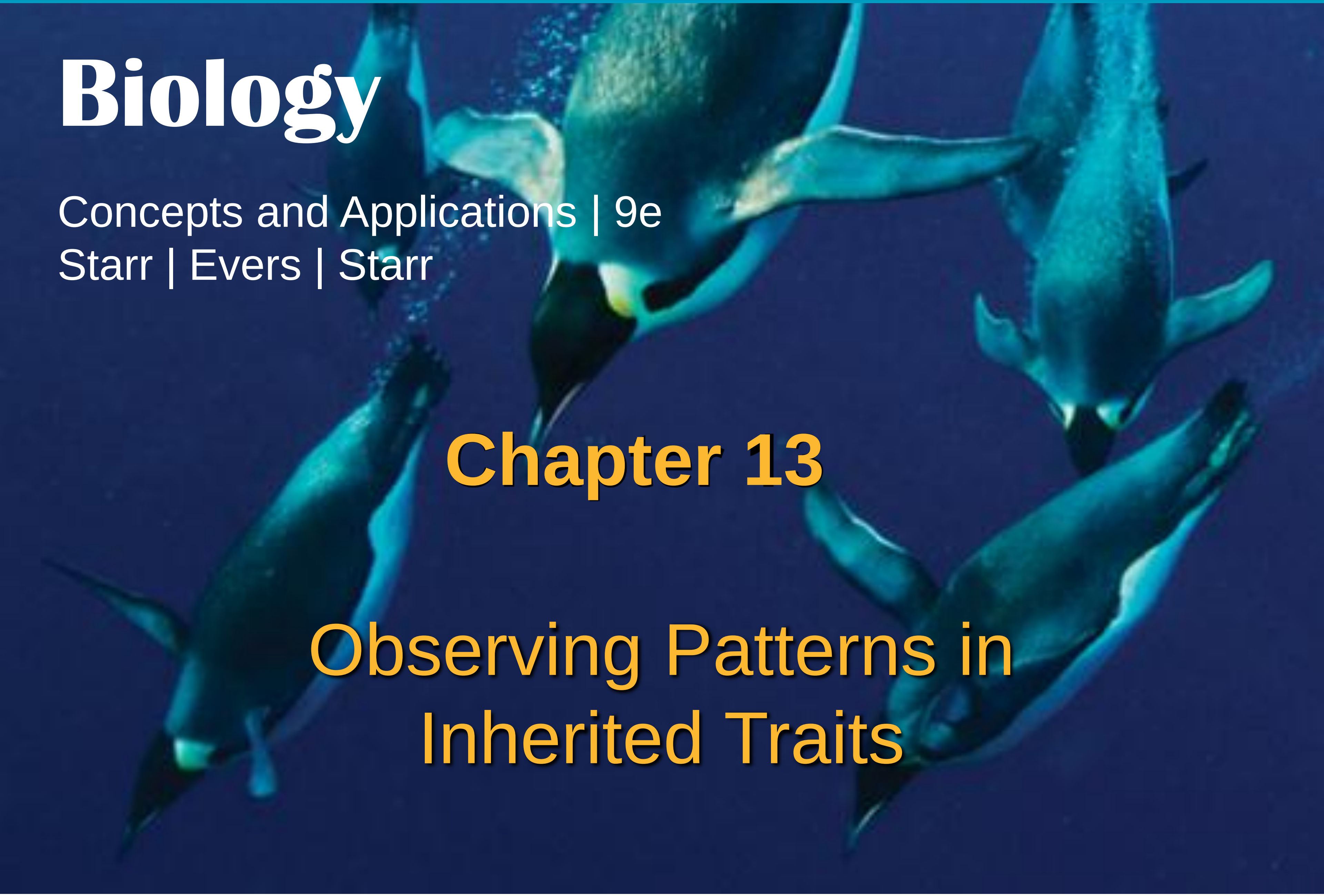

Biology

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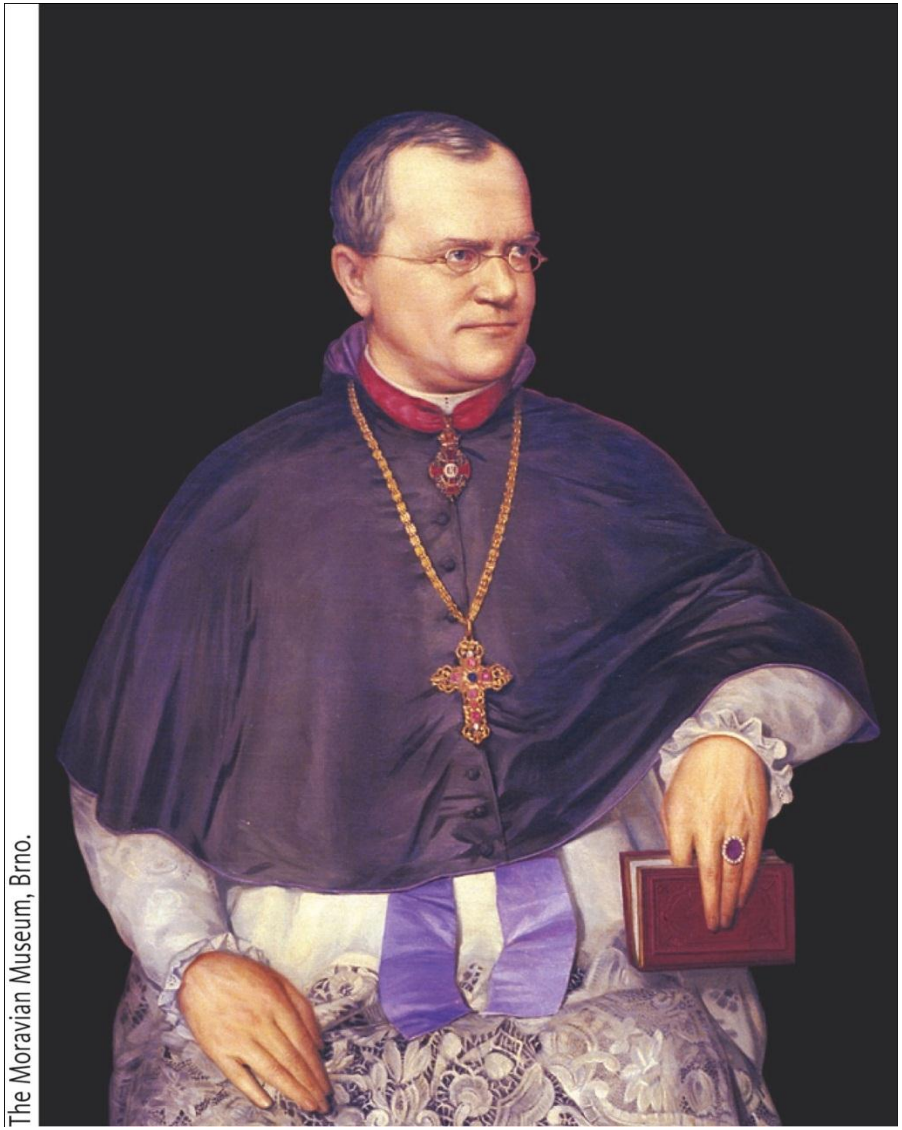
Chapter 13

Observing Patterns in Inherited Traits

13.1 How Do Alleles Contribute to Traits?

- Blending inheritance
  - 19th century idea
  - Failed to explain how traits disappear over several generations and then reappear unaltered generations later
  - Charles Darwin did not accept this idea

Mendel's Experiments



Mendel's Experiments (cont'd.)

- Gregor Mendel
  - Started breeding thousands of pea plants
  - Kept detailed record of how traits passed from one generation to the next
  - Began to formulate how inheritance works

Mendel's Experiments (cont'd.)

- Garden pea plant is self-fertilizing
  - The flowers produce male and female gametes
- The experiments
  - Controlled the pairings between individuals with specific traits and observed traits of their offspring
  - Cross fertilized plants and collected seeds
  - Recorded traits of new pea plants

Animation: Crossing garden pea plants

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Mendel's Experiments (cont'd.)

- The experiments (cont'd.)
  - Started with garden pea plants that "bred true" for a particular trait – the trait stayed the same generation after generation
  - Cross-fertilized pea plants with different traits and offspring appeared in predictable patterns
  - Concluded that hereditary information is passed in discrete units

Inheritance in Modern Terms

- Individuals share certain traits because their chromosomes carry the same genes
- The DNA sequence of each gene occurs at a specific location
- The location of a gene on a particular chromosome is called a locus

Animation: Genetic terms

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Inheritance in Modern Terms (cont'd.)

- An individual carrying identical alleles for a gene are *homozygous*
- An individual carrying two different alleles of a gene is *heterozygous*
- Hybrids are *heterozygous* offspring of a cross between individuals that breed true for different forms of a trait

