BIG IDEA III

Living systems store, retrieve, transmit and respond to information essential to life processes.

Enduring Understanding 3.A

Heritable information provides for continuity of life.

Essential Knowledge 3.A.1

DNA, and in some cases RNA, is the primary source of heritable information.

PowerPoint® Lecture Presentations for

Biology

Eighth Edition

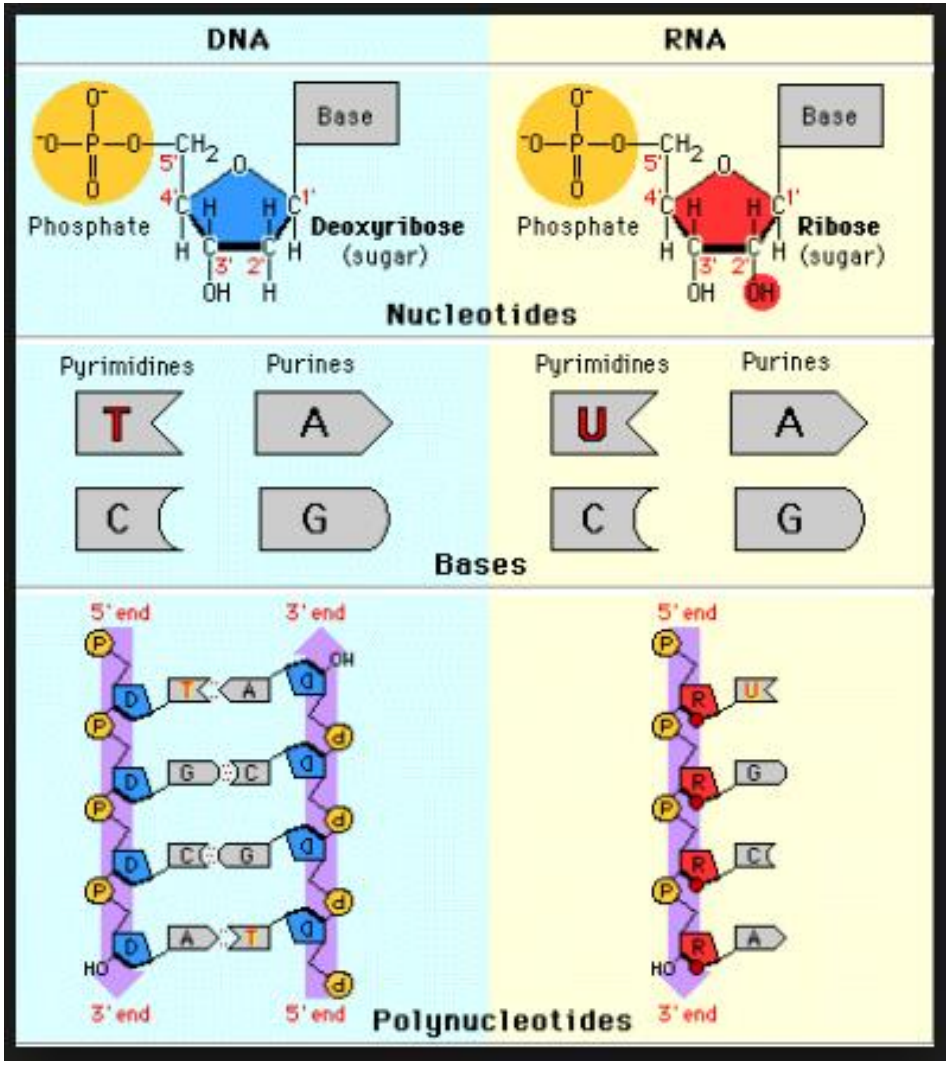
Neil Campbell and Jane Reece

Lectures by Chris Romero, updated by Erin Barley with contributions from Joan Sharp

Essential Knowledge 3.A.1: *DNA, and in some cases RNA, is the primary source of heritable information.*

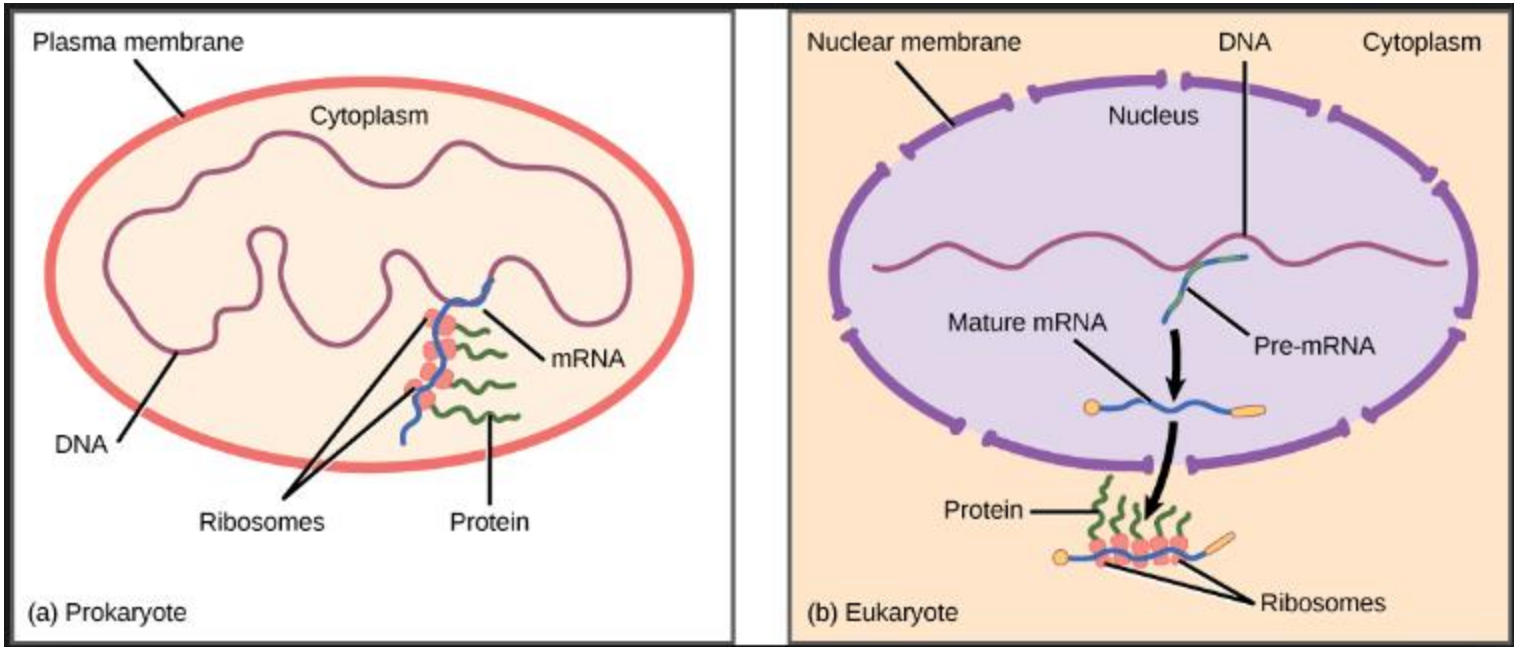
- **Learning Objectives:**
  - **(3.1)** The student is able to **construct scientific explanations** that use the structures and mechanisms of DNA and RNA to support the claim that DNA and, in some cases, that RNA are the primary sources of heritable information.
  - **(3.2)** The student is able to **justify the selection of data** from historical investigations that support the claim that DNA is the source of heritable information.
  - **(3.3)** The student is able to **describe representations and models** that illustrate how genetic information is copied for transmission between generations.
  - **(3.4)** The student is able to **describe representations and models** illustrating how genetic information is translated into polypeptides.
  - **(3.5)** The student can **justify the claim** that humans can manipulate heritable information by identifying *at least two* commonly used technologies.
  - **(3.6)** The student can **predict** how change in a specific DNA or RNA sequence can result in changes in gene expression.

Genetic information is stored and transmitted from one generation to the next through DNA or RNA.



Different types of organisms have different chromosome structure.

- Prokaryotic organisms have *circular chromosomes*, while eukaryotic organisms have *multiple linear chromosomes*, although in biology there are exceptions to this rule.



The proof that DNA is the carrier of genetic information involved a number of important historical experiments.

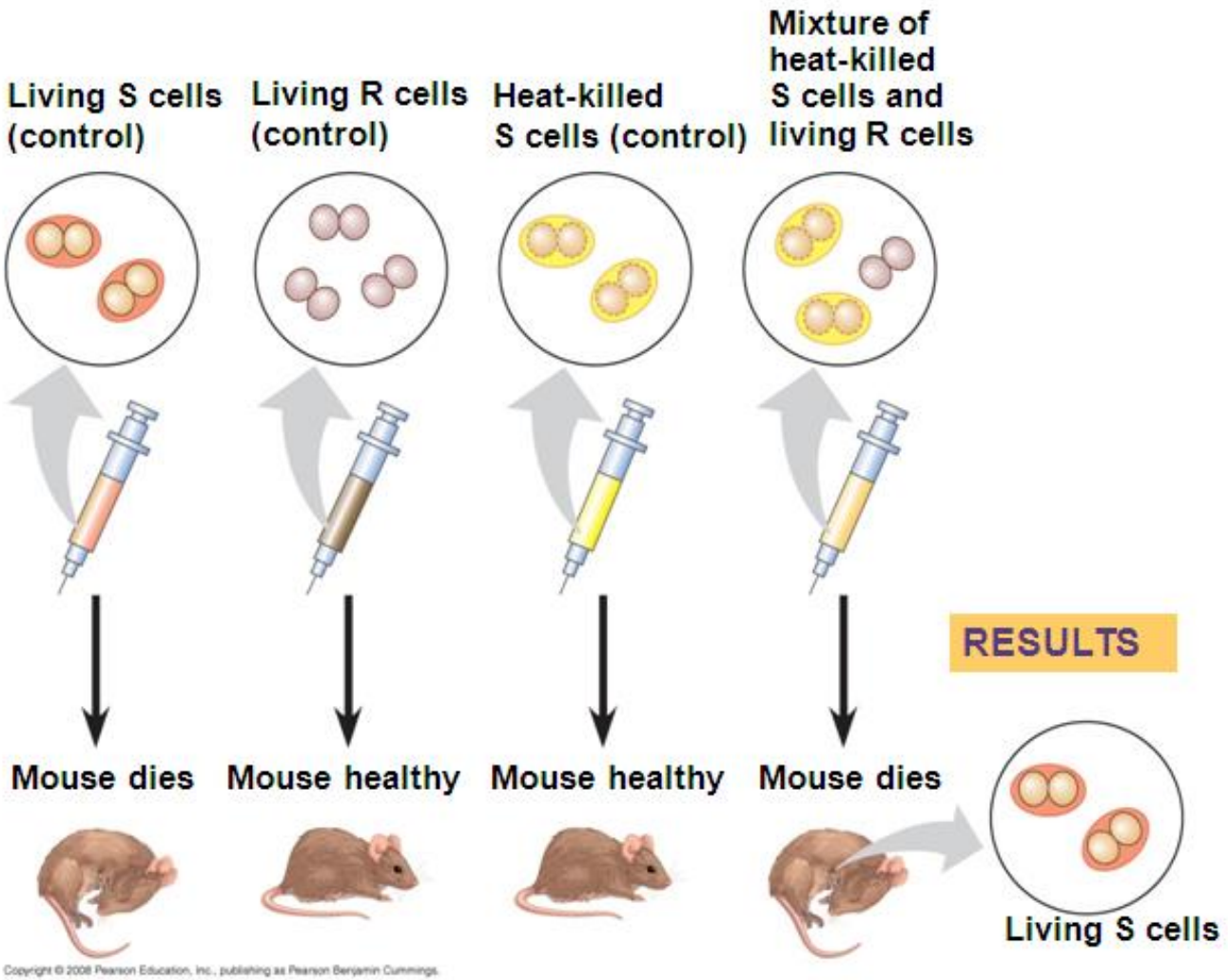
- Notable experiments include:
  1. Griffith's experiments on transforming bacteria
  2. Avery-McLeod-McCarty experiments
  3. Hershey-Chase experiment
  4. Contributions of Watson, Crick, Wilkins, and Franklin on the structure of DNA

The Search for the Genetic Material

- Early in the 20th century, the identification of the molecules of inheritance loomed as a major challenge to biologists.
  - When **T. H. Morgan's** group showed that genes are located on chromosomes, the two components of chromosomes— *DNA and protein* —became ***candidates*** for the genetic material.
  - The role of DNA in heredity was first discovered by studying bacteria and the viruses that infect them.

Figure 16.2: Evidence that Bacteria Can Be Transformed

http://nortonbooks.com/college/biology/animations/ch12a01.htm



Avery, McCarty and MacLeod Experiments

http://www.sinauer.com/cooper5e/animation0401.html

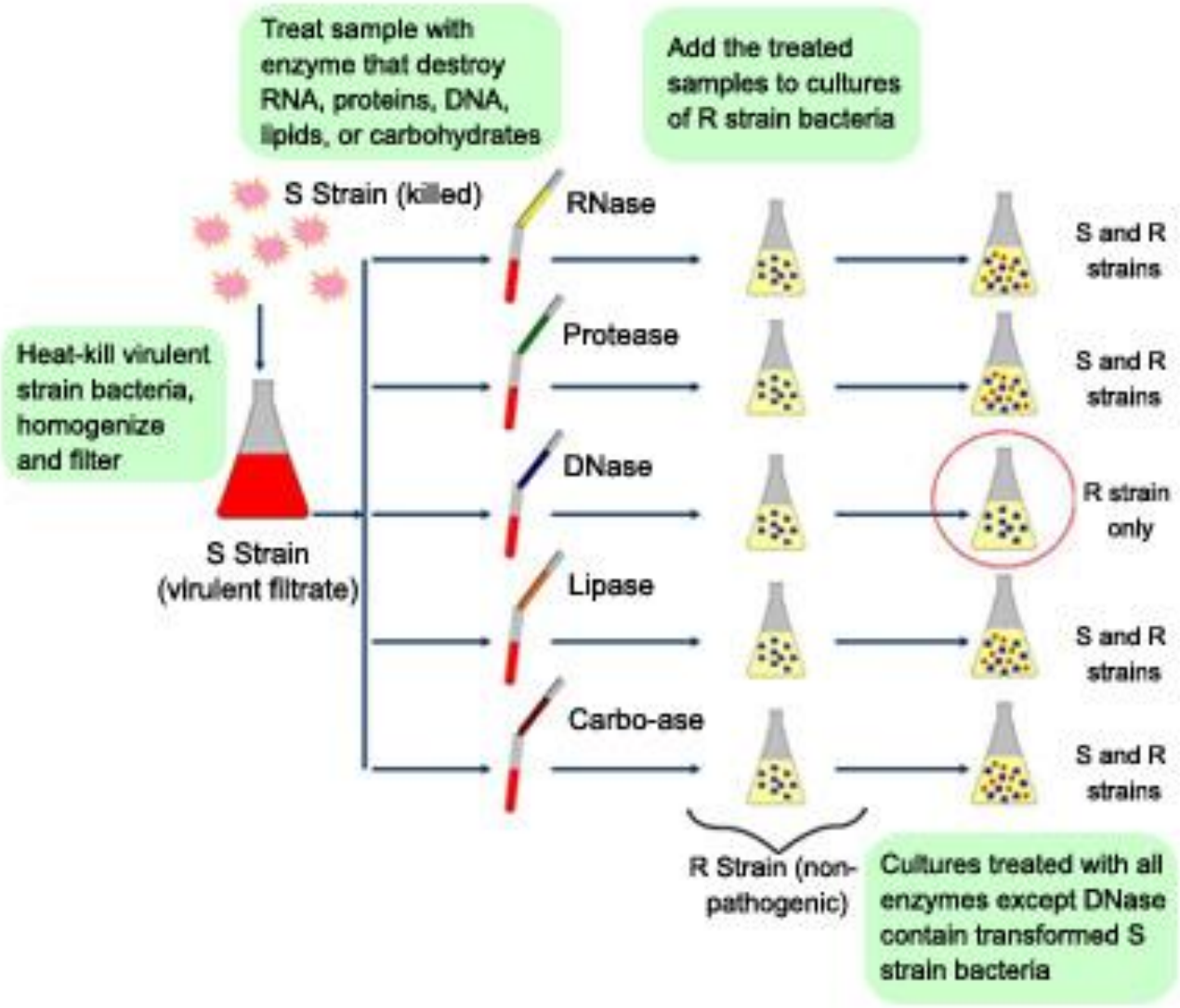
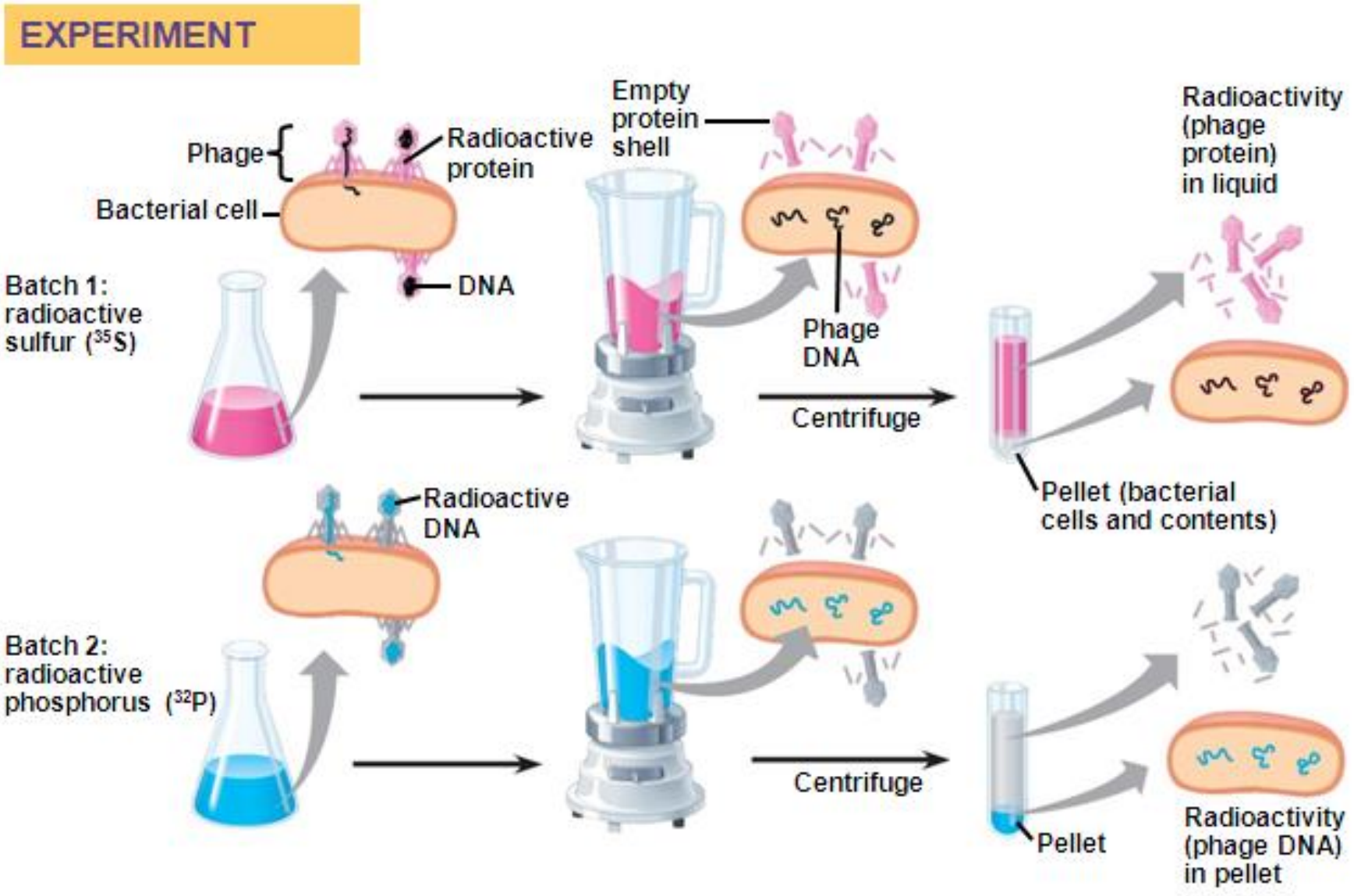


Fig. 16-4: The Hershey Chase Experiment

http://nortonbooks.com/college/biology/animations/ch12a02.htm



Additional Evidence that DNA is the Genetic Material of Cells

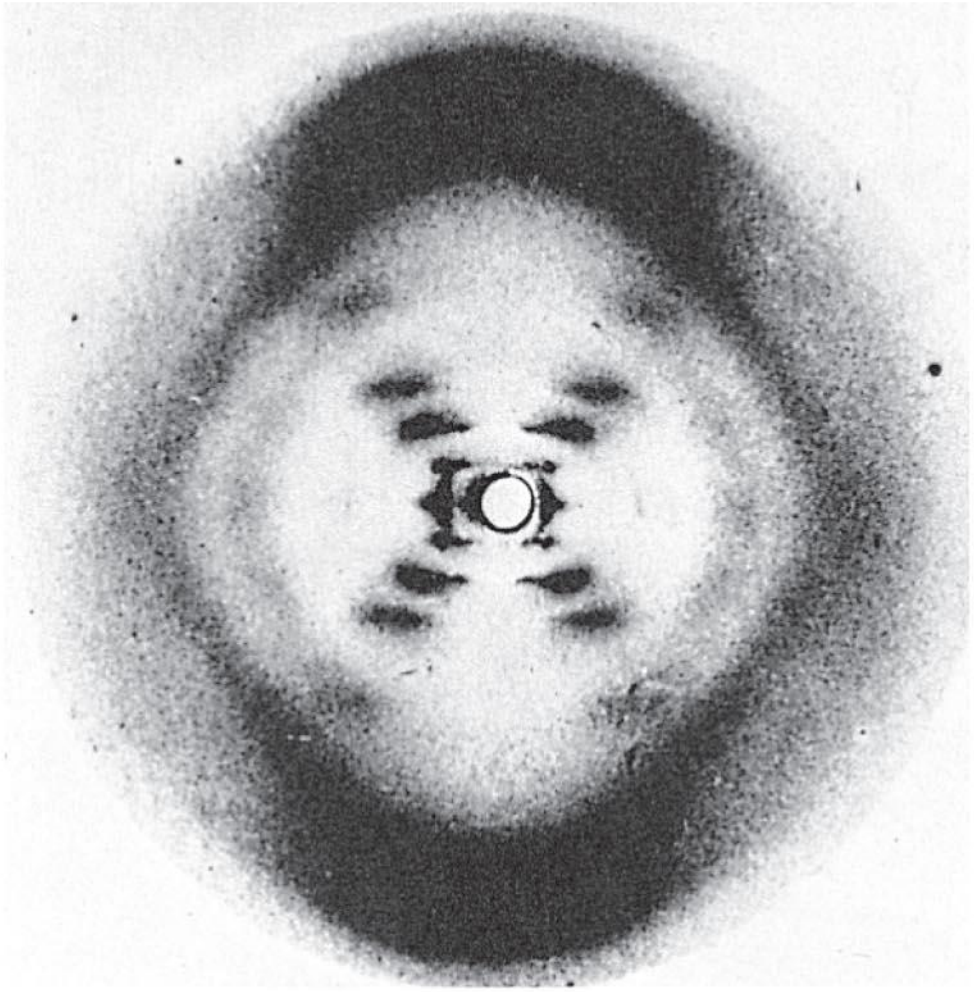
- Further evidence that DNA is the genetic material of cells came from the laboratory of biochemist **Erwin Chargaff**.
- It was already known that DNA is a polymer of nucleotides, each consisting of a nitrogenous base, a sugar, and a phosphate group.
- In 1950, Chargaff reported that DNA composition varies from one species to the next after analyzing the base composition of DNA from a number of different organisms.
- This evidence of diversity made DNA a more credible candidate for the genetic material.
- Chargaff also noticed a peculiar regularity in the ratios of nucleotide bases within a single species.
- *Chargaff's rules* state that in any species there is an equal number of A and T bases, and an equal number of G and C bases. The basis for these rules, however, remained unexplained until the discovery of the double helix structure of DNA.

Building a Structural Model of DNA

- After most biologists became convinced that DNA was the genetic material, the challenge was to determine how its structure accounts for its role.
- **Maurice Wilkins** and **Rosalind Franklin** were using a technique called X-ray crystallography to study molecular structure.
- Franklin produced a picture of the DNA molecule using this technique.

