



Genetics [part 1]

IF3211 Domain Specific Computation

School of Electrical Engineering and Informatics ITB

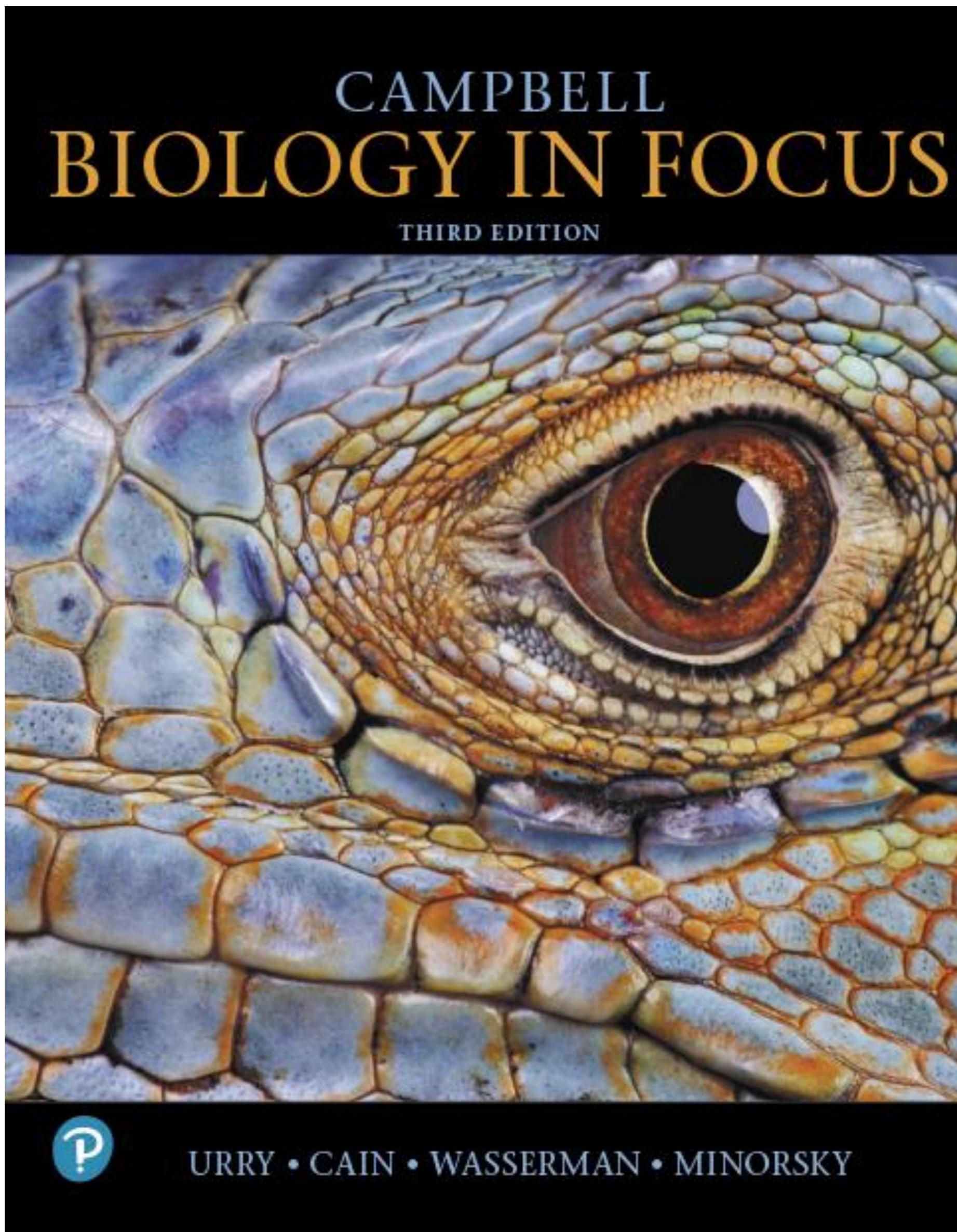
Content

- Cell Cycle
- Meiosis & Sexual Life Cycle
- Mendel & Gene Idea
- Tools

Cell Cycle

Campbell Biology in Focus

Third Edition



Chapter 9

The Cell Cycle

Lecture Presentations by
Kathleen Fitzpatrick and Nicole Tunbridge,
Simon Fraser University

Overview: The Key Roles of Cell Division (1 of 2)

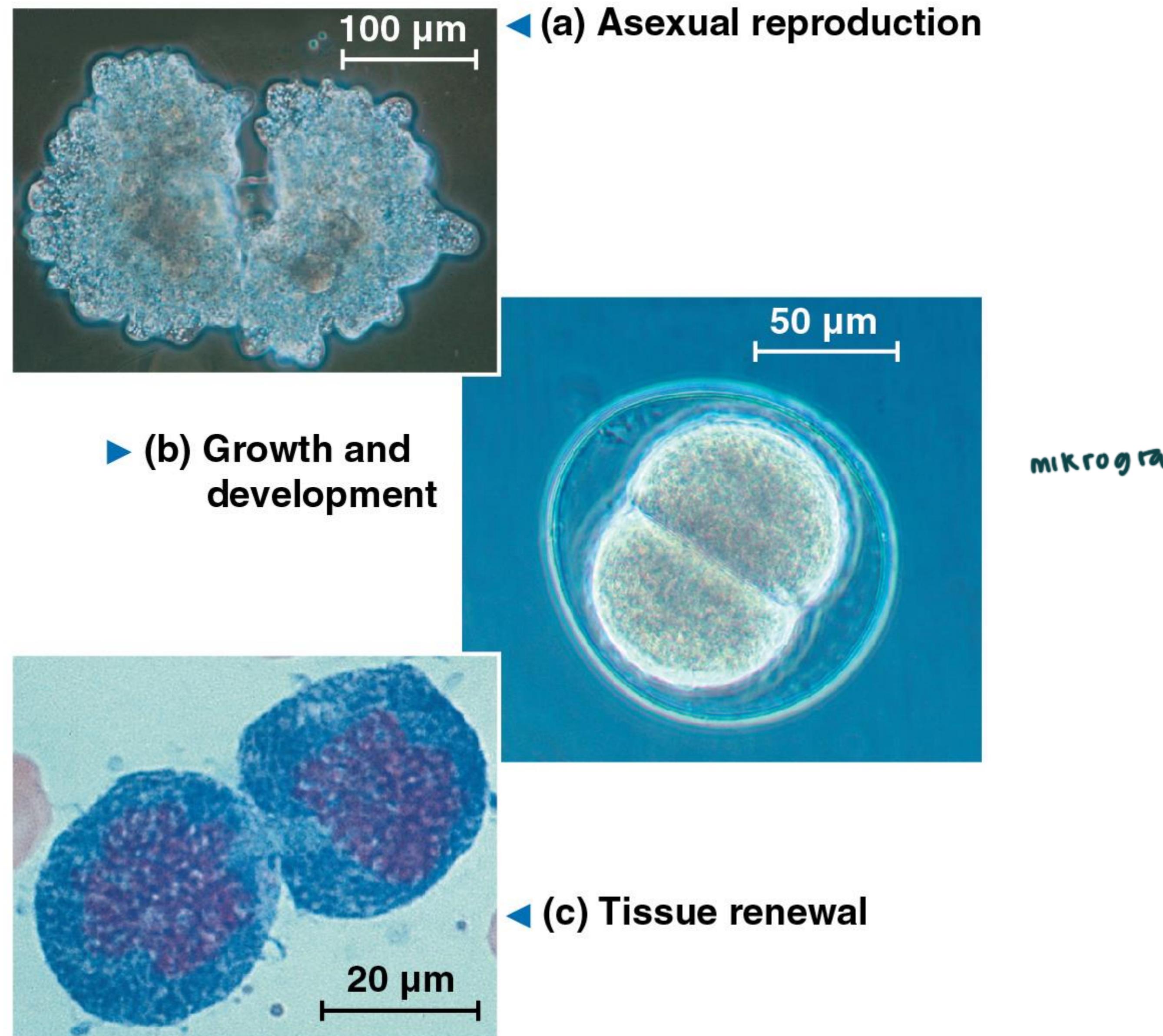
- The ability of organisms to produce more of their own kind best distinguishes living things from nonliving matter
- The continuity of life is based on the reproduction of cells, or **cell division**

Overview: The Key Roles of Cell Division (2 of 2)

- In unicellular organisms, division of one cell reproduces the entire organism
- Cell division enables multicellular eukaryotes to develop from a single cell and, once fully grown, to renew, repair, or replace cells as needed
- Cell division is an integral part of the **cell cycle**, the life of a cell from its formation to its own division

Figure 9.2

The Functions of Cell Division



Concept 9.1: Most Cell Division Results in Genetically Identical Daughter Cells

- Most cell division results in the distribution of identical genetic material—DNA—to two daughter cells
- DNA is passed from one generation of cells to the next with remarkable fidelity

Cellular Organization of the Genetic Material (1 of 2)

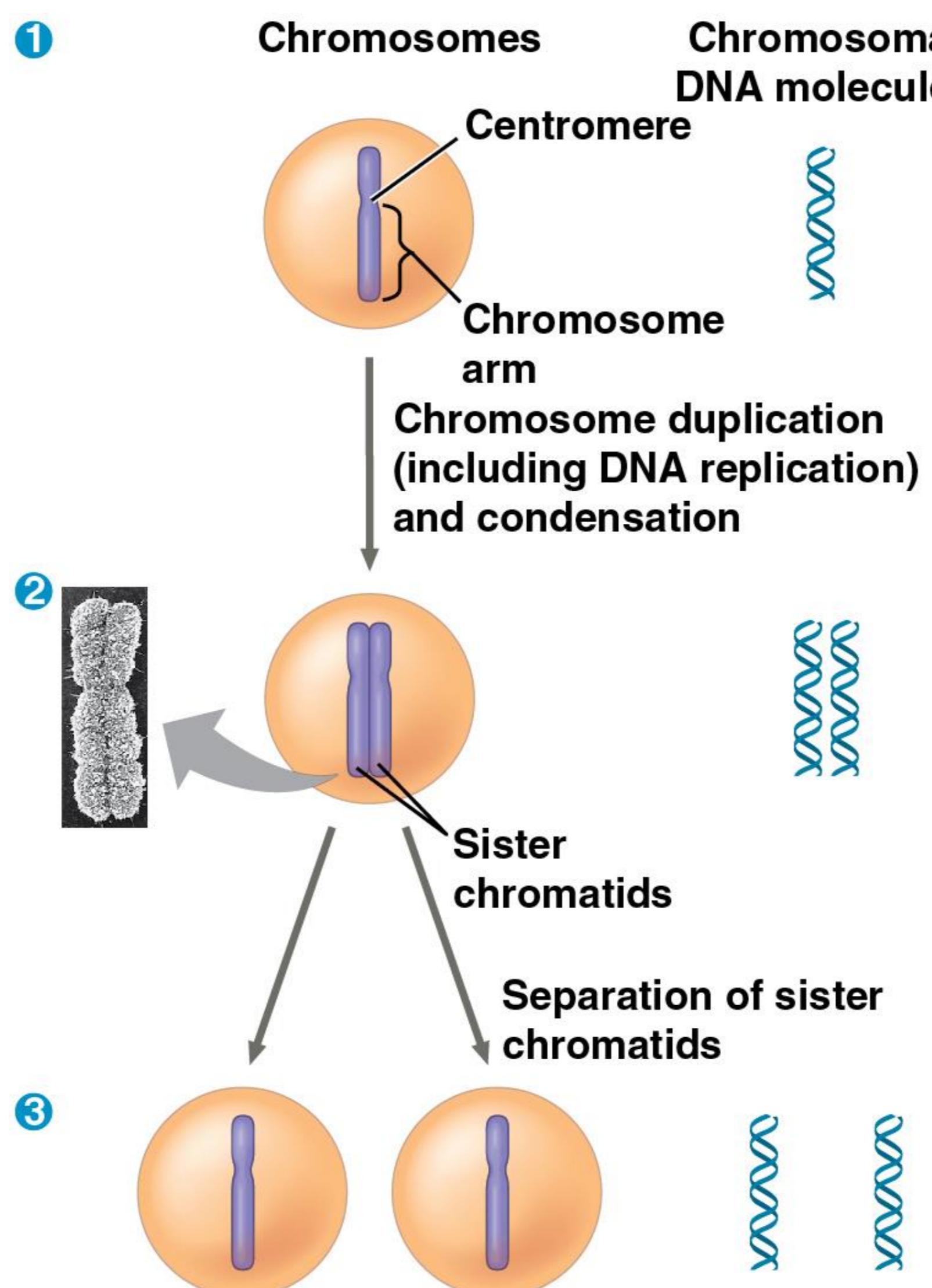
- All the DNA in a cell constitutes the cell's **genome**
- A genome can consist of a single DNA molecule (common in prokaryotic cells) or a number of DNA molecules (common in eukaryotic cells)
- DNA molecules in a cell are packaged into **chromosomes**

Cellular Organization of the Genetic Material (2 of 2)

- Eukaryotic chromosomes consist of **chromatin**, a complex of DNA and protein
- Every eukaryotic species has a characteristic number of chromosomes in each cell nucleus
- **Somatic cells** (nonreproductive cells) of humans, for instance, have 46 chromosomes, 2 sets of 23
- **Gametes** (reproductive cells: sperm and eggs) have half as many chromosomes as somatic cells, 23 chromosomes in the case of humans

Figure 9.5

Chromosome Duplication and Distribution During Cell Division



Phases of the Cell Cycle (3 of 3)

- Mitosis is conventionally divided into five phases
 - **Prophase**
 - **Prometaphase**
 - **Metaphase**
 - **Anaphase**
 - **Telophase**
- Cytokinesis overlaps the latter stages of mitosis

proses pembelahan sel
(sel sdh terbagi mnjd 2)

Figure 9.7 (1 of 2)

