Biology and Society: Our Longest-Running Genetic Experiment: Dogs

- People have selected and mated dogs with preferred traits for more than 15,000 years.
  - Over thousands of years, such genetic tinkering has led to the incredible variety of body types and behaviors in dogs today.
  - The biological principles underlying genetics have only recently been understood.

In an Abbey Garden

- Mendel studied garden peas because they
  - Are easy to grow
  - Come in many readily distinguishable varieties
  - Are easily manipulated
  - Can self-fertilize! Very important!

Figure 9.2

Flower Anatomy

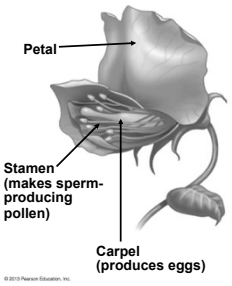


Figure 9.3-3

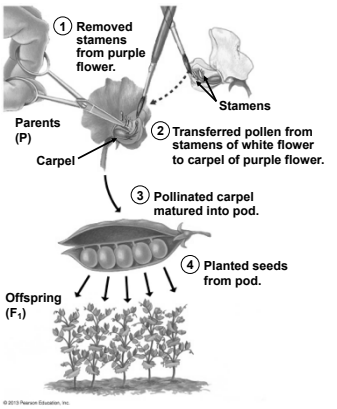
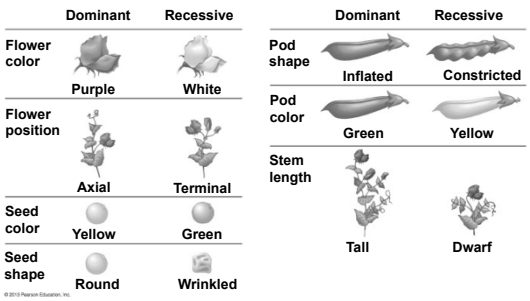


Figure 9.4

The 7characters studied by Mendel



In an Abbey Garden

- A **character** is a heritable feature that varies among individuals.
  - Ex: Hair Color, Eye Color, Fresckles
- A **trait** is a variant of a character.
  - Ex: Blonde/Red/Brown/Black, Blue/Brown/Hazel, Present/Absent
- Each of the characters Mendel studied occurred in two distinct traits.

In an Abbey Garden

- Mendel
  - created purebred varieties of plants

    and
  - crossed two different purebred varieties.

In an Abbey Garden

- **Hybrids** are the offspring of two different purebred varieties.
  - The parental plants are the **P generation**.
  - Their hybrid offspring are the **F_1 generation**.
  - A cross of the F_1 plants forms the **F_2 generation**.

Mendel's Law of Segregation

- Mendel performed many experiments.
- He tracked the inheritance of characters that occur as two alternative traits.

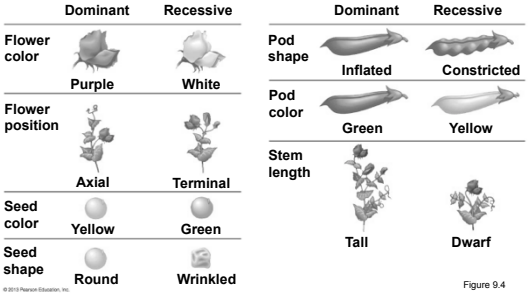


Figure 9.4

Mendel's Law of Segregation

Which characters are paired?

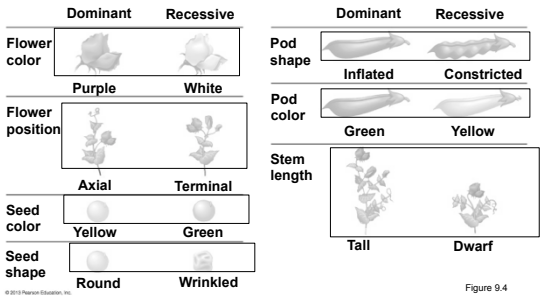
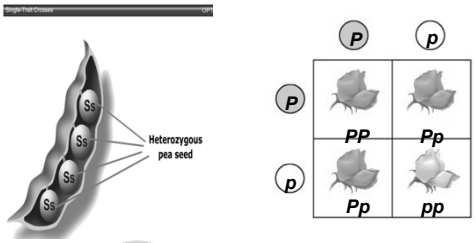


Figure 9.4

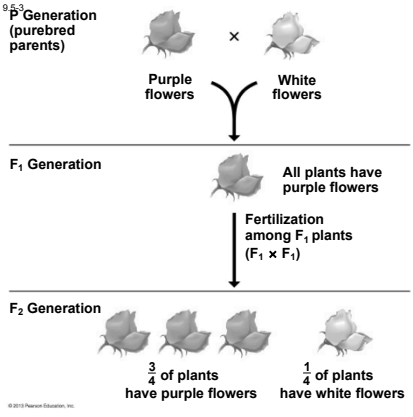
Monohybrid Crosses

– A **monohybrid cross** is a cross between purebred parent plants that differ in only one character.