Fig. 16-6

(a) Rosalind Franklin



(b) Franklin's X-ray diffraction photograph of DNA

Fig. 16-1

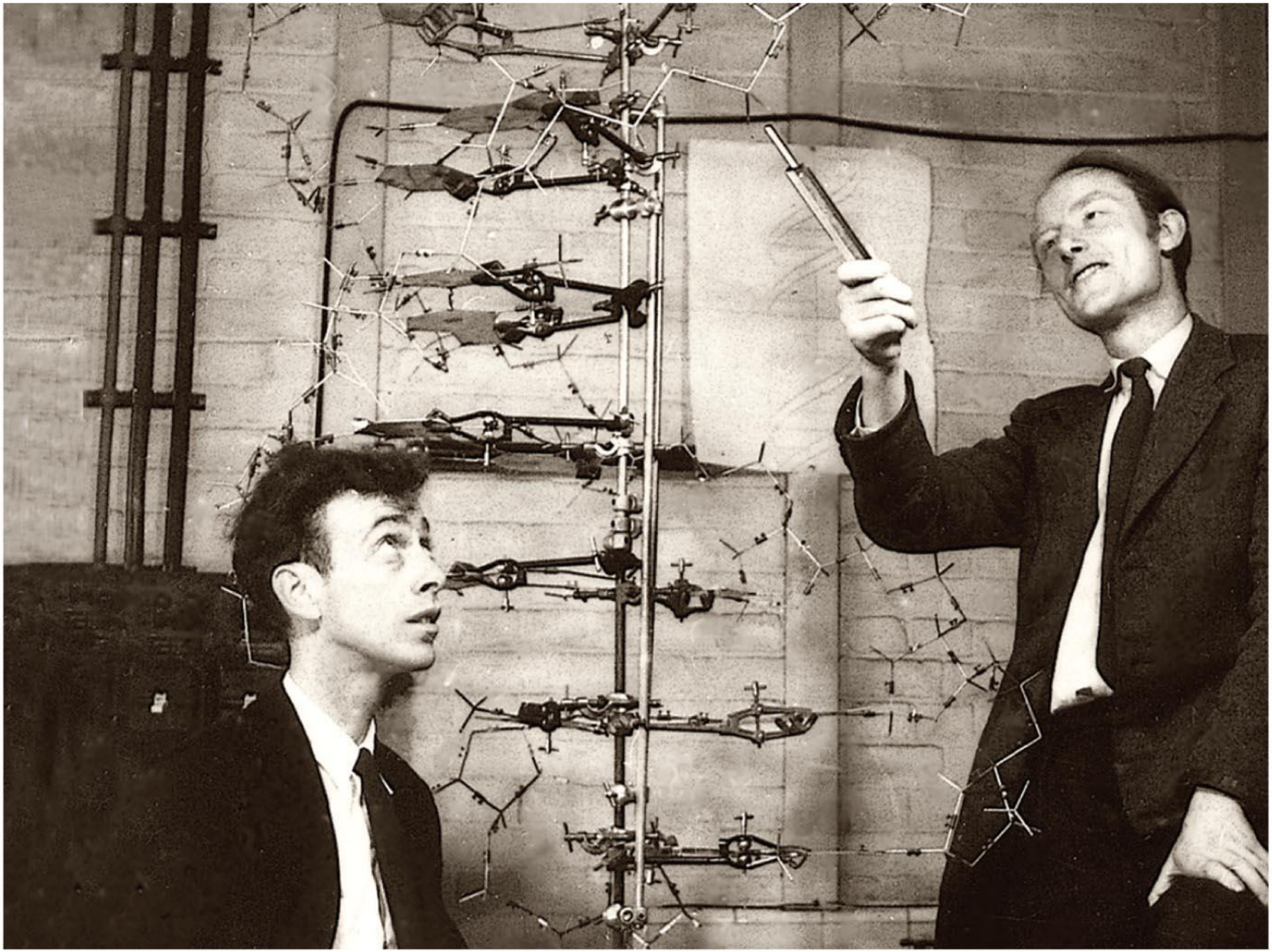
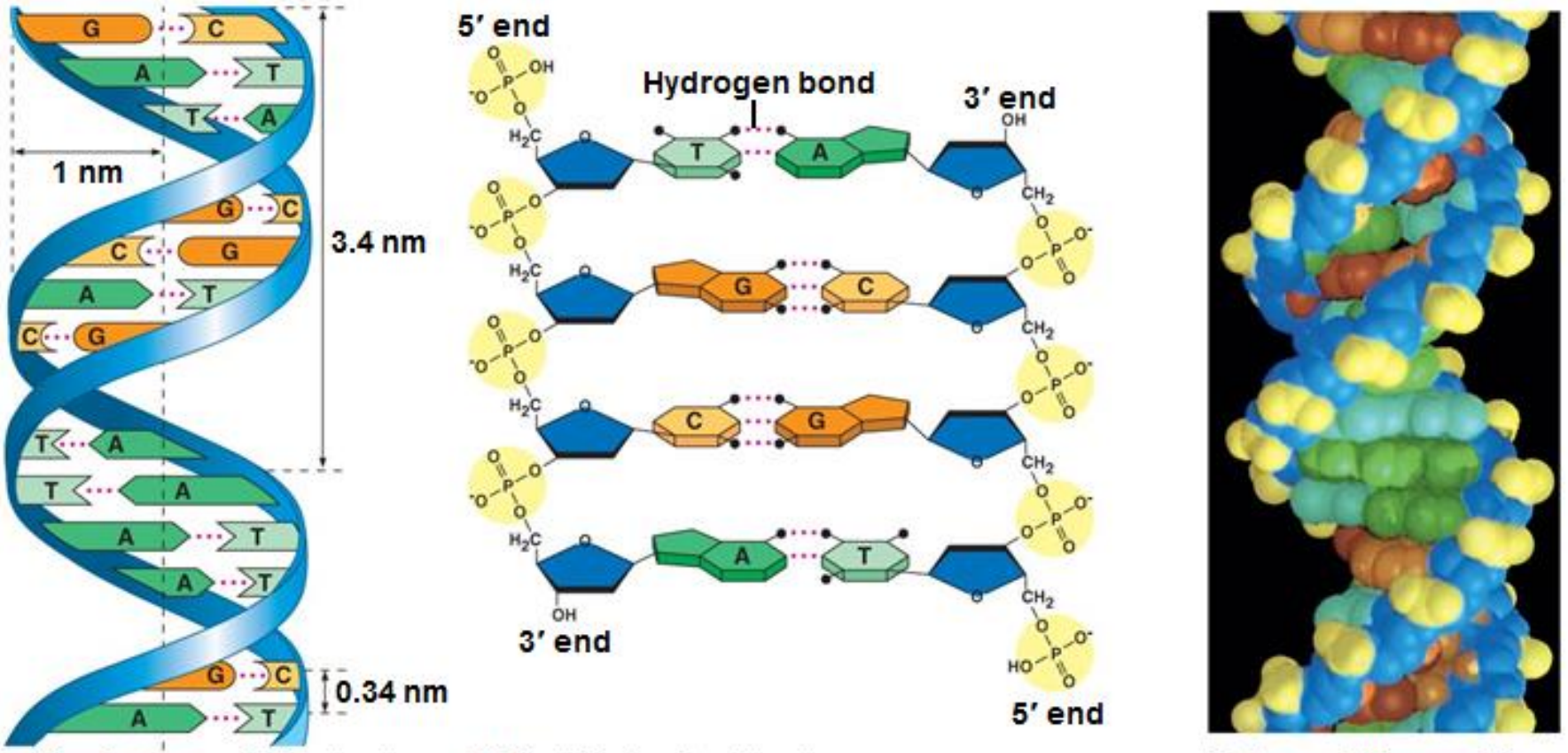
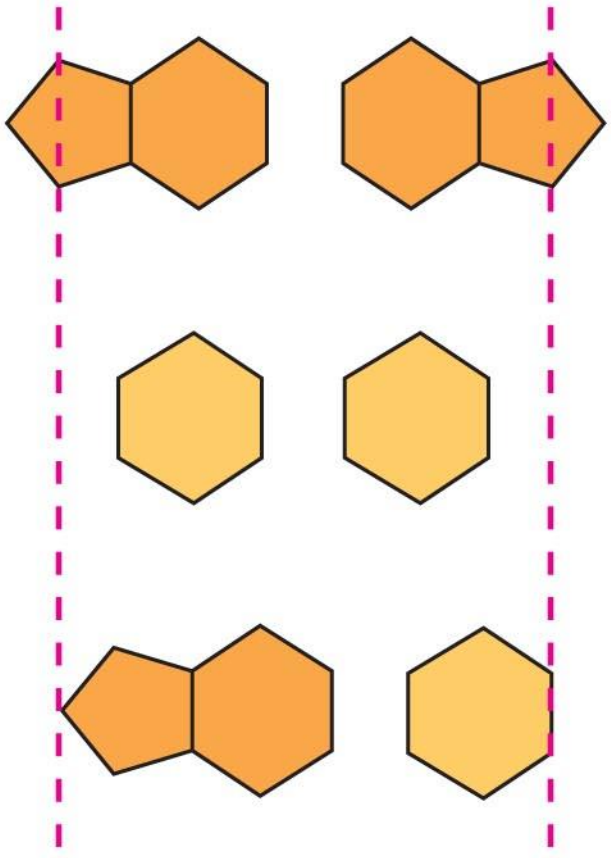


Figure 16.7: The Double Helix



How was the Structure of DNA Determined?

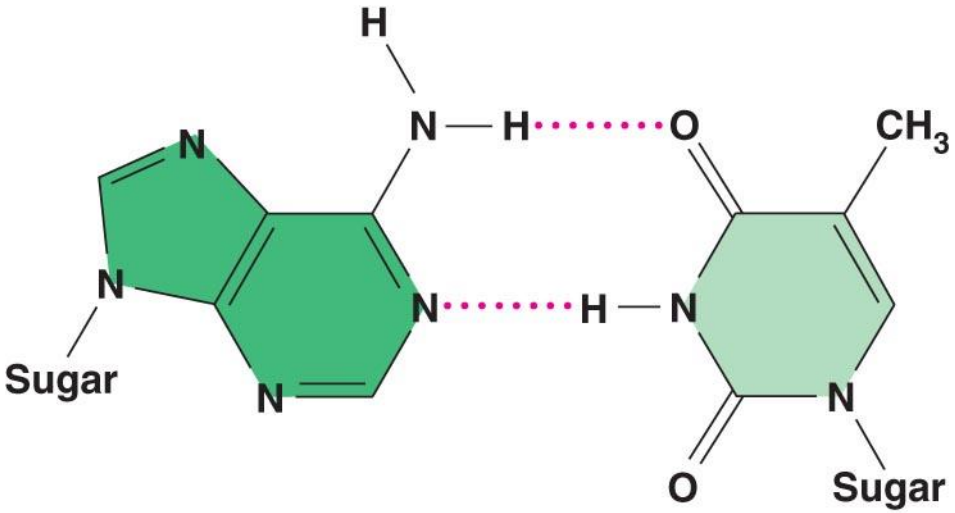


Purine + purine: too wide

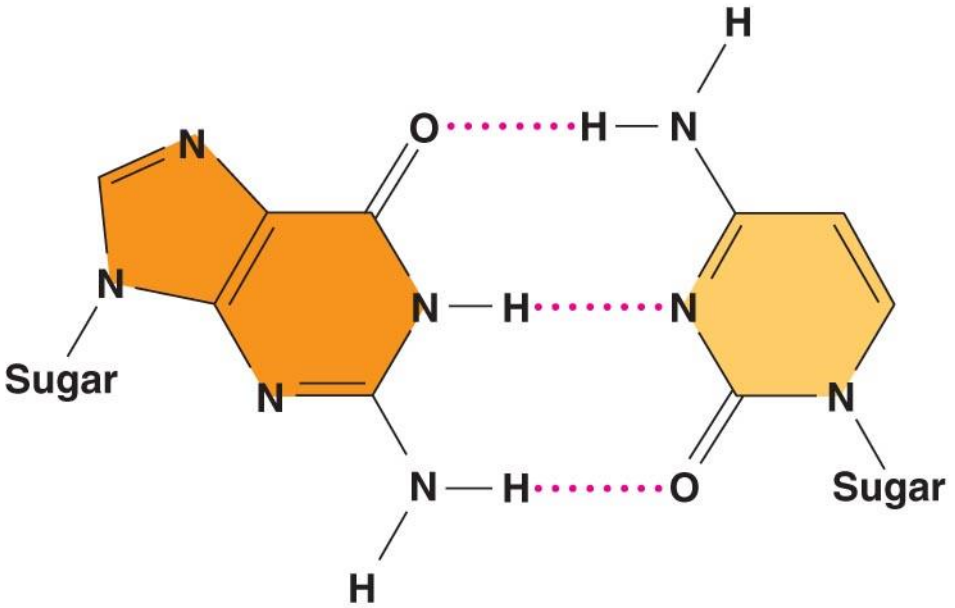
Pyrimidine + pyrimidine: too narrow

Purine + pyrimidine: width consistent with X-ray data

Fig. 16-8



Adenine (A) **Thymine (T)**



Guanine (G) **Cytosine (C)**

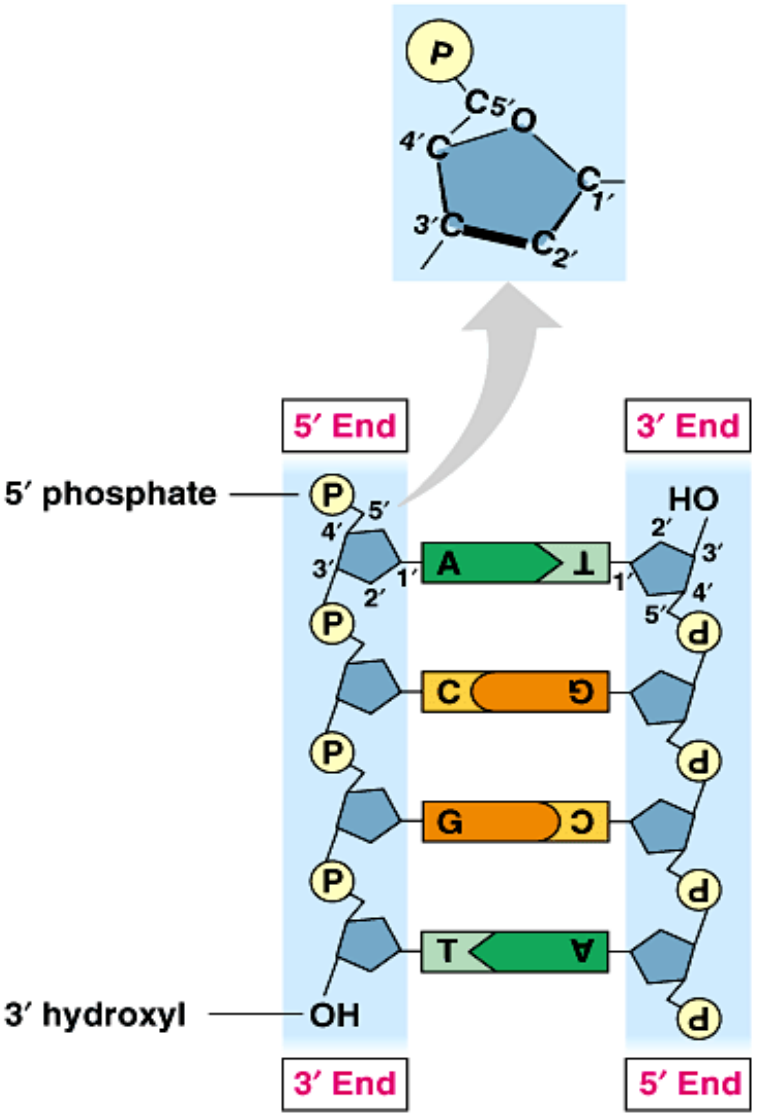
Figure 16.12 Antiparallel Arrangement of DNA Helix

The two DNA strands are *ANTIPARALLEL* – that is, their sugar-phosphate backbones run in opposite directions.

The 5’ → 3’ direction of one strand runs counter to the 5’ → 3’ direction of the other strand.

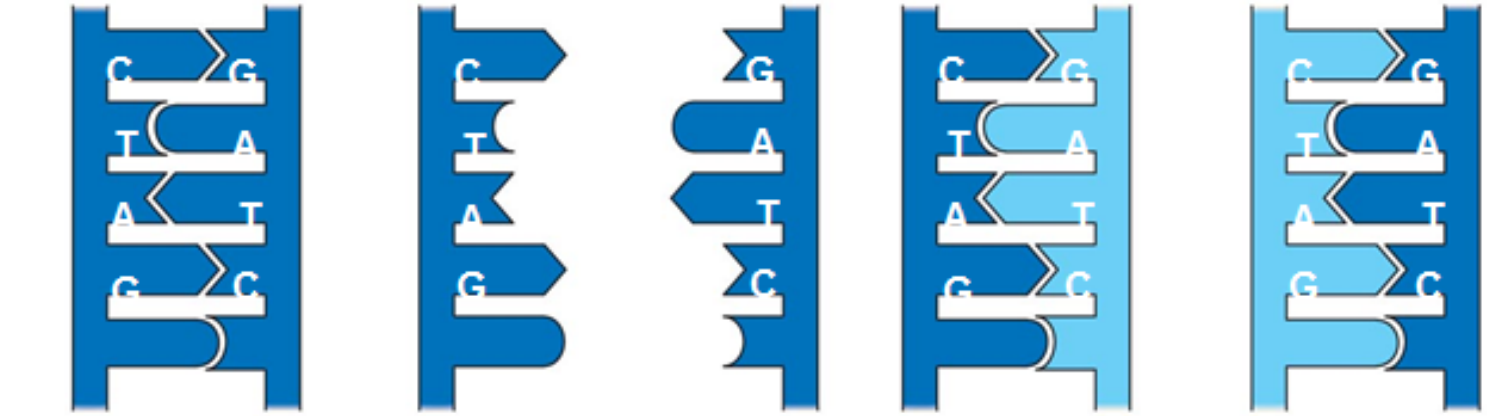
Notice in the figure that a nucleotide’s phosphate group is attached to the 5’ carbon of deoxyribose.

Notice also that the phosphate group of one nucleotide is joined to the 3’ carbon of the adjacent nucleotide.



DNA replication ensures continuity of hereditary information.

- Replication is a ***semiconservative process***; that is, one strand serves as the template for a new, ***complementary strand***.
- Replication requires DNA polymerase plus many other ***essential cellular enzymes***, occurs ***bidirectionally***, and differs in the production of the ***leading and lagging strands***.



(a) Parent molecule

(b) Separation of strands

(c) "Daughter" DNA molecules, each consisting of one parental strand and one new strand

Table 16.1 Bacterial DNA Replication Proteins and Their Functions

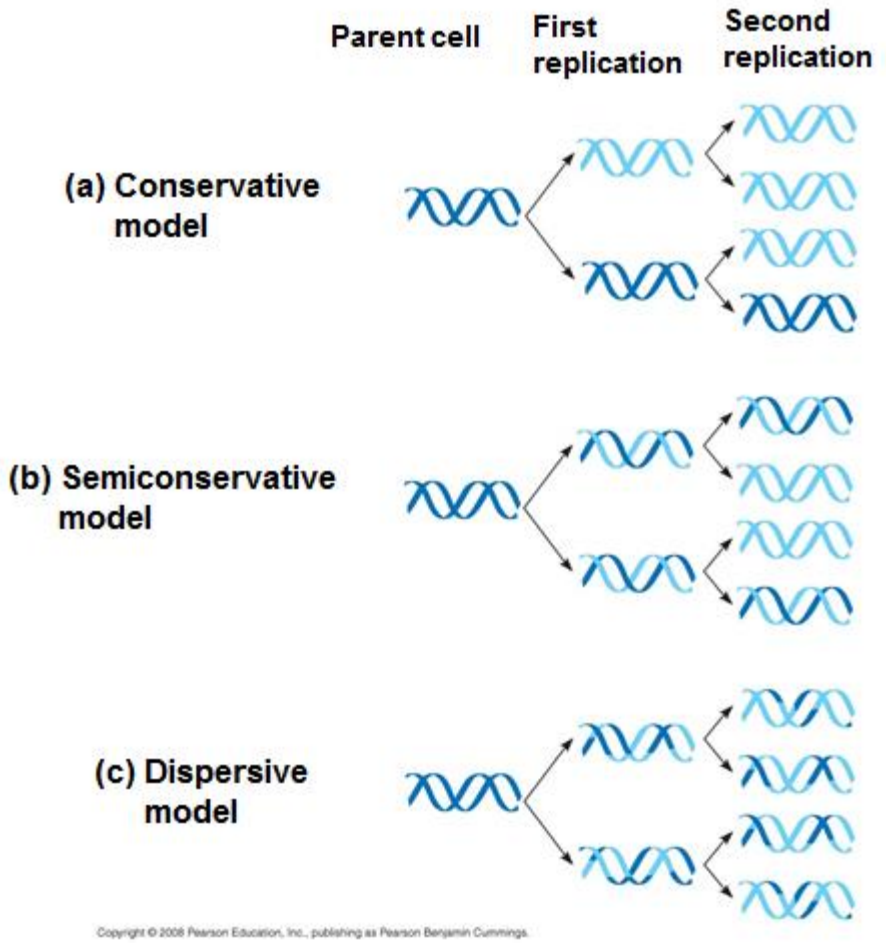
Protein	Function
Helicase	Unwinds parental double helix at replication forks
Single-strand binding protein	Binds to and stabilizes single-stranded DNA until it can be used as a template
Topoisomerase	Relieves "overwinding" strain ahead of replication forks by breaking, swiveling, and rejoining DNA strands
Primase	Synthesizes an RNA primer at 5′ end of leading strand and of each Okazaki fragment ofl agging strand
DNA pol III	Using parental DNA as a template, synthesizes new DNA strand by covalently adding nucleotides to the 3′ end of a pre-existing DNA strand or RNA primer
DNA pol I	Removes RNA nucleotides of primer from 5′ end and replaces them with DNA nucleotides
DNA ligase	Joins 3′ end of DNA that replaces primer to rest of leading strand and joins Okazaki fragments of lagging strand

The Basic Principle: Base Pairing to a Template Strand

- The relationship between structure and function is manifest in the double helix:
  - Watson and Crick noted that the specific base pairing suggested a possible copying mechanism for genetic material.
  - Since the two strands of DNA are *complementary*, each strand acts as a *template* for building a new strand in replication.
  - In DNA replication, the parent molecule unwinds, and two new daughter strands are built following the *rules of complimentary base pairing*.

Figure 16.9: Replication is a Semi-Conservative Process

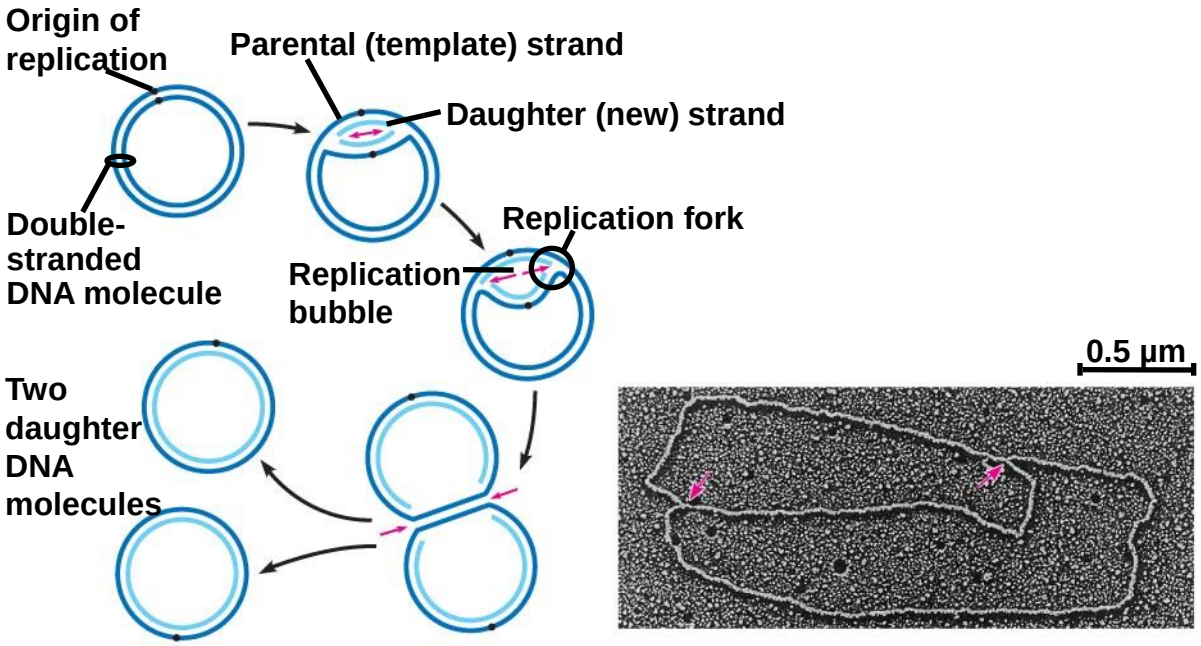
http://www.wiley.com/college/pratt/0471393878/student/animations/dna_replication/index.html



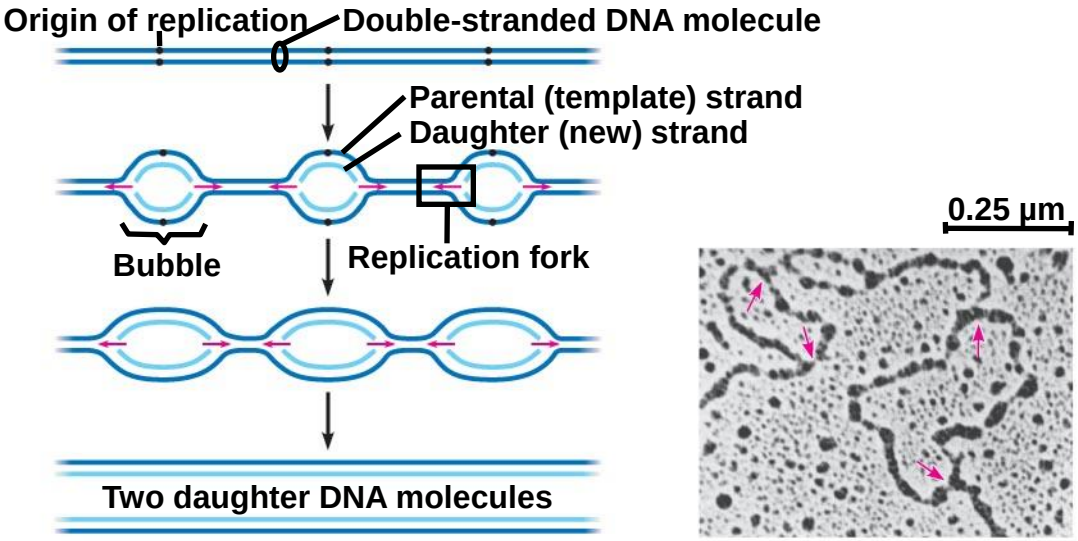
DNA Replication: *A Closer Look*

http://highered.mcgraw-hill.com/sites/0072943696/student_view0/chapter3/animation__dna_replication__quiz_1_.html

- Replication begins at special sites called *origins of replication*, where the two DNA strands are separated, opening up a *replication "bubble"*.
- A eukaryotic chromosome may have hundreds or even thousands of origins of replication.
- Replication proceeds in both directions from each origin, until the entire molecule is copied.



(a) Origins of replication in *E. coli*



(b) Origins of replication in eukaryotes

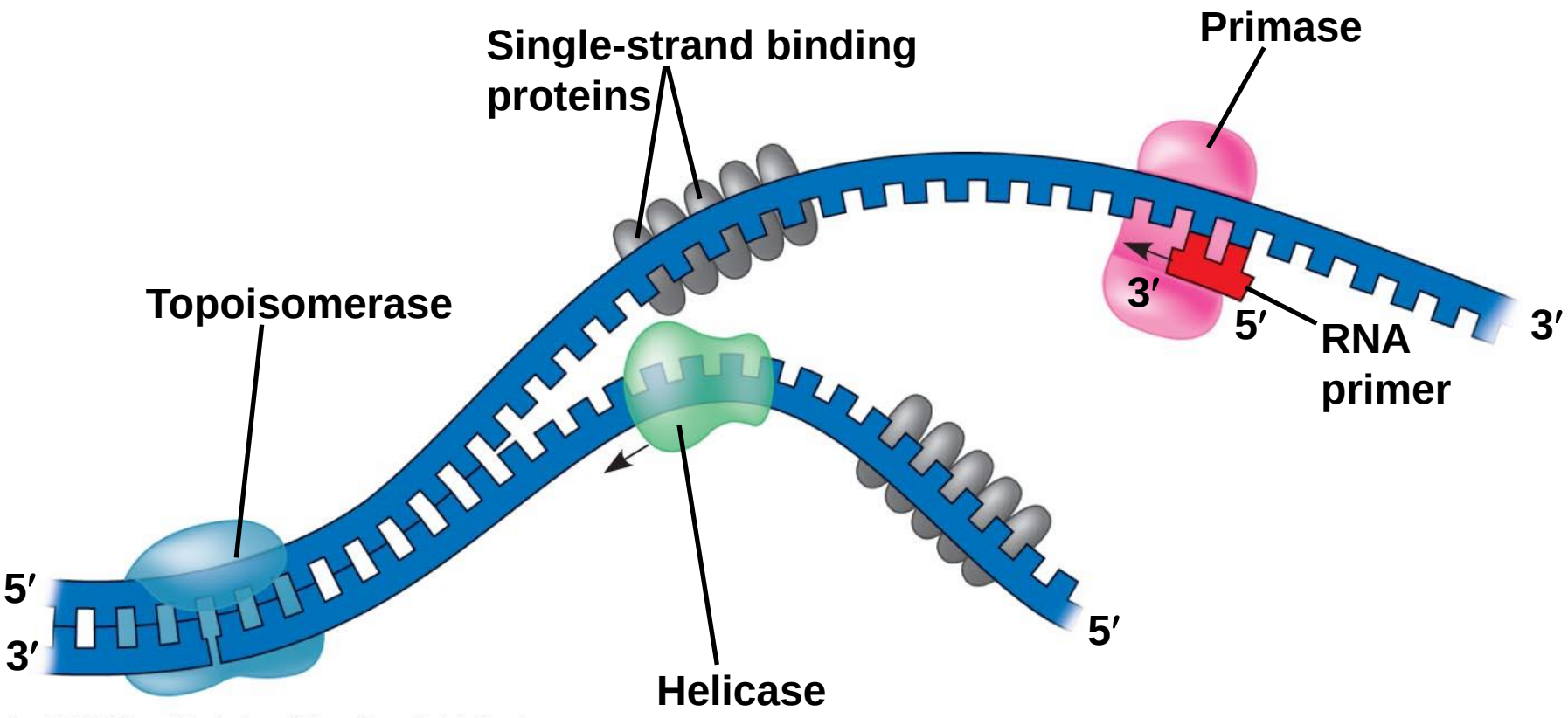
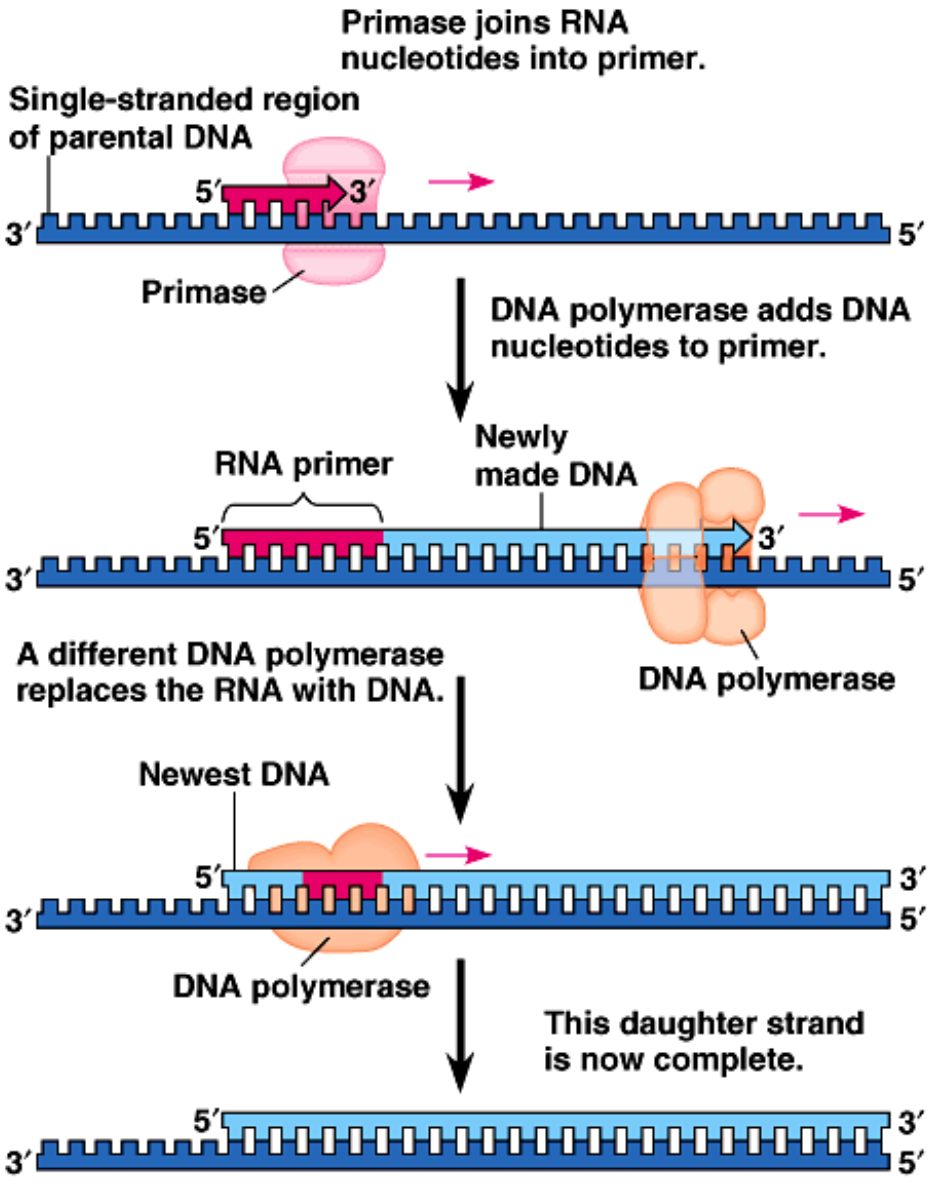