Exploring Mitosis in an Animal Cell

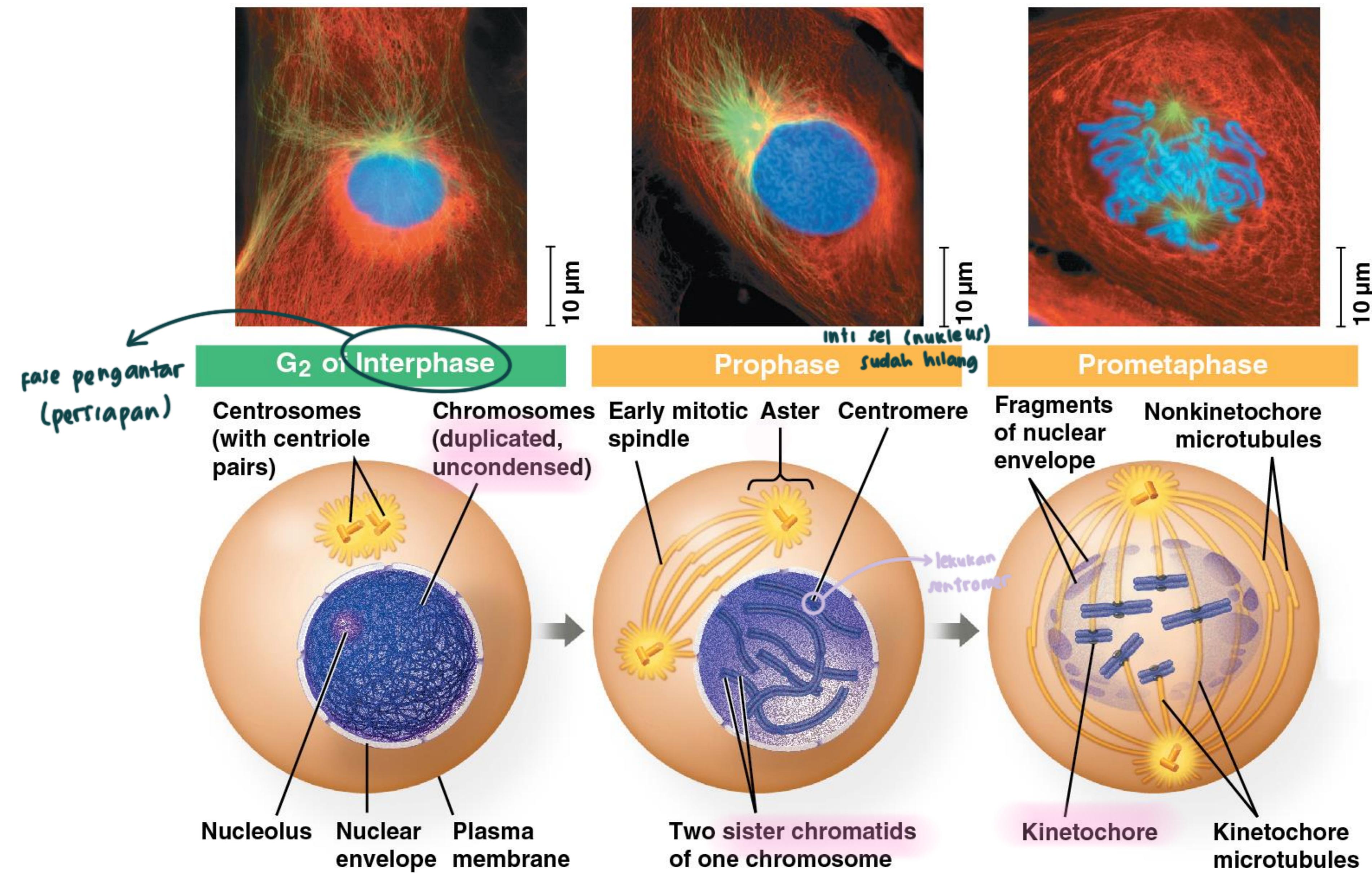
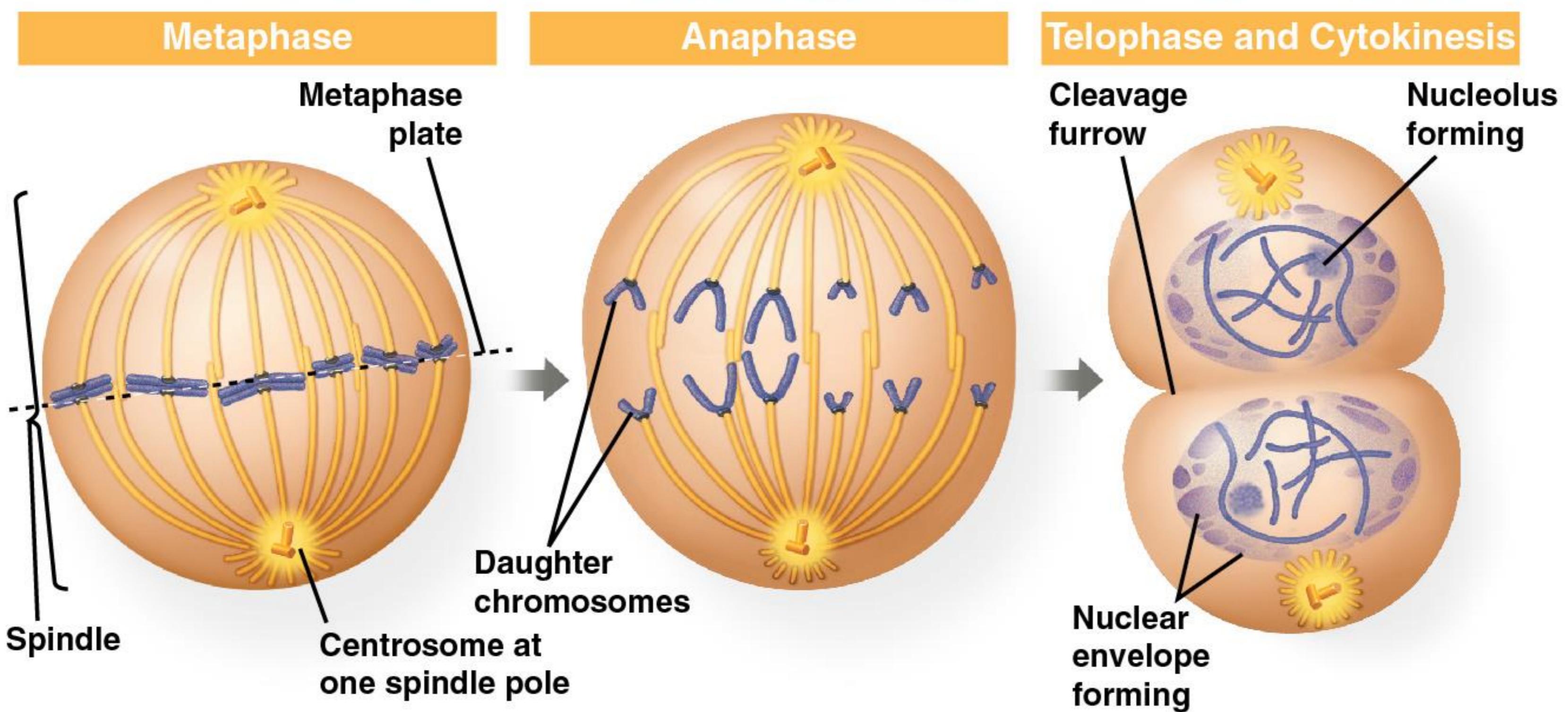
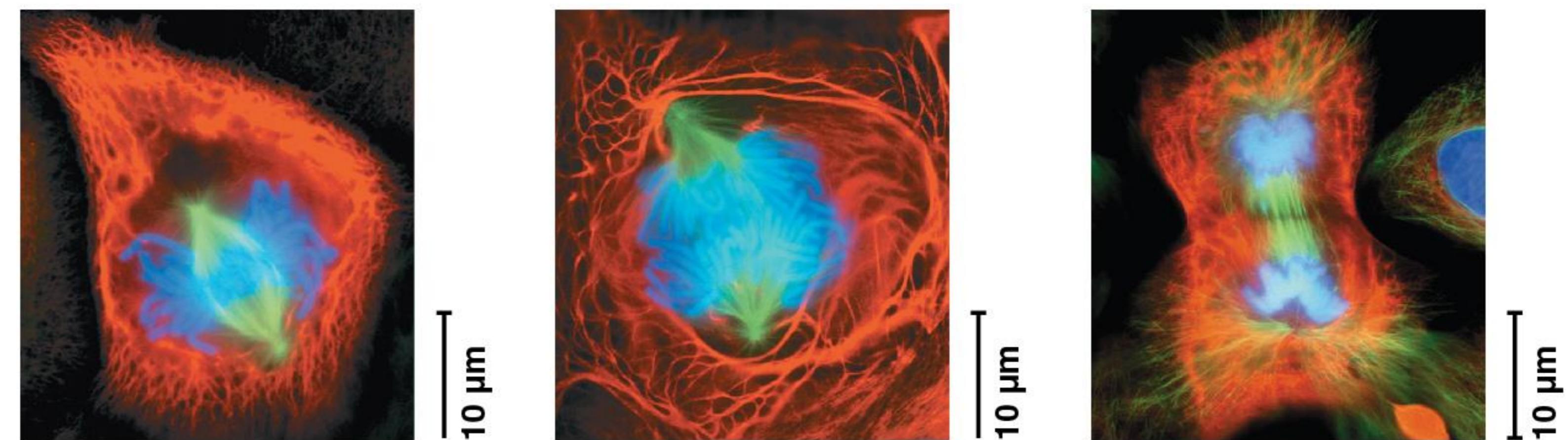


Figure 9.7 (2 of 2)

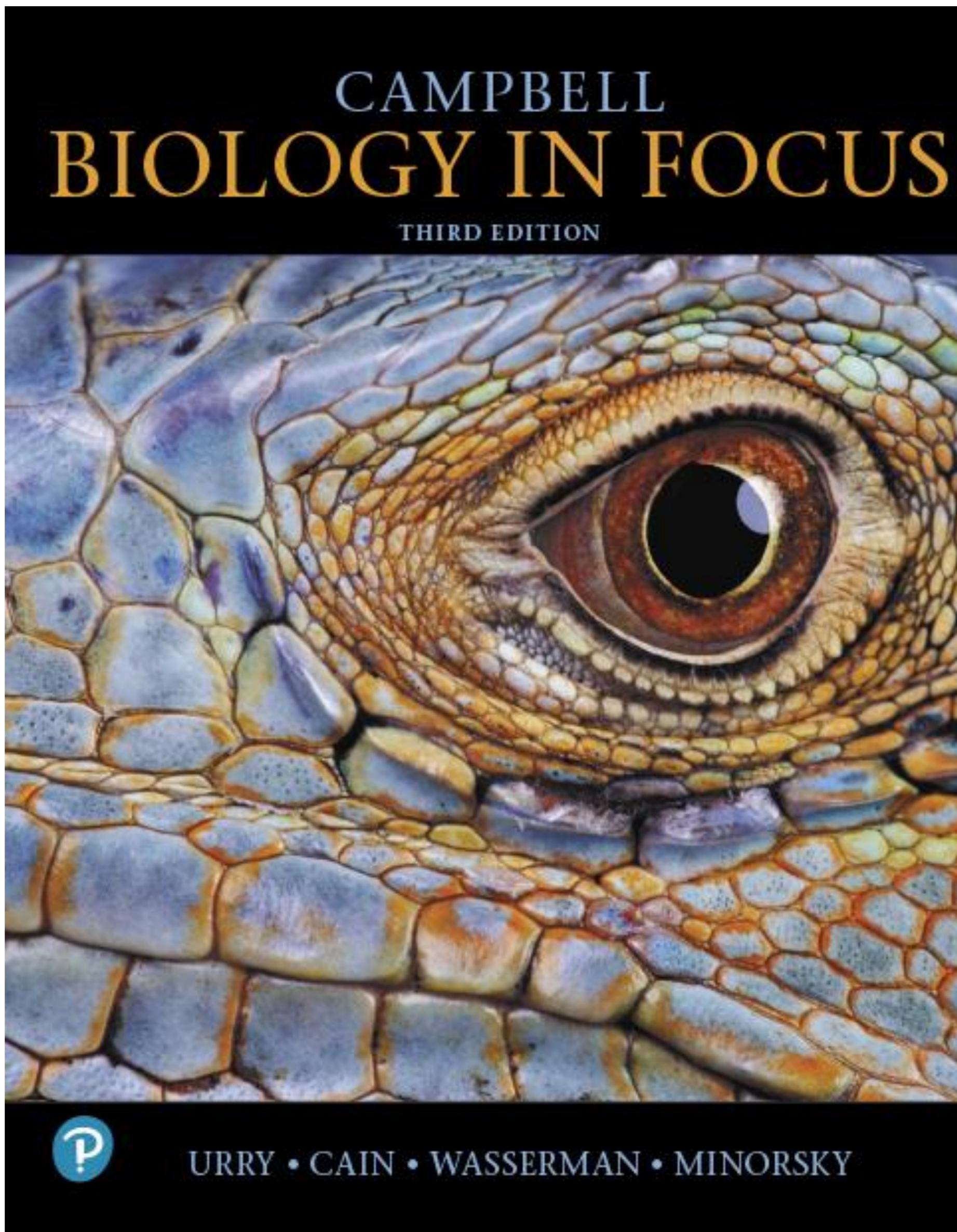
Exploring Mitosis in an Animal Cell



Meiosis & Sexual Life Cycle

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Chapter 10

Meiosis and Sexual Life
Cycles

Lecture Presentations by
Kathleen Fitzpatrick and Nicole Tunbridge,
Simon Fraser University

Overview: Variations on a Theme

- Offspring resemble their parents more than they do unrelated individuals
- **Heredity** is the transmission of traits from one generation to the next
- **Variation** is demonstrated by the differences in appearance that offspring show from parents and siblings
- **Genetics** is the scientific study of heredity and variation

Figure 10.1

What Accounts for Family Resemblance?



Concept 10.1: Offspring Acquire Genes from Parents by Inheriting Chromosomes

- In a literal sense, children do not inherit particular physical traits from their parents

Inheritance of Genes (1 of 2)

- **Genes** are the units of heredity and are made up of segments of DNA
- Genes are passed to the next generation via reproductive cells called **gametes** (sperm and eggs)

Inheritance of Genes (2 of 2)

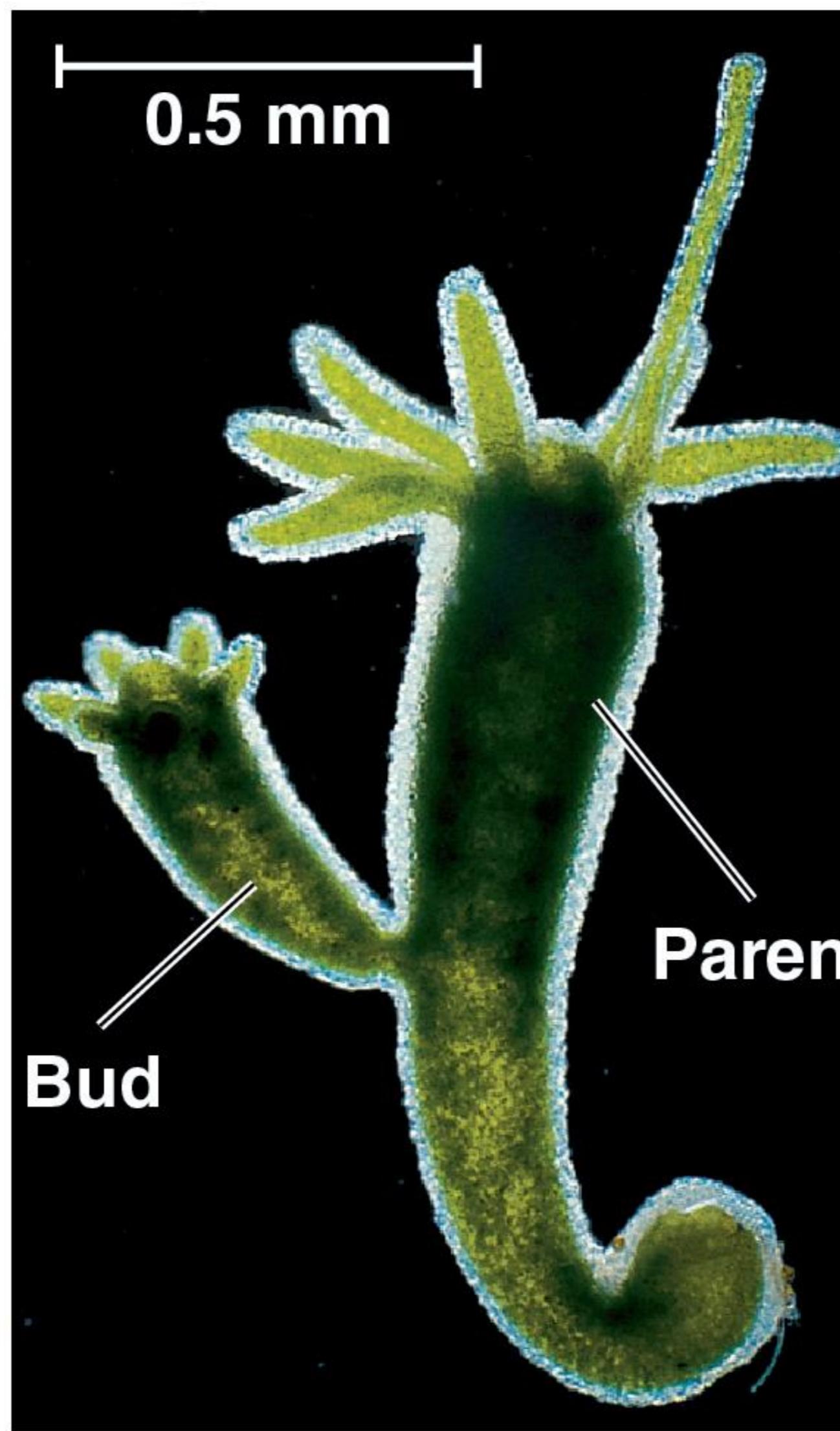
- Most DNA is packaged into chromosomes
- For example, humans have 46 chromosomes in their **somatic cells**, the cells of the body except for gametes and their precursors
- Each gene has a specific position, or **locus**, on a certain chromosome

