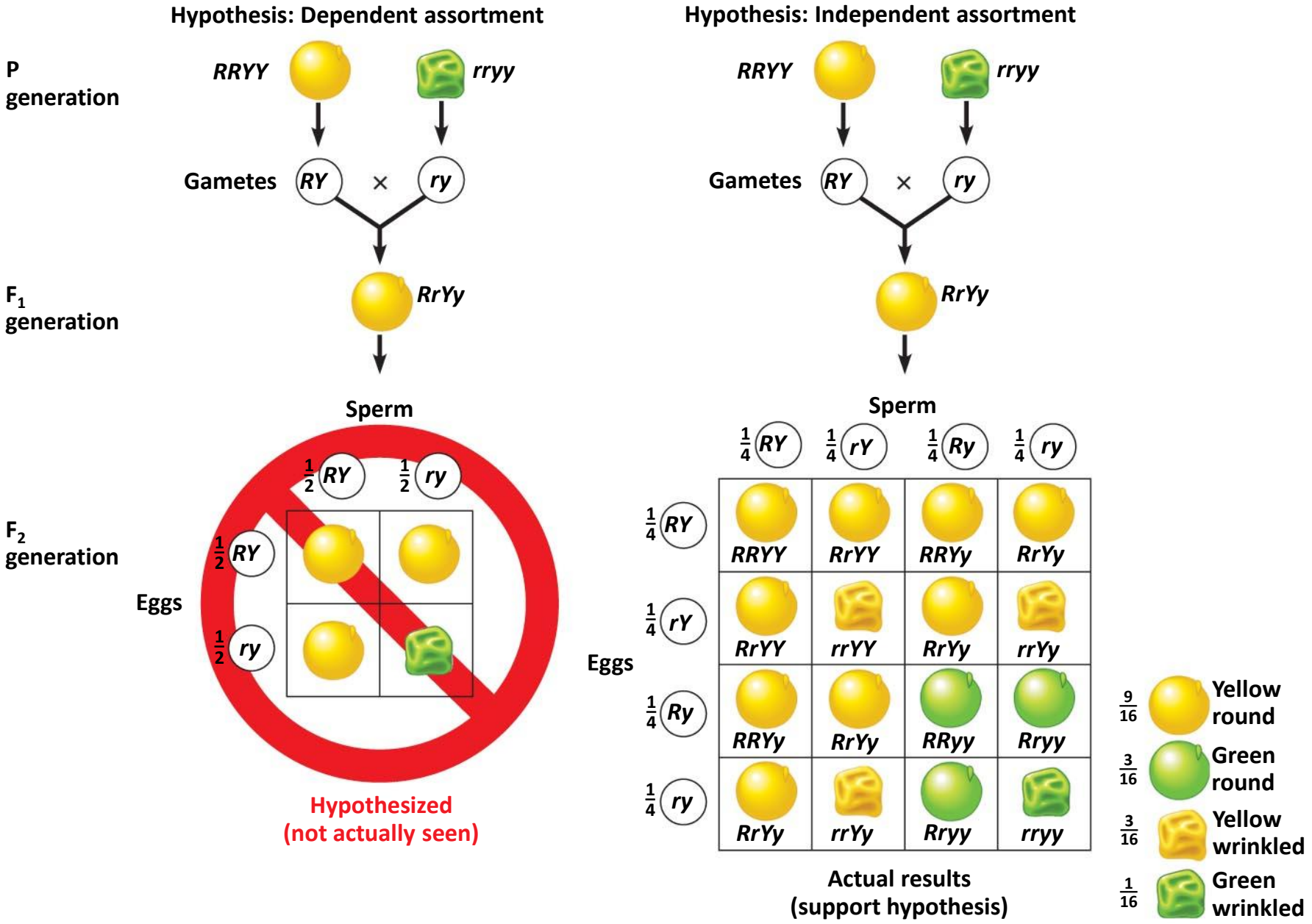
Heterozygous

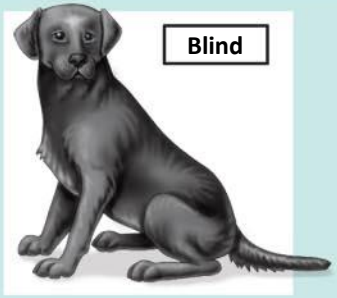
9.5 The law of independent assortment is revealed by tracking two characters at once