Figure 16.14 Priming DNA synthesis with RNA

http://bcs.whfreeman.com/thelifewire/content/chp11/1102002.html



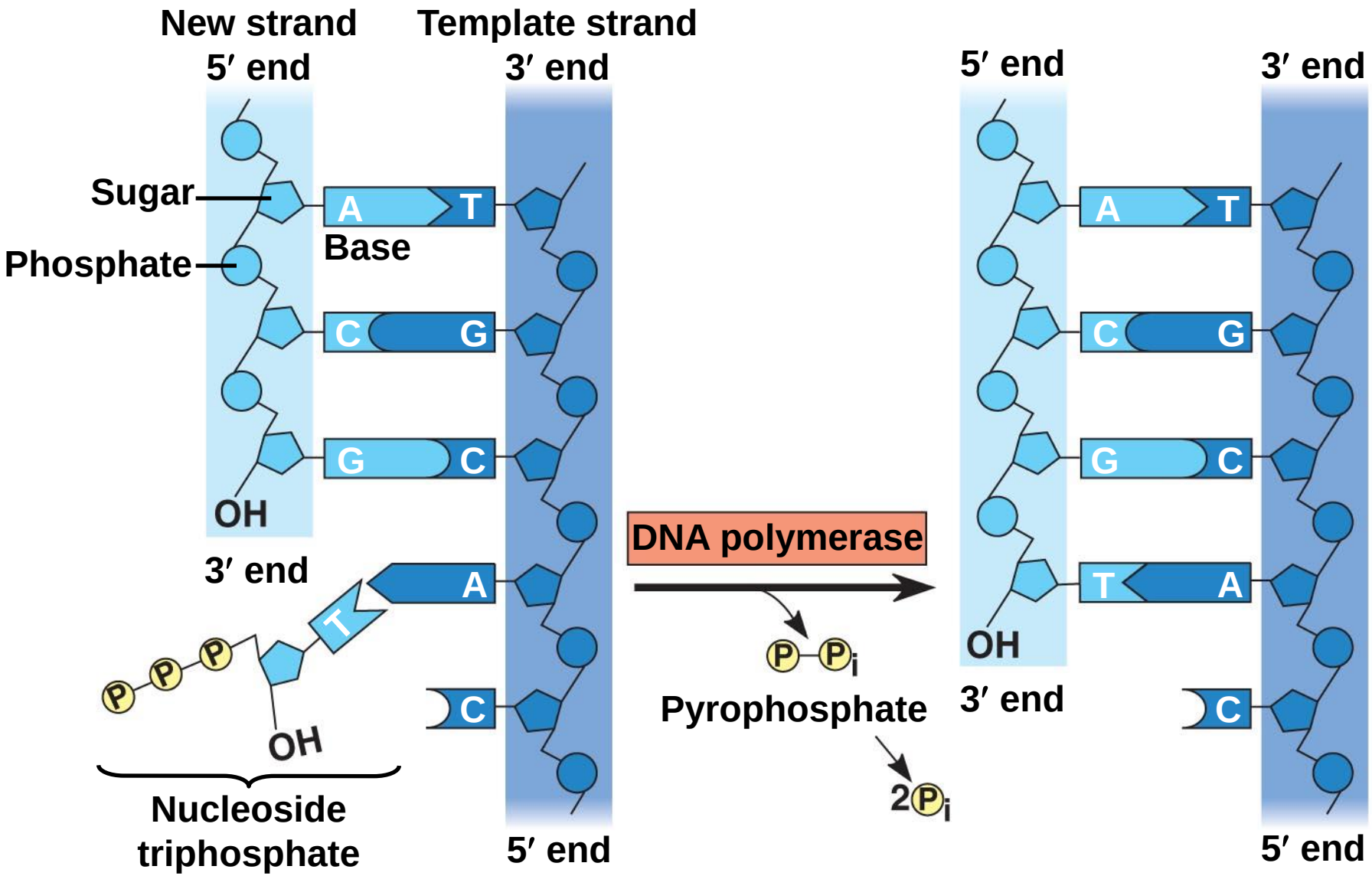
DNA polymerase cannot initiate a polynucleotide strand; it can only add to the 3’ end of an already-started strand.

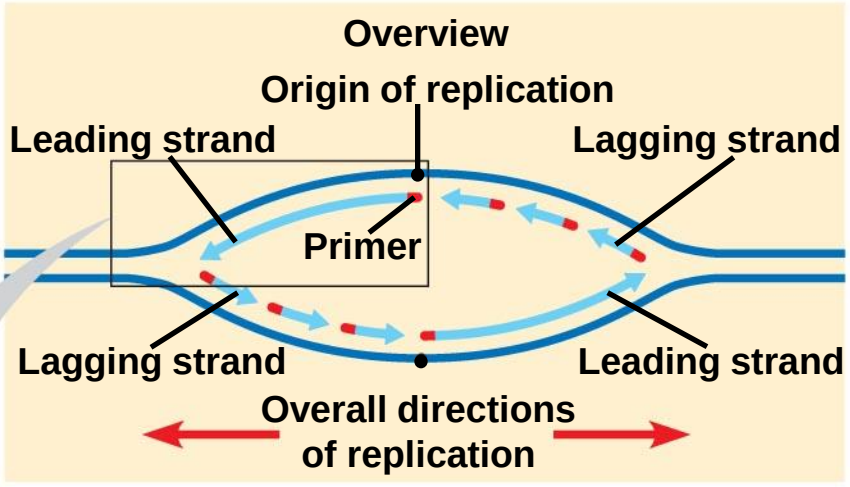
The initial nucleotide strand is s short *RNA Primer*. The primer is a short segment of RNA synthesized by the enzyme *primase*.

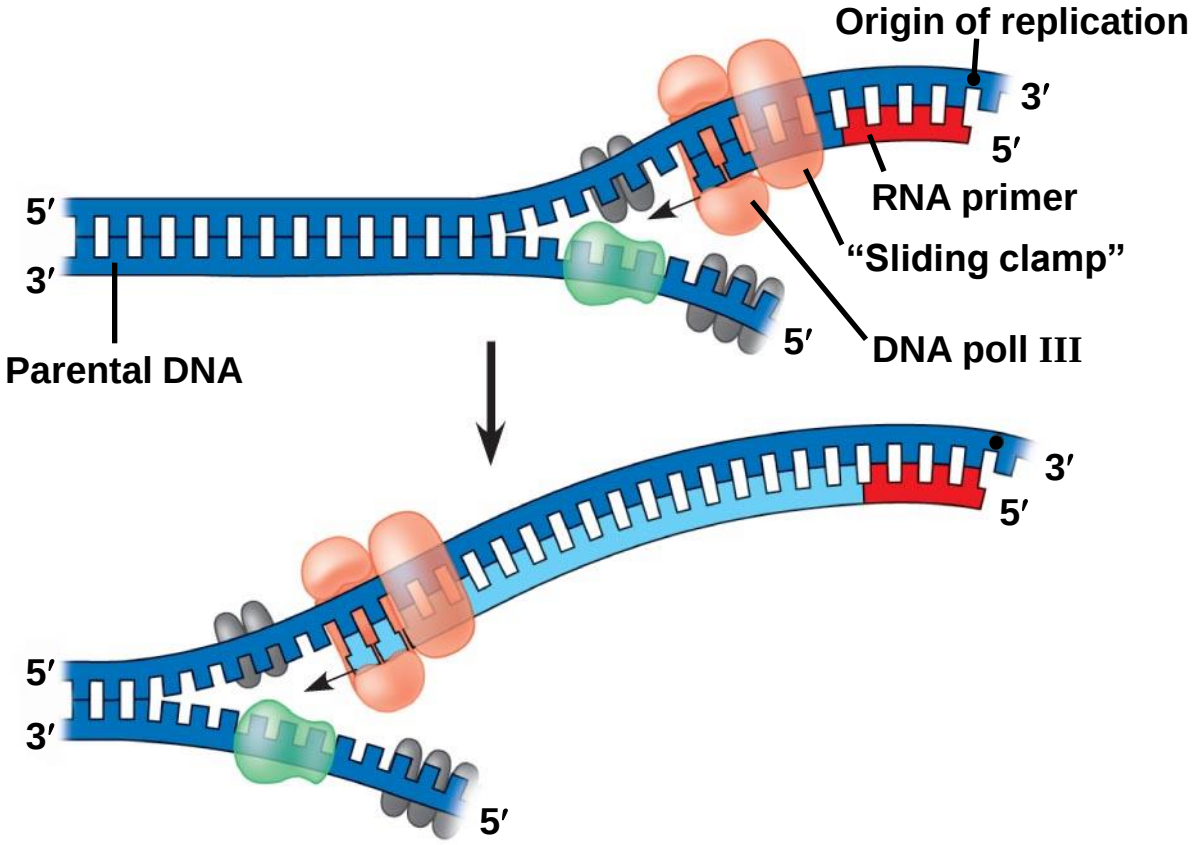
Primase can start an RNA strand from scratch and adds RNA nucleotides one at a time using the parental DNA as a template.

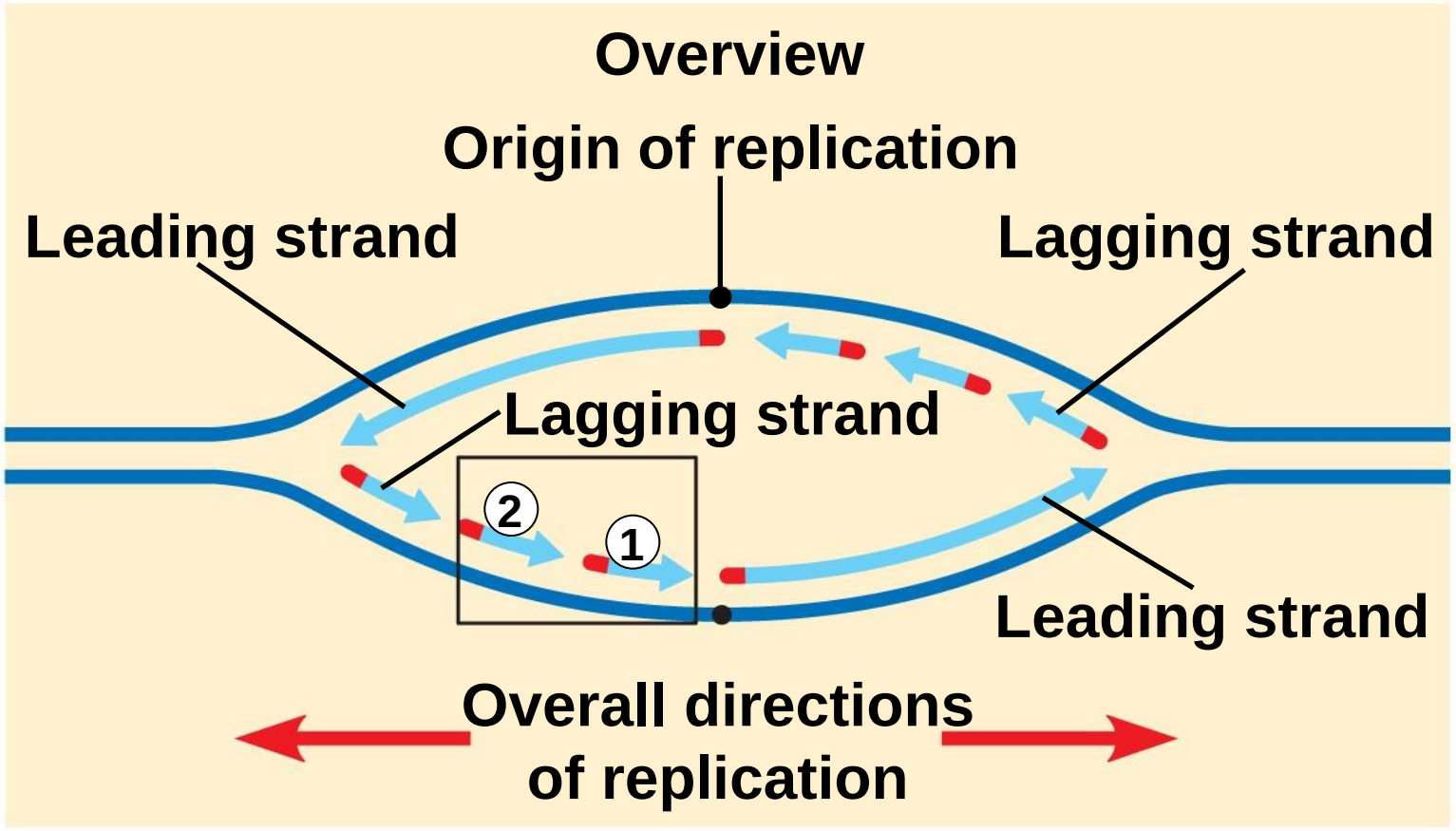
The primer is short (5–10 nucleotides long), and the 3′ end serves as the starting point for the new DNA strand.

Each primer is eventually replaced by DNA.

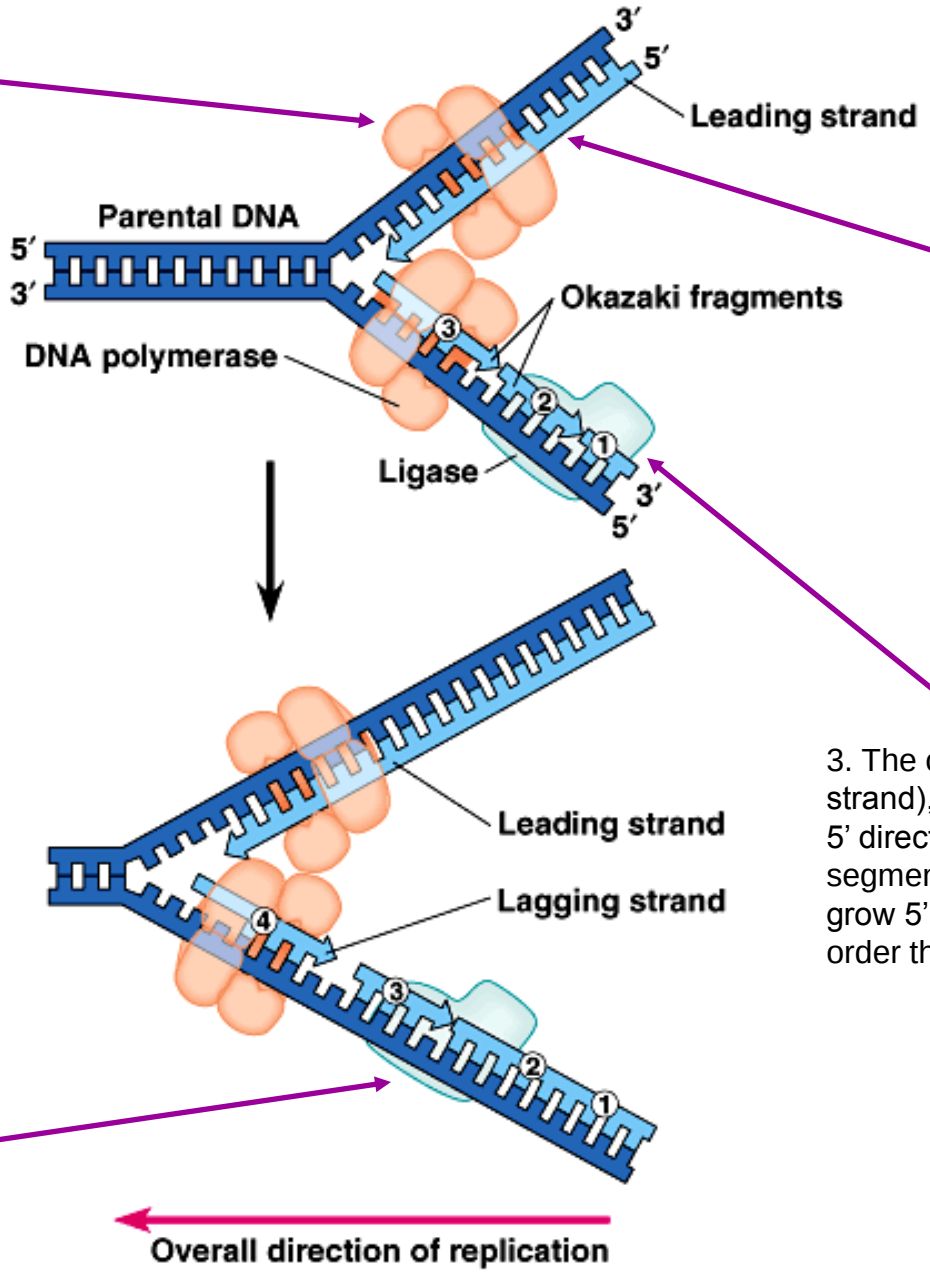








1. DNA polymerase elongates DNA strands only in the 5’ → 3’ direction.

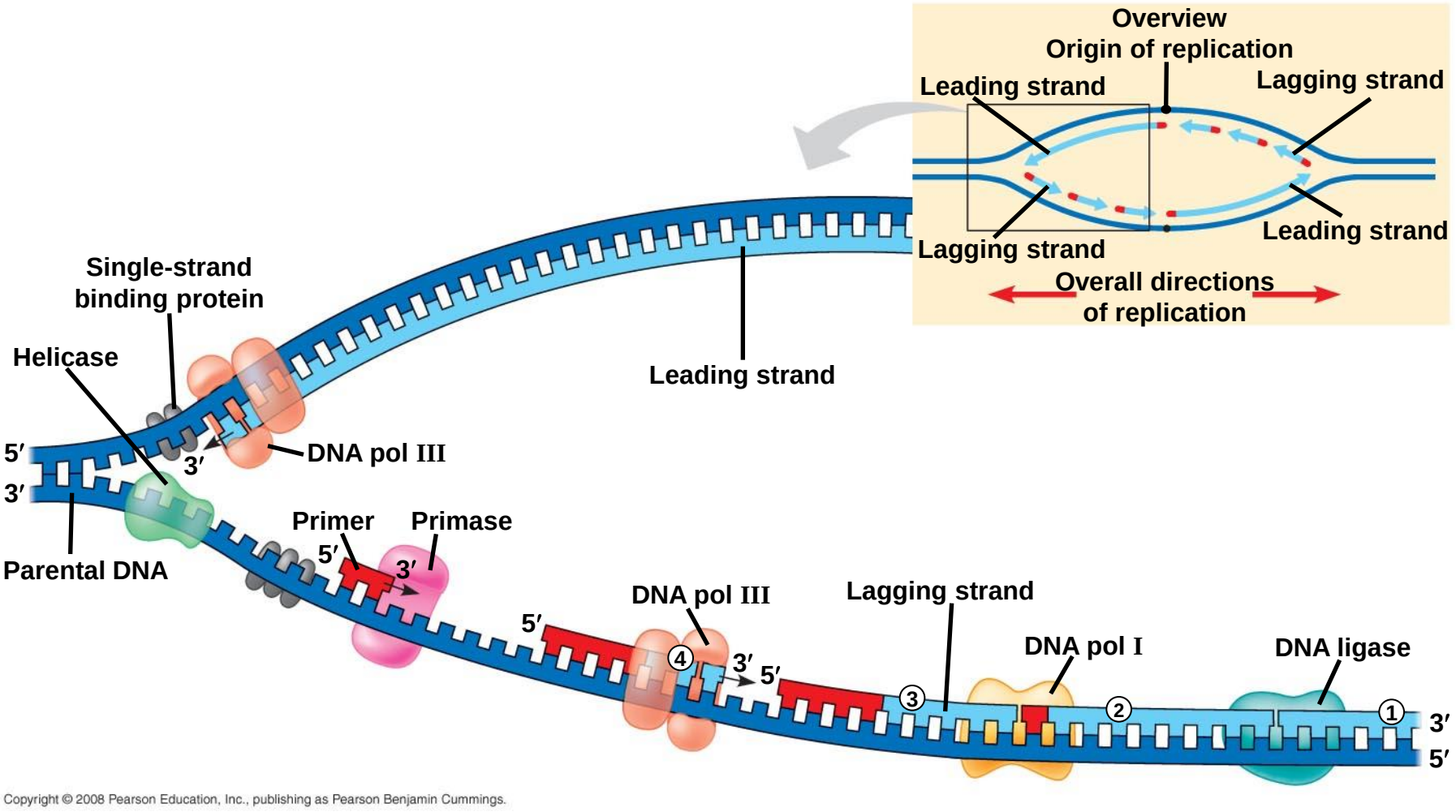


2. One new strand (leading strand) can therefore elongate continuously 5’ → 3’ as the replication for progresses.

3. The other new strand (lagging strand), must grow in an overall 3’ → 5’ direction by addition of short segments (Okazaki fragments) that grow 5’ → 3’ (numbered here in the order they were made).

4. Ligase connects the Okazaki fragments.

http://highered.mcgraw-hill.com/olc/dl/120076/bio23.swf



Proofreading and Repairing DNA

http://nortonbooks.com/college/biology/animations/ch12a05.htm

- DURING the replication process, DNA polymerases proofread newly made DNA, replacing any incorrect nucleotides.
- BUT - some incorrect pairs can evade the proofreading mechanism, or arise after replication and these must be correct AFTER replication is complete.
  - In *mismatch repair* of DNA, repair enzymes correct errors in base pairing
  - In *nucleotide excision repair*, a *nuclease* cuts out and replaces damaged stretches of DNA

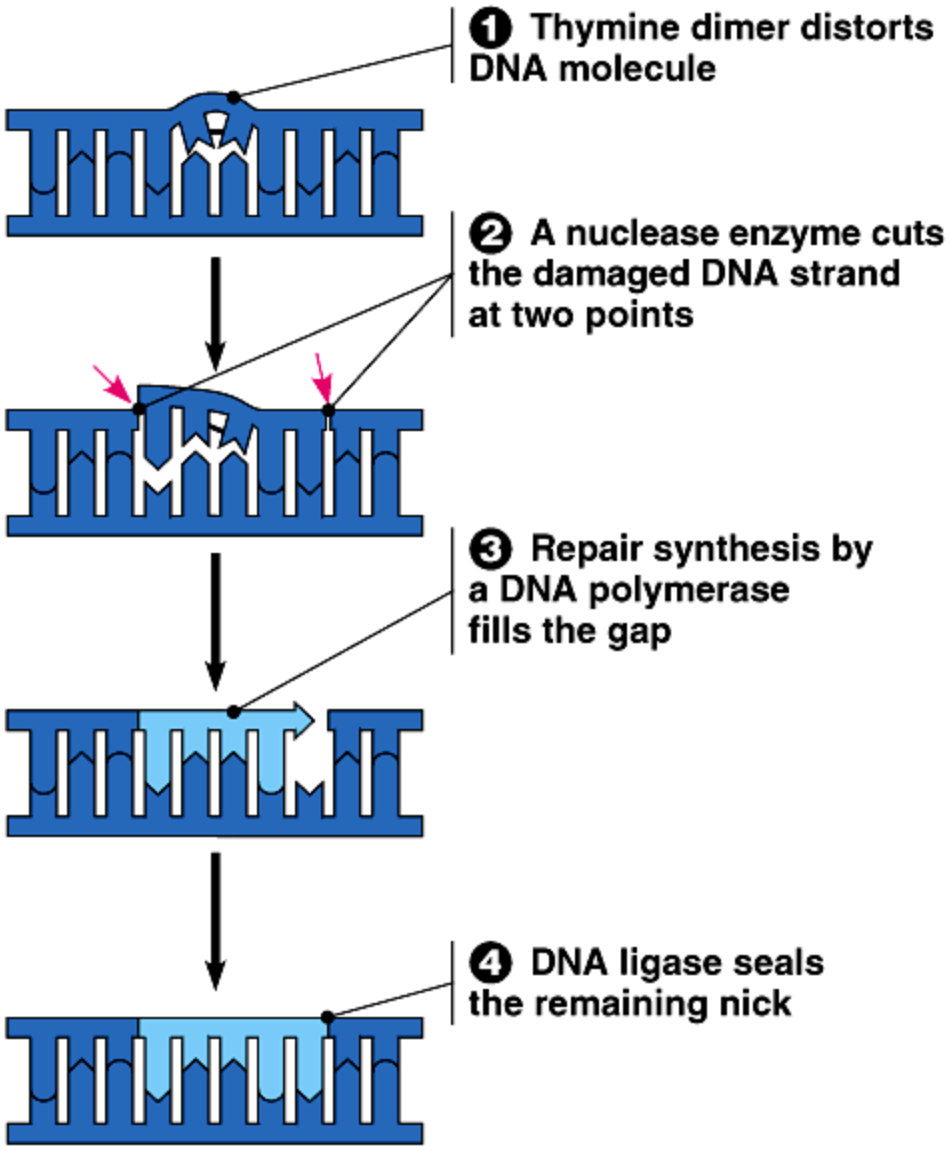
Figure 16.17 Nucleotide Excision Repair of DNA Damage

A team of enzymes detects and repairs damaged DNA in *nucleotide excision repair.*

Repair enzymes (*nucelases*) can excise damaged DNA regions from the DNA and replace them with a normal segment.