Comparison of Asexual and Sexual Reproduction

- In **asexual reproduction**, a single individual passes genes to its offspring without the fusion of gametes
- A **clone** is a group of genetically identical individuals from the same parent, produced asexually
- In **sexual reproduction**, two parents give rise to offspring that have unique combinations of genes inherited from the two parents

Figure 10.2

Asexual Reproduction in Two Multicellular Organisms



(a) Hydra

dgn membangun kuncup bambu
x tetap menempel pd dirinya



(b) Redwoods

Concept 10.2: Fertilization and Meiosis Alternate in Sexual Life Cycles

- A **life cycle** is the generation-to-generation sequence of stages in the reproductive history of an organism

Sets of Chromosomes in Human Cells (1 of 5)

- Human somatic cells have 23 pairs of chromosomes
- A **karyotype** is an ordered display of the pairs of chromosomes from a cell
- The two chromosomes in each pair are called **homologous chromosomes**, or **homologs**
- Chromosomes in a **homologous pair** are the same length and shape and carry genes controlling the same inherited characters

Figure 10.3

Research Method

Application

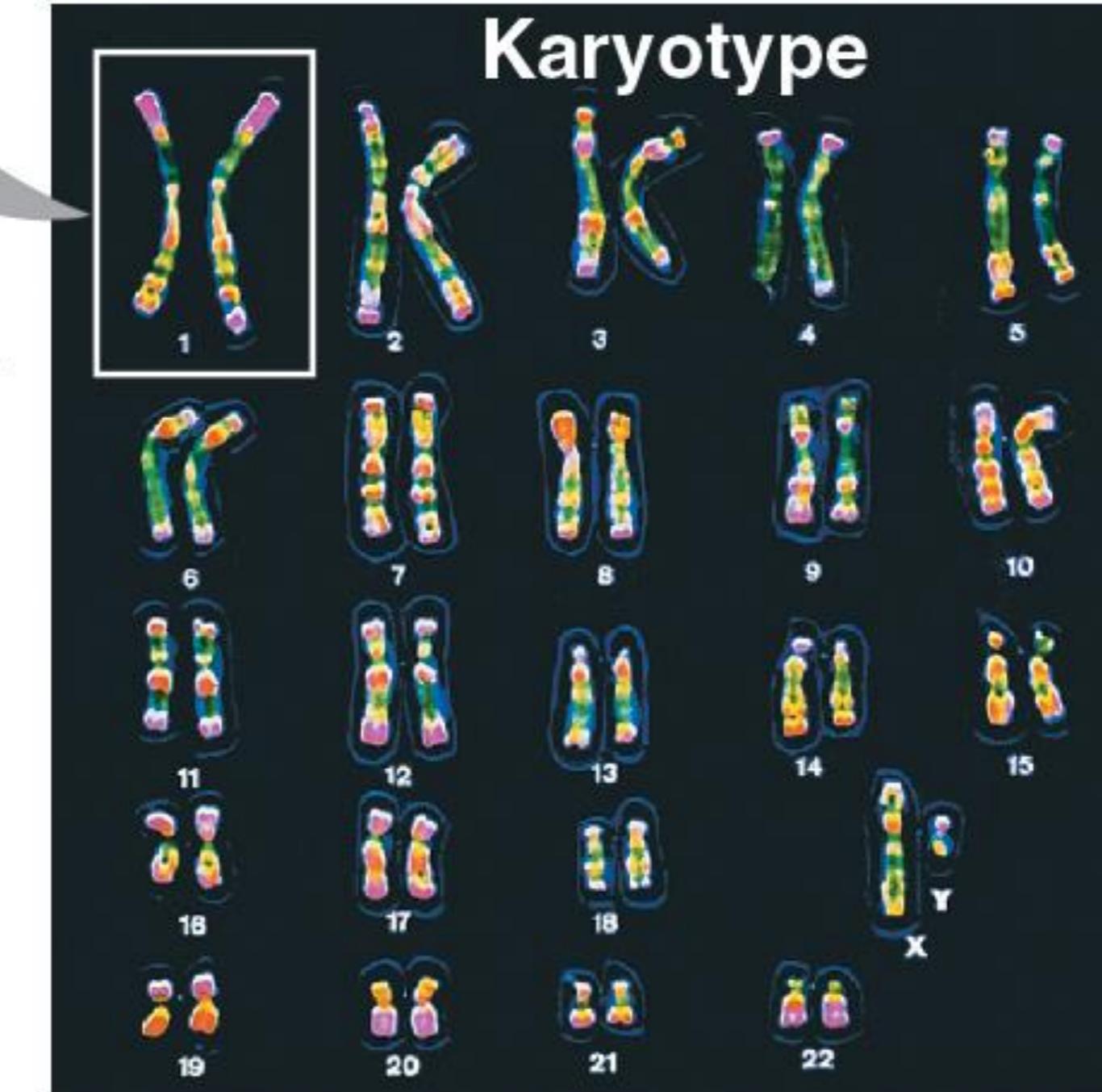


Technique

Pair of homologous chromosomes



Karyotype



Sets of Chromosomes in Human Cells (2 of 5)

- The chromosomes referred to as X and Y are an exception for the general pattern of homologous chromosomes in human somatic cells
- Typically, human females have a homologous pair of X chromosomes (XX), and human males have one X and one Y chromosome
- Due to their role in sex determination, the X and Y chromosomes are called **sex chromosomes**
- The remaining 22 pairs of chromosomes are called **autosomes**

Sets of Chromosomes in Human Cells (3 of 5)

- Each pair of homologous chromosomes includes one chromosome from each parent
- The 46 chromosomes in a human somatic cell are two sets of 23: one from each parent
- A **diploid cell** ($2n$) has two sets of chromosomes
- For humans, the diploid number is 46 ($2n = 46$)

Sets of Chromosomes in Human Cells (5 of 5)

- A gamete (sperm or egg) contains a single set of chromosomes and is **haploid** (n)
- For humans, the haploid number is 23 ($n = 23$)
- Each set of 23 consists of 22 autosomes and a single sex chromosome
- In an unfertilized egg (ovum), the sex chromosome is X
- In a sperm cell, the sex chromosome may be either X or Y

Concept 10.3: Meiosis Reduces the Number of Chromosome Sets from Diploid to Haploid

- Like mitosis, meiosis is preceded by the duplication of chromosomes
- Meiosis then takes place in two sets of cell divisions, called **meiosis I** and **meiosis II**
- The two cell divisions result in four daughter cells, rather than the two daughter cells in mitosis
- Each daughter cell has only half as many chromosomes as the parent cell

Figure 10.8

Exploring Meiosis in an Animal Cell

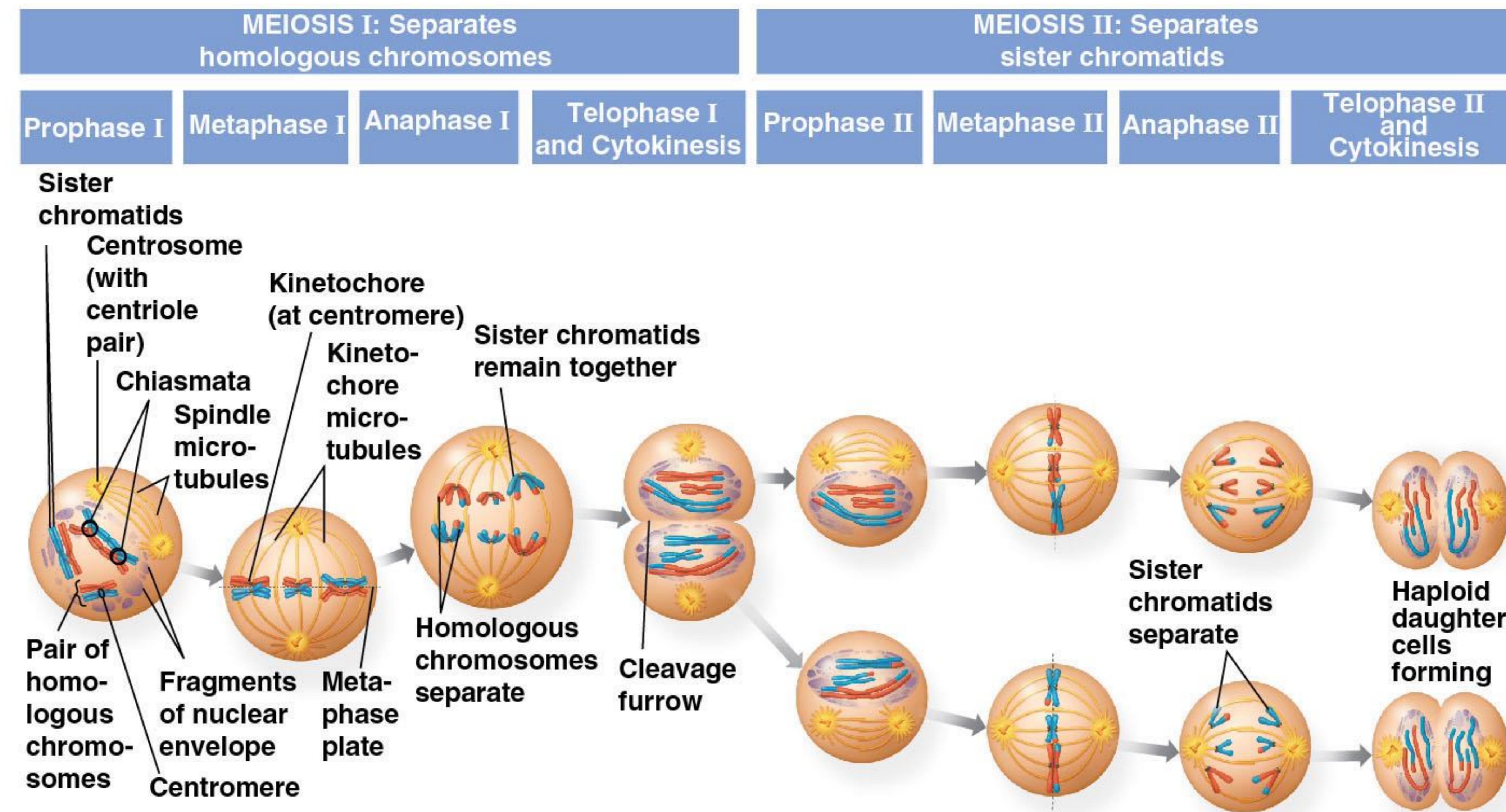


Figure 10.10 (1 of 2)

A Comparison of Mitosis and Meiosis

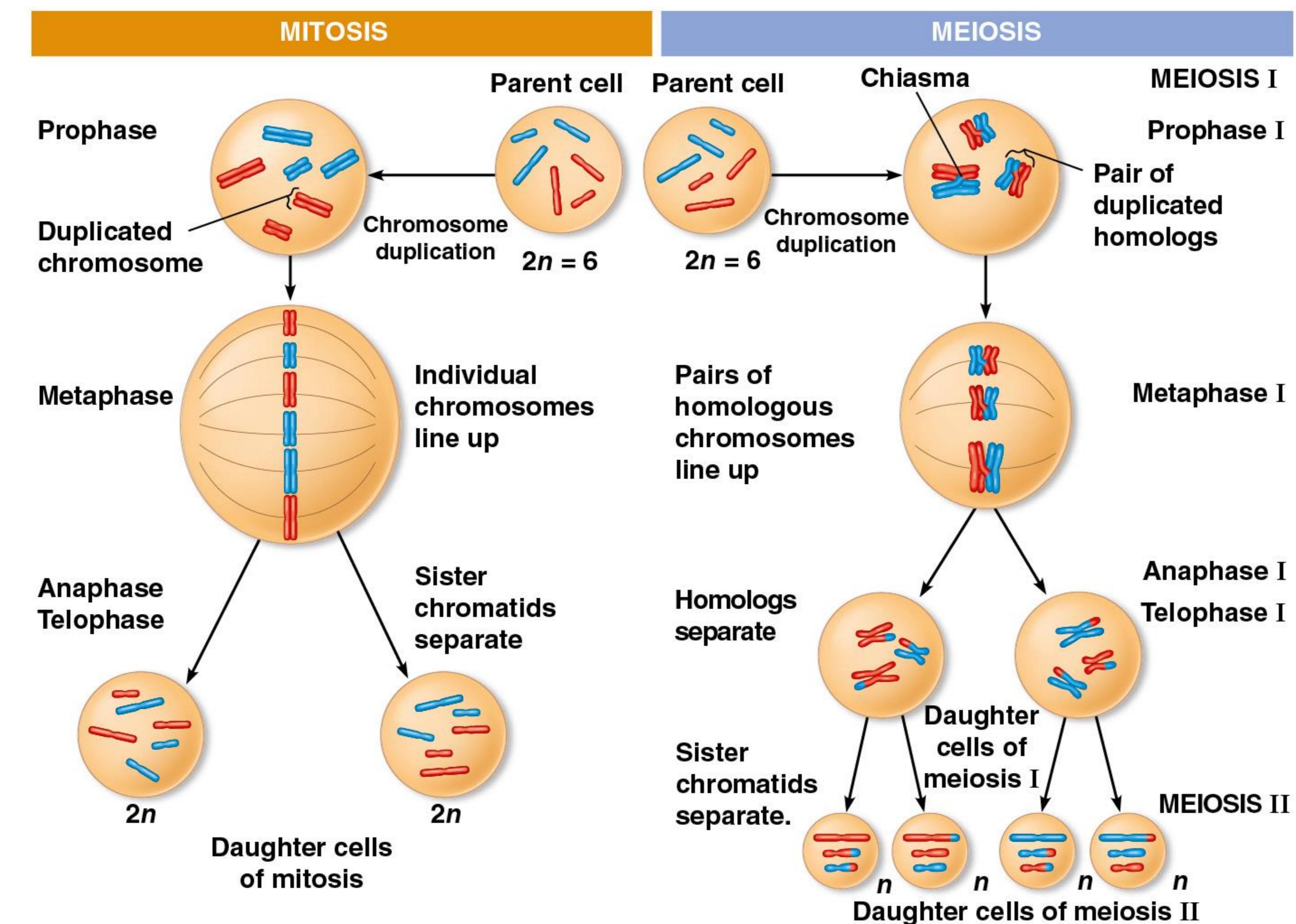


Figure 10.10 (2 of 2)