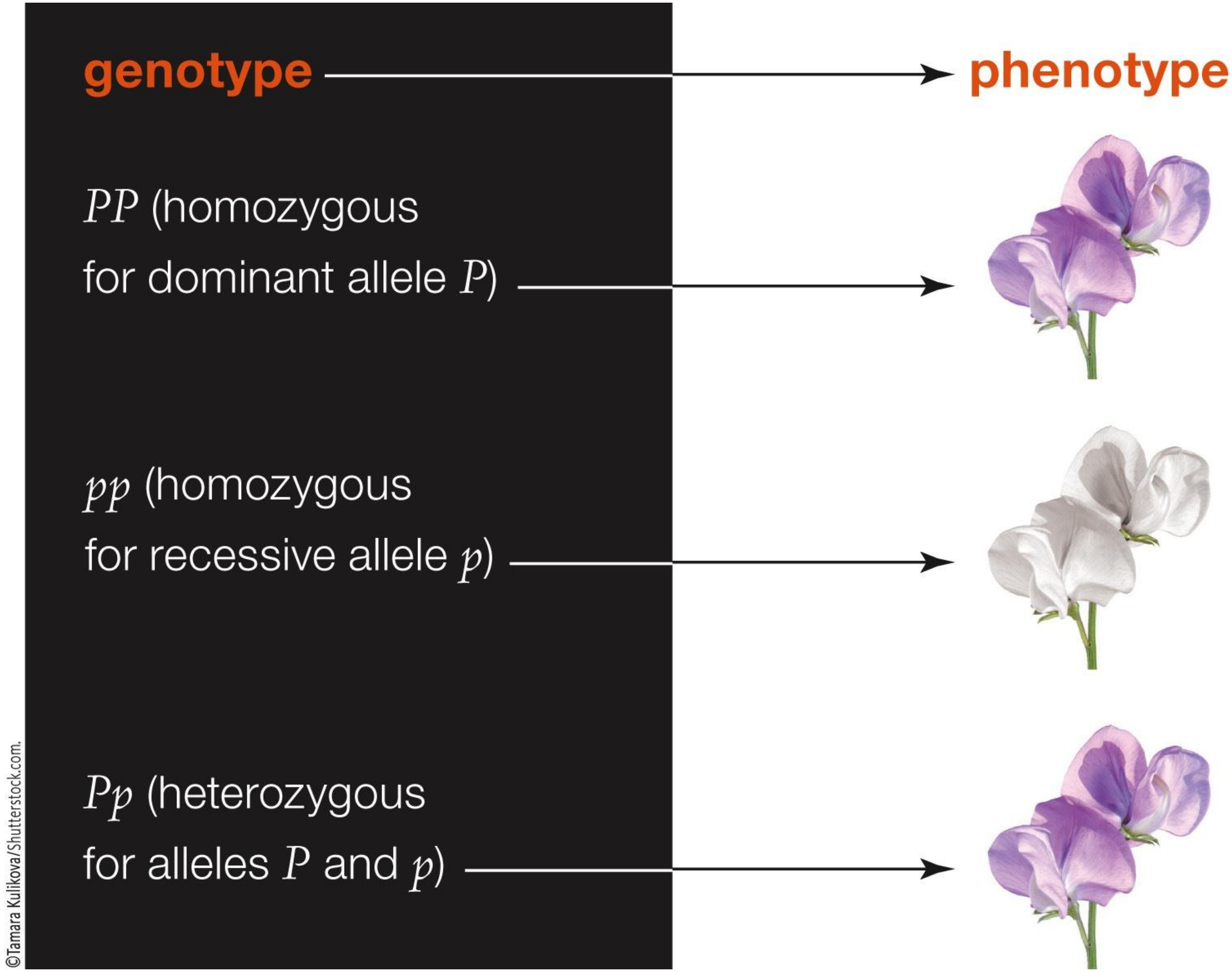
Inheritance in Modern Terms (cont’d.)

- The particular set of alleles that an individual carries is their *genotype*
- The observable traits, such as flower color, make up an individual’s *phenotype*

Inheritance in Modern Terms (cont'd.)

- An allele is *dominant* when its effect masks that of a recessive allele paired with it
  - A dominant allele is represented by italic capital letters such as (*A*)
  - A recessive allele is represented by italic lowercase letters such as (*a*)

Inheritance in Modern Terms (cont'd.)



13.2 How Are Alleles Distributed Into Gametes?

- A homozygous pea plant with two alleles (*PP*) has purple flowers, and one with two alleles (*pp*) has white flowers
- If these homozygous plants are crossed (*PP* × *pp*), all offspring will be heterozygous

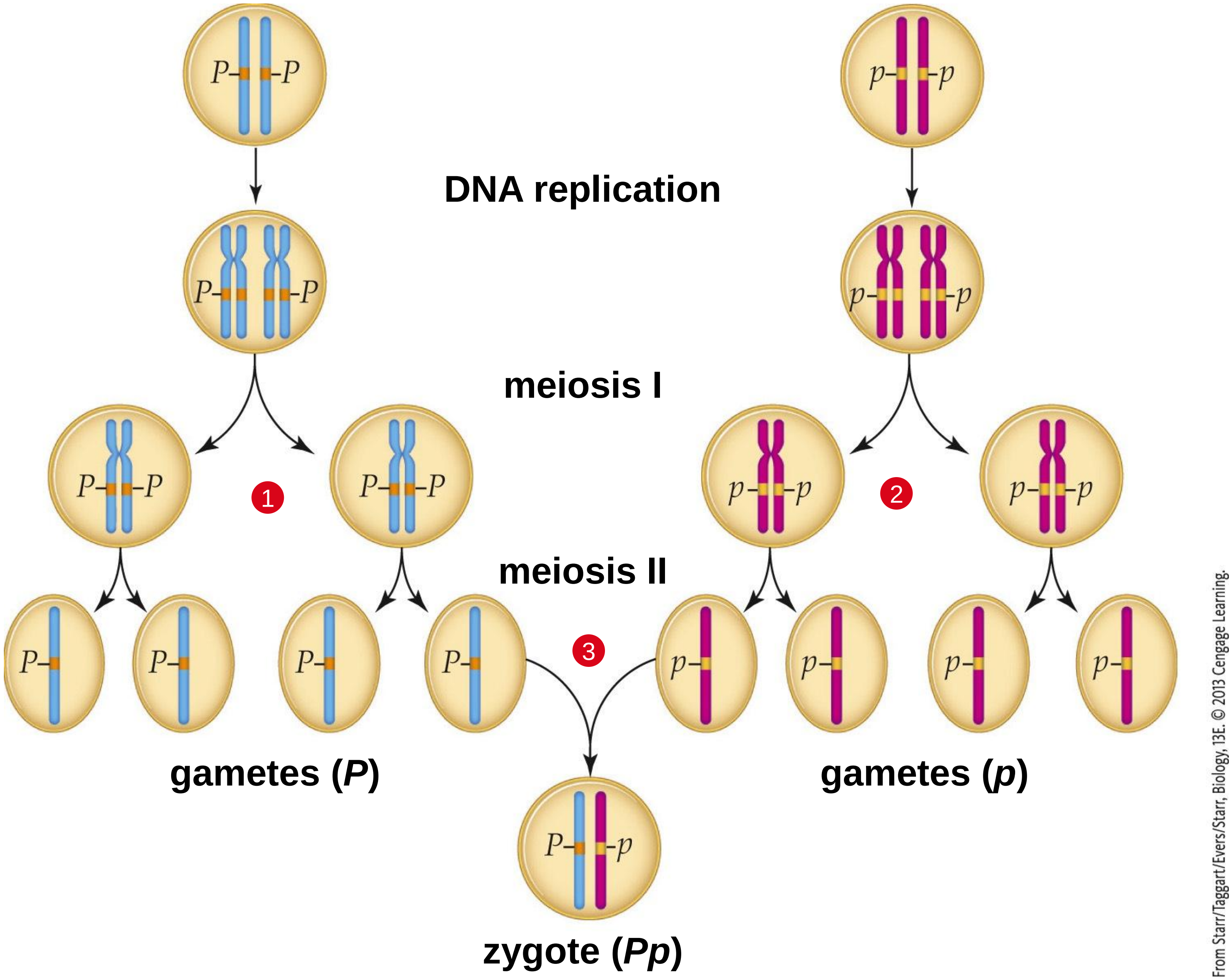
How Are Alleles Distributed Into Gametes? (cont'd.)