Good Animation:

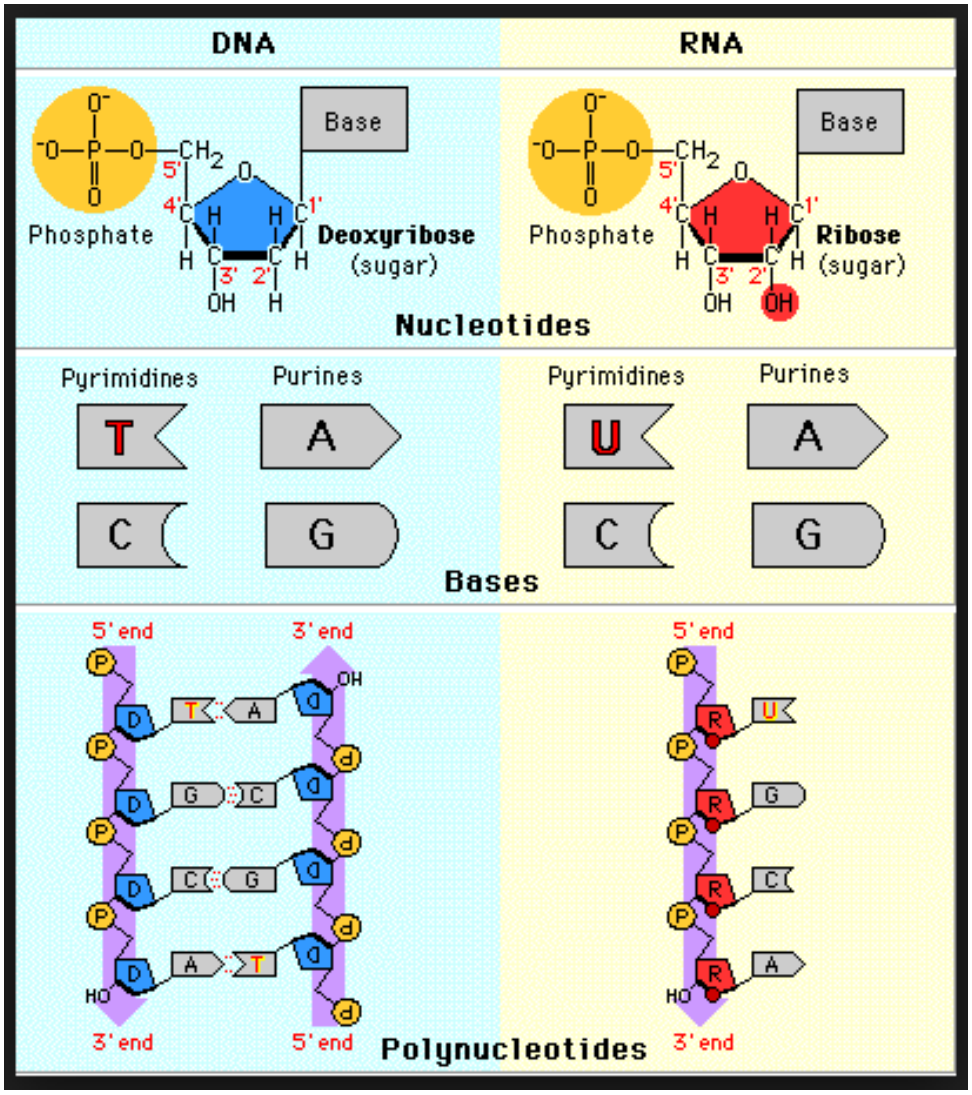
http://nortonbooks.com/college/biology/animations/ch12a05.htm



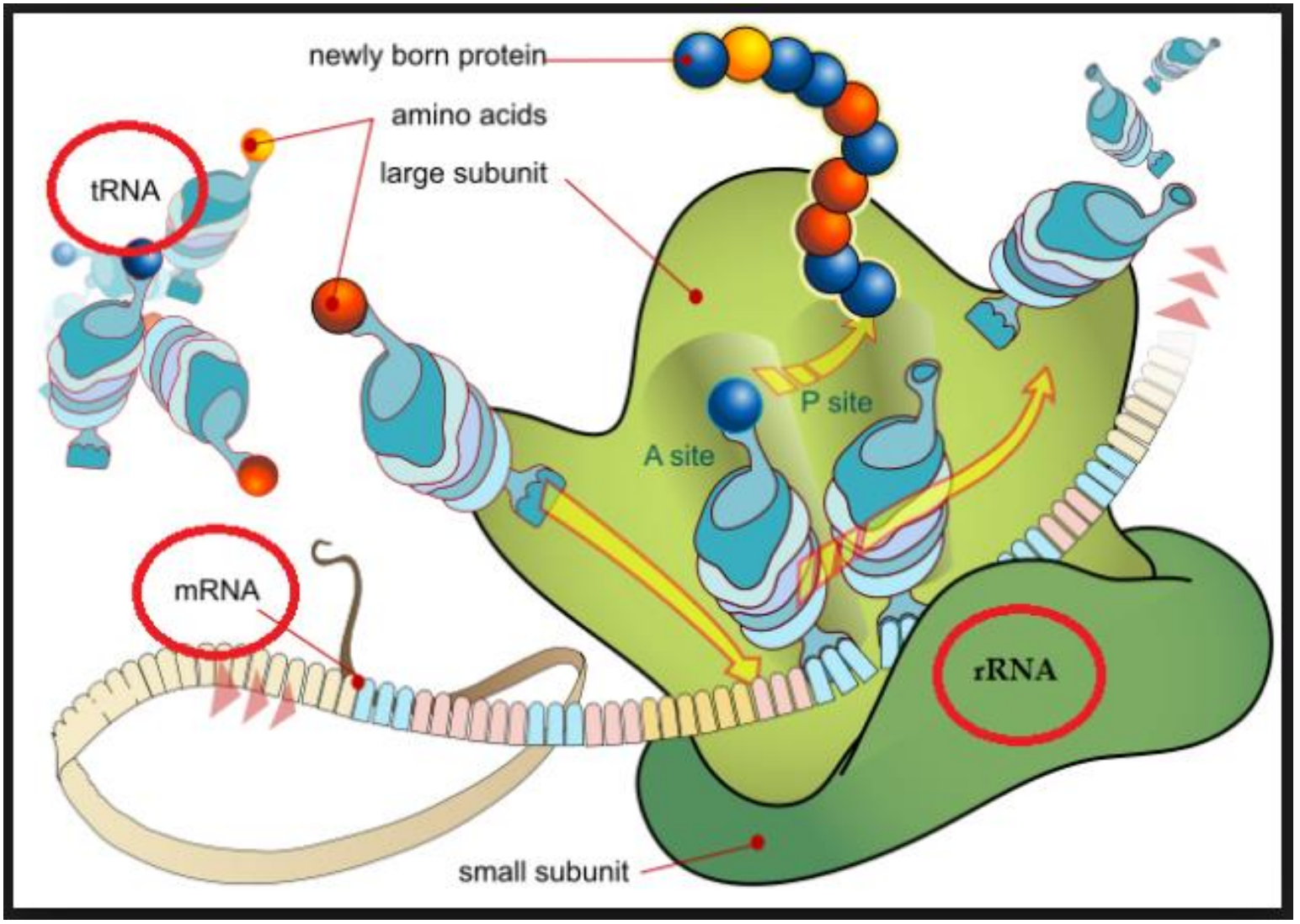
Viral replication differs from other reproductive strategies.

- In living organisms, the flow of genetic information is from DNA to RNA, and translated into proteins. Replication involves copying a template strand of DNA into a complementary new strand of DNA.
- Genetic information in retroviruses is a special case and has an alternate flow of information: from RNA to DNA, made possible by reverse transcriptase, and enzyme that copies the viral RNA genome into DNA.
- This DNA is then integrated into the host genome and becomes transcribed and translated for the assembly of new viral progeny.
- We will return to this concept while discussing 3.C.3.

DNA and RNA molecules have structural similarities and differences that define function.



The sequence of the RNA bases, together with the structure of the RNA molecule, determines RNA function.



Overview: The Flow of Genetic Information

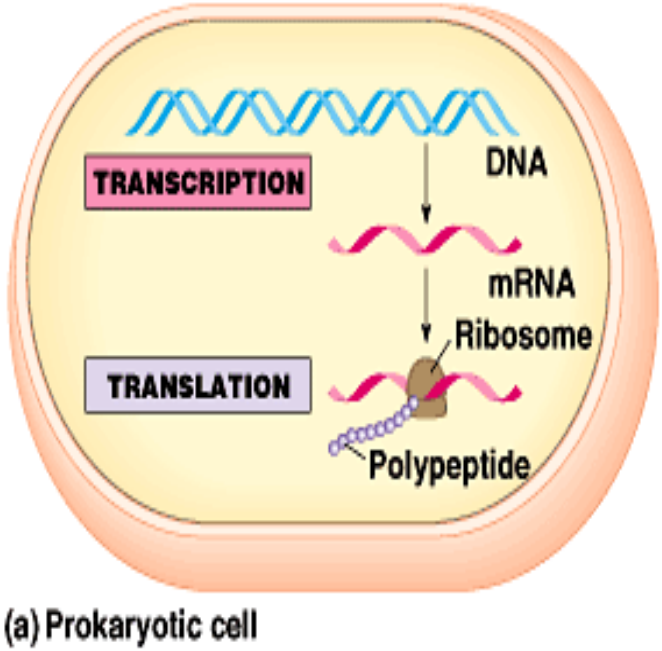
- The information content of DNA is in the form of specific sequences of nucleotides
  - The DNA inherited by an organism leads to specific traits by dictating the synthesis of proteins
  - Genetic information flows from a sequence of nucleotides in a gene (DNA) to a sequence of amino acids for a trait (protein)
- Proteins are the links between genotype and phenotype
- **Gene expression**, the process by which DNA directs protein synthesis, includes two stages: *transcription* and *translation*

Genetic information flows from a sequence of nucleotides in a gene to a sequence of amino acids in a protein.

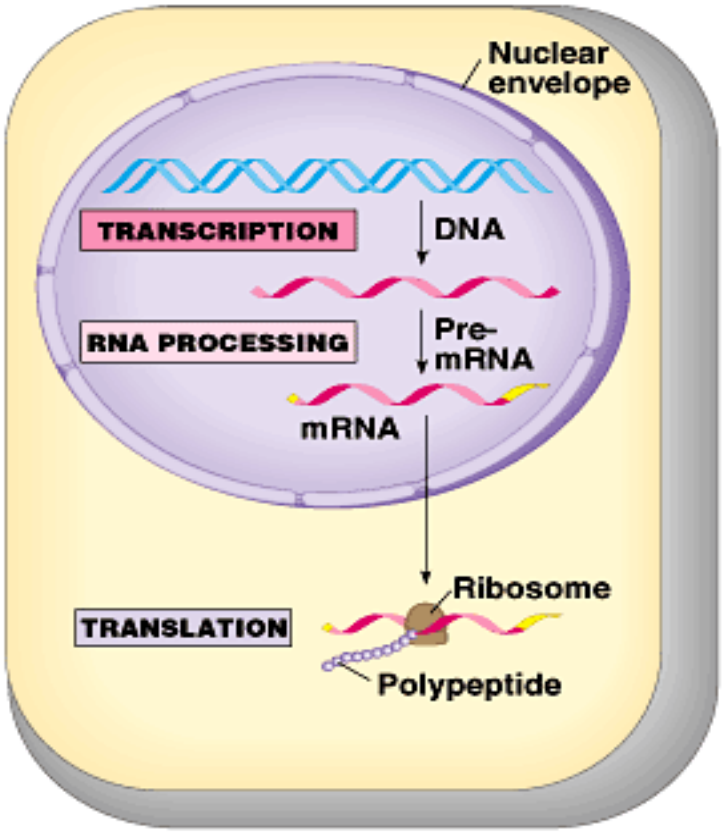
- The process begins when the enzyme RNA-polymerase reads the DNA molecule in the 3’ to 5’ direction and synthesizes complementary mRNA molecules that determine the order of amino acids in the polypeptide.
- In eukaryotic cells the mRNA transcript undergoes a series of enzyme-regulated modifications before being sent to the ribosome to build the polypeptide:
  - Addition of a poly-A tail;
  - Addition of a GTP cap;
  - Excision of introns
- In prokaryotic organisms, transcription is coupled to translation of the message. Translation involves energy and many steps, including initiation, elongation and termination.

Prokaryotic v. Eukaryotic Gene Expression

http://highered.mcgraw-hill.com/olc/dl/120077/bio25.swf

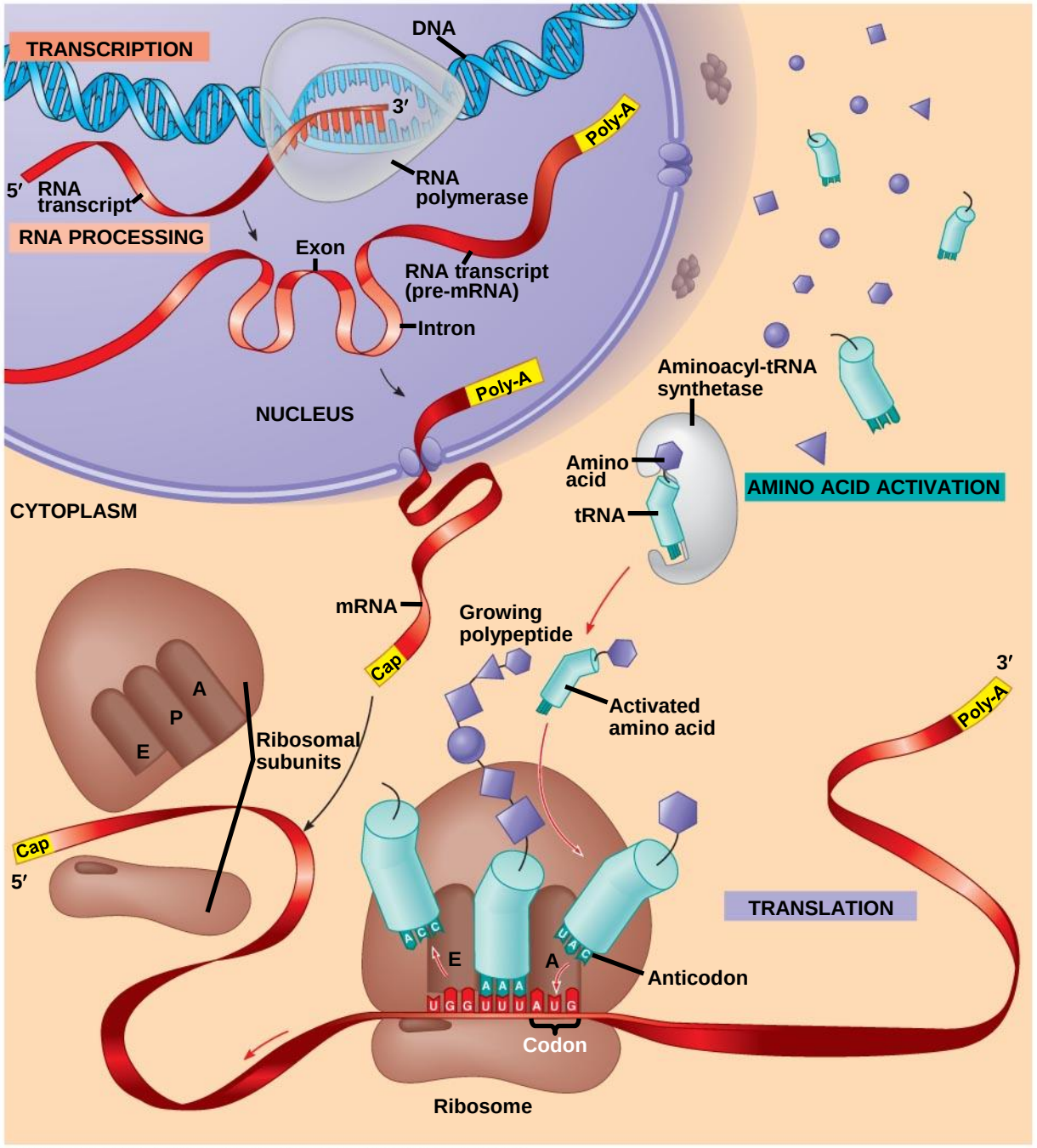


(a) Prokaryotic cell

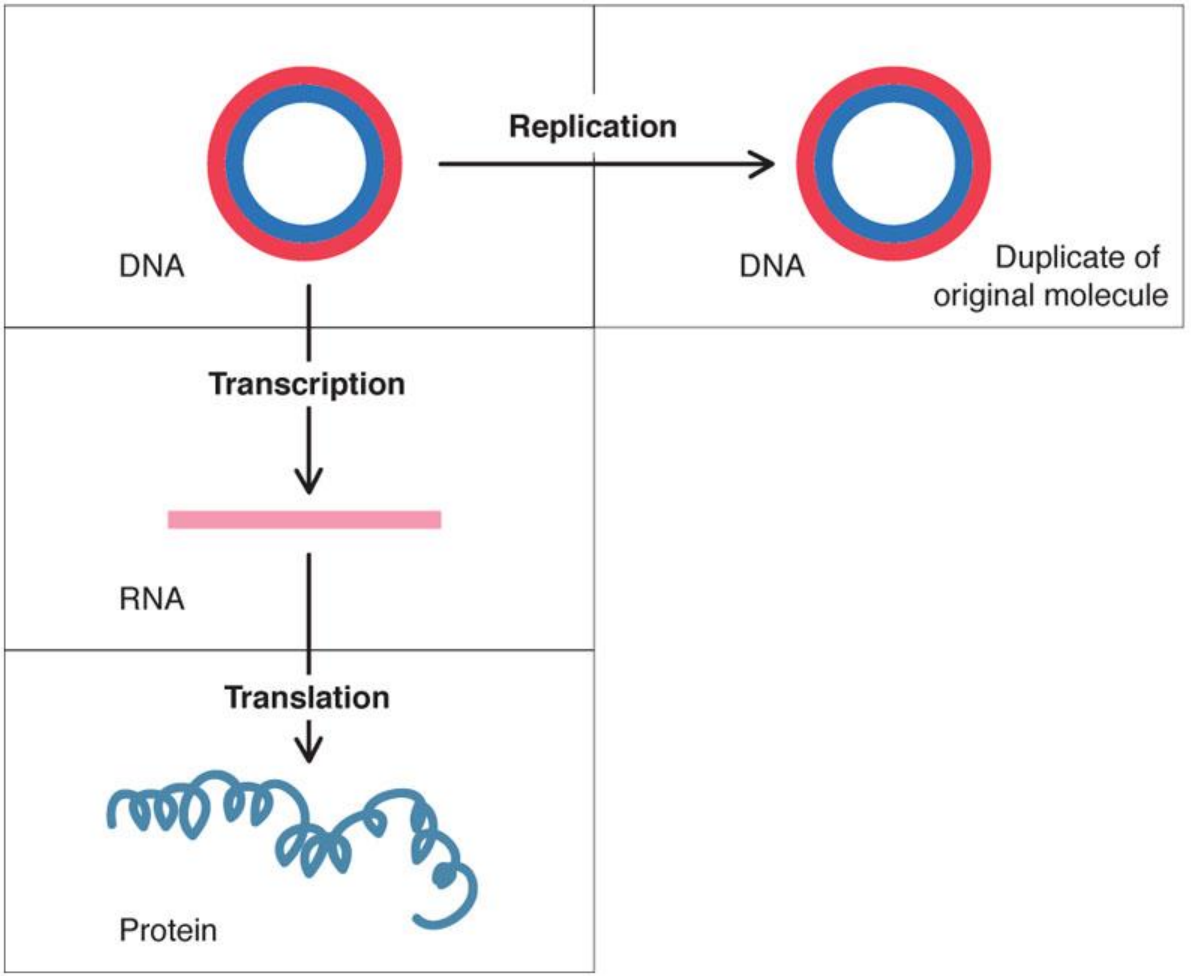


(b) Eukaryotic cell

Fig. 17-25



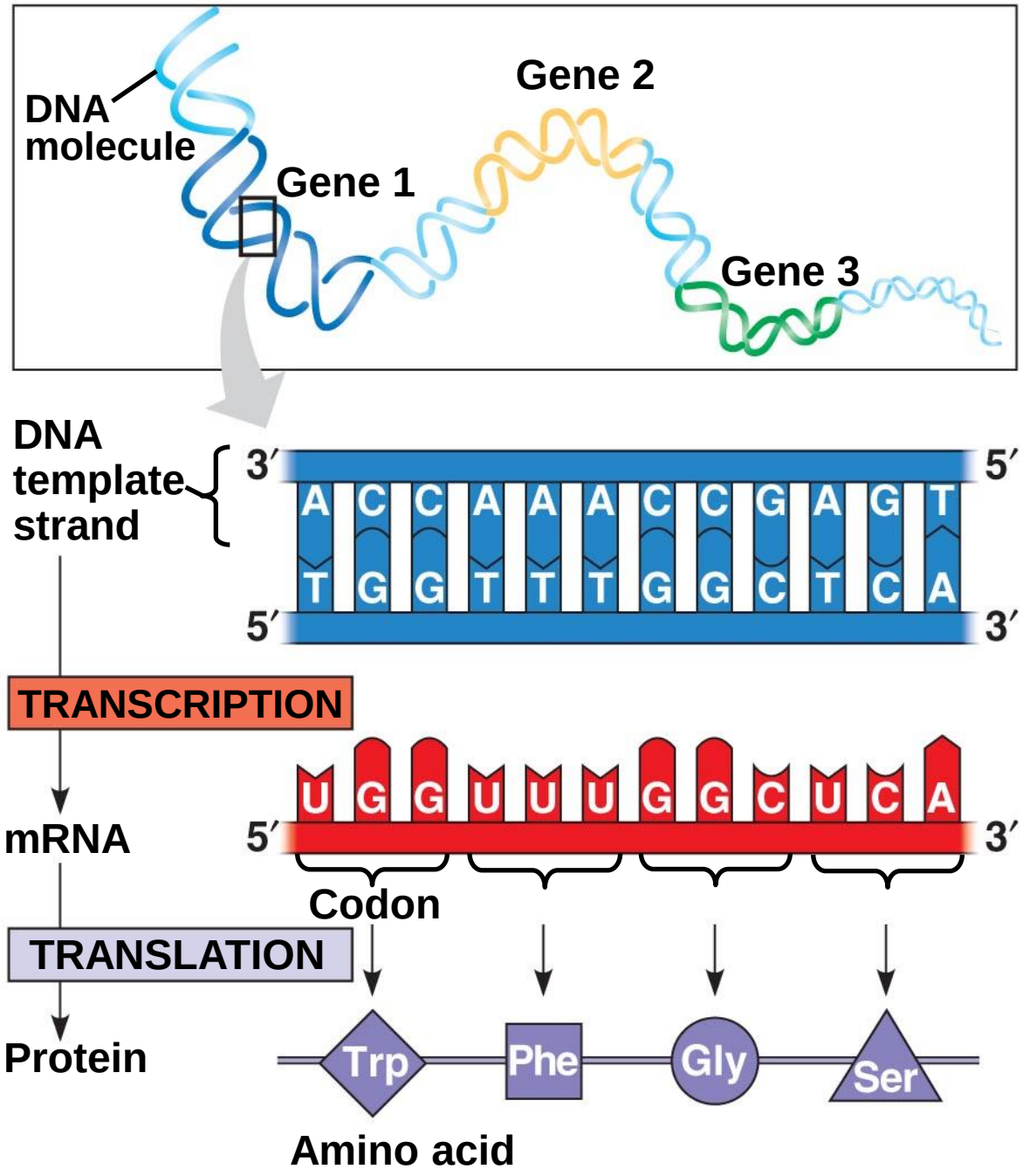
Basic Principles of Transcription and Translation



Codons: Triplets of Bases

- The flow of information from gene to protein is based on a **triplet code**: a series of nonoverlapping, three-nucleotide words.
- These triplets are the smallest units of uniform length that can code for all the amino acids.
- Example: *AGT* at a particular position on a DNA strand results in the placement of the amino acid *serine* at the corresponding position of the polypeptide to be produced.

Fig. 17-4



Cracking the Code

- All 64 codons were deciphered by the mid-1960s.
- Of the 64 triplets, 61 code for amino acids; 3 triplets are "stop" signals to end translation.
- The genetic code is *redundant* but *not ambiguous*; **no codon specifies more than one amino acid**…but a particular amino acid can be specified by more than one codon.
- Codons must be read in the correct **reading frame** (correct groupings) in order for the specified polypeptide to be produced.