A Comparison of Mitosis and Meiosis Summary

Property	Mitosis (occurs in both diploid and haploid cells)	Meiosis (can only occur in diploid cells)
DNA replication	Occurs during interphase, before mitosis begins	Occurs during interphase before meiosis I, but not meiosis II
Number of divisions	One, including prophase, prometaphase, metaphase, anaphase, and telophase	Two, each including prophase, metaphase, anaphase, and telophase
Synapsis of homologous Chromosomes	Does not occur	Occurs during prophase I along with crossing over between nonsister chromatids; resulting chiasmata hold pairs together due to sister chromatid cohesion
Number of daughter cells and genetic composition	Two, each genetically identical to the parent cell, with the same number of chromosomes	Four, each haploid (n); genetically different from the parent cell and from each other
Role in animals, fungi, and plants	Enables multicellular animal, fungus, or plant (gametophyte or sporophyte) to arise from a single cell; produces cells for growth, repair, and, in some species, asexual reproduction; produces gametes in the plant gametophyte	Produces gametes (in animals) or spores (in fungi and in plant sporophytes); reduces number of chromosome sets by half and introduces genetic variability among the gametes or spores

Concept 10.4: Genetic Variation Produced in Sexual Life Cycles Contributes to Evolution

- Mutations (changes in an organism's DNA) are the original source of genetic diversity
- Mutations create different versions of genes called alleles
- Reshuffling of alleles during sexual reproduction produces genetic variation

Origins of Genetic Variation Among Offspring

- The behavior of chromosomes during meiosis and fertilization is responsible for most of the variation that arises in each generation
- Three mechanisms contribute to genetic variation
 - Independent assortment of chromosomes
 - Crossing over
 - Random fertilization

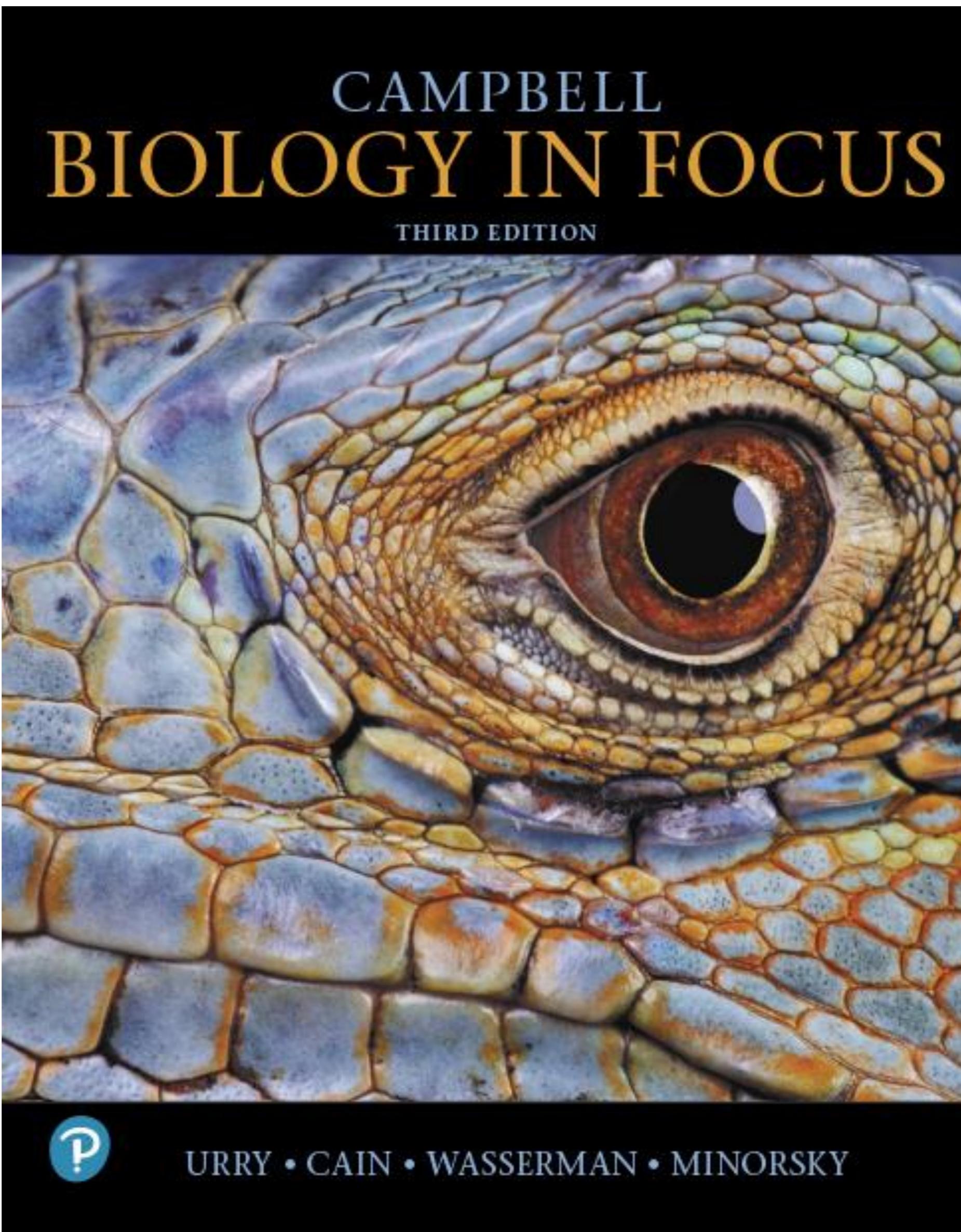
The Evolutionary Significance of Genetic Variation within Populations

- Natural selection results in the accumulation of genetic variations favored by the environment
- Sexual reproduction contributes to the genetic variation in a population, which originates from mutations

Mendel & Gene Idea

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Chapter 11

Mendel and the Gene Idea

Lecture Presentations by
Kathleen Fitzpatrick and Nicole Tunbridge,
Simon Fraser University

Overview: Drawing from the Deck of Genes (1 of 2)

- What principles account for the passing of traits from parents to offspring?
- The “blending” hypothesis is the idea that genetic material from the two parents blends together (the way blue and yellow paint blend to make green)

Overview: Drawing from the Deck of Genes (2 of 2)

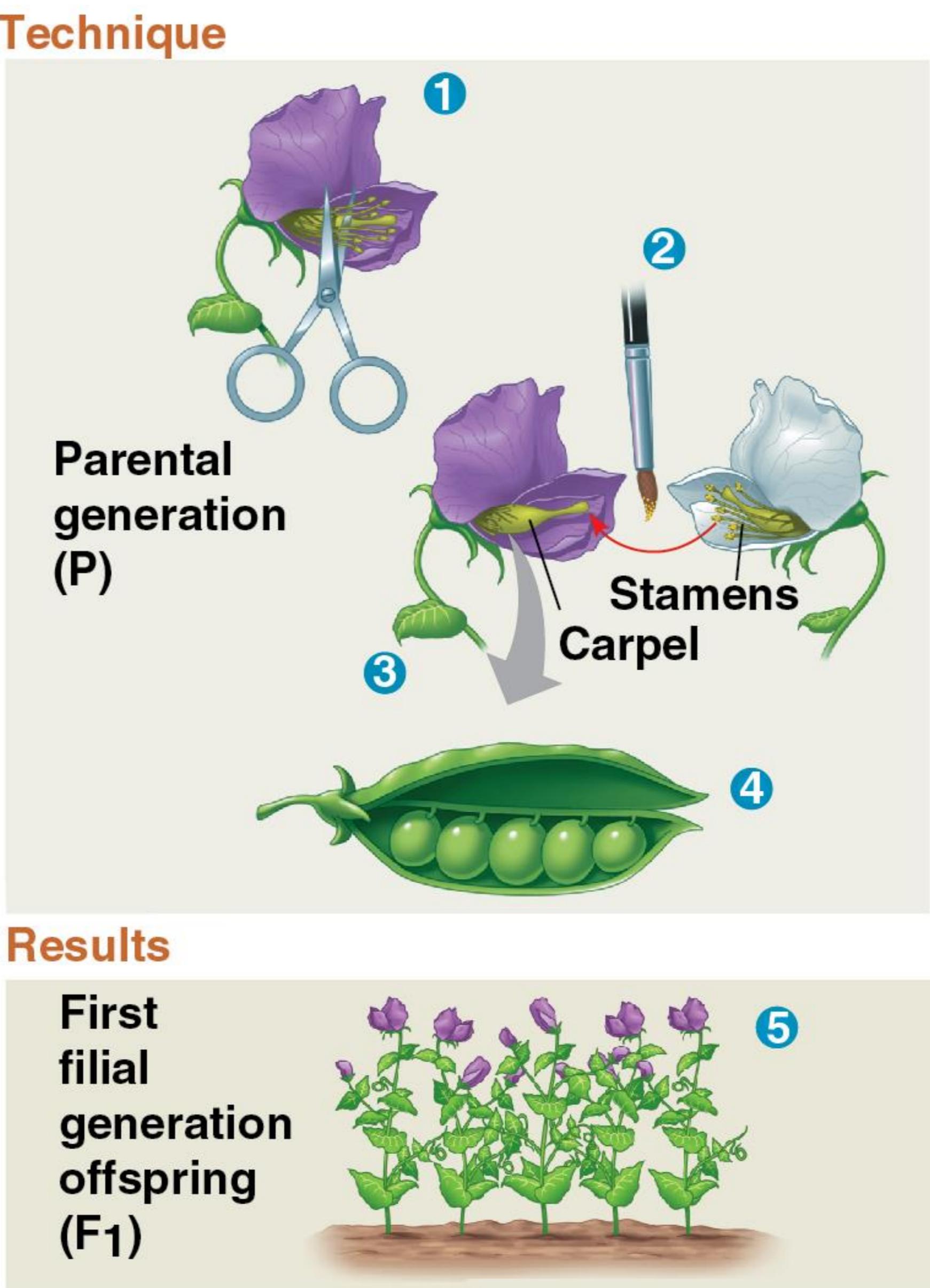
- The “particulate” hypothesis is the idea that parents pass on discrete heritable units (genes)
- Mendel documented a particulate mechanism through his experiments with garden peas

Mendel's Experimental, Quantitative Approach (1 of 3)

- Mendel probably chose to work with peas because
 - There are many varieties with distinct heritable features, or **characters** (such as flower color); character variants (such as purple or white flowers) are called **traits**
 - He could strictly control mating between plants

Figure 11.2

Research Method: Crossing Pea Plants



Mendel's Experimental, Quantitative Approach (2 of 3)

- Mendel chose to track only characters that occurred in two distinct alternative forms
- He also used varieties that were **true-breeding** (plants that produce offspring of the same variety when they self-pollinate)

Mendel's Experimental, Quantitative Approach (3 of 3)

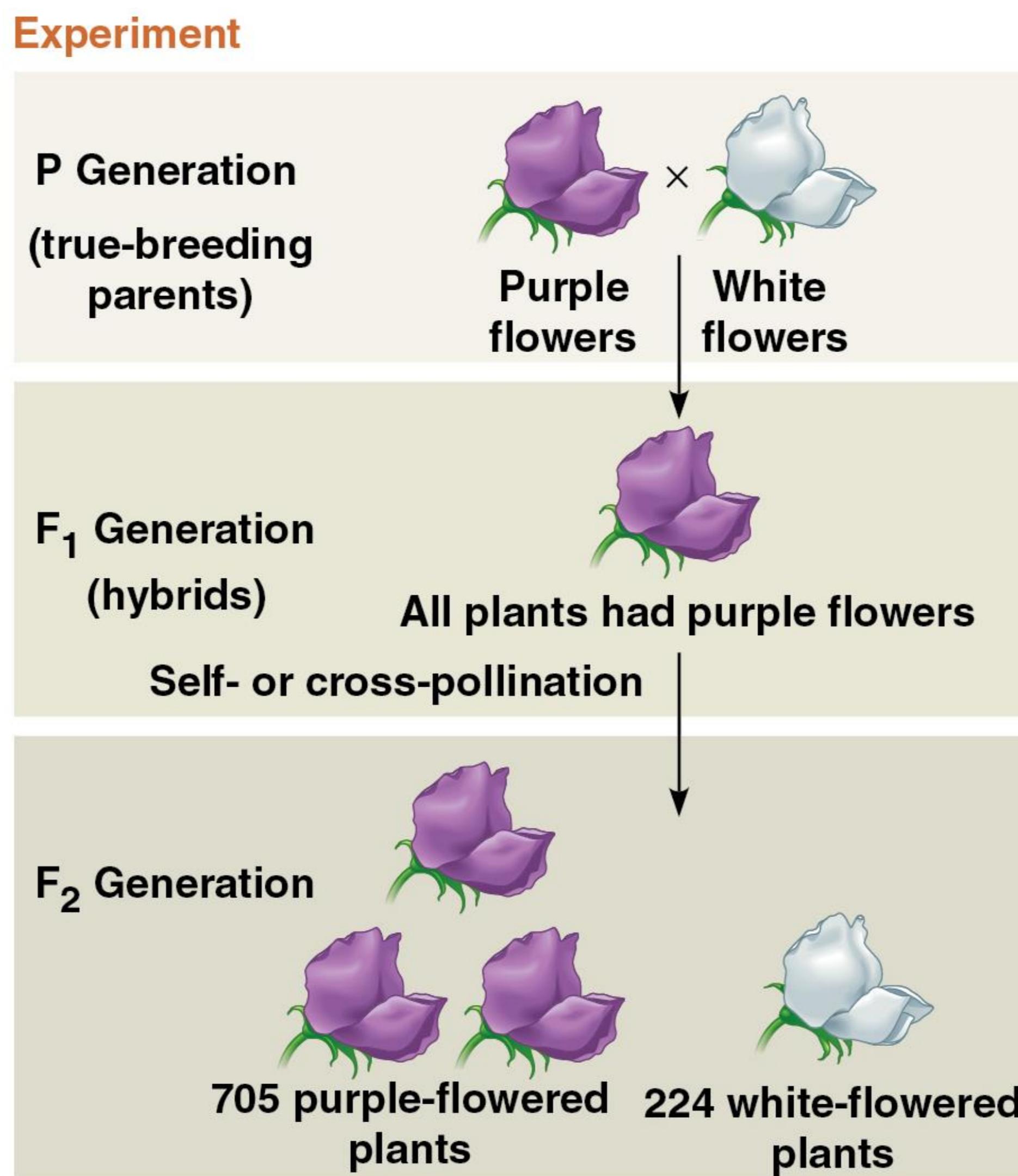
- In a typical experiment, Mendel mated two contrasting, true-breeding varieties, a process called **hybridization**
- The true-breeding parents are the **P generation**
- The hybrid offspring of the P generation are called the **F₁ generation**
- When F₁ individuals self-pollinate or cross-pollinate with other F₁ hybrids, the **F₂ generation** is produced

The Law of Segregation (1 of 3)

- When Mendel crossed contrasting, true-breeding white- and purple-flowered pea plants, all of the F_1 hybrids were purple
- When Mendel crossed the F_1 hybrids, many of the F_2 plants had purple flowers, but some had white
- Mendel discovered a ratio of about 3:1 purple to white flowers in the F_2 generation

Figure 11.3

Inquiry: When F_1 Hybrid Pea Plants Self- or Cross-Pollinate, Which Traits Appear in the F_2 Generation?