- The allele for purple (*P*) is dominant over the allele for white (*p*)
- Therefore, the heterozygote (*Pp*) will have purple flowers

How Are Alleles Distributed Into Gametes? (cont'd.)

- When homozygous dominant and homozygous recessive plants are crossed (*PP* × *pp*), only one outcome is possible
- All first generation (F_1) offspring will be heterozygous
  - Genotype = *Pp*
  - Phenotype = purple

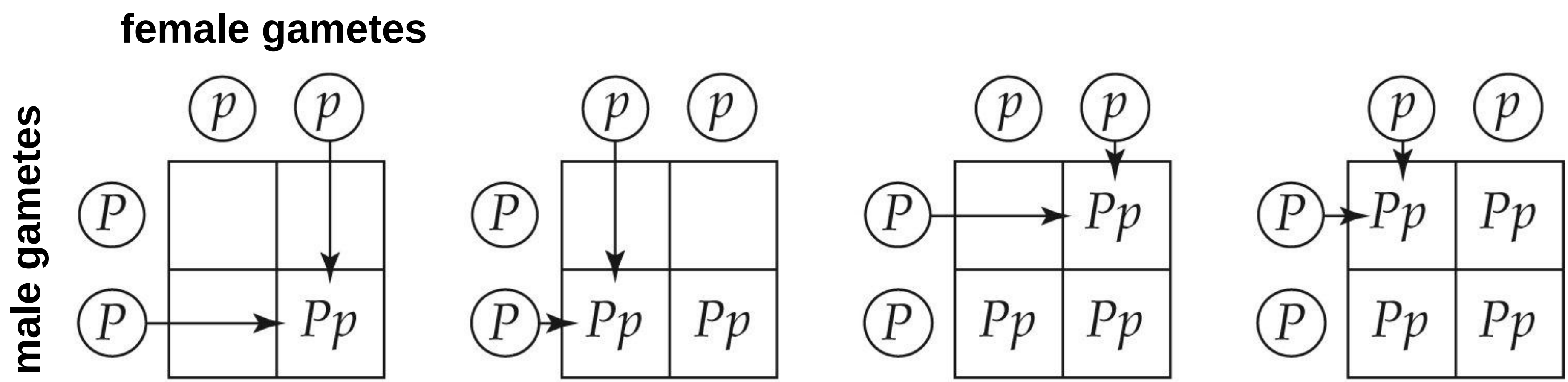
How Are Alleles Distributed Into Gametes? (cont'd.)



How Are Alleles Distributed Into Gametes? (cont'd.)

- Punnett square
  - A grid used to predict the genetic and phenotypic outcome of a cross

How Are Alleles Distributed Into Gametes? (cont'd.)



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How Are Alleles Distributed Into Gametes? (cont'd.)

- Testcross
  - Breeding experiments used to determine genotype
  - An individual that has a dominant trait (but an unknown genotype) is crossed with one that is homozygous recessive

How Are Alleles Distributed Into Gametes? (cont’d.)

- Testcross (cont’d.)
  - If all offspring have dominant trait, than the unknown genotype is homozygous for dominant allele
  - If any offspring have recessive trait, then it is heterozygous

How Are Alleles Distributed Into Gametes? (cont'd.)

- Monohybrid cross
  - Breeding experiment in which individuals identically heterozygous for one gene are crossed
  - Frequency of traits among offspring offers information about the dominance relationship between the alleles
  - First generation = F1
  - Second generation = F2

How Are Alleles Distributed Into Gametes? (cont'd.)

TABLE 13.1

Mendel's Seven Pea Plant Traits

Trait	Dominant Form	Recessive Form
Seed Shape	Round	Wrinkled
Seed Color	Yellow	Green
Pod Texture	Smooth	Wrinkled
Pod Color	Green	Yellow
Flower Color	Purple	White
Flower Position	Along Stem	At Tip
Stem Length	Tall	Short

How Are Alleles Distributed Into Gametes? (cont'd.)