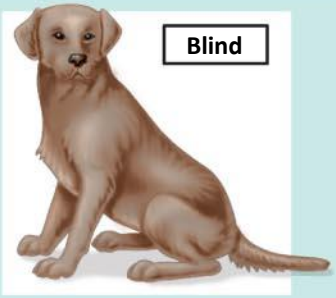
- Example of a **dihybrid cross**
  - Parental generation: round yellow seeds × wrinkled green seeds
  - F_1 generation: all plants with round yellow seeds
  - F_2 generation: 9/16 of plants with round yellow seeds
    3/16 of plants with round green seeds
    3/16 of plants with wrinkled yellow seeds
    1/16 of plants with wrinkled green seeds
- Mendel needed to explain
  - Why nonparental combinations were observed
  - Why a 9:3:3:1 ratio was observed among the F_2 offspring

9.5 The law of independent assortment is revealed by tracking two characters at once

- **Law of independent assortment**
  - Each pair of alleles segregates independently of the other pairs of alleles during gamete formation
  - For genotype *RrYy*, four gamete types are possible: *RY*, *Ry*, *rY*, and *ry*







Phenotypes	**Black coat, normal vision**	**Black coat, blind (PRA)**	**Chocolate coat, normal vision**	**Chocolate coat, blind (PRA)**
Genotypes	***B_N_***	***B_nn***	***bbN_***	***bbnn***

Mating of heterozygotes (black, normal vision)

BbNn × ***BbNn***

Phenotypic ratio of offspring				
	9 black coat, normal vision	**3 black coat, blind (PRA)**	**3 chocolate coat, normal vision**	**1 chocolate coat, blind (PRA)**

9.6 Geneticists use the testcross to determine unknown genotypes

- **Testcross**
  - Mating between an individual of unknown genotype and a homozygous recessive individual
  - Will show whether the unknown genotype includes a recessive allele
  - Used by Mendel to confirm true-breeding genotypes

Testcross:

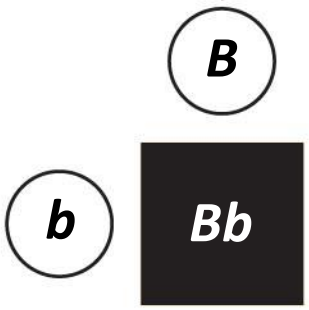
×

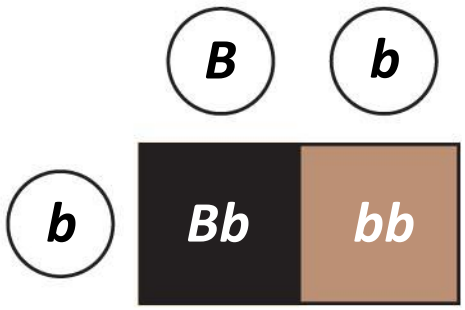
Genotypes ***B_*** ***bb***

Two possibilities for the black dog:

BB **or** ***Bb***

Gametes

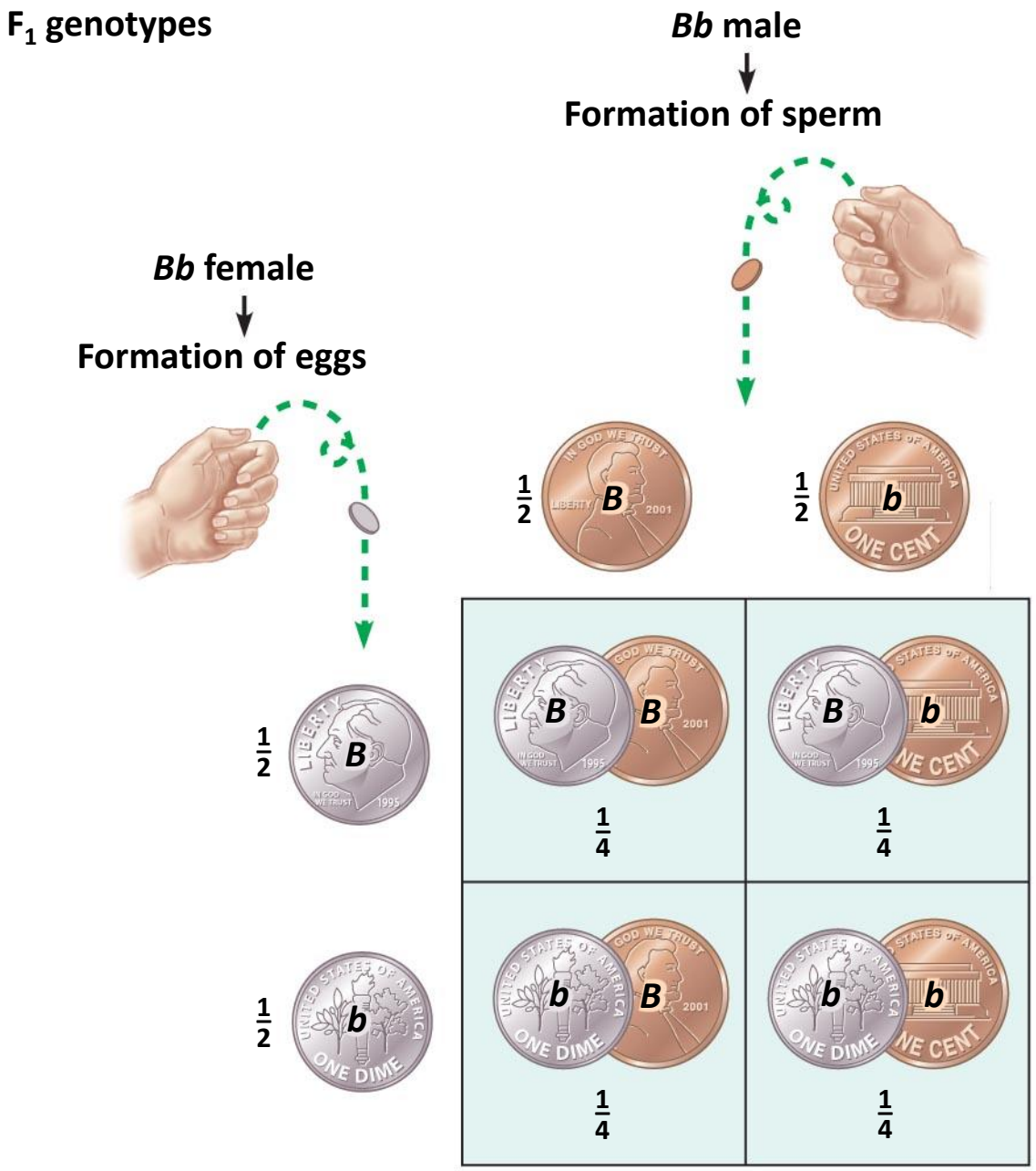




Offspring **All black** **1 black : 1 chocolate**

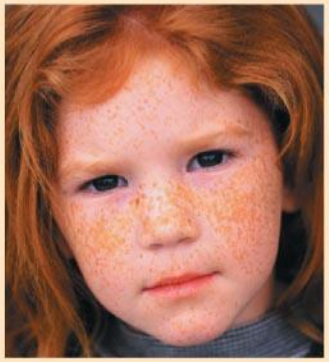
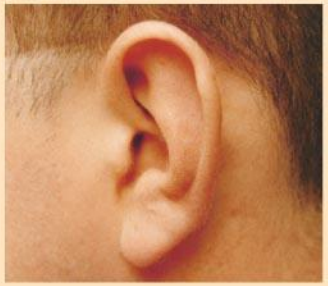
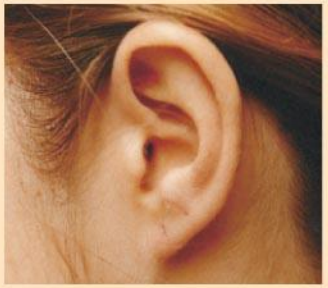
9.7 Mendel's laws reflect the rules of probability

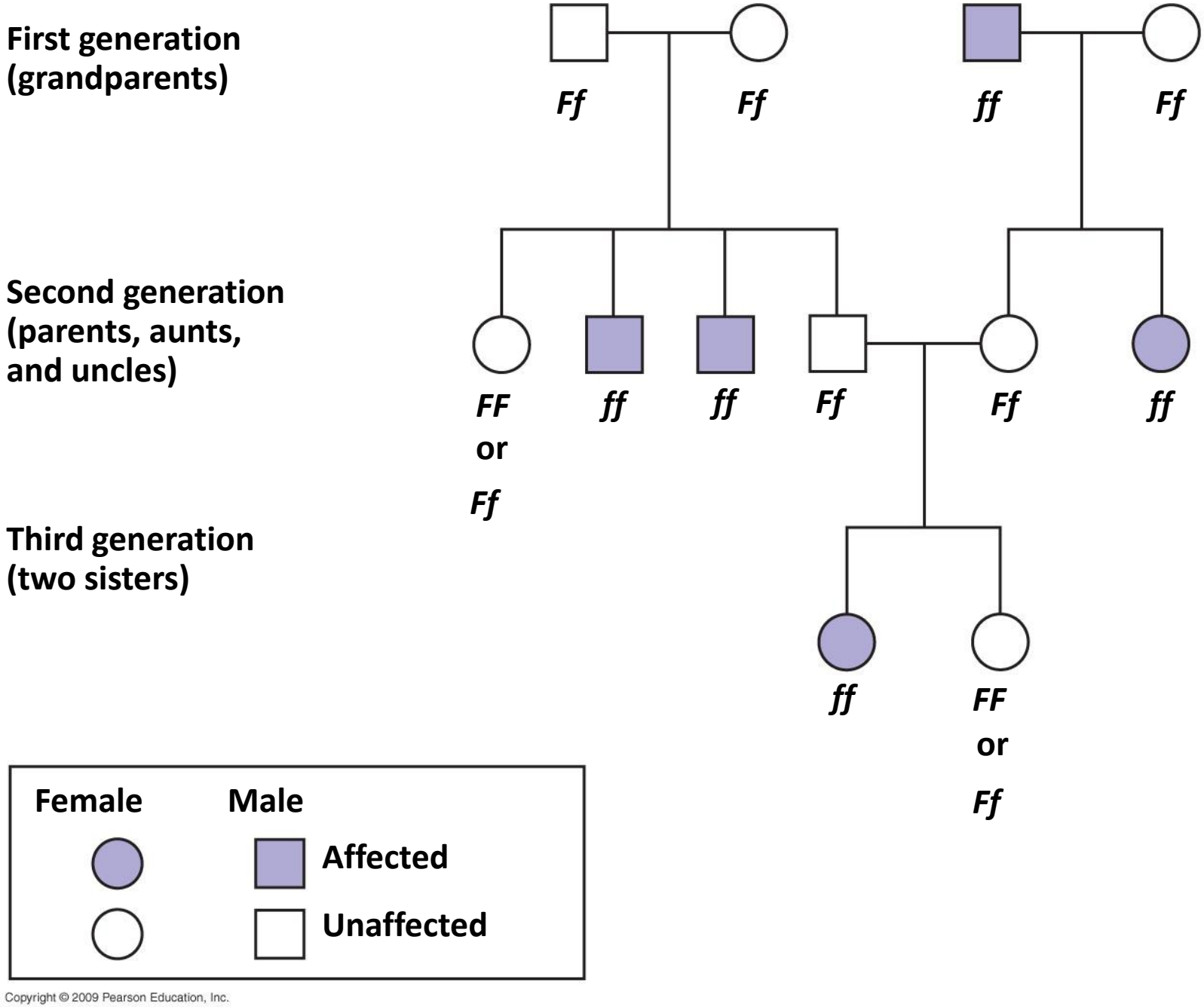
- The probability of a specific event is the number of ways that event can occur out of the total possible outcomes.
- **Rule of multiplication**
  - Multiply the probabilities of events that must occur together
- **Rule of addition**
  - Add probabilities of events that can happen in alternate ways