The Law of Segregation (2 of 3)

- Mendel reasoned that in the F_1 plants, the heritable factor for white flowers was hidden or masked in the presence of the purple-flower factor
- He called the purple flower color a dominant trait and the white flower color a recessive trait
- The factor for white flowers was not diluted or destroyed because it reappeared in the F_2 generation

The Law of Segregation (3 of 3)

- Mendel observed the same pattern of inheritance in six other pea plant characters, each represented by two traits
- What Mendel called a “heritable factor” is what we now call a gene

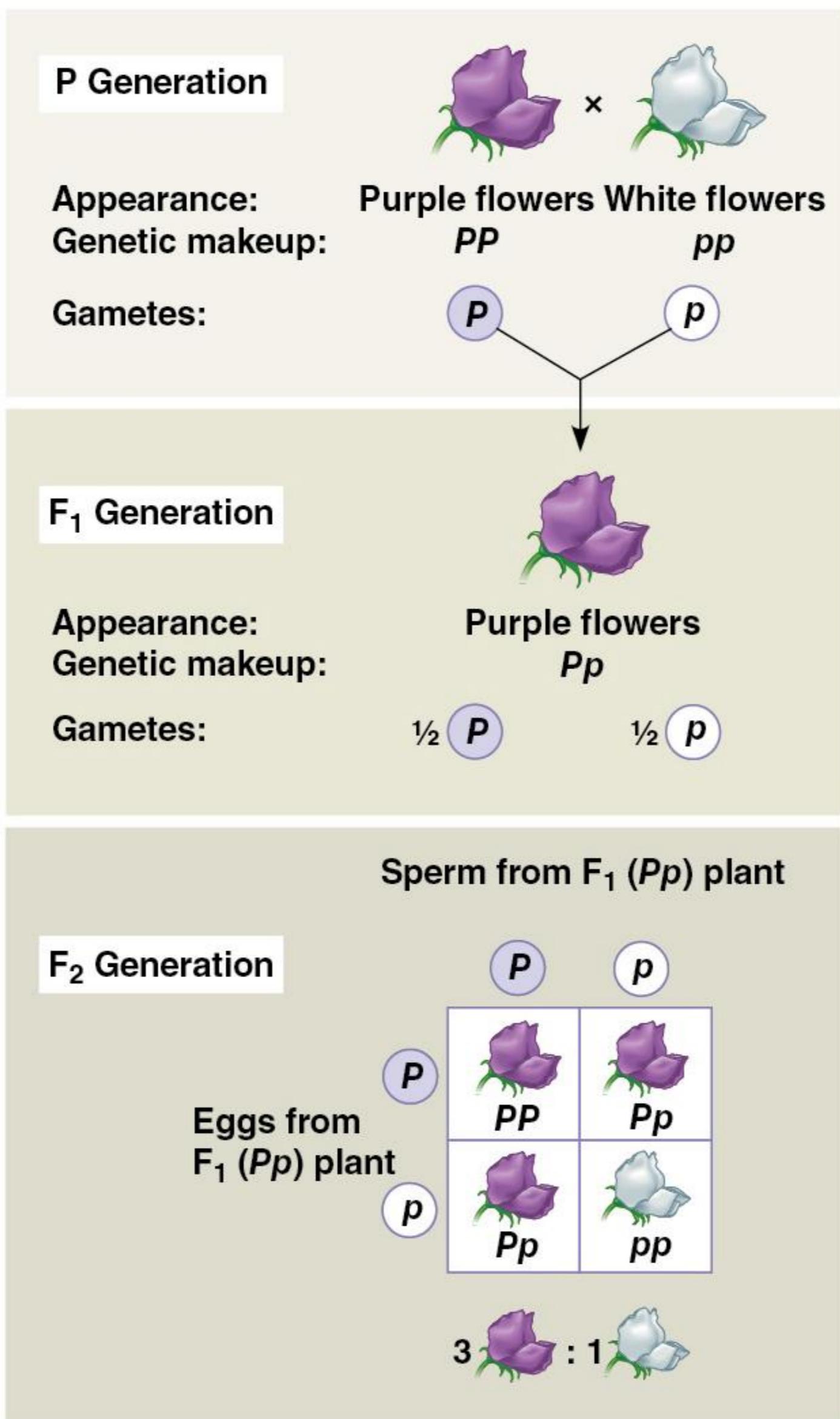
Table 11.1

The Results of Mendel's F1 Crosses for Seven Characters in Pea Plants

Character	Dominant Trait	x	Recessive Trait	F ₂ Generation	
				Dominant: Recessive	Ratio
Flower color	Purple	x	White	705:224	3.15:1
					
Seed color	Yellow	x	Green	6,022:2,001	3.01:1
					
Seed shape	Round	x	Wrinkled	5,474:1,850	2.96:1
					
Pod color	Green	x	Yellow	428:152	2.82:1
					
Pod shape	Inflated	x	Constricted	882:299	2.95:1
					
Flower position	Axial	x	Terminal	651:207	3.14:1
					
Stem length	Tall	x	Dwarf	787:277	2.84:1
					

Figure 11.5

Mendel's Law of Segregation



Useful Genetic Vocabulary (1 of 2)

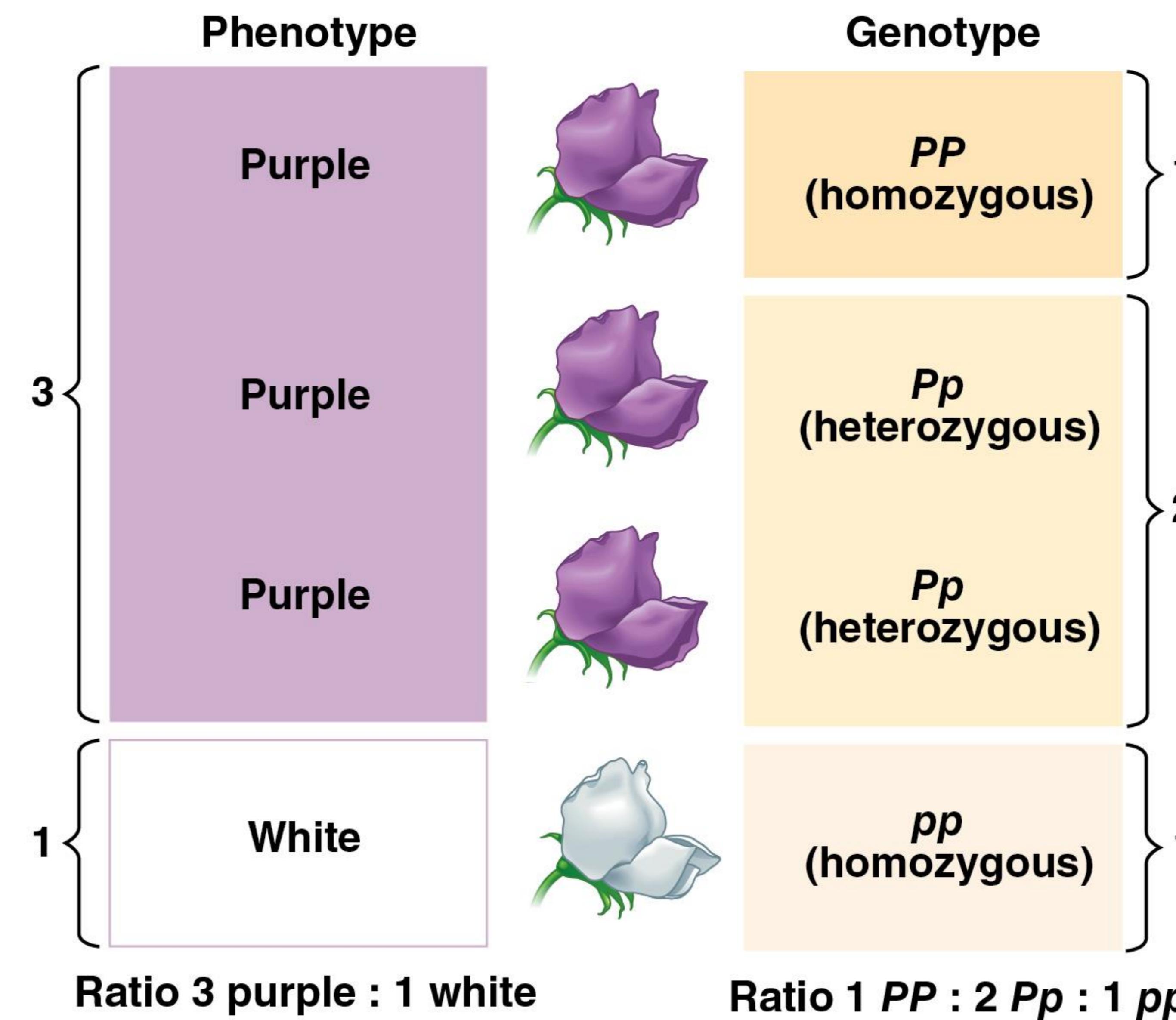
- An organism with two identical alleles for a character is said to be **homozygous** for the gene controlling that character
- An organism that has two different alleles for a gene is said to be **heterozygous** for the gene controlling that character
- Unlike **homozygotes**, **heterozygotes** are not true-breeding

Useful Genetic Vocabulary (2 of 2)

- Because of the effects of dominant and recessive alleles, an organism's traits do not always reveal its genetic composition
- Therefore, we distinguish between an organism's **phenotype**, or physical appearance, and its **genotype**, or genetic makeup
- In the example of flower color in pea plants, PP and Pp plants have the same phenotype (purple) but different genotypes

Figure 11.6

Phenotype Versus Genotype

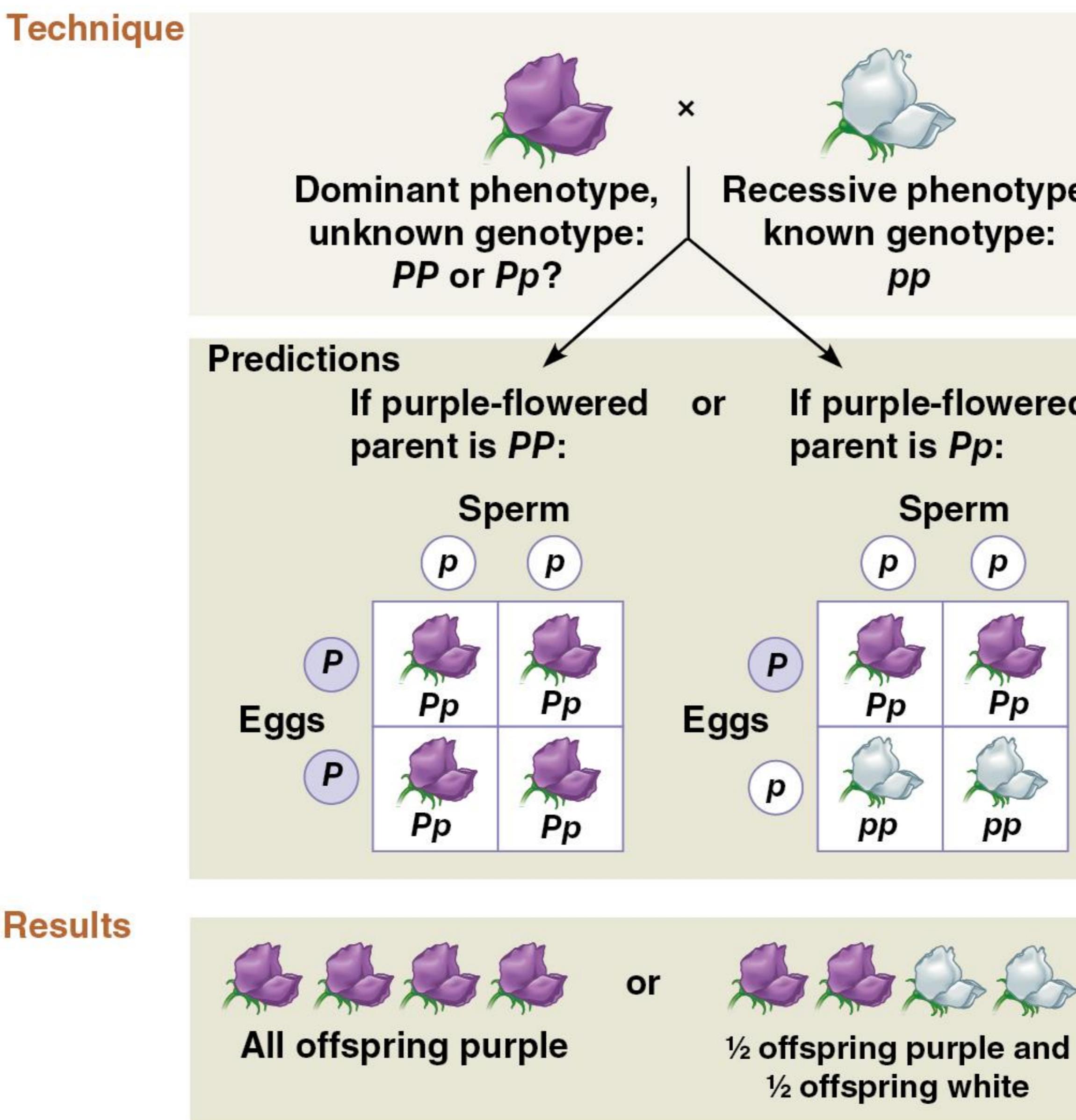


The Testcross

- We cannot tell the genotype of an individual with a dominant phenotype, such as purple flowers
- Such an individual could be either homozygous dominant or heterozygous
- Breeding the individual with an recessive homozygote is called a **testcross** because it can reveal the genotype of that organism
- If any offspring display the recessive phenotype, the mystery parent must be heterozygous

Figure 11.7

Research Method: The Testcross



Concept 11.2: Probability Laws Govern Mendelian Inheritance

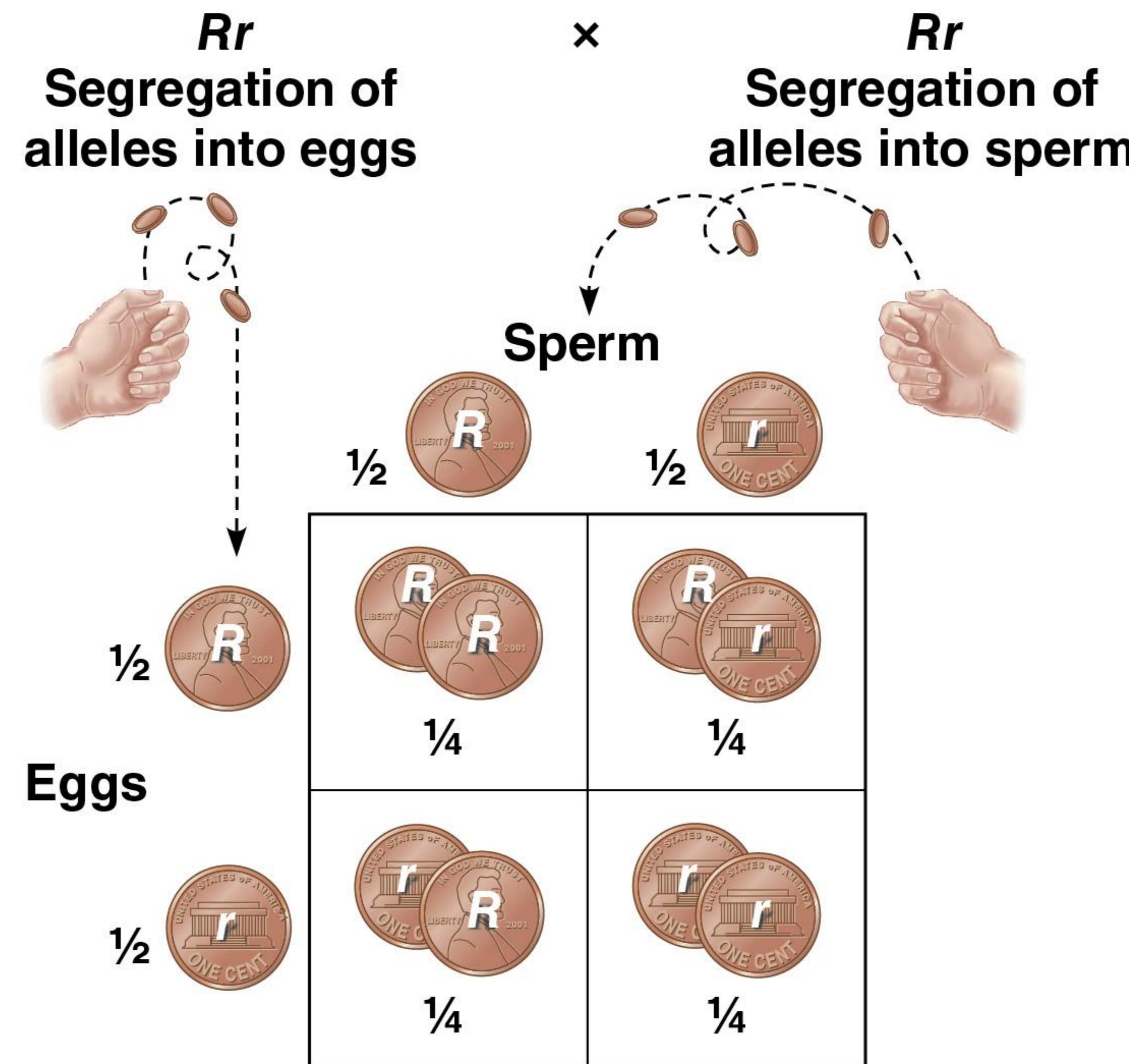
- Mendel's laws of segregation and independent assortment reflect the rules of probability
- The outcome of one coin toss has no impact on the outcome of the next toss
- In the same way, the alleles of one gene segregate into gametes independently of another gene's alleles