- In a monohybrid cross between two *Pp* plants (*Pp* × *Pp*), the two types of gametes can meet in four possible ways:
  - Sperm *P* meets egg *P* → zygote genotype *PP*
  - Sperm *P* meets egg *p* → zygote genotype *Pp*
  - Sperm *p* meets egg *P* → zygote genotype *Pp*
  - Sperm *p* meets egg *p* → zygote genotype *pp*

How Are Alleles Distributed Into Gametes? (cont'd.)

- The probability that second-generation (F_2) offspring of (*Pp* × *Pp)* will have purple flowers
  - A ration of 3 purple to 1 white, or 3:1

Animation: Monohybrid cross

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How Are Alleles Distributed Into Gametes? (cont'd.)

- Law of segregation
  - The 3:1 phenotype ratios in F_2 offspring of monohybrid crosses became the basis of Mendel's law of segregation
  - Diploid cells carry pairs of genes on each pair of homologous chromosomes.
  - The two genes of each pair are separated from each other during meiosis so that they end up on different gametes

13.3 How Are Gene Pairs Distributed Into Gametes?

- When homologous chromosomes separate during meiosis, either one of the pair can end up in a particular nucleus
- Gene pairs on one chromosome get sorted into gametes independently of gene pairs on other chromosomes
- Punnett squares can be used to predict inheritance

How Are Gene Pairs Distributed Into Gametes? (cont'd.)

- *Dihybrid cross*
  - Individuals identically heterozygous for alleles of two genes (dihybrids) are crossed, and the traits of the offspring are observed
  - Frequency of traits among the offspring offers information about the dominance relationships between the paired alleles

How Are Gene Pairs Distributed Into Gametes? (cont'd.)

- One parent plant that breeds true for purple flowers and tall stems (*PPTT*) is crossed with one that breeds true for white flowers and short stems (*pptt*)
- Each plant makes only one type of gamete (*PT* or *pt*)
- All F_1 offspring will be dihybrids (*PpTt*) and have purple flowers and tall stems

How Are Gene Pairs Distributed Into Gametes? (cont'd.)

- The result of two F_1 plants crossing: a *dihybrid cross (PpTt × PpTt)*
- Four types of gametes can combine in sixteen possible ways

How Are Gene Pairs Distributed Into Gametes? (cont'd.)

- In F_2 plants, four phenotypes result in a ratio of 9:3:3:1
  - Nine tall with purple flowers
  - Three short with purple flowers
  - Three tall with white flowers
  - One short with white flowers

Animation: Dihybrid cross

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How Are Gene Pairs Distributed Into Gametes? (cont'd.)

- Law of independent assortment
  - Mendel discovered the 9:3:3:1 ratio in his dihybrid experiments
    - Each trait still kept its individual 3:1 ratio
    - Each trait sorted into gametes independently of other traits
  - During meiosis, members of a pair of genes on homologous chromosomes get distributed into gametes *independently* of other gene pairs

Contribution of Crossovers

- How two genes get sorted into gametes depends on if they are found on same chromosome
  - Random assortment
  - Genes on one chromosome assort into gametes independent of genes on other chromosomes

ANIMATION: Independent assortment

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Contribution of Crossovers (cont'd.)

- *Linkage* group – all genes on a chromosome
  - Genes that are far apart on a chromosome tend to assort into gametes independently
  - Genes very close together on a chromosome are *linked*
    - They do not assort independently because crossing over rarely happens between them

13.4 Are All Genes Inherited in a Mendelian Pattern?

- Simple dominance
  - A dominant allele fully masks the expression of a recessive one
- Other patterns of inheritance are not so simple:
  - Codominance
  - Incomplete dominance
  - Epistasis
  - Pleiotropy