PLAY Blast Animation: Single-Trait Crosses

Figure 9.5-3



Monohybrid Crosses

- Mendel developed four hypotheses from the monohybrid cross, listed here using modern terminology (including "gene" instead of "heritable factor").

1. The alternative versions of genes are called **alleles.**
2. For each inherited character, an organism inherits two alleles, one from each parent.
   - An organism is **homozygous** for that gene if both alleles are identical. PP or pp
   - An organism is **heterozygous** for that gene if the alleles are different. Pp
3. If two alleles of an inherited pair differ,
   - Then one determines the organism's appearance and is called the **dominant allele** and P
   - The other has no noticeable effect on the organism's appearance and is called the **recessive allele**. p
4. Gametes carry only one allele for each inherited character.
   - The two alleles for a character segregate (separate) from each other during the production of gametes.
   - This statement is called the **law of segregation**.

Monohybrid Crosses

Figure 9.6

- Do Mendel's hypotheses account for the 3:1 ratio he observed in the F_2 generation? Yes
- A **Punnett square** highlights
  - The four possible combinations of gametes and
  - The four possible offspring in the F_2 generation.

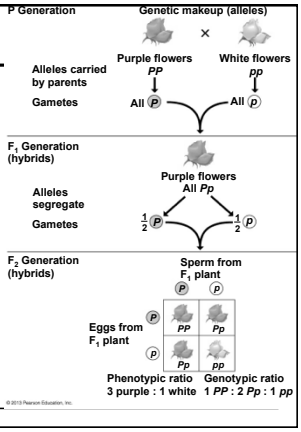


Figure 9.6a

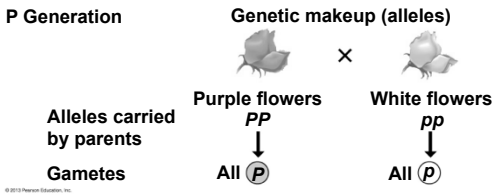


Figure 9.6b

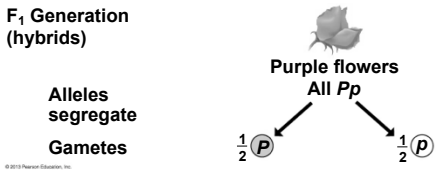
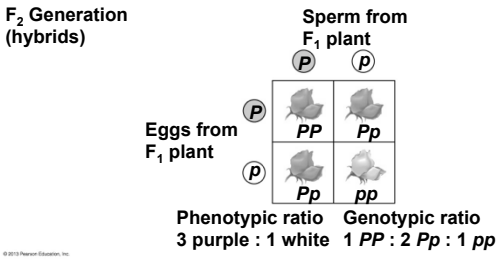
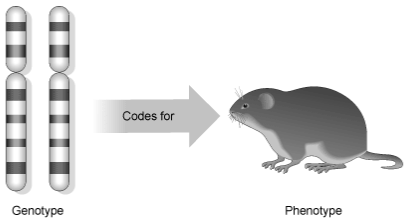


Figure 9.6c



Monohybrid Crosses

- Geneticists distinguish between an organism's physical appearance and its genetic makeup.
  - An organism's physical appearance is its **phenotype**.
  - An organism's genetic makeup is its **genotype**.



Genetic Alleles and Homologous Chromosomes

- A gene **locus** is a specific location of a gene along a chromosome.
- Homologous chromosomes have alleles (alternate versions) of a gene at the same locus.