9.8 CONNECTION: Genetic traits in humans can be tracked through family pedigrees

- A **pedigree**
  - Shows the inheritance of a trait in a family through multiple generations
  - Demonstrates dominant or recessive inheritance
  - Can also be used to deduce genotypes of family members

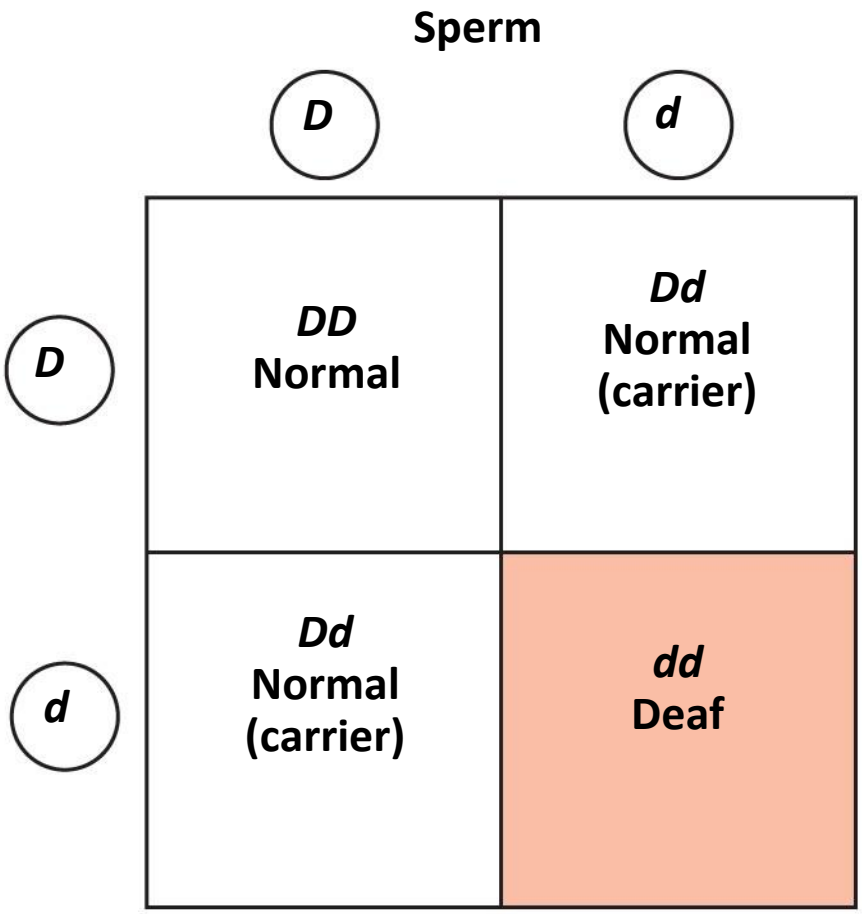
Dominant Traits	Recessive Traits
	
Freckles	No freckles
	
Widow's peak	Straight hairline
	
Free earlobe	Attached earlobe



Many inherited disorders in humans are controlled by a single gene

- Inherited human disorders show
  - Recessive inheritance
    - Two recessive alleles are needed to show disease
    - Heterozygous parents are carriers of the disease-causing allele
    - Probability of inheritance increases with inbreeding, mating between close relatives
  - Dominant inheritance
    - One dominant allele is needed to show disease
    - Dominant lethal alleles are usually eliminated from the population