The Multiplication and Addition Rules Applied to Monohybrid Crosses (1 of 2)

- The **multiplication rule** states that the probability that two or more independent events will occur together is the product of their individual probabilities
- This can be applied to an F_1 monohybrid cross
- Segregation in a heterozygous plant is like flipping a coin: Each gamete has a $\frac{1}{2}$ chance of carrying the dominant allele and a $\frac{1}{2}$ chance of carrying the recessive allele

Figure 11.9

Segregation of Alleles and Fertilization as Chance Events



The Multiplication and Addition Rules Applied to Monohybrid Crosses (2 of 2)

- The **addition rule** states that the probability that any one of two or more mutually exclusive events will occur is calculated by adding together their individual probabilities
- It can be used to figure out the probability that an F_2 plant from a monohybrid cross will be heterozygous rather than homozygous

Solving Complex Genetics Problems with the Rules of Probability (1 of 3)

- We can apply the rules of probability to predict the outcome of crosses involving multiple characters
- A dihybrid or other multicharacter cross is equivalent to two or more independent monohybrid crosses occurring simultaneously
- In calculating the chances for various genotypes, each character is considered separately, and then the individual probabilities are multiplied

Solving Complex Genetics Problems with the Rules of Probability (2 of 3)

- For example, if we cross F_1 heterozygotes of genotype $YyRr$, we can calculate the probability of different genotypes among the F_2 generation

Probability of $YYRR = \frac{1}{4}$ (probability of YY) $\times \frac{1}{4}$ (RR) = $\frac{1}{16}$

Probability of $YyRR = \frac{1}{2}(Yy) \times \frac{1}{4} (RR) = \frac{1}{8}$

Solving Complex Genetics Problems with the Rules of Probability (3 of 3)

- For example, for the cross $PpYyRr \times Ppyyrr$, we can calculate the probability of offspring showing at least two recessive traits

$$ppyyRr \quad \frac{1}{4} (\text{probability of } pp) \times \frac{1}{2} (yy) \times \frac{1}{2} (Rr) = \frac{1}{16}$$

$$ppYyrr \quad \frac{1}{4} (pp) \times \frac{1}{2} (Yy) \times \frac{1}{2} (rr) = \frac{1}{16}$$

$$Ppyyrr \quad \frac{1}{2} (Pp) \times \frac{1}{2} (yy) \times \frac{1}{2} (rr) = \frac{2}{16}$$

$$PPyyrr \quad \frac{1}{4} (PP) \times \frac{1}{2} (yy) \times \frac{1}{2} (rr) = \frac{1}{16}$$

$$ppyyrr \quad \frac{1}{4} (pp) \times \frac{1}{2} (yy) \times \frac{1}{2} (rr) = \frac{1}{16}$$

$$\text{Chance of at least two recessive traits} \qquad \qquad \qquad = \frac{6}{16} = \frac{3}{8}$$

Concept 11.3: Inheritance Patterns Are Often More Complex Than Predicted by Simple Mendelian Genetics

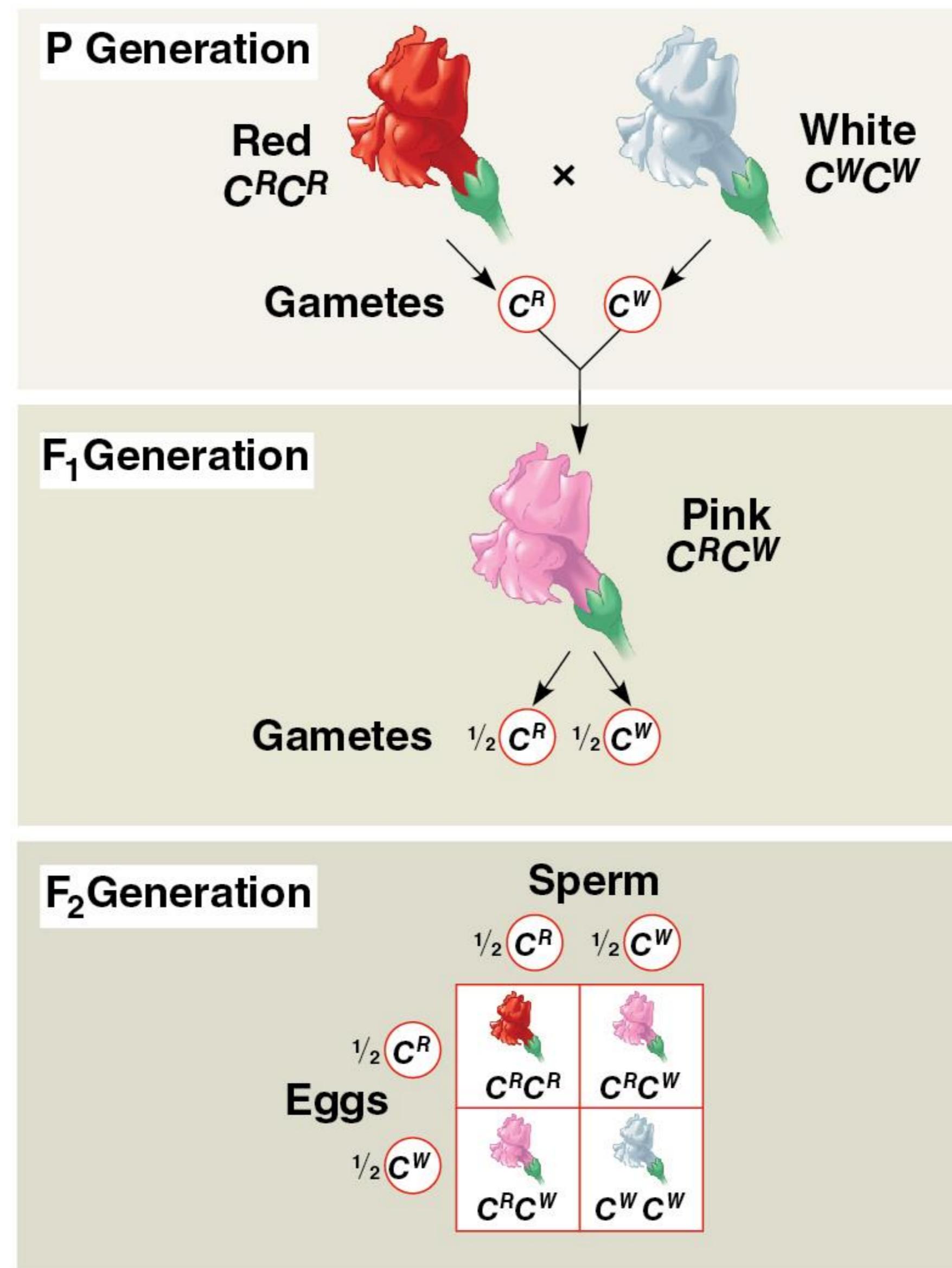
- Few heritable characters are determined as simply as the traits Mendel studied
- However, the basic principles of segregation and independent assortment apply even to more complex patterns of inheritance

Degrees of Dominance (1 of 4)

- **Complete dominance** occurs when phenotypes of the heterozygote and dominant homozygote are indistinguishable
- In **incomplete dominance**, the phenotype of F_1 hybrids is somewhere between the phenotypes of the two parental varieties
- In **codominance**, two dominant alleles affect the phenotype in separate, distinguishable ways

Figure 11.10

Incomplete Dominance in Snapdragon Color

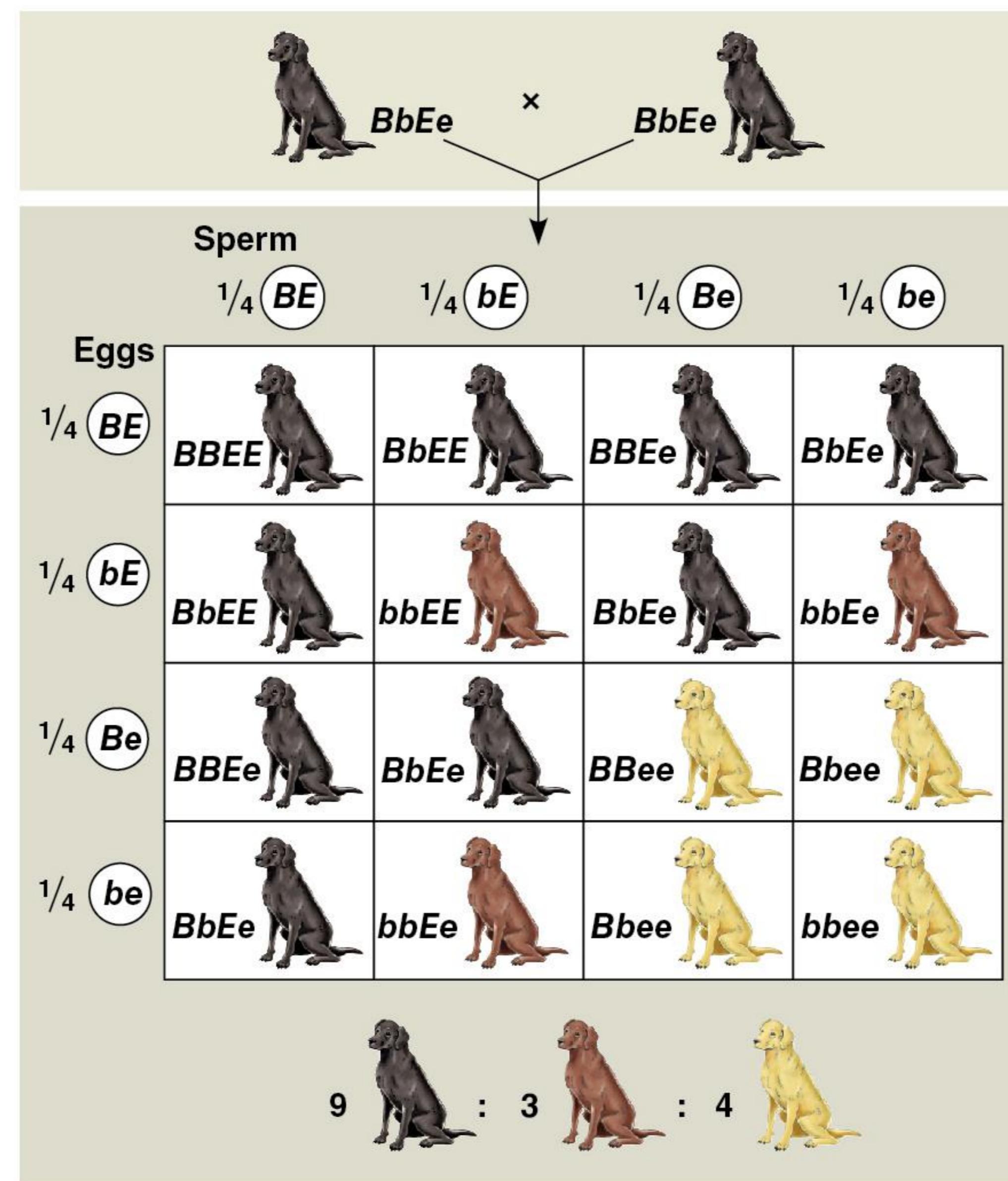


Epistasis

- In **epistasis**, a gene at one locus alters the phenotypic expression of a gene at a second locus
- For example, in Labrador retrievers and many other mammals, coat color depends on two genes
- One gene determines the pigment color (with alleles *B* for black and *b* for brown)
- The other gene (with alleles *C* for color and *c* for no color) determines whether the pigment will be deposited in the hair

Figure 11.12

An Example of Epistasis



Concept 11.4: Many Human Traits Follow Mendelian Patterns of Inheritance

- Humans are not good subjects for genetic research
 - Generation time is too long
 - Parents produce relatively few offspring
 - Breeding experiments would be unethical
- However, basic Mendelian genetics endures as the foundation of human genetics