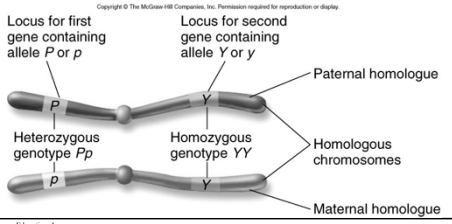
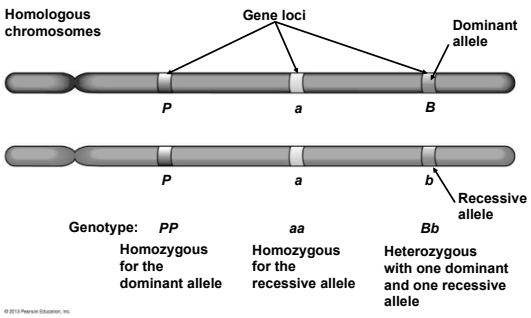
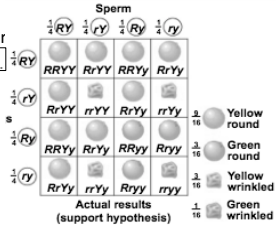


Figure 9.7

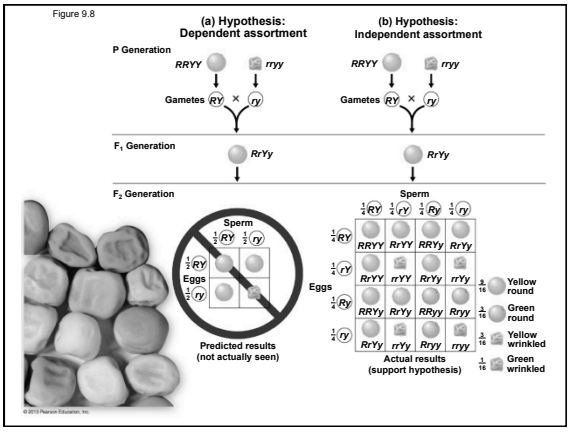


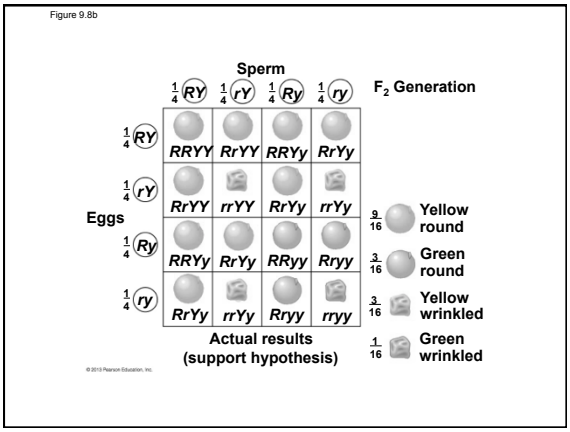
Mendel's Law of Independent Assortment

- A **dihybrid cross** is the mating of parental varieties differing in two characters.
- What would result from a dihybrid cross? Two hypotheses are possible:
  1. Dependent assortment or
  2. Independent assortment.