Fig. 17-5

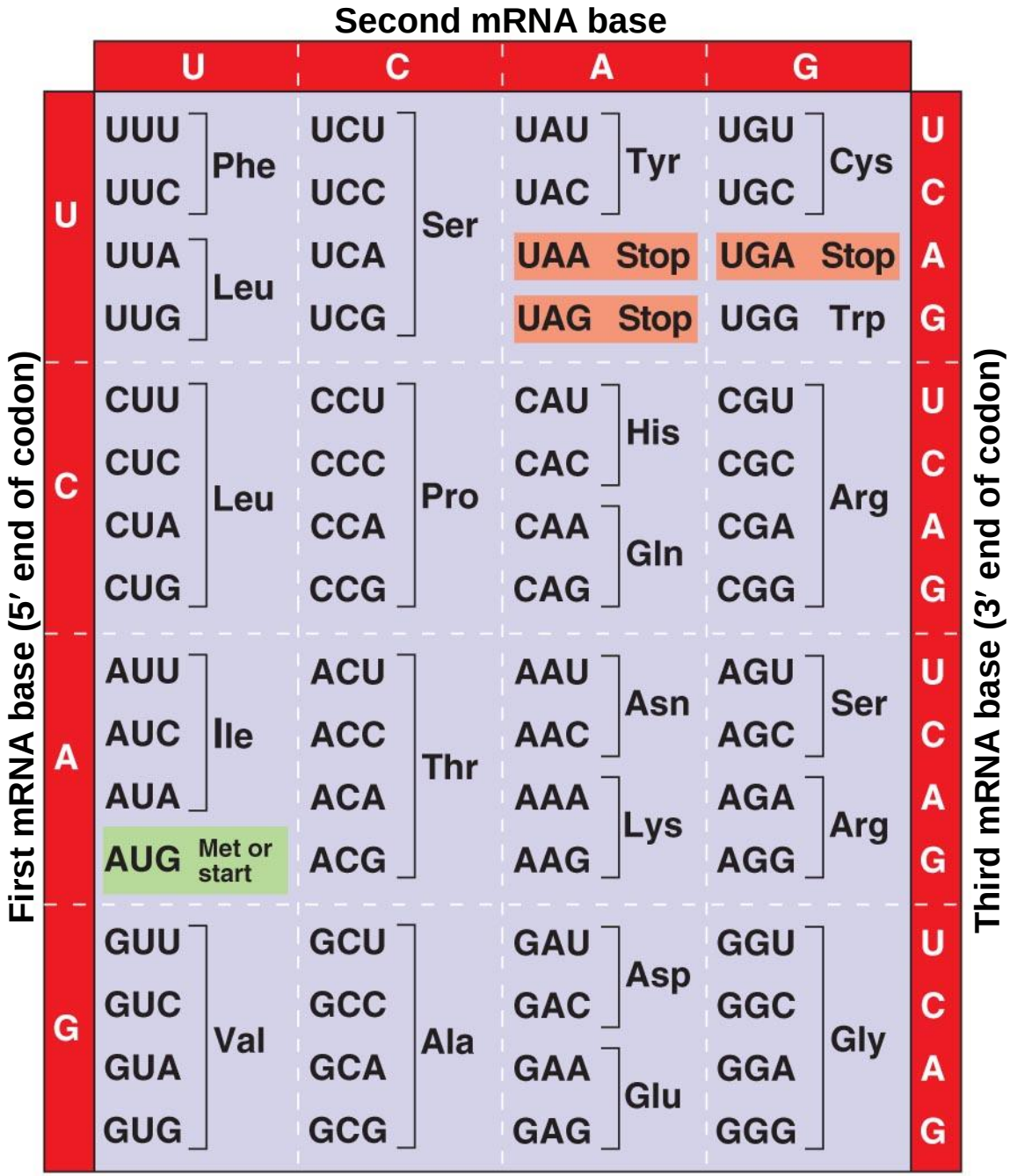
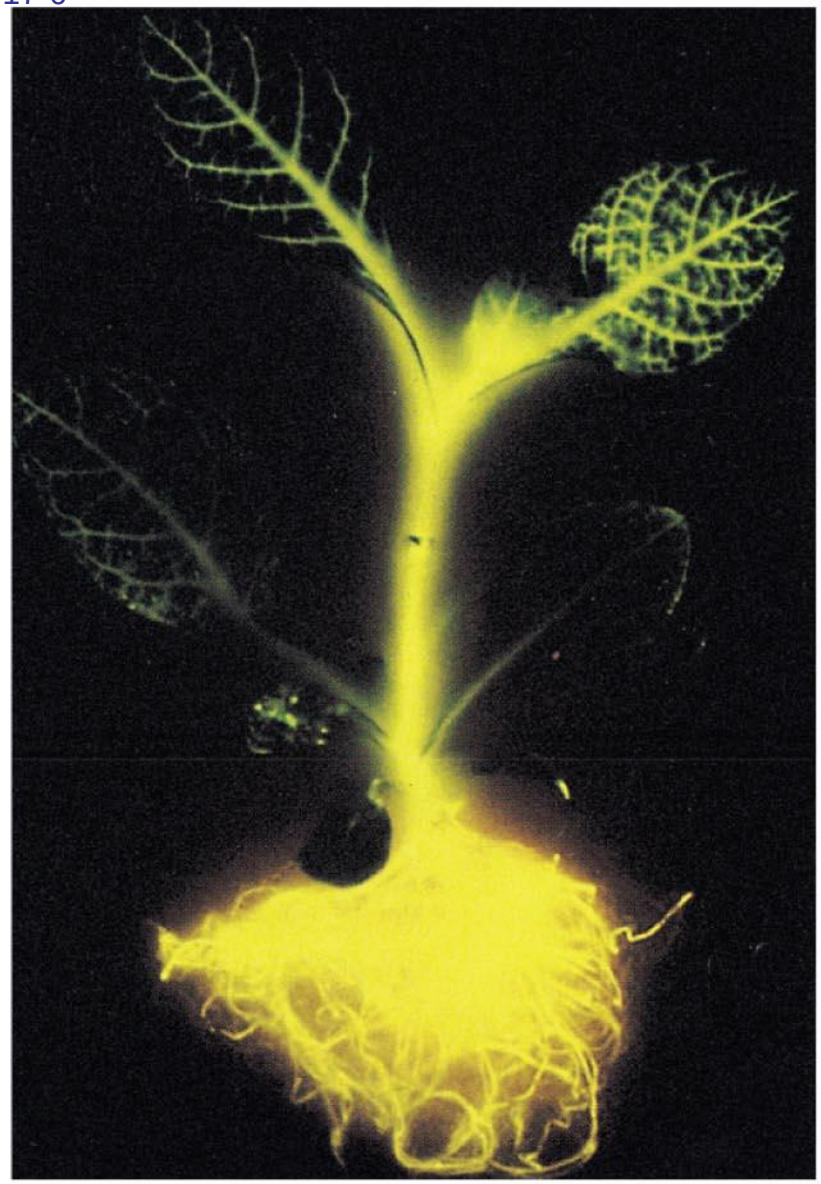


Fig. 17-6



(a) Tobacco plant expressing a firefly gene

(b) Pig expressing a jellyfish gene

Transcription

http://bcs.whfreeman.com/thelifewire/content/chp12/1202001.html

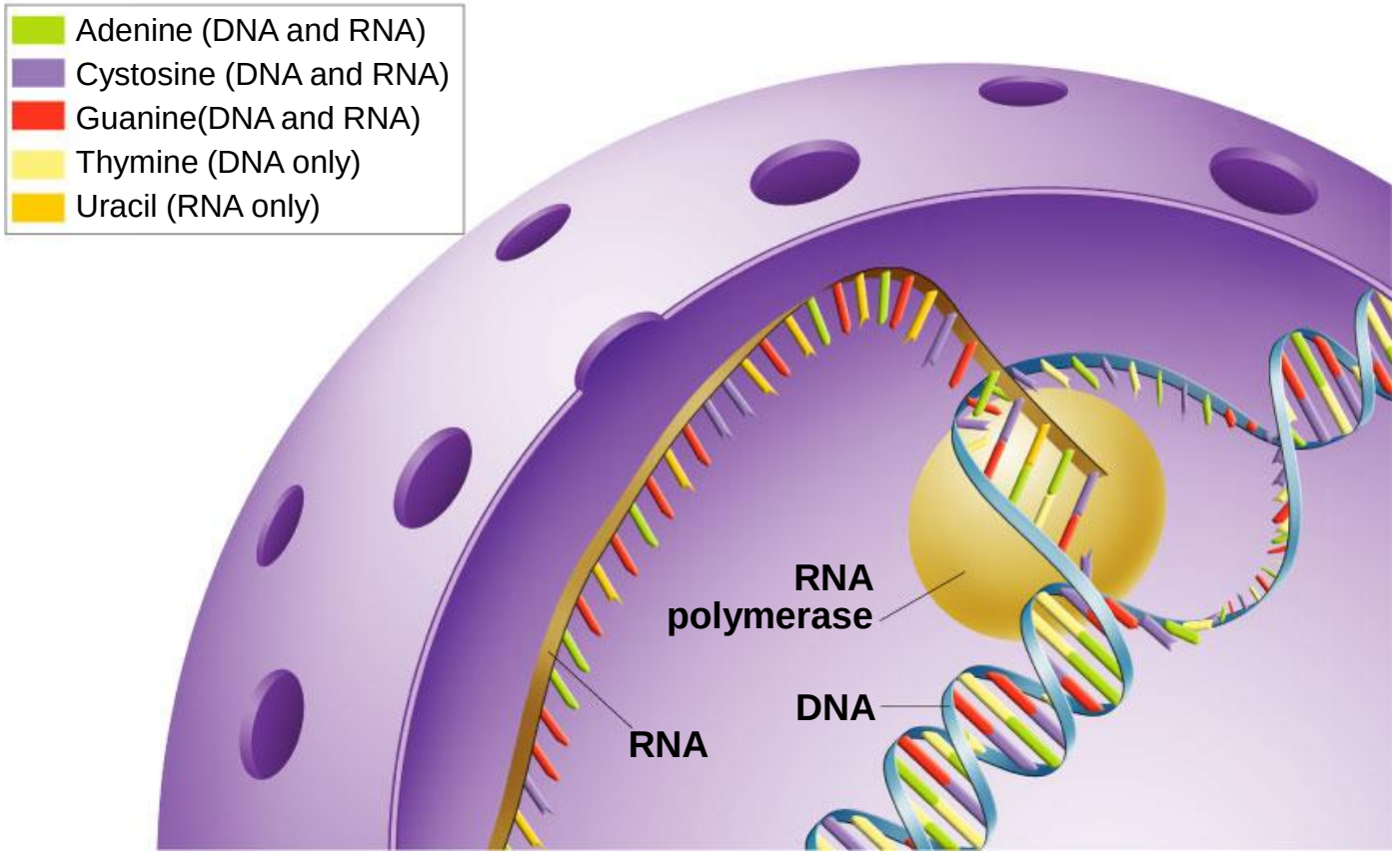
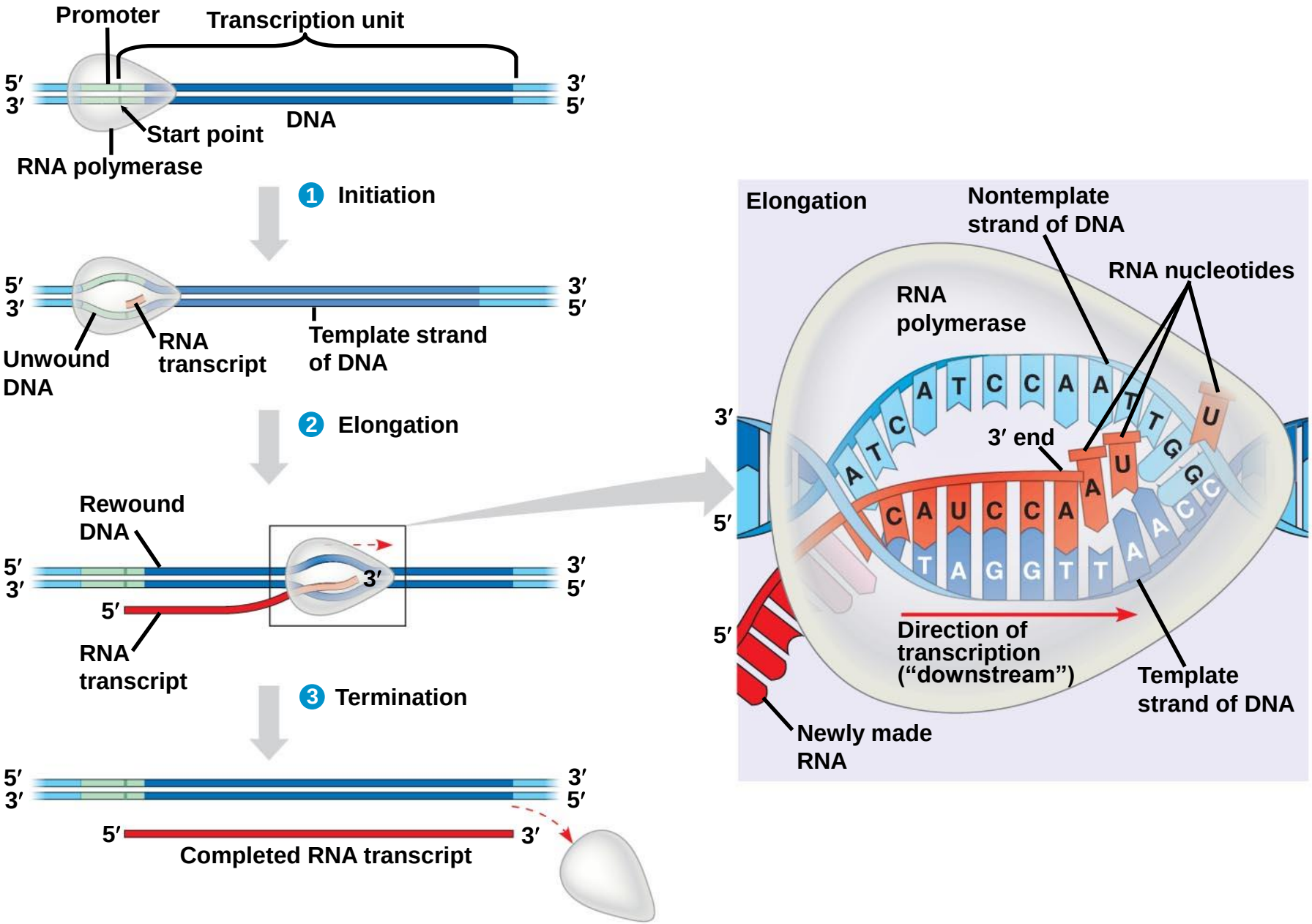


Fig. 17-7 – The Stages of Transcription



RNA Modification

- Enzymes in the eukaryotic nucleus modify pre-mRNA before the genetic messages are dispatched to the cytoplasm.
- During RNA processing, both ends of the primary transcript are usually altered.
- Also, usually some interior parts of the molecule are cut out, and the other parts spliced together.

Fig. 17-9

A modified guanosine triphosphate added to the 5’ end.

50 to 250 adenine nucleotides added to the 3’ end created by cleavage downstream of the termination signal.

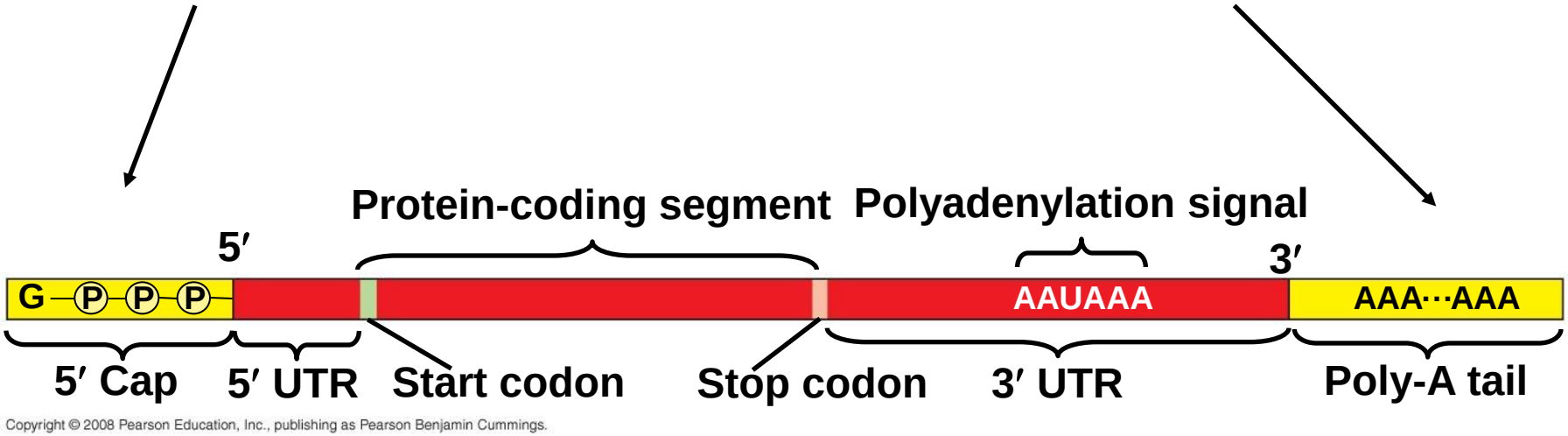
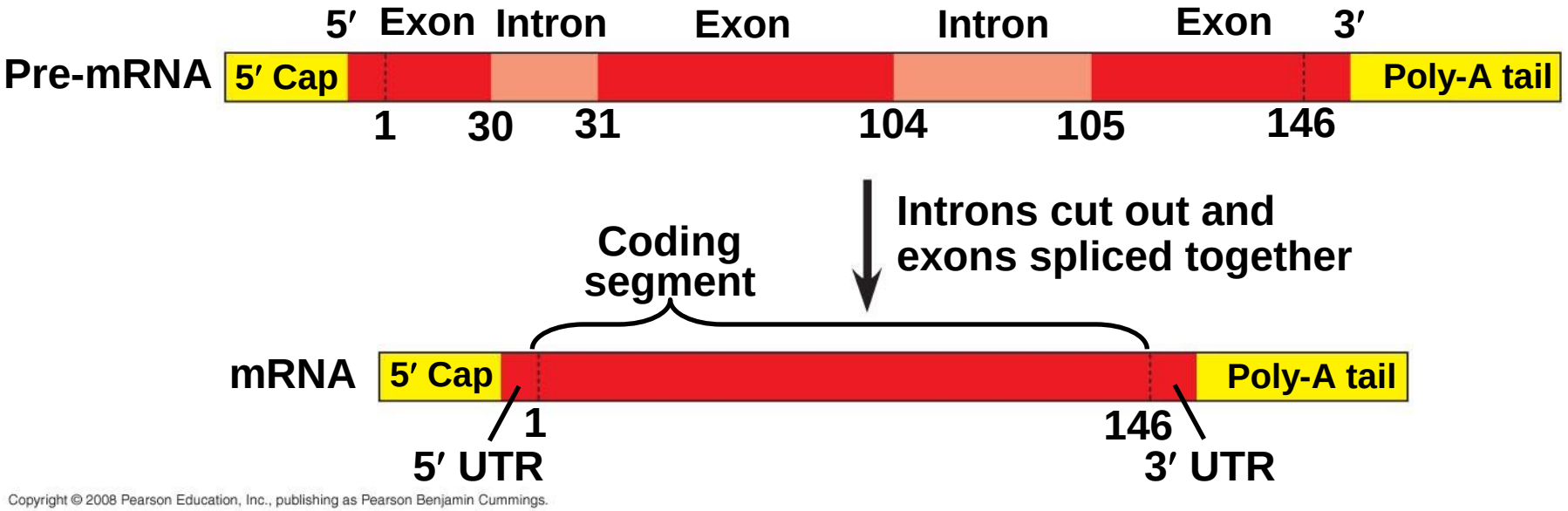


Fig. 17-10



The Functional and Evolutionary Importance of Introns

- Some genes can encode more than one kind of polypeptide, depending on which segments are treated as exons during RNA splicing.
- Such variations are called ***alternative RNA splicing***.
- Because of alternative splicing, the number of different proteins an organism can produce is much greater than its number of genes.

Translation

- Translation is the RNA-directed synthesis of a polypeptide.
- Generally: is the reading of the codons on the mRNA strand and the sequencing of them into an amino acid sequence – polypeptide.
- The Players:
  - **mRNA**: already processed within the nucleus via transcription, will be the template for the sequence of amino acids.
  - **tRNA**: transfers amino acids from the cytoplasm to the ribosome.
  - **rRNA (ribosome)**: adds amino acids together from the tRNA and in the sequence of the mRNA.

Fig. 17-13

http://highered.mcgraw-hill.com/olcweb/cgi/pluginpop.cgi?it=swf::535::535::/sites/dl/free/0072437316/120077/micro06.swf::Protein%20Synthesis

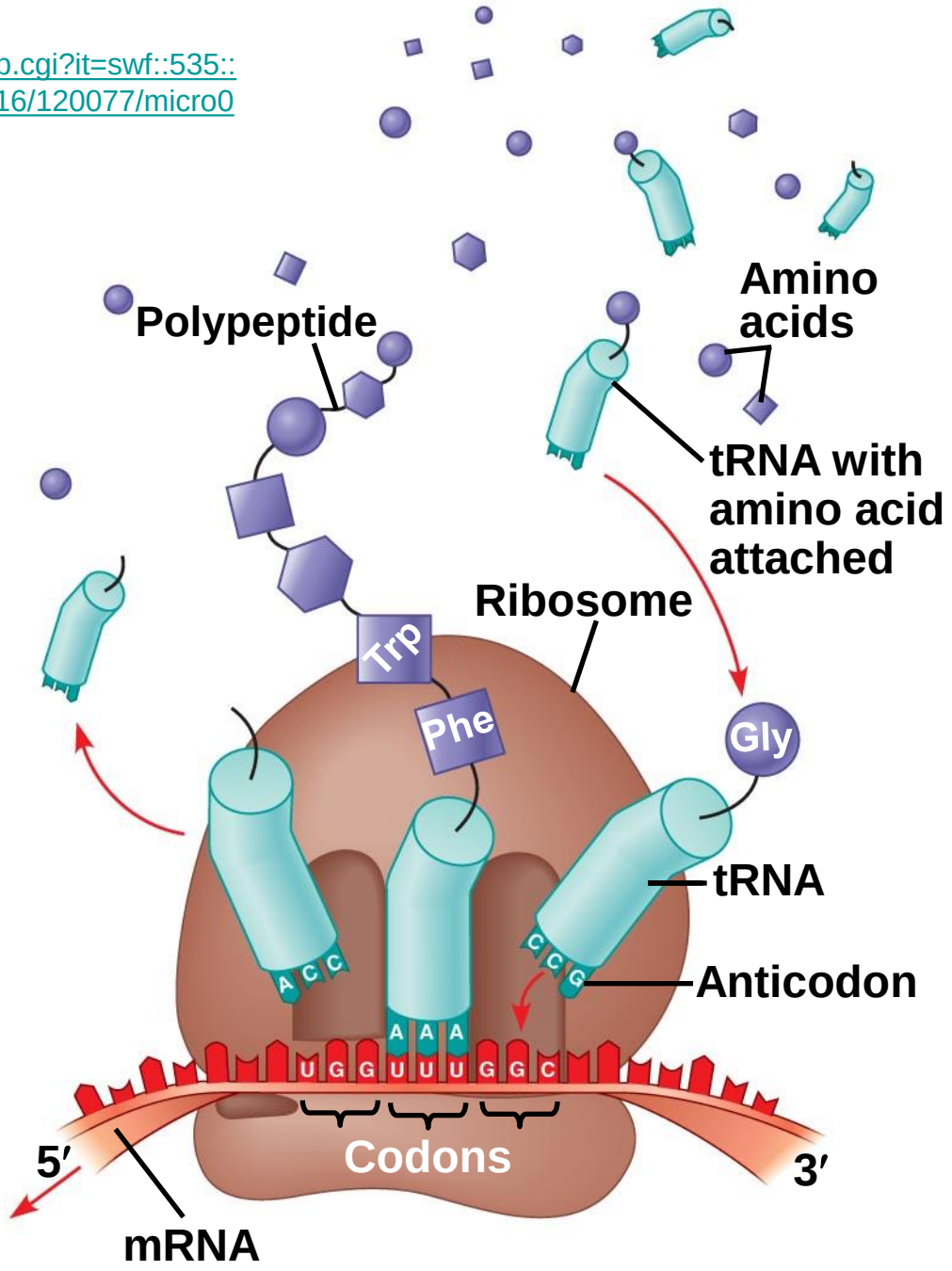
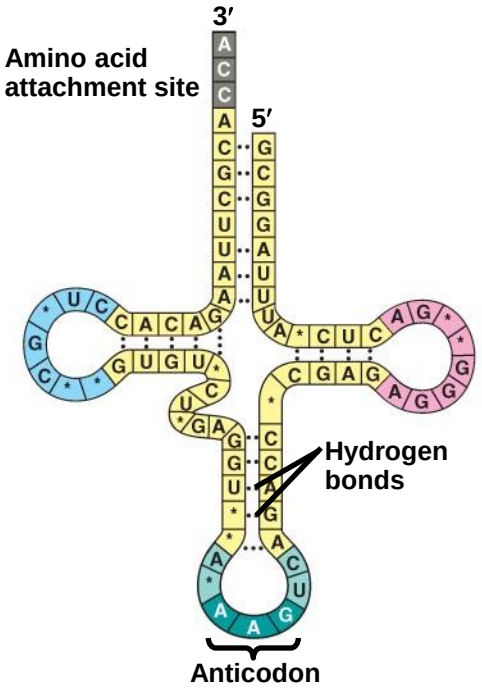
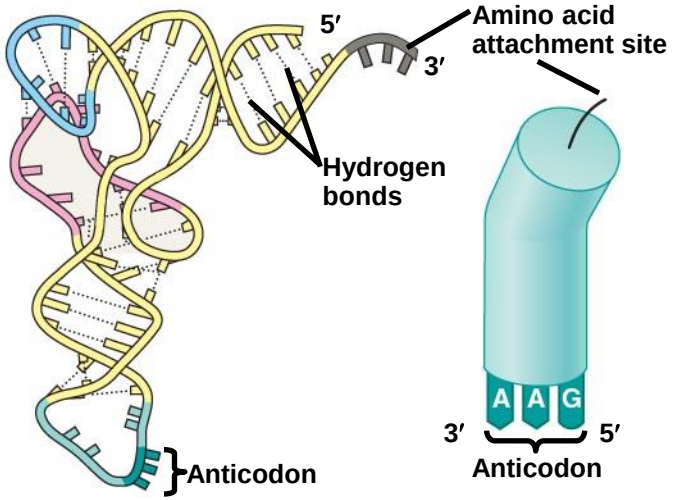


Fig. 17-14



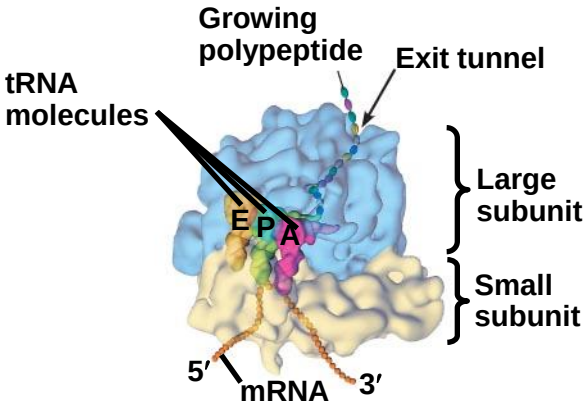
(a) Two-dimensional structure



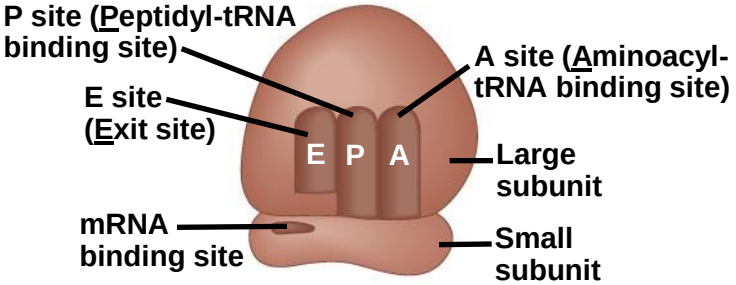
(b) Three-dimensional structure

(c) Symbol used in this book

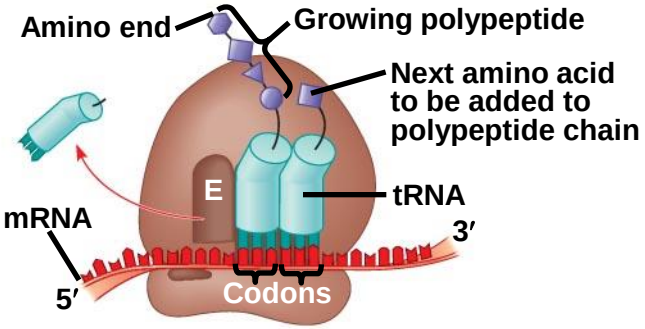
Fig. 17-16



(a) Computer model of functioning ribosome



(b) Schematic model showing binding sites



(c) Schematic model with mRNA and tRNA

http://highered.mcgraw-hill.com/olc/dl/120077/micro06.swf

Building a Polypeptide

- The three stages of translation:
  - Initiation (requires energy “GTP”)
  - Elongation (requires energy “GTP”)
  - Termination
- All three stages require protein “factors” that aid in the translation process