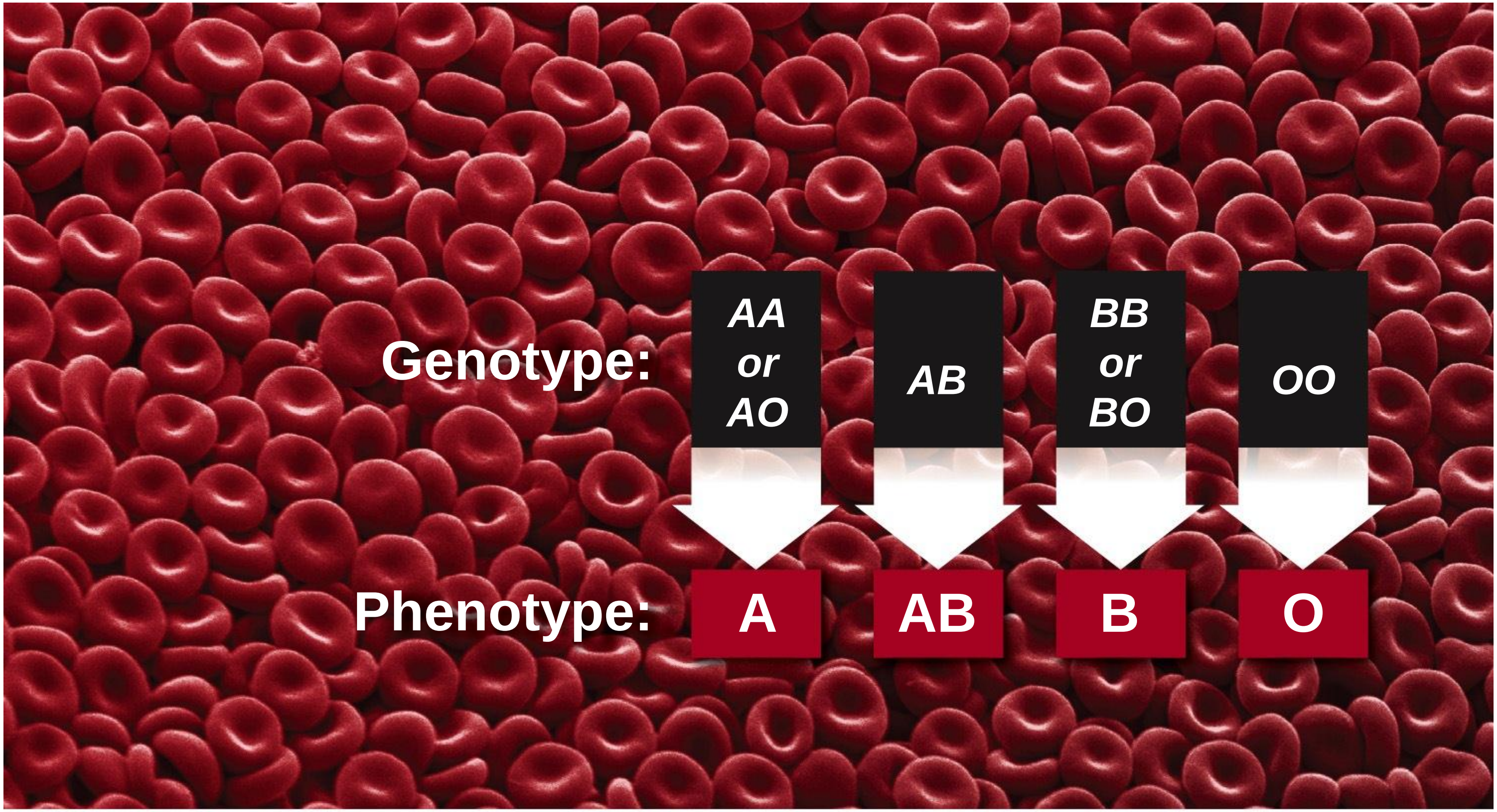
Are All Genes Inherited in a Mendelian Pattern? (cont'd.)

- Codominance
  - Two alleles that are both fully expressed in heterozygous individuals
  - Multiple allele systems – gene for which three or more alleles persist in a population
  - Example: an ABO gene for blood type

Are All Genes Inherited in a Mendelian Pattern? (cont'd.)

- Which two of the three alleles of the *ABO* gene you have determines your blood type
  - The *A* and the *B* allele are codominant when paired
    - Genotype *AB* = blood type AB
  - The *O* allele is recessive when paired with either *A* or *B*
    - Genotype *AA or AO* = blood type A
    - Genotype *BB or BO*= blood type B
    - Genotype *OO* = blood type O

Are All Genes Inherited in a Mendelian Pattern? (cont'd.)



photo, Annie Cavanagh/Wellcome Images; art, © Cengage Learning

Are All Genes Inherited in a Mendelian Pattern? (cont'd.)

- Incomplete dominance
  - One allele is not fully dominant over another
  - The heterozygous phenotype is between the two homozygous phenotypes

Are All Genes Inherited in a Mendelian Pattern? (cont'd.)

- In snapdragons, one allele (*R*) encodes an enzyme that makes a red pigment, and allele (*r*) makes no pigment
  - *RR* = red; *Rr* = pink; *rr* = white
- A cross between red and white (*RR* × *rr*) yields pink (*Rr*)
- A cross between two pink (*Rr* × *Rr*) yields red, pink, and white in a 1:2:1 ratio

ANIMATION: Incomplete dominance

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Are All Genes Inherited in a Mendelian Pattern? (cont'd.)

- Epistasis: the effect in which a trait is influenced by the products of multiple genes
  - Example: Fur color in dogs