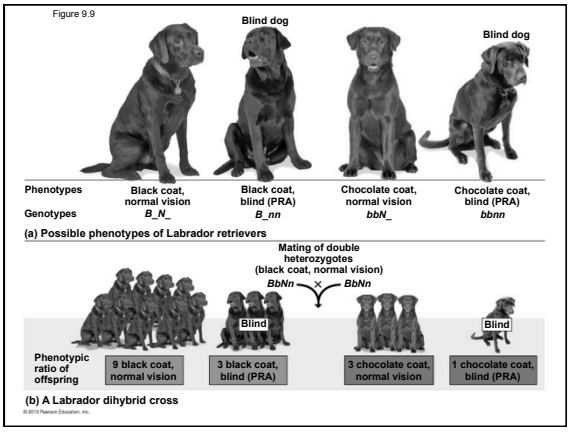
PLAY Blast Animation: Two-Trait Crosses

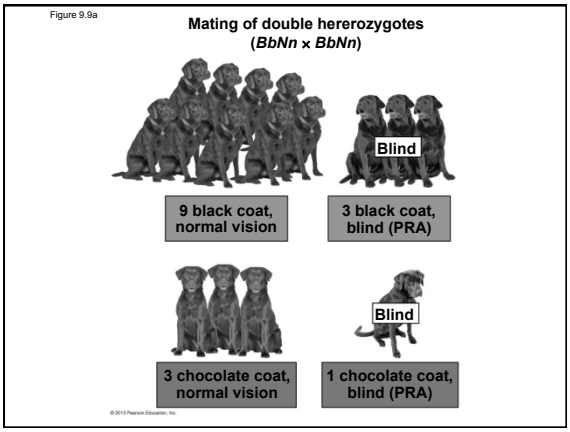




Mendel's Law of Independent Assortment

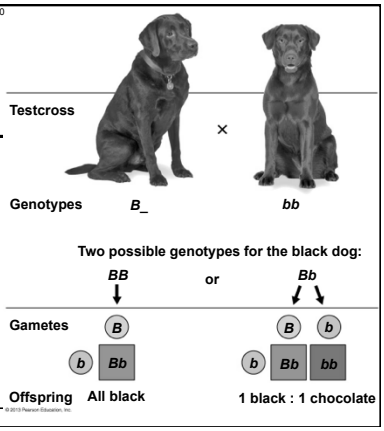
- Mendel's dihybrid cross supported the hypothesis that each pair of alleles segregates **independently** of the other pairs during gamete formation.
- Thus, the inheritance of one character has no effect on the inheritance of another.
- This is called Mendel's **law of independent assortment**.
- Independent assortment is also seen in two hereditary characters in Labrador retrievers.

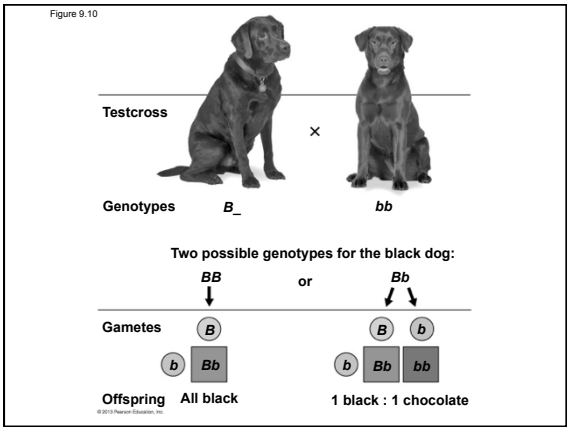




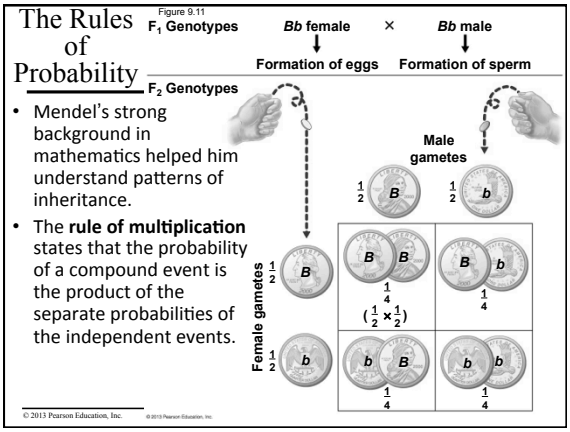
Using a Testcross to Determine an Unknown Genotype

- A **testcross** is a mating between
  - an individual of dominant phenotype (but unknown genotype) and
  - a homozygous recessive individual.





The Rules of Probability

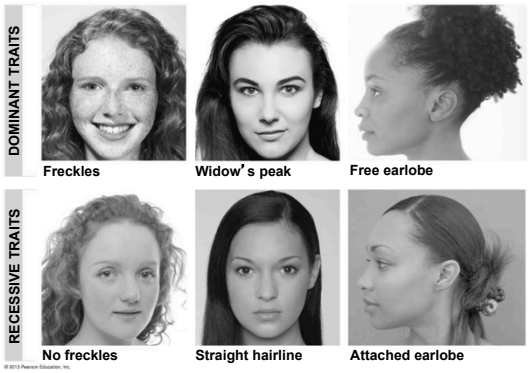


- Mendel's strong background in mathematics helped him understand patterns of inheritance.
- The **rule of multiplication** states that the probability of a compound event is the product of the separate probabilities of the independent events.

Family Pedigrees

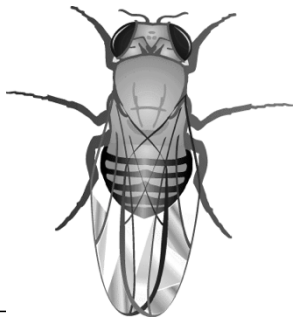
– Mendel's principles apply to the inheritance of many human traits.

Figure 9.12



Family Pedigrees

- Dominant traits are not necessarily
  - normal or
  - more common.
- **Wild-type traits** are
  - those seen most often in nature and
  - not necessarily specified by dominant alleles.