Parents **Normal** ***Dd*** **Normal** ***Dd***



Offspring

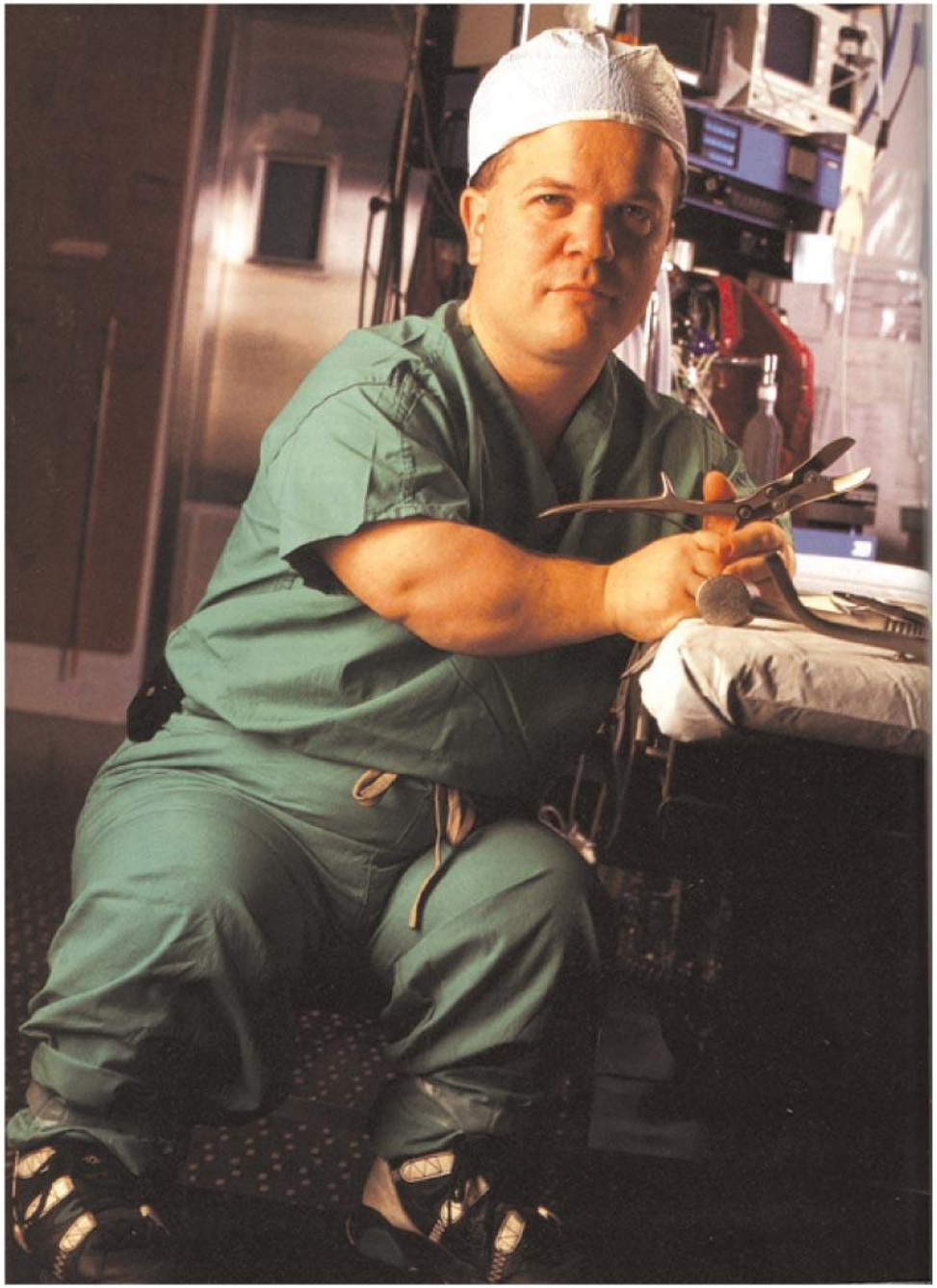


TABLE 9.9 SOME AUTOSOMAL DISORDERS IN HUMANS

Disorder	Major Symptoms	Incidence	Comments
Recessive disorders			
Albinism	Lack of pigment in skin, hair, and eyes	$\frac{1}{22,000}$	Prone to skin cancer
Cystic fibrosis	Excess mucus in lungs, digestive tract, liver; increased susceptibility to infections; death in early childhood unless treated	$\frac{1}{2,500}$ Caucasians	See Module 9.9
Galactosemia	Accumulation of galactose in tissues; mental retardation; eye and liver damage	$\frac{1}{100,000}$	Treated by eliminating galactose from diet
Phenylketonuria (PKU)	Accumulation of phenylalanine in blood; lack of normal skin pigment; mental retardation	$\frac{1}{10,000}$ in U.S. and Europe	See Module 9.10
Sickle-cell disease	Sickled red blood cells; damage to many tissues	$\frac{1}{400}$ African-Americans	See Module 9.13
Tay-Sachs disease	Lipid accumulation in brain cells; mental deficiency; blindness; death in childhood	$\frac{1}{3,500}$ Jews from central Europe	See Module 4.11
Dominant disorders			
Achondroplasia	Dwarfism	$\frac{1}{25,000}$	See Module 9.9
Alzheimer's disease (one type)	Mental deterioration; usually strikes late in life	Not known	
Huntington's disease	Mental deterioration and uncontrollable movements; strikes in middle age	$\frac{1}{25,000}$	See Module 9.9
Hypercholesterolemia	Excess cholesterol in blood; heart disease	$\frac{1}{500}$ are heterozygous	See Module 9.11