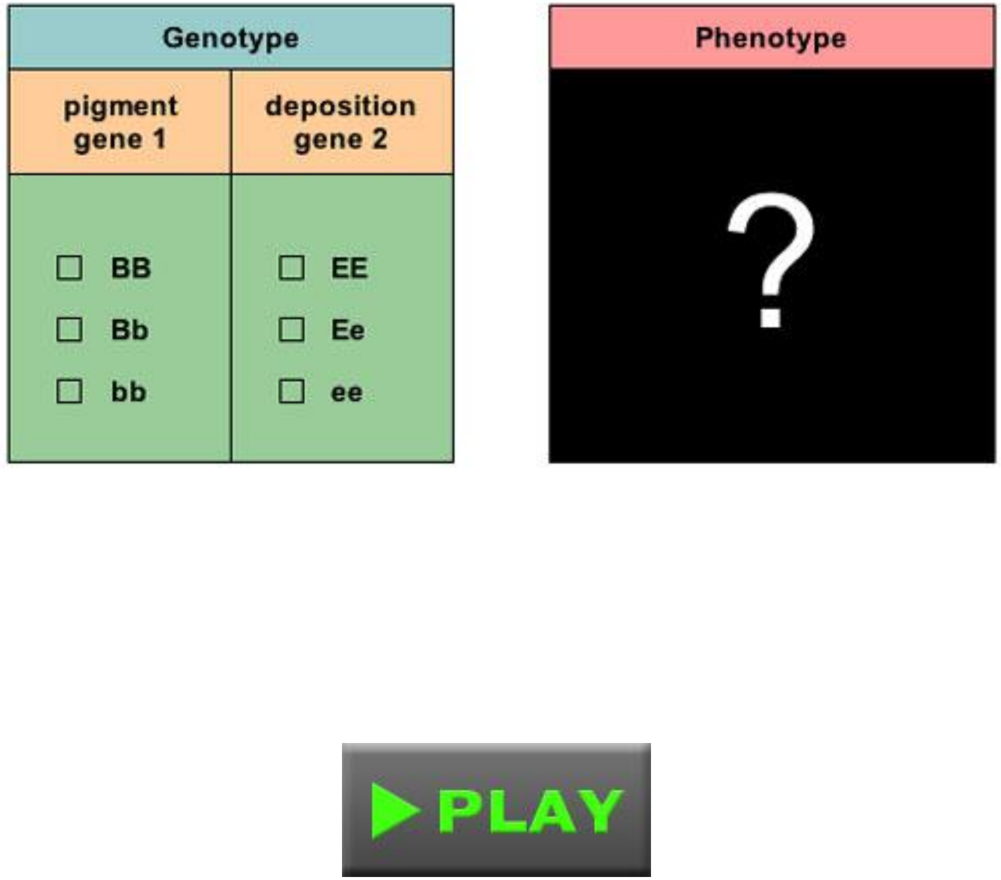
Are All Genes Inherited in a Mendelian Pattern? (cont'd.)

- A dominant allele (*B*) specifies black fur, and its recessive partner (*b*) specifies brown fur
- A dominant allele of a different gene (*E*) causes color to be deposited in fur and its recessive partner (*e*) reduces color

Are All Genes Inherited in a Mendelian Pattern? (cont'd.)

- A dog with an *E* and a *B* allele has black fur
- A dog with an *E* allele and homozygous for b is brown
- A dog homozygous for the *e* allele has yellow fur regardless of its *B* or *b* alleles

Animation: Dog Color



Are All Genes Inherited in a Mendelian Pattern? (cont'd.)

- Pleiotropy
  - A gene whose product influences multiple traits
  - Mutations in pleiotropic genes are associated with complex genetic disorders
    - Sickle-cell anaemia
    - Cystic fibrosis
    - Marfan syndrome

Are All Genes Inherited in a Mendelian Pattern? (cont'd.)


Courtesy of The Family of Haris Charalambous and the University of Toledo.

13.5 Does The Environment Affect Phenotype?

- Epigenetic research is revealing that environment can influence phenotype
- Some examples of environmental effects
  - Seasonal changes in coat color
  - Effect of altitude on yarrow
  - Alternative phenotypes in water fleas
  - Psychiatric disorders

Animation: Coat color in the Himalayan rabbit

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13.6 Do All Traits Occur in Distinct Forms?

- Phenotype often results from complex interactions among gene products and the environment
- Many traits show a continuous range of variation

Do All Traits Occur in Distinct Forms? (cont'd.)

- Short tandem repeats
  - Some genes have regions of DNA in which a series of two to six nucleotides is repeated hundreds or thousands of times in a row
    - Example: 12 alleles of homeotic gene that influence face length in dogs

Do All Traits Occur in Distinct Forms? (cont'd.)

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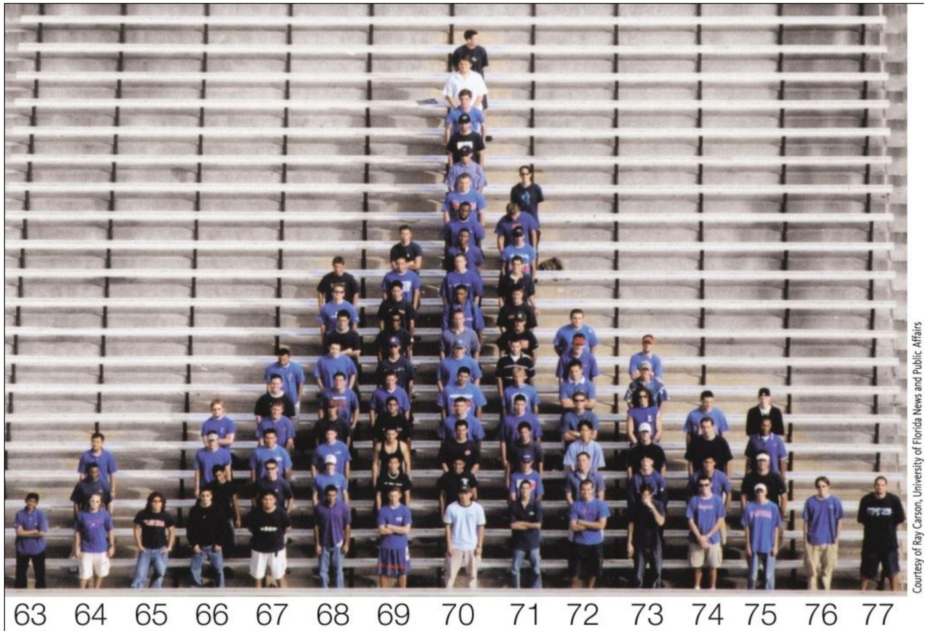
Do All Traits Occur in Distinct Forms? (cont'd.)