Recessive Disorders

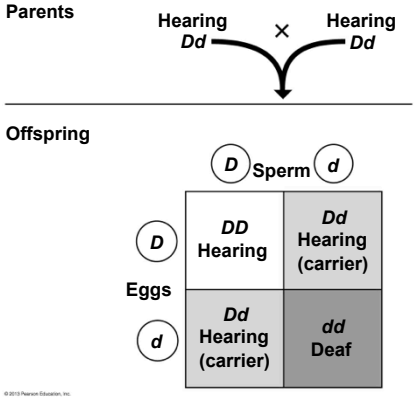
- Most human genetic disorders are recessive.
- Individuals who have the recessive allele but appear normal are **carriers** of the disorder.

Table 9.1

Table 9.1 Some Autosomal Disorders in People

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	$\frac{1}{1,800}$ European Americans
Phenylketonuria (PKU)	Accumulation of phenylalanine in blood; lack of normal skin pigment; mental retardation unless treated	$\frac{1}{100,000}$ in U.S. and Europe
Sickle-cell disease	Sickled red blood cells; damage to many tissues	$\frac{1}{500}$ African Americans
Tay Sachs disease	Lipid accumulation in brain cells; mental deficiency; blindness; death in childhood	$\frac{1}{3,600}$ European Jews
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	$\frac{1}{500}$

Figure 9.14



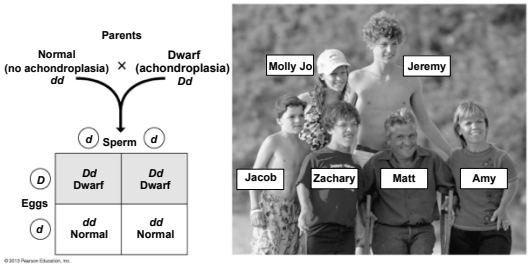
Recessive Disorders

- **Cystic fibrosis** is
  - the most common lethal genetic disease in the United States and
  - caused by a recessive allele carried by about one in 31 Americans.
- Prolonged geographic isolation of certain populations can lead to inbreeding, the mating of close relatives.
  - Inbreeding increases the chance of offspring that are homozygous for a harmful recessive trait.

Dominant Disorders

- Some human genetic disorders are dominant.
  - **Achondroplasia** is a form of dwarfism.
    - The homozygous dominant genotype causes death of the embryo.
    - Thus, only heterozygotes have this disorder.
  - **Huntington's disease**, which leads to degeneration of the nervous system, does not usually begin until middle age.

Figure 9.16



The Process of Science: What Is the Genetic Basis of Coat Variation in Dogs?

- **Observation**: Dogs come in a wide variety of physical types.
- **Question**: What is the genetic basis for canine coats?
- **Hypothesis**: A comparison of genes of a wide variety of dogs with different coats would identify the genes responsible.

The Process of Science:
What Is the Genetic Basis of Coat Variation in Dogs?

- **Prediction**: Mutations in just a few genes account for the coat appearance.
- **Experiment**: Compared DNA sequences of 622 dogs from dozens of breeds.
- **Results**: Three genes in different combinations produced seven different coat appearances, from very short hair to full, thick, wired hair.

Figure 9.18-2

VARIATIONS ON MENDEL'S LAWS

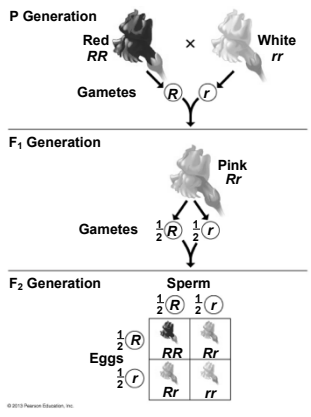
- Some patterns of genetic inheritance are not explained by Mendel's laws.
  1. **Incomplete Dominance:** F_1 hybrids have an appearance **between** the phenotypes of the two parents.

     I remember Incomplete Dominance in the form of an example like so:
     RED Flower x WHITE Flower ---> PINK Flower

  2. **Codominance**: F_1 hybrids have an appearancein which **both** the phenotypes of the two parents appear.