Fig. 17-17

INITIATION OF TRANSLATION

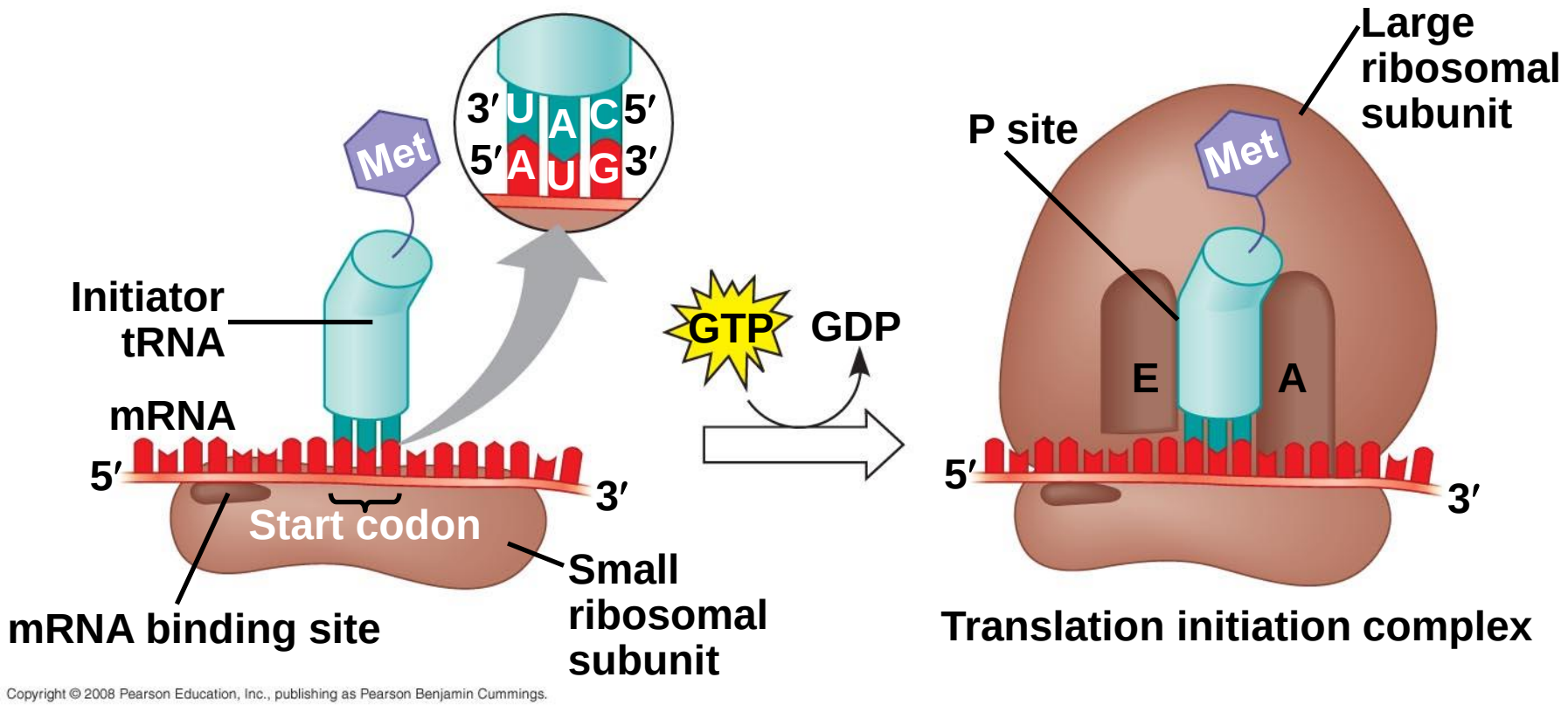


Fig. 17-18-4

ELONGATION OF TRANSLATION

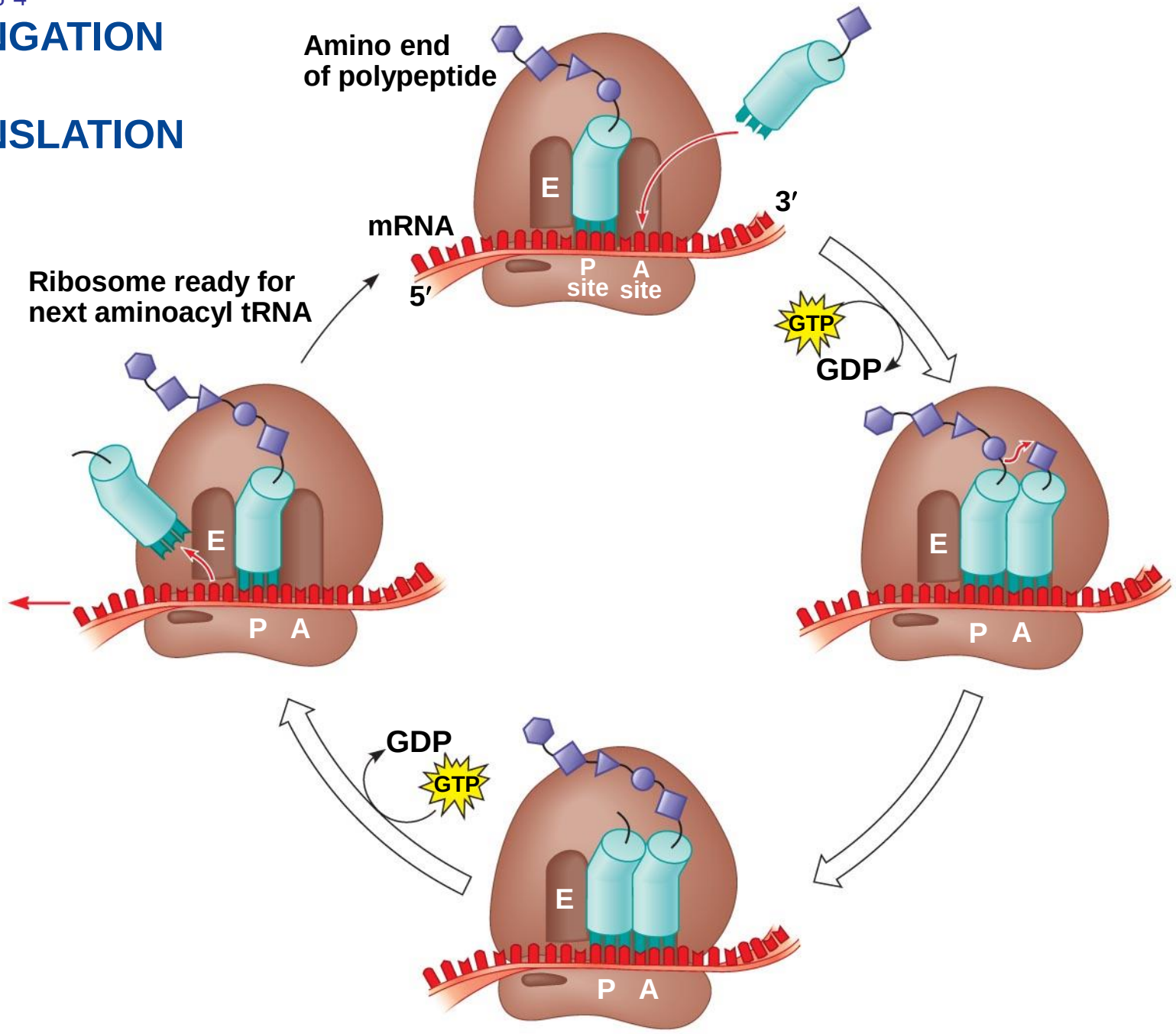


Fig. 17-19-3

TERMINATION OF TRANSLATION

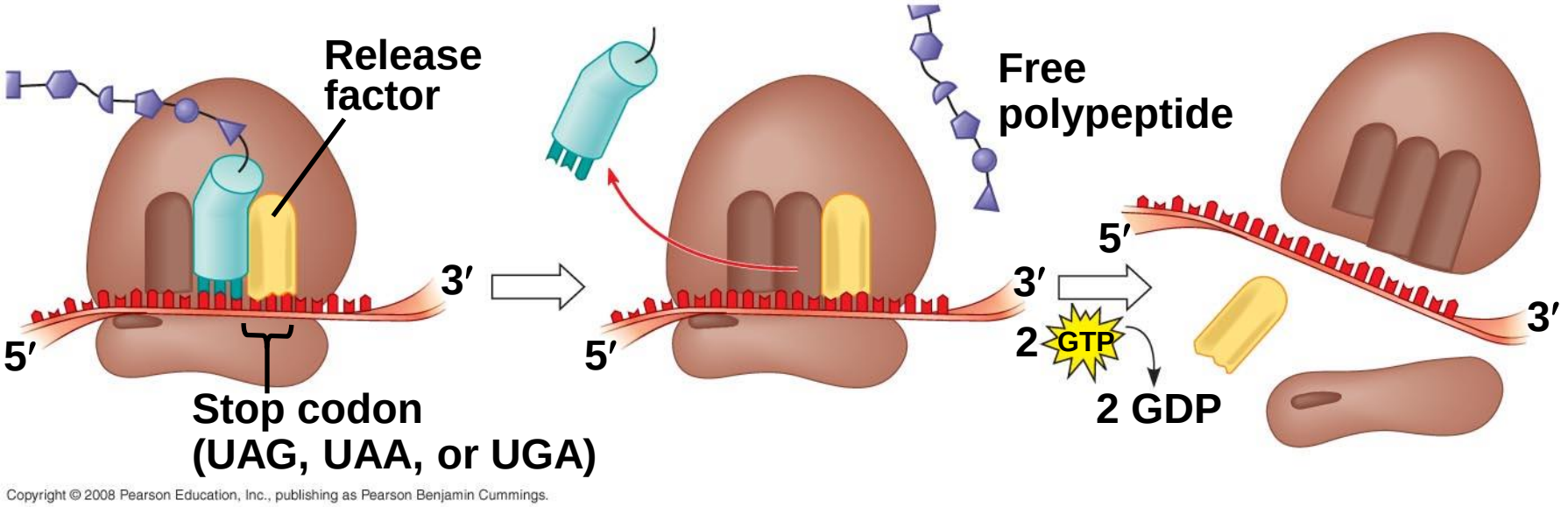
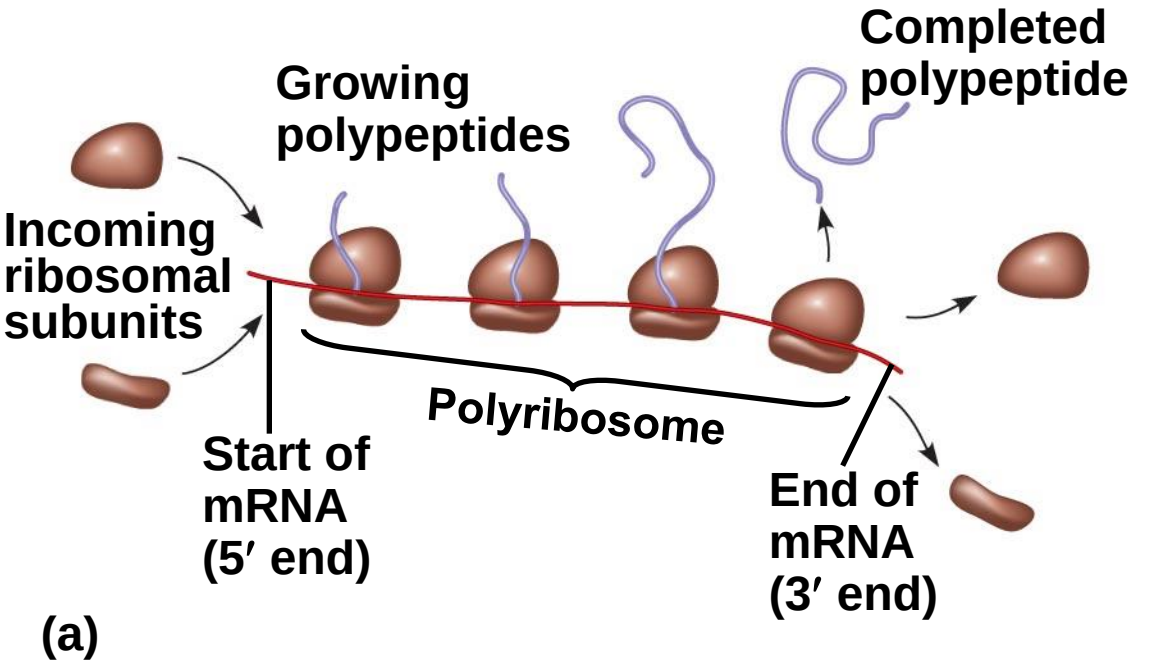
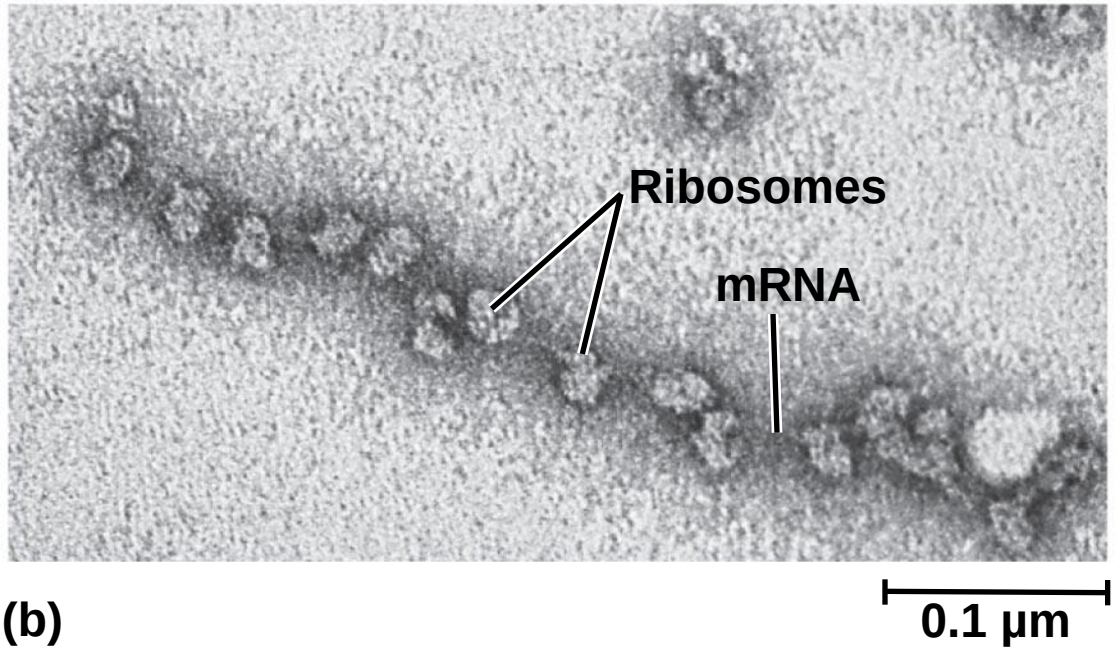


Fig. 17-20





Completing and Targeting the Functional Protein

- Often translation is not sufficient to make a functional protein…THEREFORE…polypeptide chains are modified after translation.
- Completed proteins are targeted to specific sites in the cell:
  - During and after synthesis, a polypeptide chain spontaneously coils and folds into its three-dimensional shape.
  - Proteins may also require post-translational modifications before doing their job.
  - Some polypeptides are activated by enzymes that cleave them.
  - Other polypeptides come together to form the subunits of a protein.

Targeting Polypeptides to Specific Locations

- Two populations of ribosomes are evident in cells: free ribosomes (in the cytosol) and bound ribosomes (attached to the ER).
- Free ribosomes mostly synthesize proteins that function in the cytosol.
- Bound ribosomes make proteins of the endomembrane system and proteins that are secreted from the cell.
- Ribosomes are identical and can switch from free to bound.

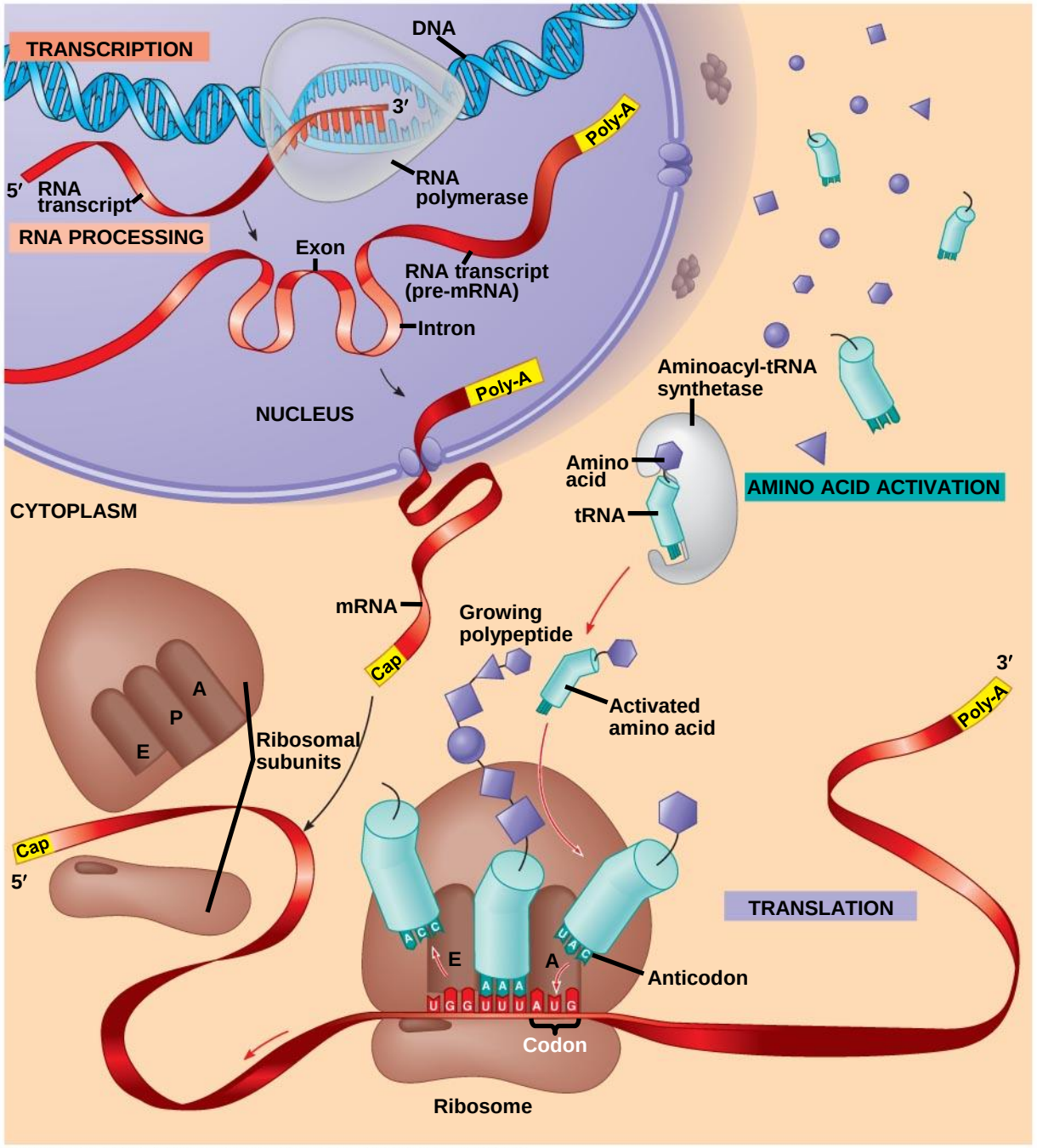
Review – PROTEIN SYNTHESIS

- **Transcription**
  - DNA → mRNA (in nucleus)
  - In eukaryotes will have RNA processing (in nucleus).
- **Translation**
  - mRNA → Polypeptide (at ribosome in cytoplasm)
- **Coiling & Folding**
  - Three dimensional (in cytoplasm as translation is occurring)
  - Chaperone proteins involved

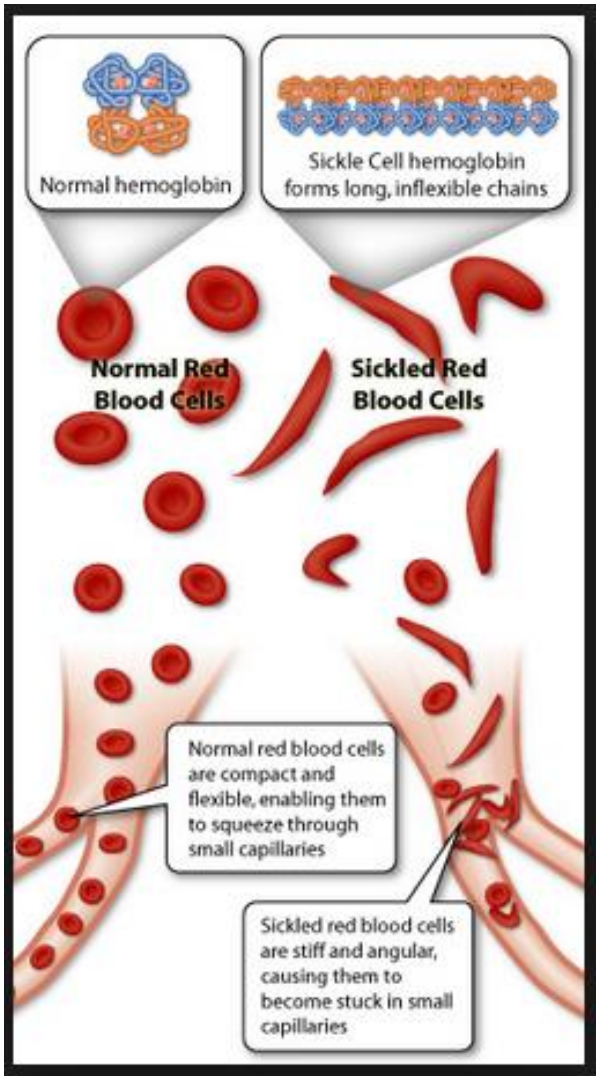
Comparing Gene Expression in Bacteria, Archaea, and Eukarya

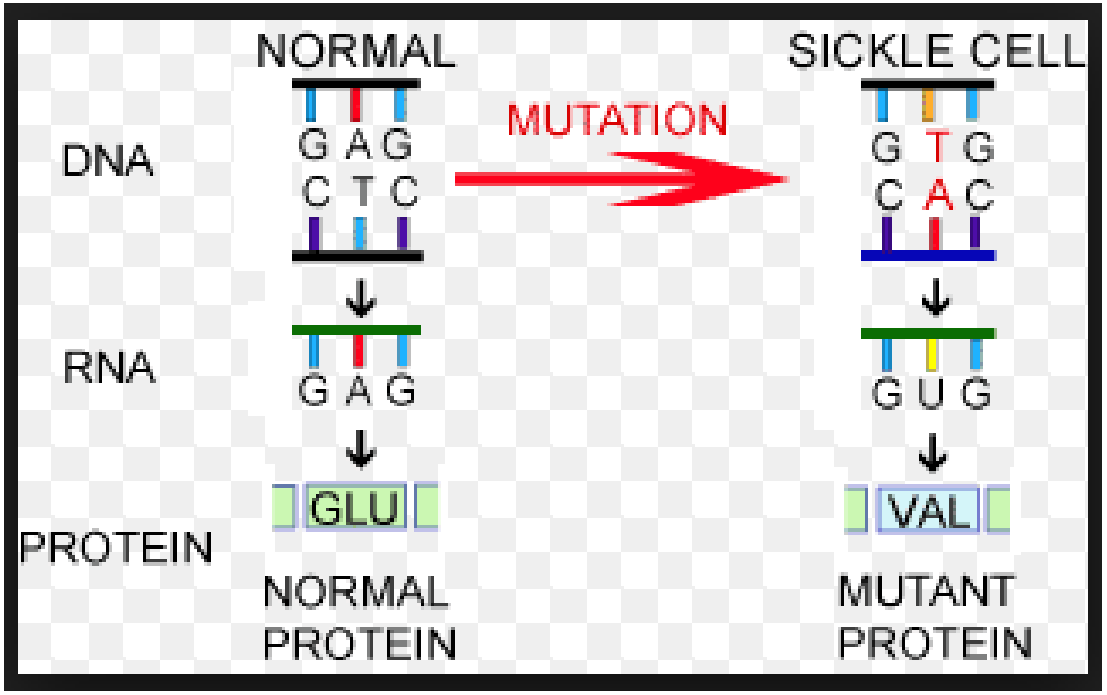
- Archaea are prokaryotes, but share many features of gene expression with eukaryotes.
- Bacteria and Eukarya differ in their RNA polymerases, termination of transcription and ribosomes; Archaea tend to resemble Eukarya in these respects.
- Bacteria can simultaneously transcribe and translate the same gene.
- In Eukarya, transcription and translation are separated by the nuclear envelope.
- In Archaea, transcription and translation are likely coupled.

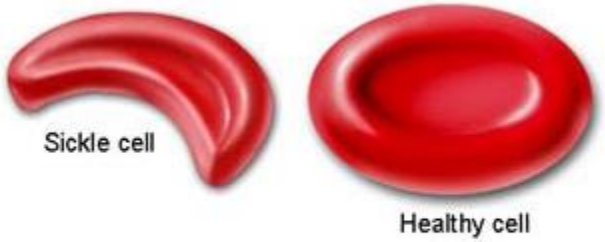
Fig. 17-25



Phenotypes are determined through protein activities.







Phenotype:	Affected, has sickle cell anemia	Normal
Genotype:	SS	AA

Genetic engineering techniques can manipulate the heritable information of DNA and, in special cases, RNA.

- ***Genetic engineering*** is the process of manipulating genes and genomes.
  - ***Recombinant DNA*** is DNA that has been artificially made, using DNA from different sources – and often different species.
  - ***Gene cloning*** is the process of producing multiple copies of specific segments of DNA.
  - ***Restriction enzymes*** are used to cut strands of DNA at specific locations (called restriction sites).

Using Restriction Enzymes to Cut DNA

http://highered.mcgraw-hill.com/olc/dl/120078/bio37.swf

- ***Restriction enzymes*** are used to cut strands of DNA at specific locations (called ***restriction sites***). They are derived from bacteria.
- When a DNA molecule is cut by restriction enzymes, the result will always be a set of ***restriction fragments***, which will have at least one single-stranded end, called a ***sticky end***.
- Sticky ends can form hydrogen bonds with complementary single-stranded pieces of DNA. These unions can be sealed with the enzyme ***DNA ligase***.

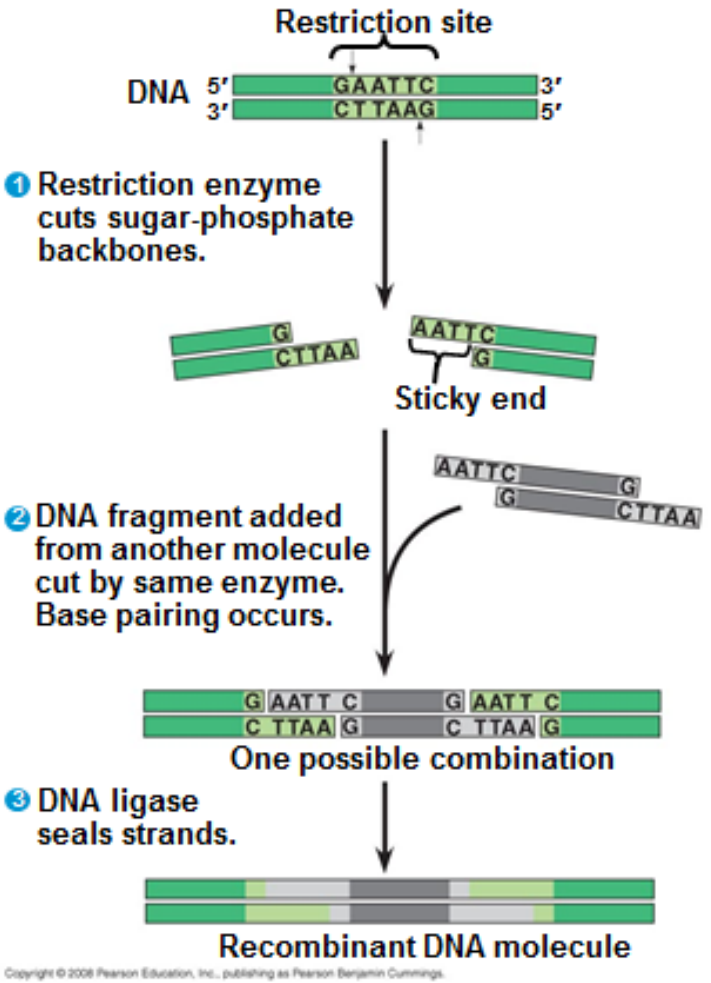
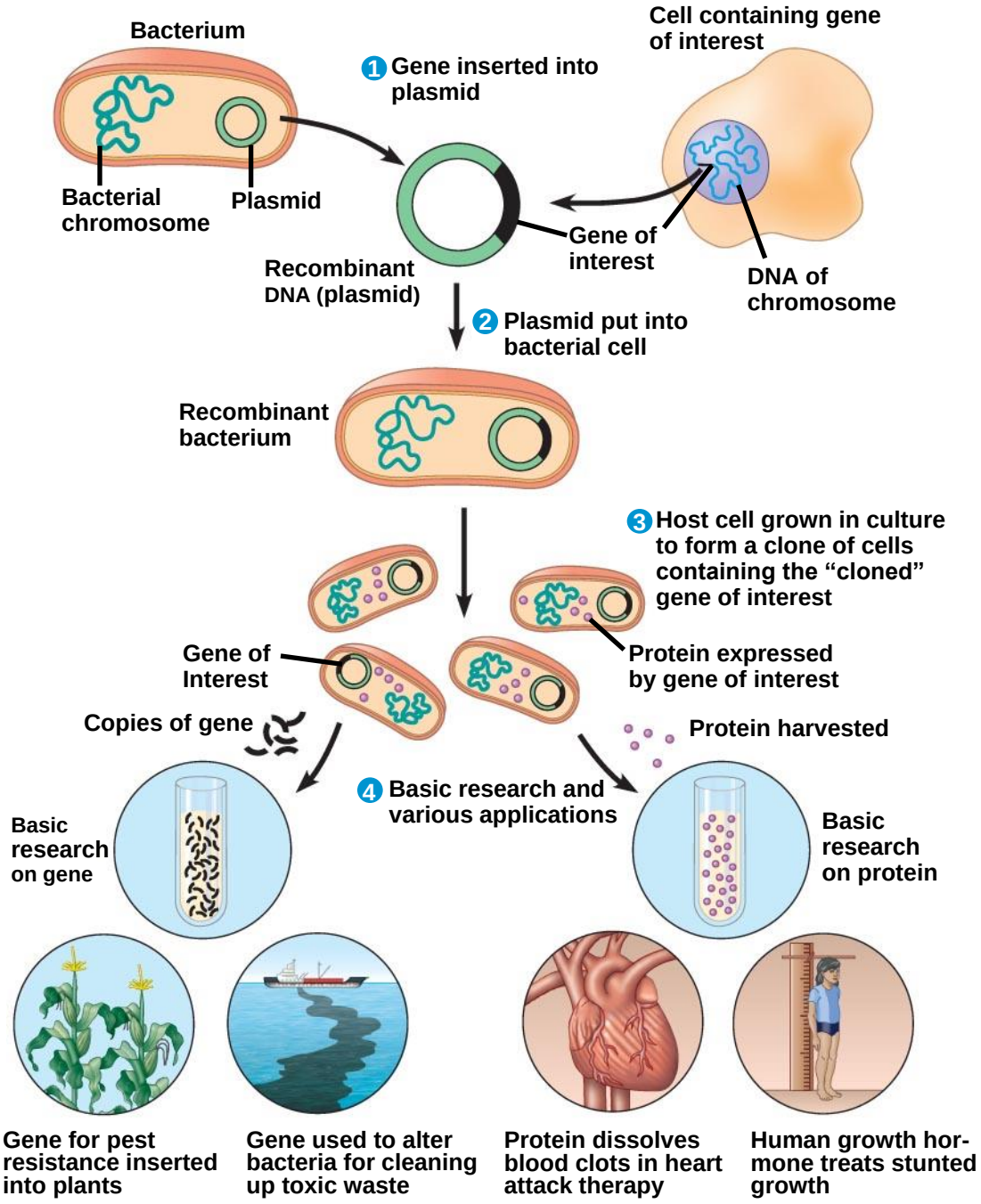


Fig. 20-2



Using Gel Electrophoresis to Separate DNA

http://www.sumanasinc.com/webcontent/animations/content/gelelectrophoresis.html

- One indirect method of rapidly analyzing and comparing genomes is **gel electrophoresis**.
- This technique uses a gel as a molecular sieve to separate nucleic acids or proteins by size.
- A current is applied that causes charged molecules to move through the gel.
- Molecules are sorted into “bands” by their size.