- Continuous variation
  - Some traits appear in two or three forms
  - Others occur in a range of small differences
  - The more genes and environmental factors that influence a trait, the more continuous the variation
  - Examples
    - Face length in dogs; human skin color; human height

Do All Traits Occur in Distinct Forms? (cont'd.)

- If a trait varies continuously, it will have bell-shaped curve

Do All Traits Occur in Distinct Forms? (cont'd.)



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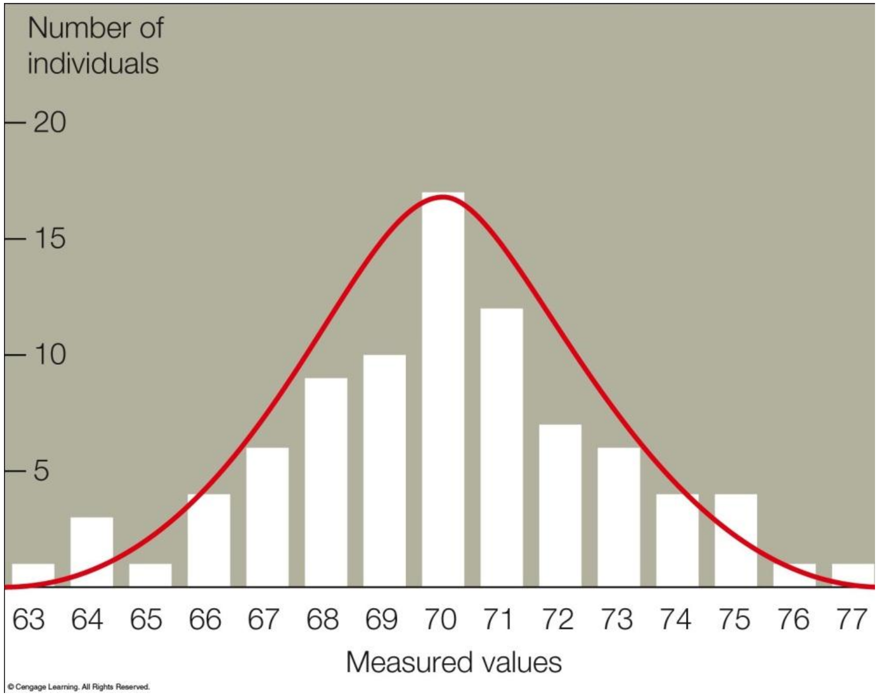

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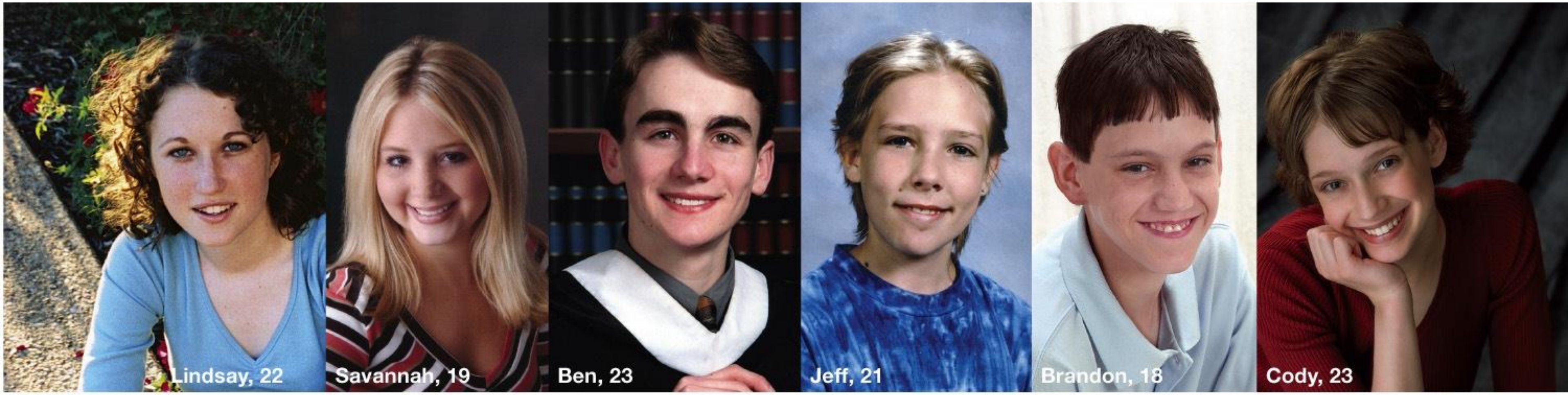
13.7 Application: Menacing Mucus

- Cystic fibrosis
  - Most common fatal genetic disorder in the U.S.
  - Most CF patients live no more than 30 years
  - The *CFTR* gene encodes a protein
    - Protein moves chloride ions out of epithelial cells
    - Binds disease-causing bacteria
  - Occurs in people homozygous for a mutated allele of *CFTR* gene

Menacing Mucus (cont'd.)



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Menacing Mucus (cont'd.)

- The allele of CF has a 3 base pair deletion
  - Called Δ*F508* because protein is missing the normal 508th amino acid
  - People with CF inherit 2 copies of ΔF508
  - The deletion causes mucus to accumulate, making breathing difficult

Menacing Mucus (cont'd.)

- ΔF508 allele is at least 50,000 years old
  - People who carry ΔF508 are probably less susceptible to typhoid fever and other bacterial diseases that begin in the intestinal tract