Recessively Inherited Disorders

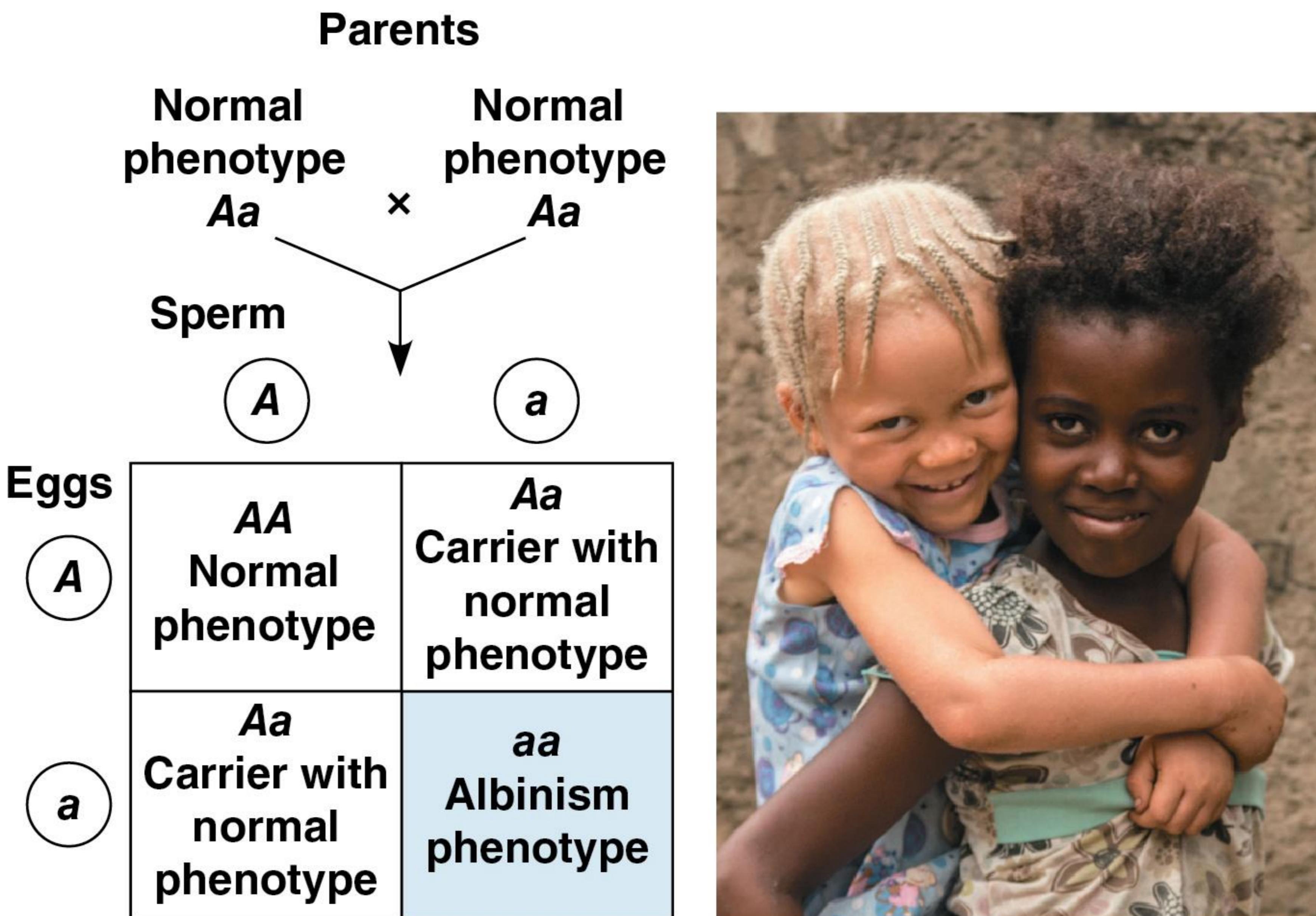
- Many genetic disorders are inherited in a recessive manner
- These range from relatively mild to life-threatening

The Behavior of Recessive Alleles (1 of 2)

- Recessively inherited disorders show up only in individuals homozygous for the allele
- **Carriers** are heterozygous individuals who carry the recessive allele but are phenotypically normal with regard to the disorder
- Most people who have recessive disorders are born to parents who are carriers of the disorder

Figure 11.15

Albinism: A Recessive Trait



Dominantly Inherited Disorders (1 of 2)

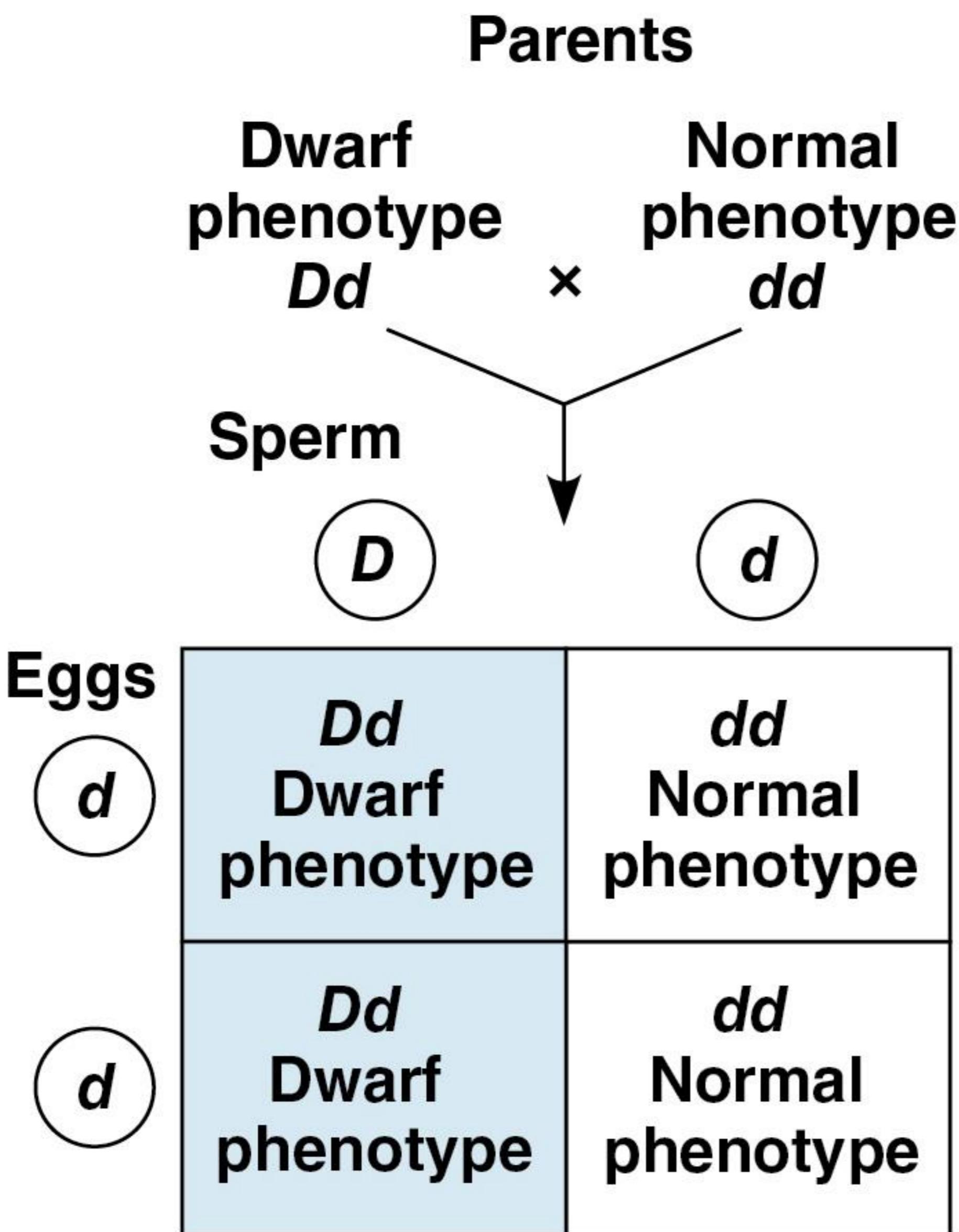
- A number of human disorders are caused by dominant alleles
- Dominant alleles that cause a lethal disease are rare and often cause the death of affected individuals before they can mature and reproduce
- A lethal dominant allele may be passed to the next generation if the lethal disease symptoms first appear after reproductive age

Dominantly Inherited Disorders (2 of 2)

- Achondroplasia is a form of dwarfism caused by a rare dominant allele
- **Huntington's disease** is a degenerative disease of the nervous system caused by a lethal dominant allele with no obvious phenotype apparent until the individual is about 35-45 years old
- Once the deterioration of the nervous system begins, the condition is irreversible and fatal

Figure 11.17

Achondroplasia: A Dominant Trait



Multifactorial Disorders

- Many diseases, such as heart disease, diabetes, cancer, alcoholism, and mental illnesses have both genetic and environmental components
- Lifestyle has a tremendous effect on phenotype for cardiovascular health and other multifactorial characters

Genetic Counseling Based on Mendelian Genetics

- Genetic counselors can provide information to prospective parents concerned about a family history for a specific disease
- Each child represents an independent event in the sense that its genotype is unaffected by the genotypes of older siblings
- Genetic counseling relies on the Mendelian model of inheritance

Copyright



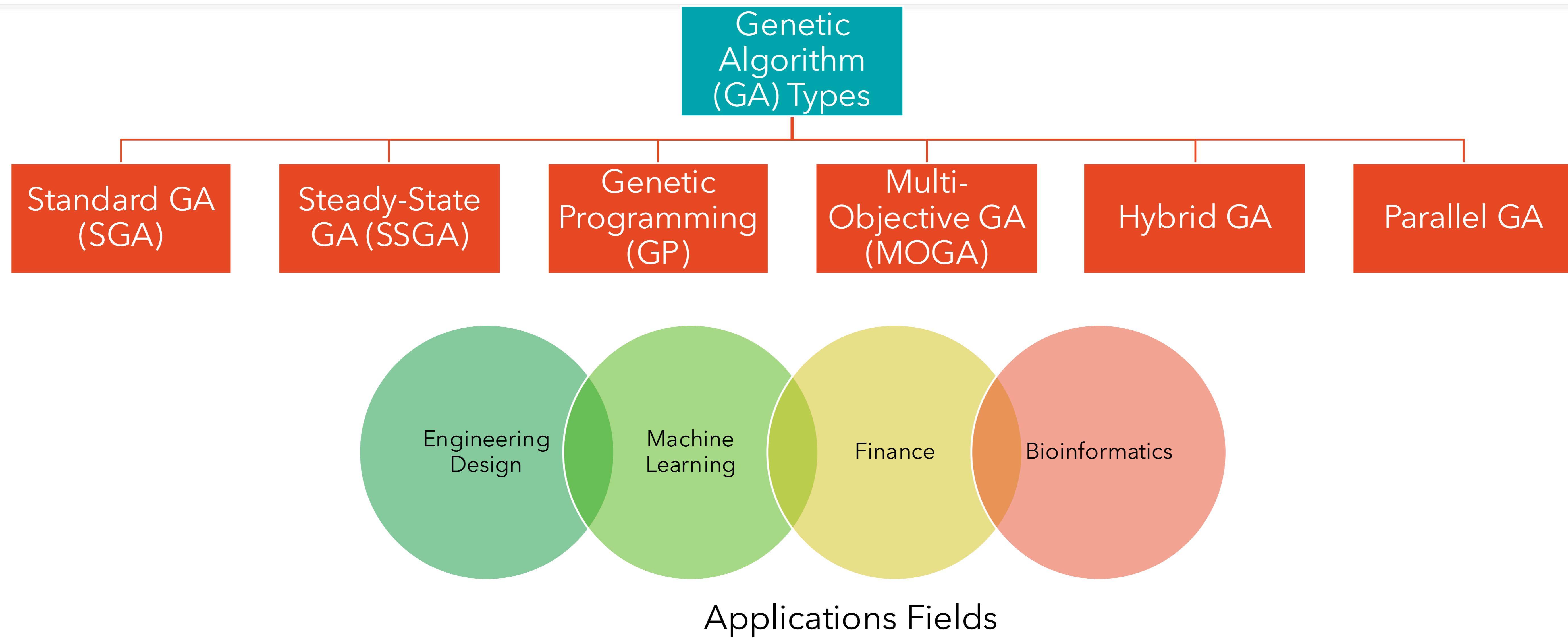
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Genetics Tools

Computing Tasks

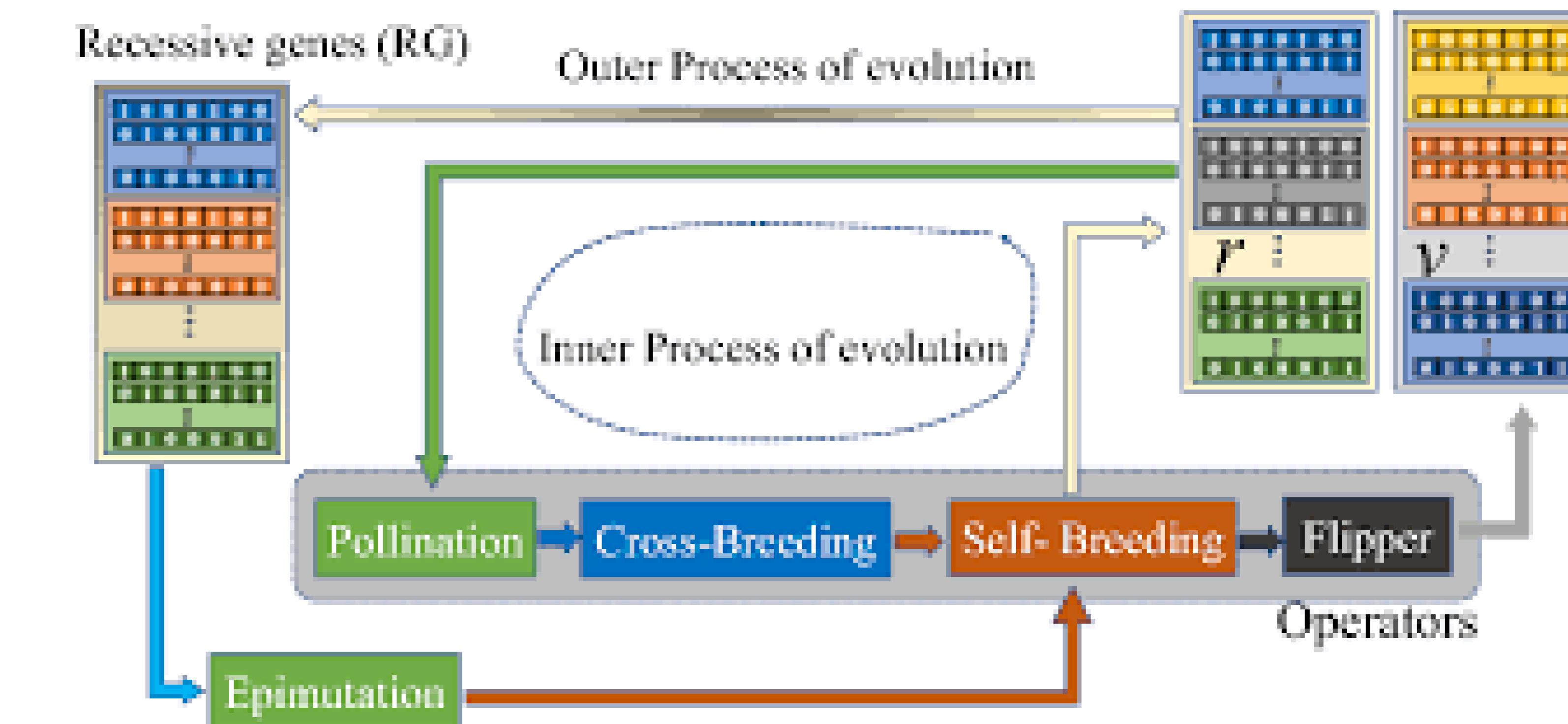
- Machine Learning for prediction/detection, analysis: Cell-Cycle Estimation
- Simulation and modeling: Cells Alive, CellProfiler, CompuCell3d
- Optimization: METO
- Bio-inspired Algorithms: GA

Genetic Algorithms



Mendelian Evolutionary Theory Optimization Algorithm (METO)

- A new algorithm inspired by plant genetics based on Mendel's inheritance laws to propose a genetically evolved evolutionary optimization algorithm.