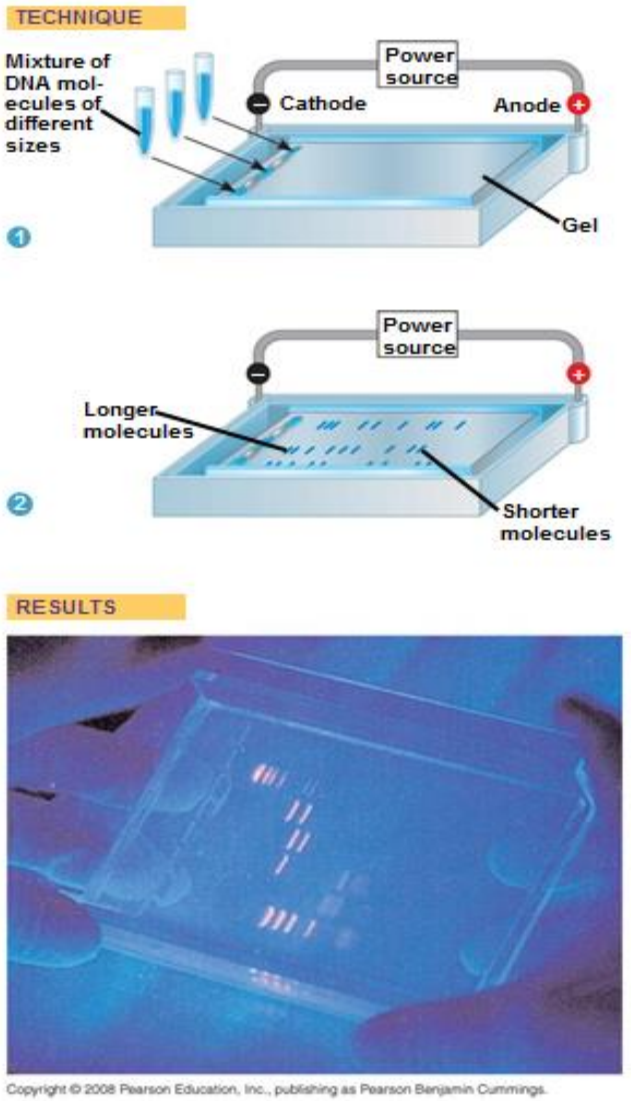
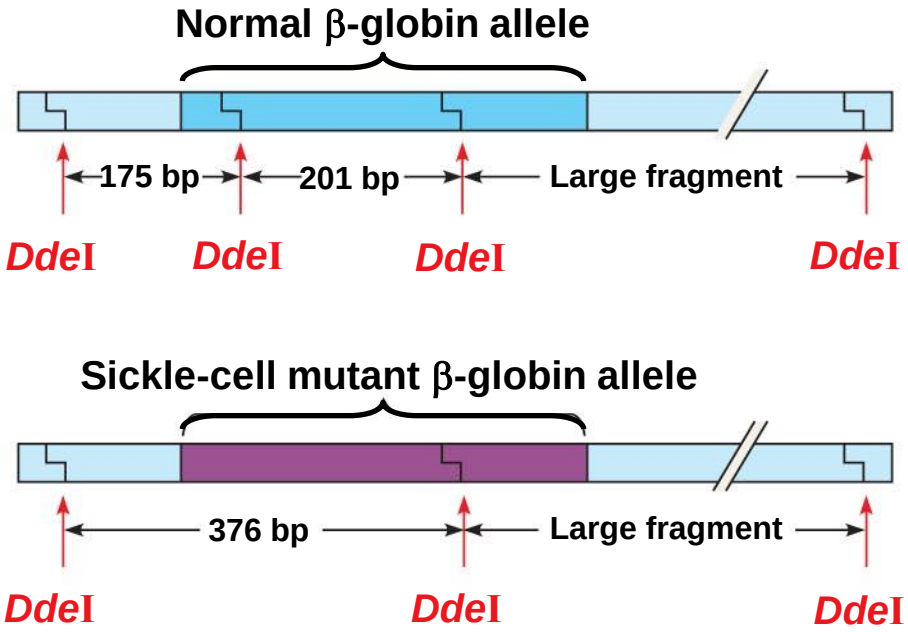
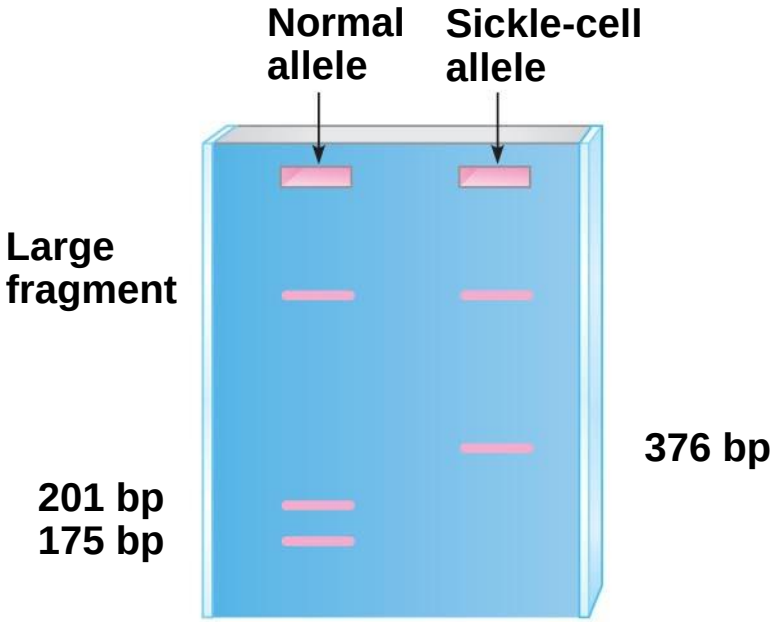


Fig. 20-10

Using Restriction Analysis



(a) *DdeI* restriction sites in normal and sickle-cell alleles of β-globin gene



(b) Electrophoresis of restriction fragments from normal and sickle-cell alleles

The products of genetic engineering include, but are not limited to, GM foods; transgenic animals; cloned animals; and pharmaceuticals.

- ***Biotechnology*** is the process of manipulating organisms of their components for the purpose of making useful products.
- Illustrative examples include:
  - Genetically modified foods;
  - Transgenic animals;
  - Cloned animals;
  - Pharmaceuticals, such as human insulin

GM Foods

- **Genetically modified foods** are foods produced from organisms that have had specific changes introduced into their DNA using the methods of genetic engineering.
- These techniques have allowed for the introduction of new crop traits as well as a far greater control over a food's genetic structure than previously afforded by methods.
- Benefits of GM Foods include:
  - Ensuring an adequate food supply for the growing human population;
  - Pest resistance or herbicide tolerance reduces the need for harmful chemicals;
  - Disease resistance and economics;
  - Cold/drought tolerance;
  - Nutritional value of commonly consumed foods.

Transgenic Animals

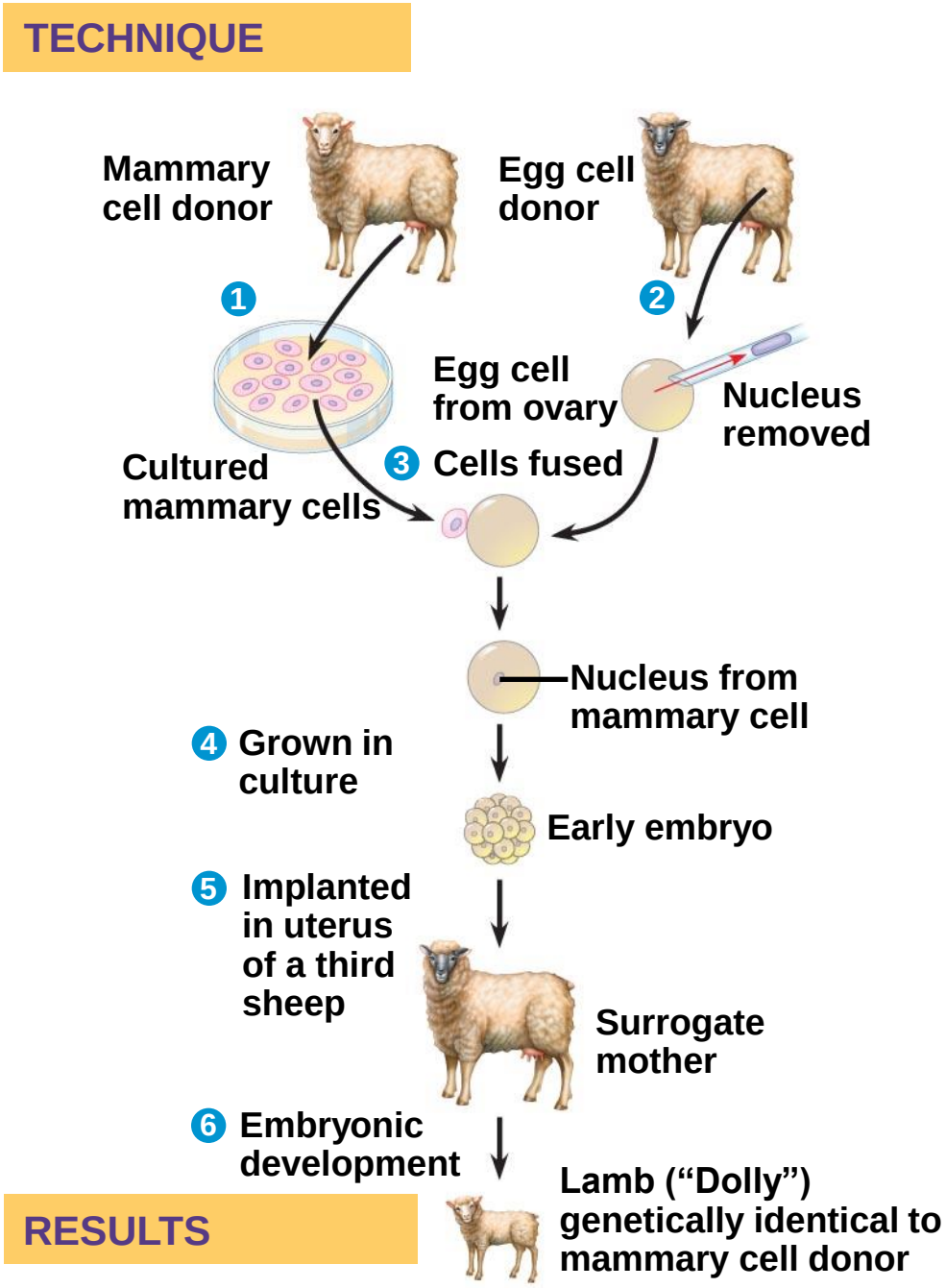
- ***Transgenic animals*** are made by introducing genes from one species into the genome of another animal.
- Transgenic animals are pharmaceutical "factories," producers of large amounts of otherwise rare substances for medical use.
- "Pharm" plants are also being developed to make human proteins for medical use.

Fig. 20-23

Cloned Animals

- In animal cloning the nucleus of an egg is removed and replaced with the diploid nucleus of a body cell.
- The major goal of most animal cloning is reproduction, but not for humans. In humans, the major goal is the production of ***stem cells***.
- A stem cell can both reproduce itself indefinitely and, under the proper conditions, produce other specialized cells.
- Stem cells have enormous potential for medical application. Embryonic stem cells are capable of differentiating into many cell types.
- The ultimate aim is to use them for the repair of damaged or disease organs, such as insulin-producing pancreatic cells for people with diabetes or certain kinds of brain cells for people with Parkinson's disease.

Fig. 20-18

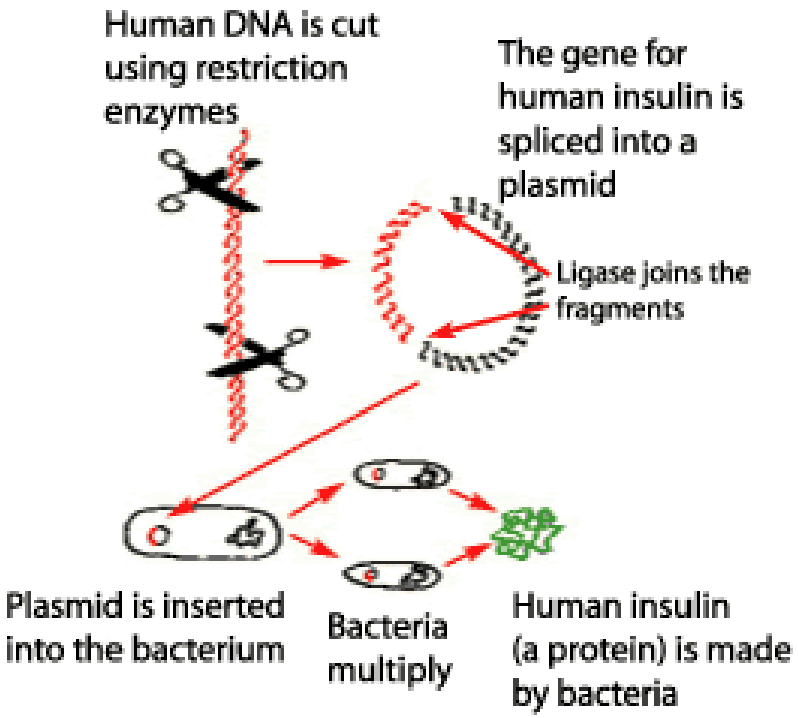


Pharmaceutical Applications

- Advances in DNA technology and genetic research are important to the development of new drugs to treat diseases.
- Host cells in culture can be engineered to secrete a protein as it is made.
- This is useful for the production of insulin, human growth hormones, and vaccines.

Pharmaceutical Products – Human Insulin

Gene splicing is used to make bacterial cells produce human insulin.



BIG IDEA III

Living systems store, retrieve, transmit and respond to information essential to life processes.

Enduring Understanding 3.A

Heritable information provides for continuity of life.

Essential Knowledge 3.A.2

In eukaryotes, heritable information is passed to the next generation via processes that include the cell cycle and mitosis or meiosis plus fertilization.

PowerPoint® Lecture Presentations for

Biology

Eighth Edition

Neil Campbell and Jane Reece

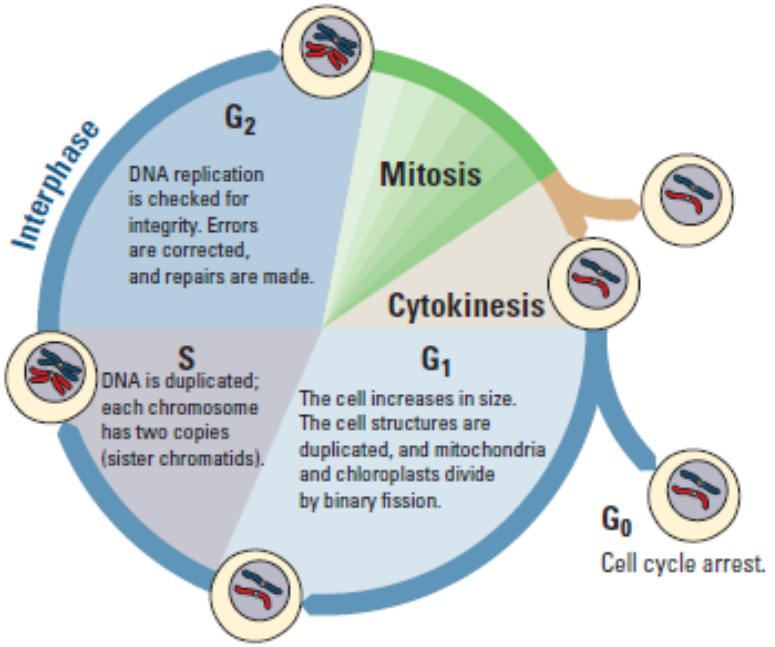
Lectures by Chris Romero, updated by Erin Barley with contributions from Joan Sharp

Essential Knowledge 3.A.2: *In eukaryotes, heritable information is passed to the next generation via processes that include the cell cycle and mitosis or meiosis plus fertilization.*

- **Learning Objectives:**
  - **(3.7)** The student can **make predictions** about natural phenomena occurring during the cell cycle.
  - **(3.8)** The student can **describe** the events that occur in the cell cycle.
  - **(3.9)** The student is able to **construct an explanation, using visual representations or narratives**, as to how DNA in chromosomes is transmitted to the next generation via mitosis, or meiosis followed by fertilization.
  - **(3.10)** The student is able to **represent the connection between** meiosis and increased genetic diversity necessary for evolution.
  - **(3.11)** The student is able to **evaluate evidence provided by data sets to support the claim** that heritable information is passed from one generation to another through mitosis, or meiosis followed by fertilization.

Mitosis alternates with interphase in the cell cycle.

- Interphase consists of three phases: G_1=growth, S=synthesis of DNA, and G_2=preparation for mitosis.
- Mitosis is division of the NUCLEUS – it consists of prophase, metaphase, anaphase, and telophase.
- Cytokinesis divides the cytoplasm and other components of the cell.

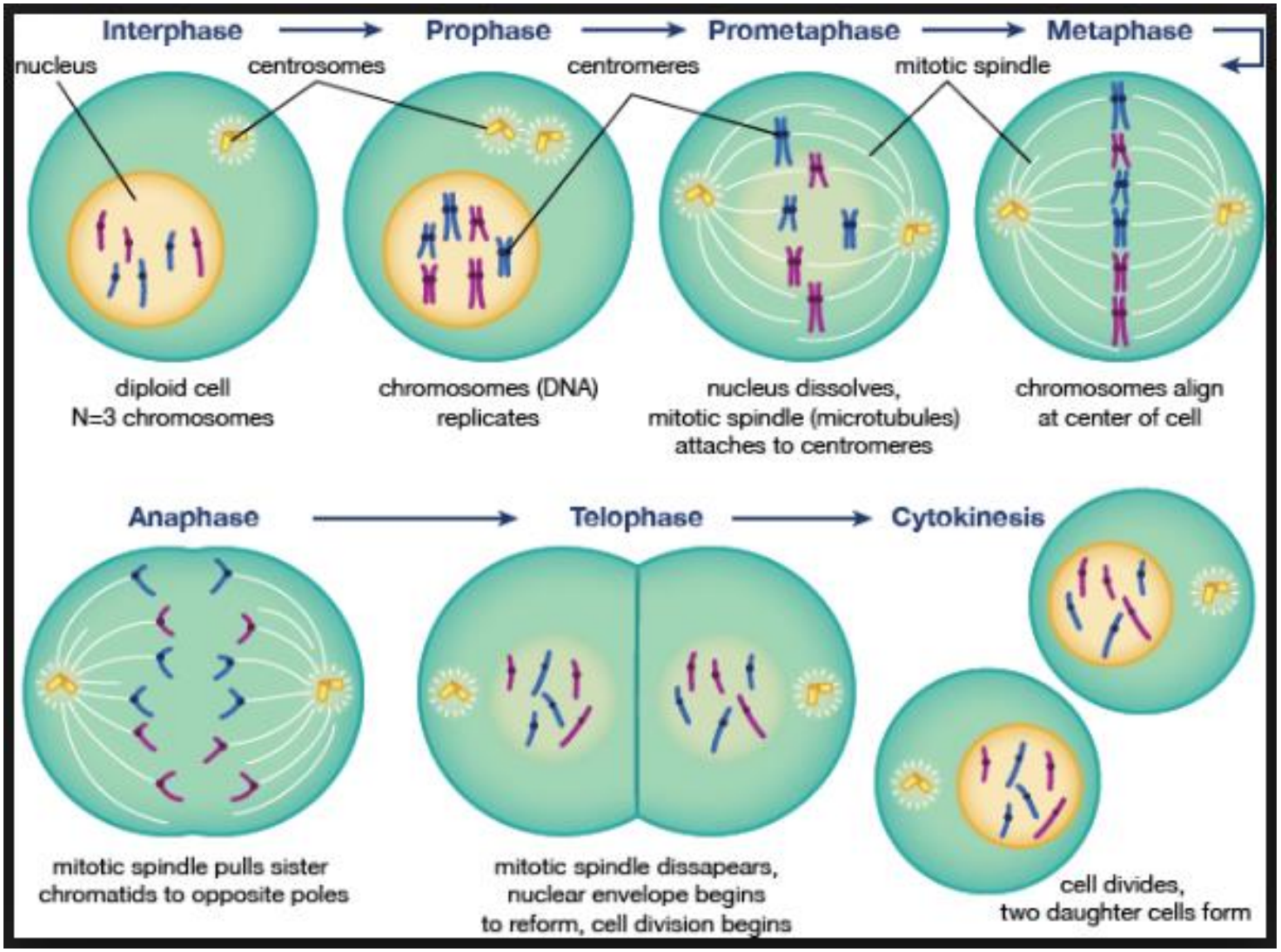


The Cell Cycle Showing G_1, S, and G_2 Phases, Mitosis, and Cytokinesis

Mitoses passes a complete genome from the parent cell to daughter cells.

- Mitosis occurs after DNA replication.
- Mitosis followed by cytokinesis produces two genetically identical daughter cells.
- Mitosis is a continuous process with observable structural features along the mitotic process. These features include replication, alignment, and separation.
- When a cell specializes, it often enters into a stage where it no longer divides, but can reenter the cell cycle when given appropriate cues. Nondividing cells may exit the cell cycle; or hold at a particular stage in the cell cycle.

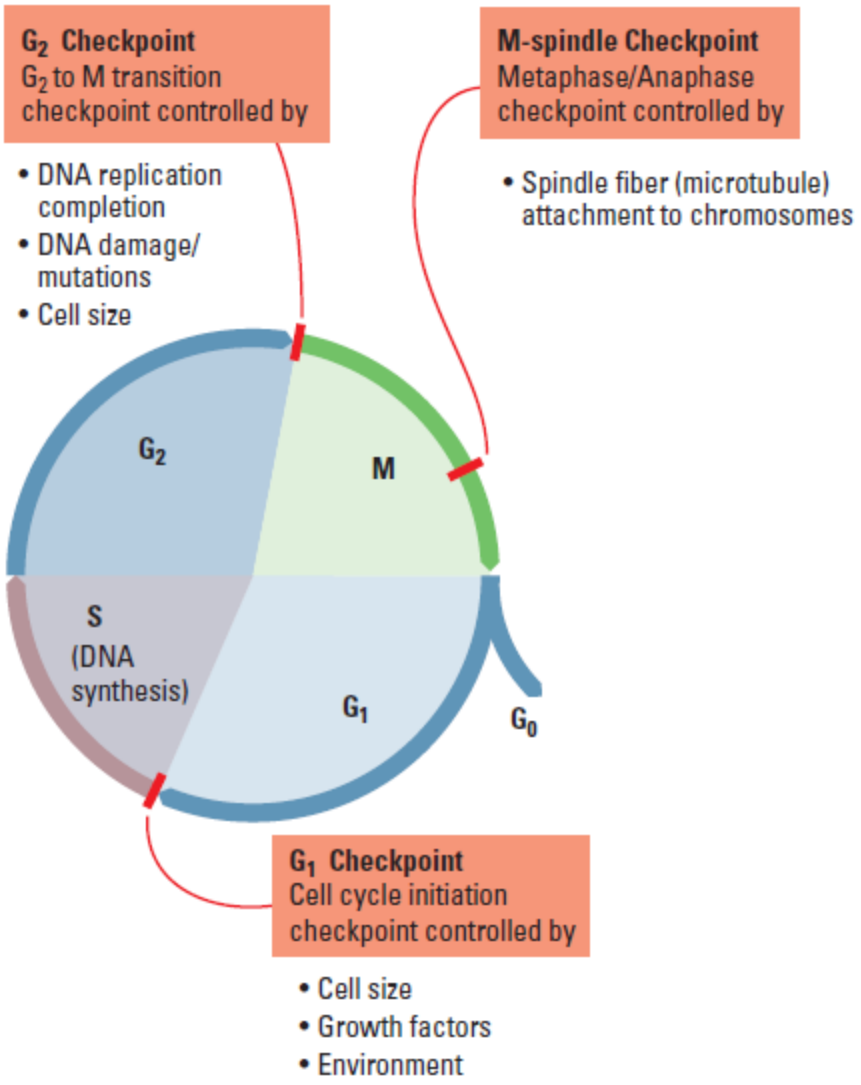
Observable Structure of Mitosis



The cell cycle is a complex set of stages that is highly regulated with checkpoints, which determine the ultimate fate of the cell.

- The cell cycle is directed by internal controls or checkpoints. Internal and external signals provide the stop-and-go signs at the checkpoints.
- A ***checkpoint*** in the cell cycle is a control point where stop and go ahead signals can regulate the cycle.
- These signals report whether crucial cellular processes that should have occurred by that point have in fact been completed correctly and thus whether or not the cell cycle should proceed.
- Checkpoints also register signals from outside the cell.

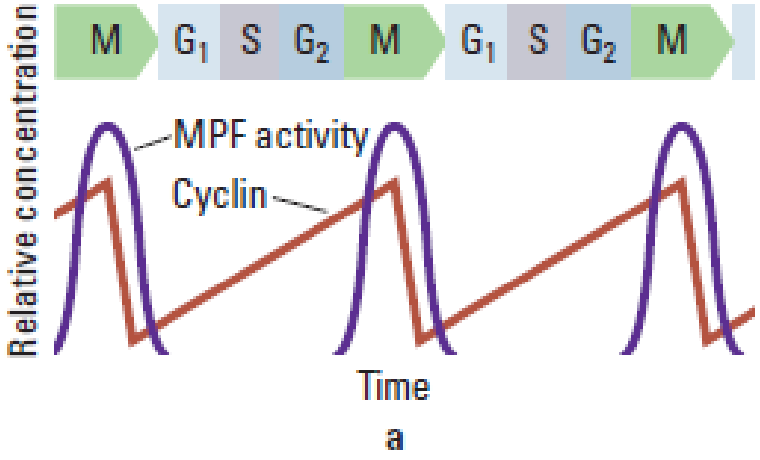
The three major checkpoints of the cell cycle are found in the G_1, G_2, and M phases.