TABLE 9.9 SOME AUTOSOMAL DISORDERS IN HUMANS

Disorder	Major Symptoms	Incidence
Recessive disorders		
Albinism	Lack of pigment in skin, hair, and eyes	$\frac{1}{22,000}$
Cystic fibrosis	Excess mucus in lungs, digestive tract, liver; increased susceptibility to infections; death in early childhood unless treated	$\frac{1}{2,500}$ Caucasians
Galactosemia	Accumulation of galactose in tissues; mental retardation; eye and liver damage	$\frac{1}{100,000}$
Phenylketonuria (PKU)	Accumulation of phenylalanine in blood; lack of normal skin pigment; mental retardation	$\frac{1}{10,000}$ in U.S. and Europe
Sickle-cell disease	Sickled red blood cells; damage to many tissues	$\frac{1}{400}$ African-Americans
Tay-Sachs disease	Lipid accumulation in brain cells; mental deficiency; blindness; death in childhood	$\frac{1}{3,500}$ Jews from central Europe
Dominant disorders		
Achondroplasia	Dwarfism	$\frac{1}{25,000}$
Alzheimer's disease (one type)	Mental deterioration; usually strikes late in life	Not known
Huntington's disease	Mental deterioration and uncontrollable movements; strikes in middle age	$\frac{1}{25,000}$
Hypercholesterolemia	Excess cholesterol in blood; heart disease	$\frac{1}{500}$ are heterozygous

9.10 CONNECTION: New technologies can provide insight into one's genetic legacy

- Genetic testing of parents
- Fetal testing: biochemical and karyotype analyses
  - **Amniocentesis**
  - **Chorionic villus sampling**
- Maternal blood test
- Fetal imaging
  - Ultrasound
  - Fetoscopy
- Newborn screening