     I remember codominance in the form of an example like so:
     red x white ---> r d & w h t s o t d

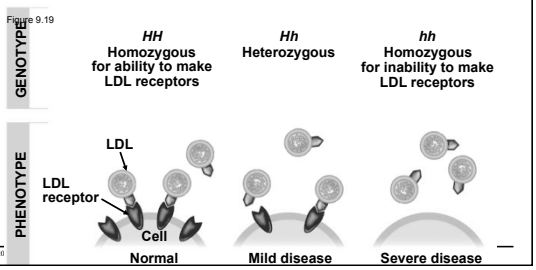
Figure 9.18-3



Incomplete Dominance in Plants and People

- **Hypercholesterolemia**
  - Is characterized by dangerously high levels of cholesterol in the blood.
  - heterozygotes have blood cholesterol levels about twice normal, and
  - homozygotes have about five times the normal amount of blood cholesterol and may have heart attacks as early as age 2.

Figure 9.19



ABO Blood Groups: An Example of Multiple Alleles and Codominance

Figure 9.20

- The **ABO blood groups** in humans are an example of multiple alleles.
- The immune system produces blood proteins called antibodies that bind specifically to foreign carbohydrates.

Blood Group (Phenotype)	Genotypes	Red Blood Cells	Antibodies Present in Blood
A	I^AI^A or I^Ai	Carbohydrate A	Anti-B
B	I^BI^B or I^Bi	Carbohydrate B	Anti-A
AB	I^AI^B		—
O	ii		Anti-A Anti-B

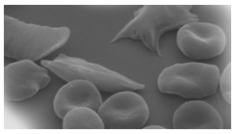
ABO Blood Groups: An Example of Multiple Alleles and Codominance

- If a donor's blood cells have a carbohydrate (A or B) that is foreign to the recipient, the blood cells may clump together, potentially killing the recipient.
- The clumping reaction is the basis of a blood-typing lab test.
- The human blood type alleles I^A and I^B are **codominant**, meaning that both alleles are expressed in heterozygous individuals who have type AB blood.

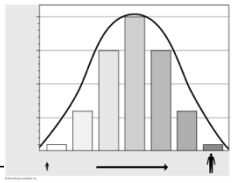
Figure 9.18-2
Page 1 of 2

VARIATIONS ON MENDEL'S LAWS

- Some patterns of genetic inheritance are not explained by Mendel's laws.
  3. **Pleiotropy** is when one gene influences several characters.
     - **EX: Sickle-cell disease**



  4. **Polygenic inheritance** is the additive effects of two or more genes on a single phenotype.



Pleiotropy and Sickle-Cell Disease

– **Pleiotropy** is when one gene influences several characters.
  - **EX: Sickle-cell disease**
    – Results in abnormal hemoglobin proteins, and
    – Causes disk-shaped red blood cells to deform into a sickle shape with jagged edges.

Figure 9.21

Individual homozygous for sickle-cell allele

↓

Sickle-cell (abnormal) hemoglobin

↓

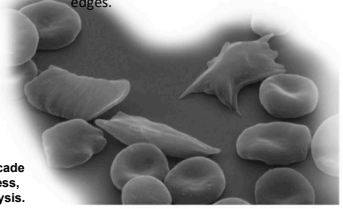
Abnormal hemoglobin crystallizes into long flexible chains, causing red blood cells to become sickle-shaped.

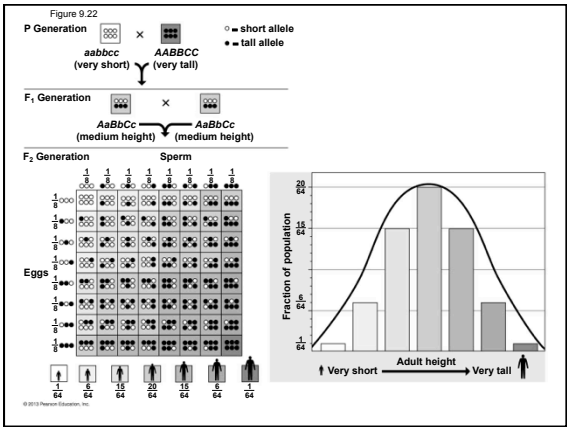
↓

Sickled cells can lead to a cascade of symptoms, such as weakness, pain, organ damage, and paralysis.

Pleiotropy and Sickle-Cell Disease

- **Pleiotropy** is when one gene influences several characters.
- **Sickle-cell disease**
  – exhibits pleiotropy,
  – results in abnormal hemoglobin proteins,
  – causes disk-shaped red blood cells to deform into a sickle shape with jagged edges.





The Role of Environment

- Many human characters result from a combination of
  - heredity and
  - environment.
- Only genetic influences are inherited.