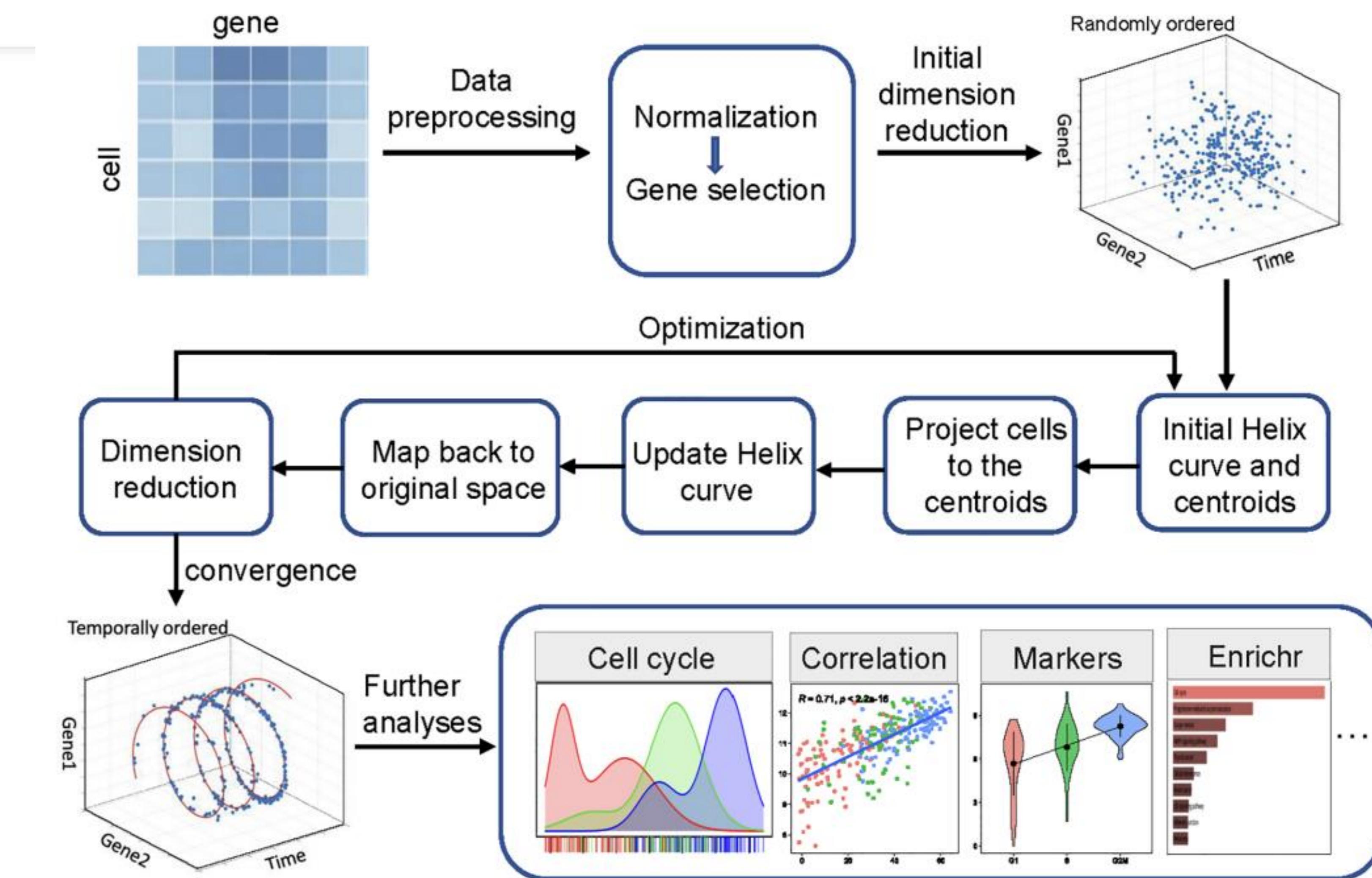


Cell-Cycle Estimation [1]

CCPE: Cell Cycle Pseudotime Estimation for Single Cell RNA-seq Data

A semi-supervised algorithm that uses 378 cell cycle genes to estimate gene expression and predict the position of cells in the cell cycle.

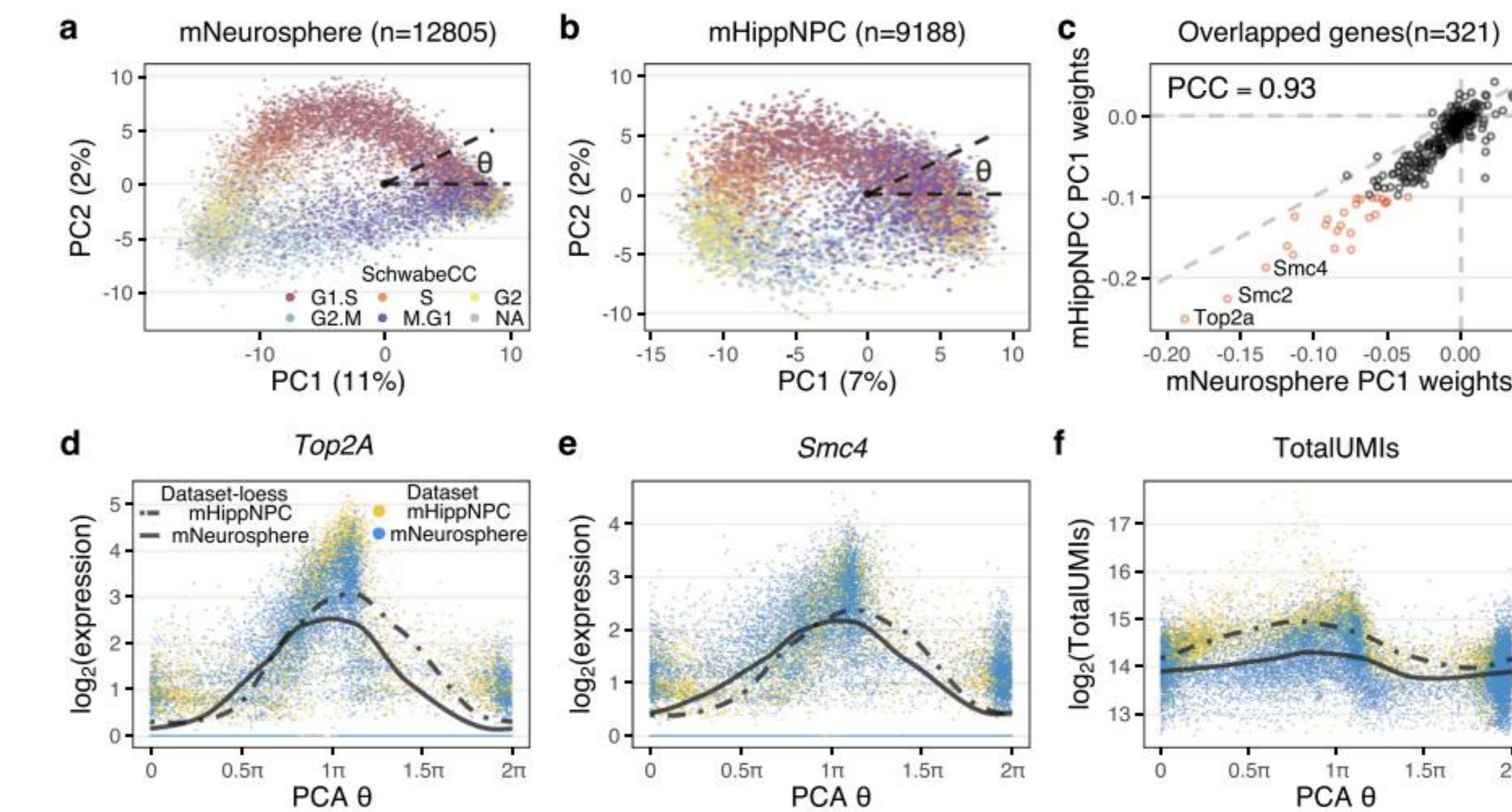
Estimation method: ANOVA



Cell-Cycle Estimation [2]

Universal Prediction of Cell-Cycle Position Using Transfer Learning

This study introduces 'tricycle', an R/Bioconductor package, which leverages key features of cell cycle biology, mathematical properties of Principal Component Analysis of periodic functions, and the use of transfer learning to predict position in the cell cycle from single-cell RNA-seq data.



The cell-cycle ellipsoid (above) and cell-cycle position (below)

Simulation Tools

- **Cell Cycle Simulation:** Using agent-based modeling to understand cell growth and division.

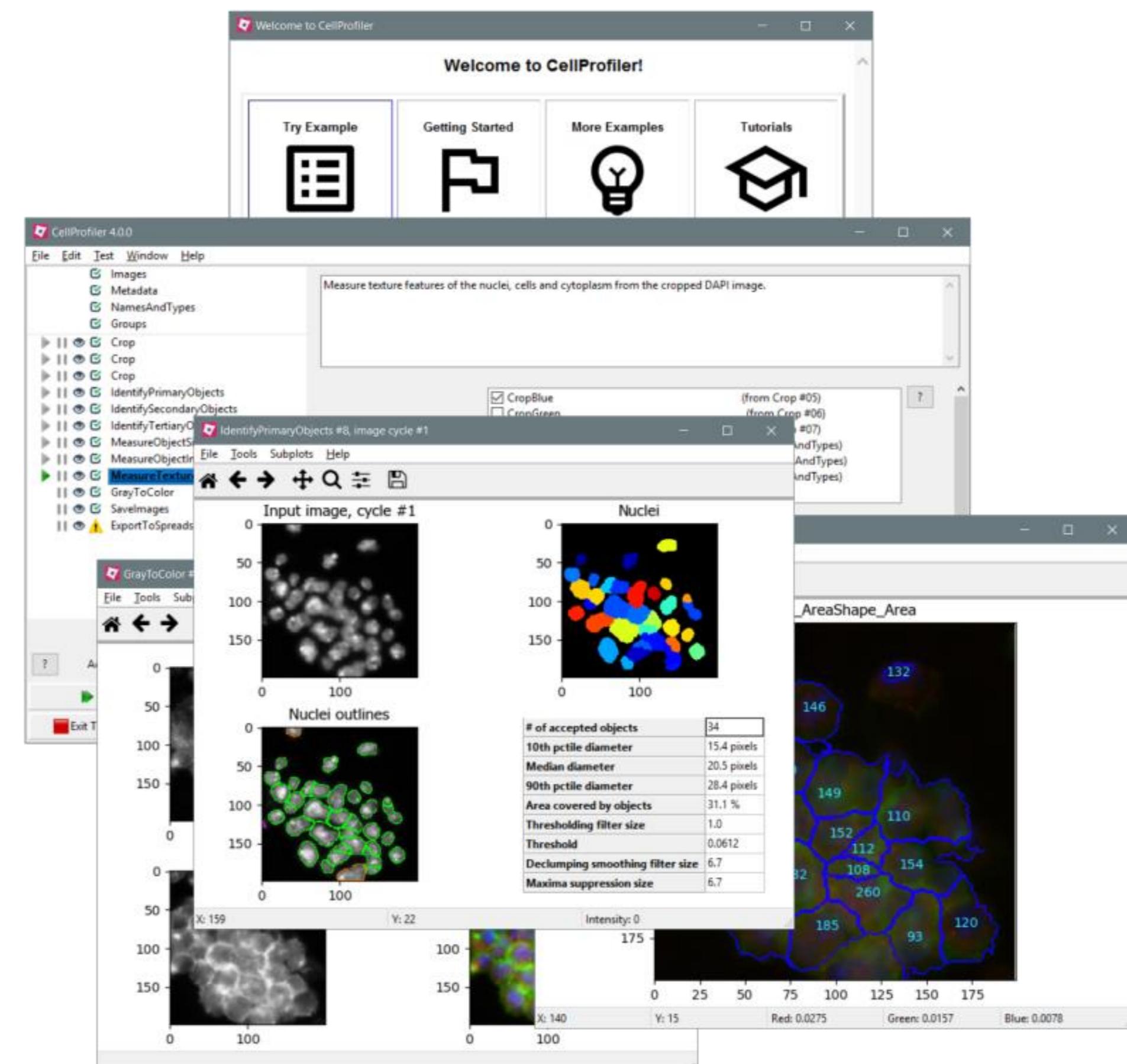
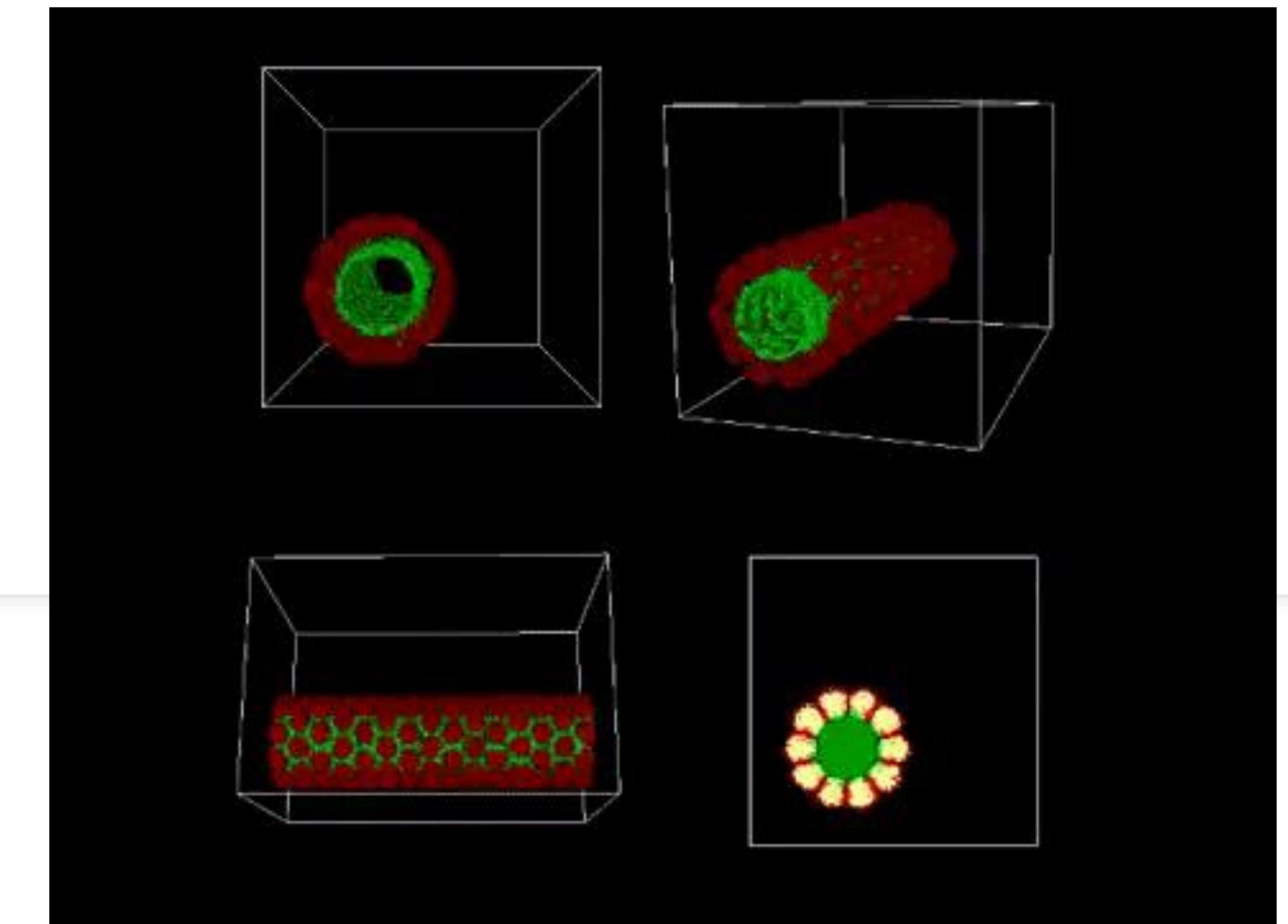
Tools: [CompuCell3D](#)

- **Cellular Abnormality Detection:** Deep learning to detect cancer cells through microscopic images.

Tools: [CellProfiler](#)

- **Cell Cycle Simulation:** Learn about cell cycle, meiosis, mitosis.

Tools: [CellsAlive](#)



Tool Exploration

Explore and understand topics: Cell Cycle, Meiosis & Sexual Life Cycle, Mendel & Gene, using at least one of the three simulation tools mentioned previously.

No need for submission.

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