The Cell Cycle Clock: Cyclins and Cyclin-Dependent Kinases

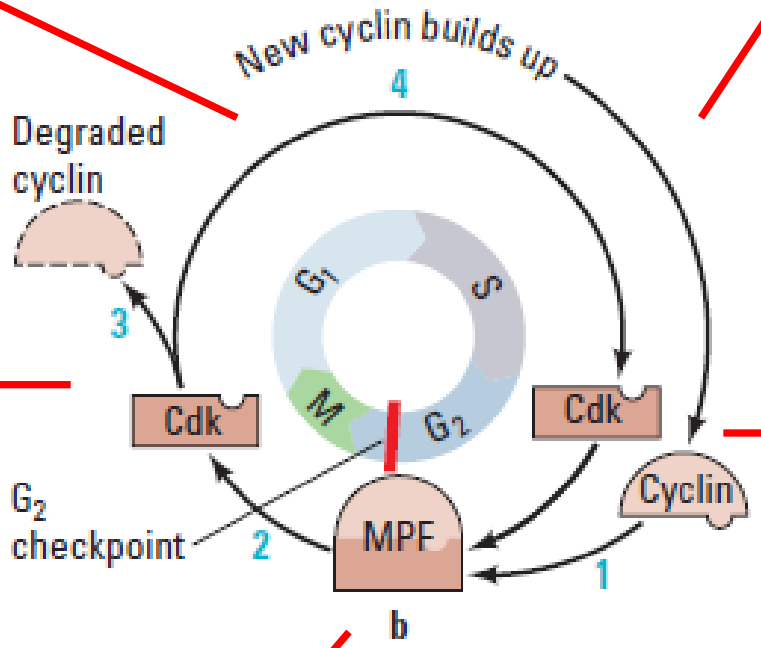
- Cell division is tightly controlled by complexes made of several specific proteins. These complexes contain enzymes called ***cyclin-dependent kinases*** (CDKs), which turn on or off the various processes that take place in cell division.
- Many of the kinases that drive the cell cycle are actually present at a constant concentration in the growing cell, but much of the time they are in an inactive form.
- To be active, a kinase much be attached to a cyclin, and is therefore called a cyclin-dependent kinase, or CDK.
- On such complex is ***mitosis-promoting factor*** (MPF), which contains cyclin A or B attached to CDK.
- MPF is *activated* when it is bound to cyclin, interacting with various other proteins that, in this case, allow the cell to proceed from G_2 into mitosis.

MPF Production During the Cell Cycle



During G_1, conditions in the cell favor degradation of cyclin, and the CDK component of MPF is recycled.

Synthesis of cyclin begins in late S phase and continues through G2. Because cyclin is protected from degradation during this stage, it accumulates.



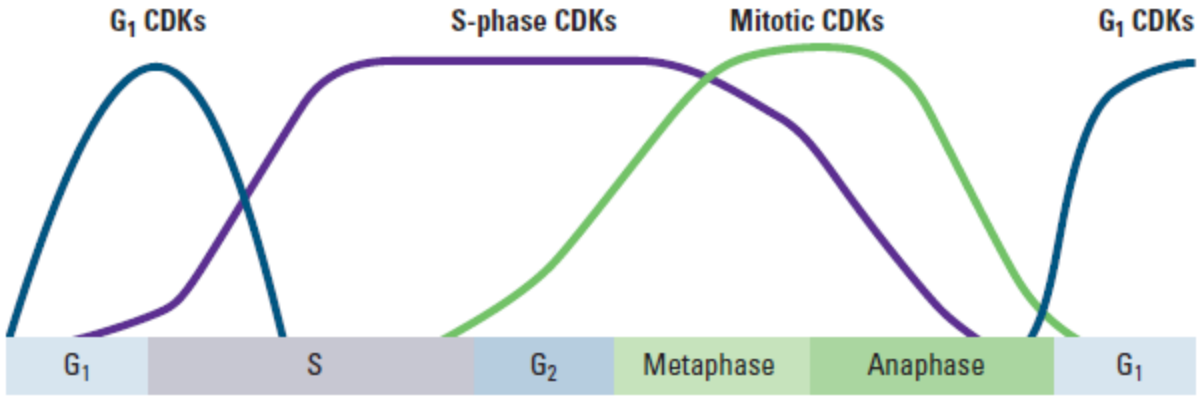
During anaphase, the cyclin component of MPF is degraded, terminating the M phase. The cell enters the G_1 phase.

Accumulated cyclin molecules combine with recycle CDK, producing enough molecules of MPF for the cell to pass the G2 checkpoint and initiate the events of mitosis.

MPF promotes mitosis by phosphorylating various proteins. MPF's activity peaks during interphase.

Levels of CDKs During the Cell Cycle

- Different CDKs are produced during the phases of the cell cycle.
- The cyclins determine which processes in cell division are turned on or off and in what order by CDK.
- As each cyclin is turned on or off, CDK causes the cell to move through the stages in the cell cycle.
- REMINDER: cyclins and CDKs do not allow the cell to progress through its cycle automatically, there are still three checkpoints a cell must pass through!

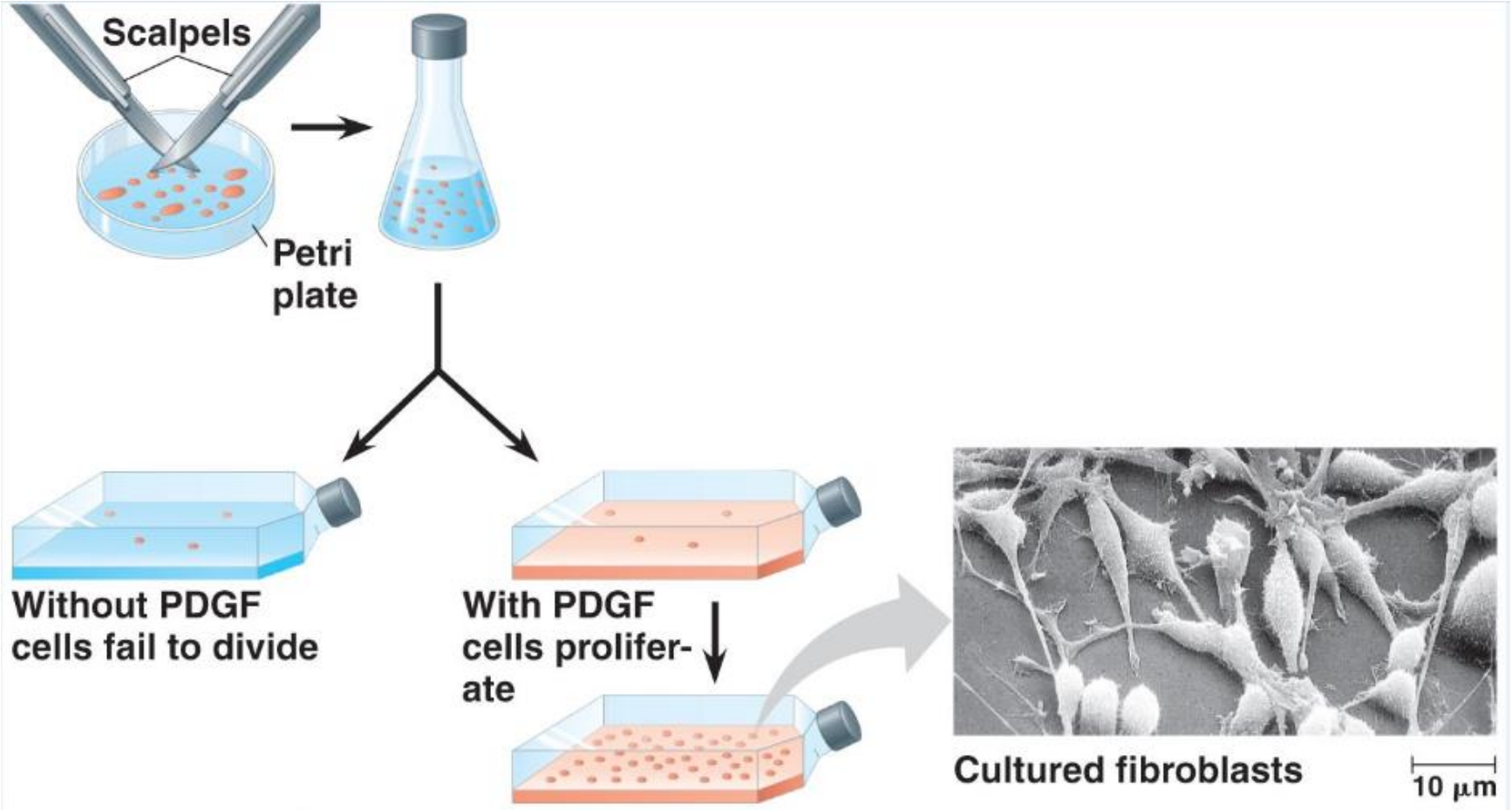


Stop and Go Signs: External Signals at the Checkpoints

- Many external factors, both chemical and physical, can influence cell division.
- For example, even when all other conditions are favorable, most types of animals cells divide in culture only if the growth medium includes specific growth factors.
- A ***growth factor*** is a protein released by certain cells that stimulates other cells to divide.
- These are local regulators – they travel short distances influence only cells in close vicinity.
- One such example is ***platelet-derived growth factor***, or PDGF.

The Action of External Factors: PGDF

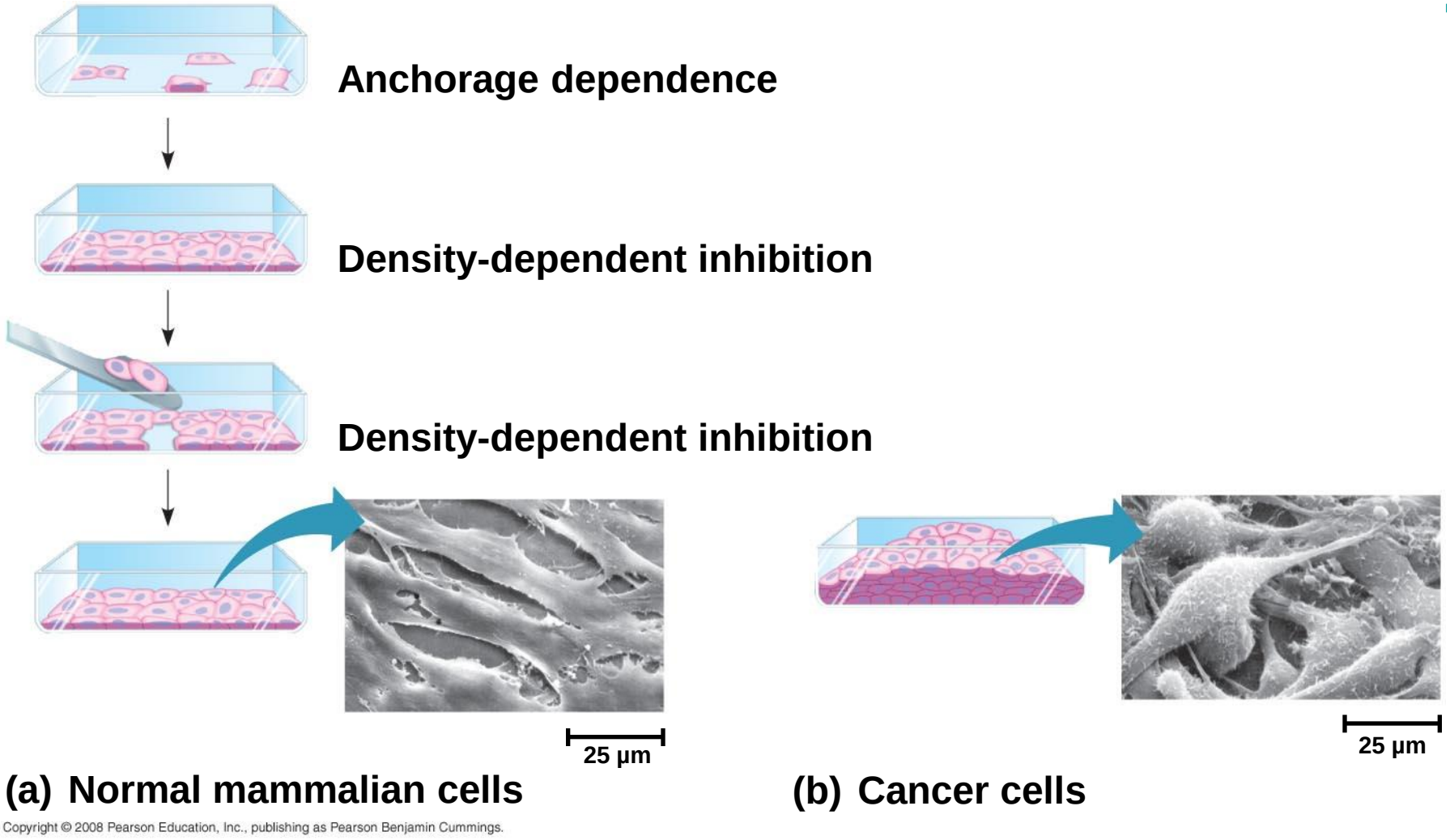
http://highered.mcgraw-hill.com/sites/9834092339/student_view0/chapter10/cell_proliferation_signaling_pathway.html



Contact Cell Cycle Control

- Contact cell cycle control mechanisms include density-dependent inhibition and anchorage dependence.
  - In **density-dependent inhibition**, crowded cells stop dividing.
  - **Anchorage dependence** is a phenomenon in which cells must be attached to a substratum (*i.e.* extracellular matrix of a tissue) in order to divide.

Fig. 12-19



Loss of Cell Cycle Controls in Cancer Cells

- The cell cycle is regulated very precisely. Mutations in cell cycle genes that interfere with proper cell cycle control are found very often in **cancer cells**.
- Cancer cells do not respond normally to the body's control:
  - They exhibit neither density-dependent inhibition nor anchorage dependence.
  - They may not need growth factors to grow and divide.
  - If and when they do stop dividing, cancer cells do so at random points in the cycle, rather than at the normal checkpoints.

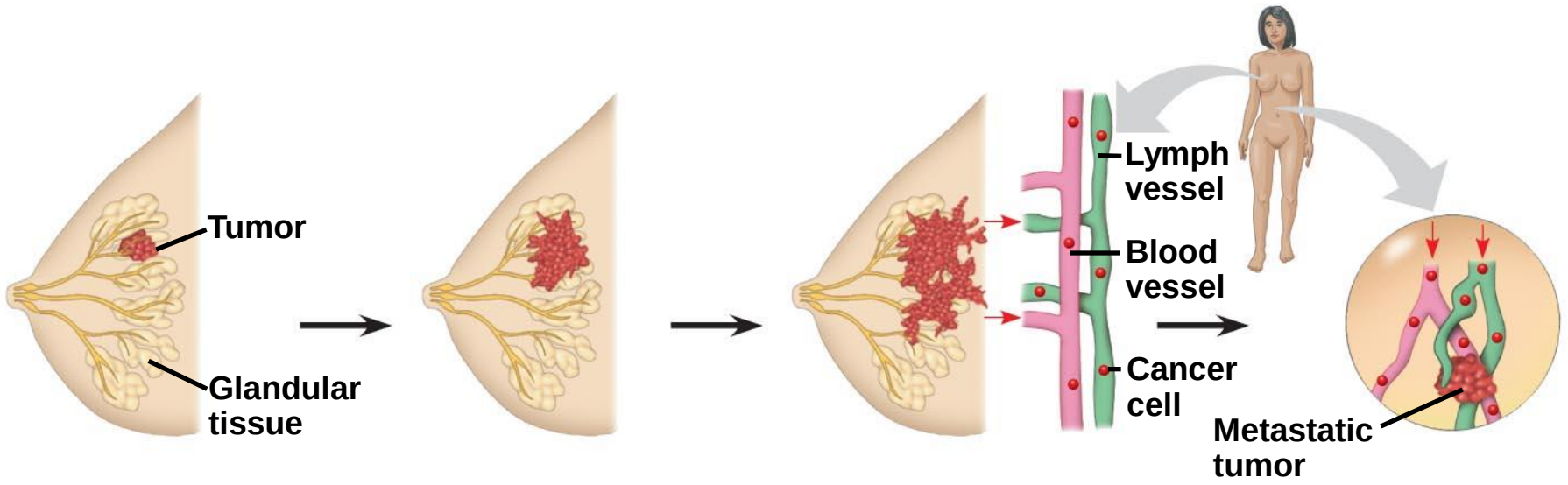
The Immortality of Cancer Cells

- Cancer cells can go on dividing indefinitely in culture if they are given a continual supply of nutrients.
- A striking example is a cell line that has been reproducing in culture since 1951.
- Cells of this line are called HeLa cells because their original source was a tumor removed from a woman named Henrietta Lacks.
- By contrast, nearly all normal mammalian cells growing in culture divide only about 20 to 50 times before they stop dividing, age, and die.

Cancer Cells and the Formation of Tumors

- A normal cell is converted to a cancerous cell by a process called **transformation.**
- Cancer cells form tumors, masses of abnormal cells within otherwise normal tissue.
- If abnormal cells remain at the original site, the lump is called a **benign tumor.**
- **Malignant tumors** invade surrounding tissues and can **metastasize**, exporting cancer cells to other parts of the body, where they may form secondary tumors.

Fig. 12-20
http://www.youtube.com/watch?v=IeUANxFVXKc



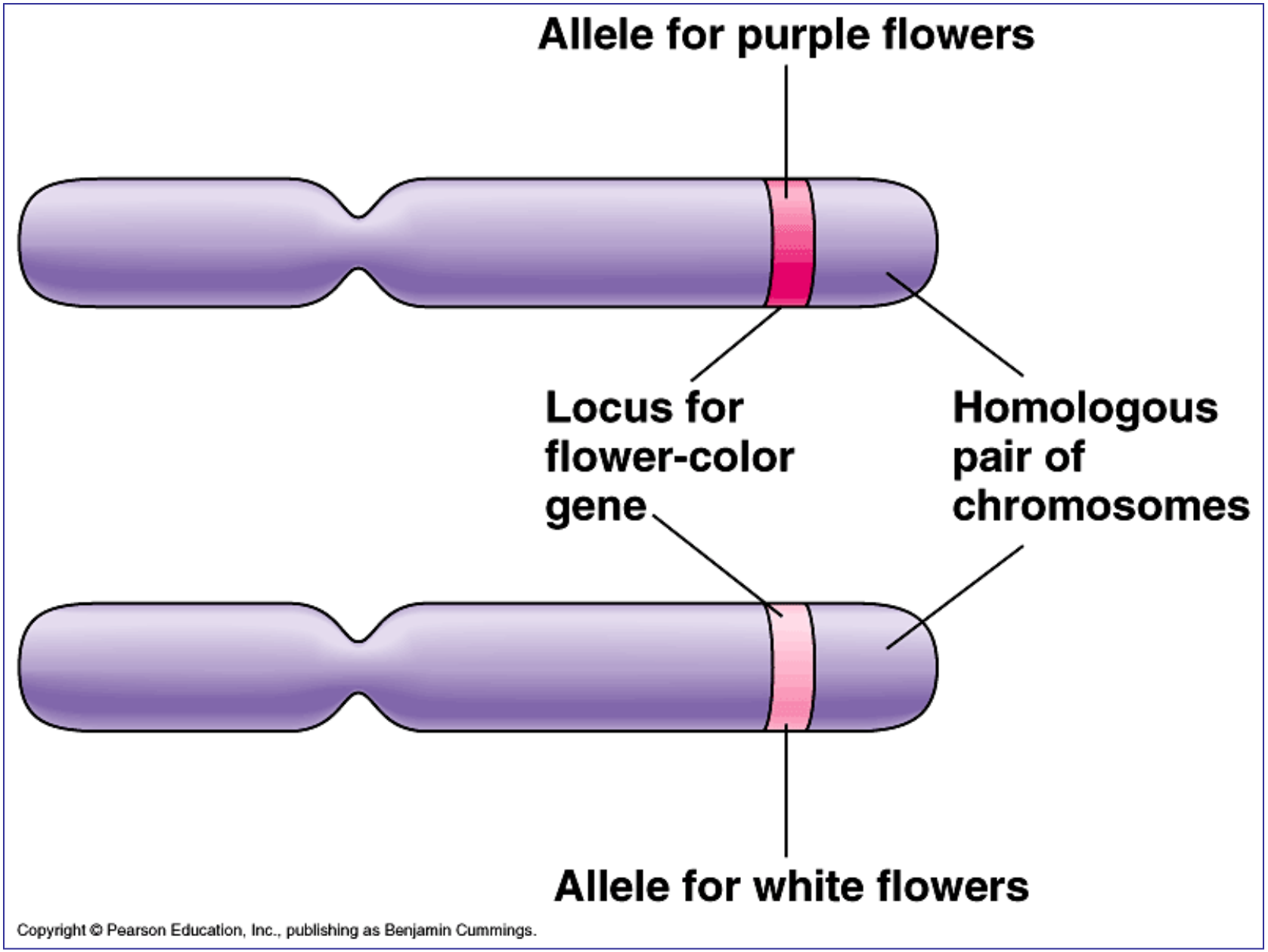
Meiosis, a reduction division, followed by fertilization ensures genetic diversity in sexually reproducing organisms.

- Meiosis ensures that each gamete receives one complete ***haploid*** (1n) set of chromosomes.
- During meiosis, ***homologous chromosomes*** are paired, with one homologue originating from the maternal parent and the other from the paternal parent.
- Orientation of the chromosome pairs is ***random*** with respect to the cell poles.
- Separation of the homologous chromosomes ensures that each gamete receives a haploid (1n) set of chromosomes composed of both maternal and paternal chromosomes.
- During meiosis, homologous chromatids exchange genetic material via "***crossing over***", which increases genetic variation in the resultant gametes.
- ***Fertilization*** involves the fusion of two gametes, increases genetic variation in populations by providing for new combinations of genetic information in the zygote, and restores the diploid number of chromosomes.

Acquiring Genes from Parents

- In a literal sense, children do not inherit particular physical traits from their parents…it is genes that are actually inherited.
  - **Genes** are the units of heredity, and are made up of segments of DNA.
  - Genes are passed to the next generation through reproductive cells called **gametes** (sperm and eggs).
  - Each gene has a specific location called a **locus** on a certain chromosome.
- Most DNA is packaged into **chromosomes**.
- One set of chromosomes is inherited from each parent.

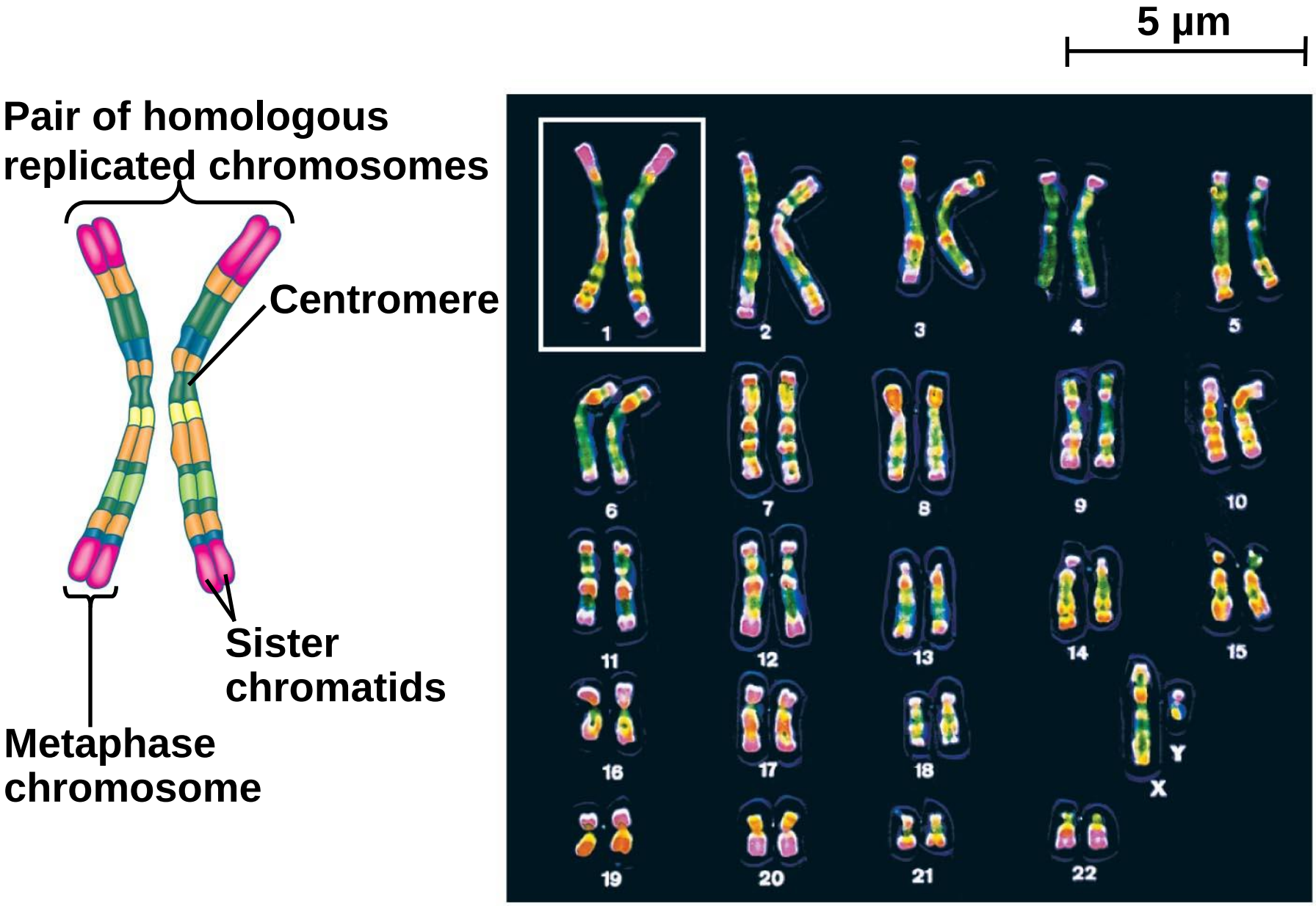
Fig. 14-4



Sets of Chromosomes in Human Cells

- Human **somatic cells** (any cell other than a gamete) 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 carry genes controlling the *same inherited characters*. ***One is inherited from the mother, and the other from the father.***

Fig. 13-3b



Homologous Chromosomes

- **HOMOLOGOUS CHROMOSOMES** are chromosome pairs of the same length, centromere position, staining pattern, and gene possession for the same characters:
  - One homologous chromosome is inherited from the organism's father and the other from the mother.
- **DIPLOID** – means "two sets"
  - This represents the cells in which the chromosomes are paired up and have a partner in size and shape.
  - A cell containing TWO sets of chromosomes (2n), one set inherited from each parent is referred to as a **DIPLOID CELL.**

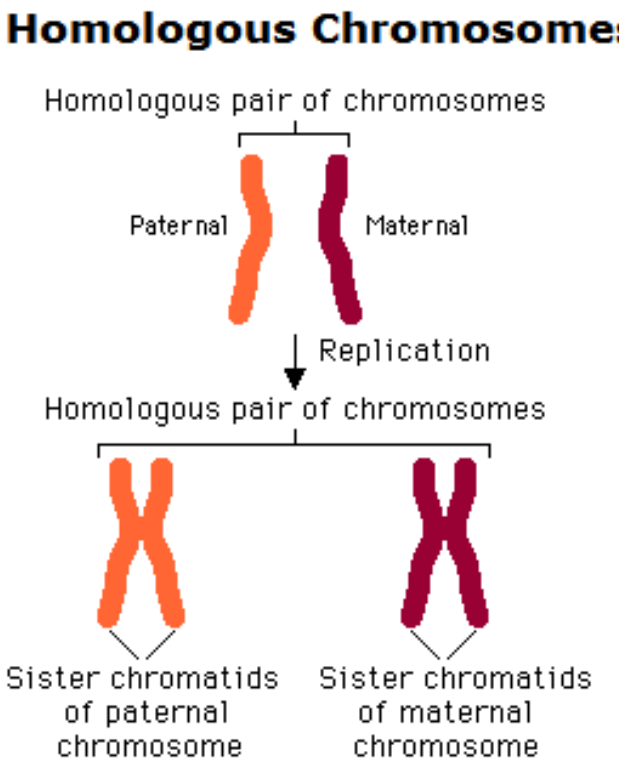
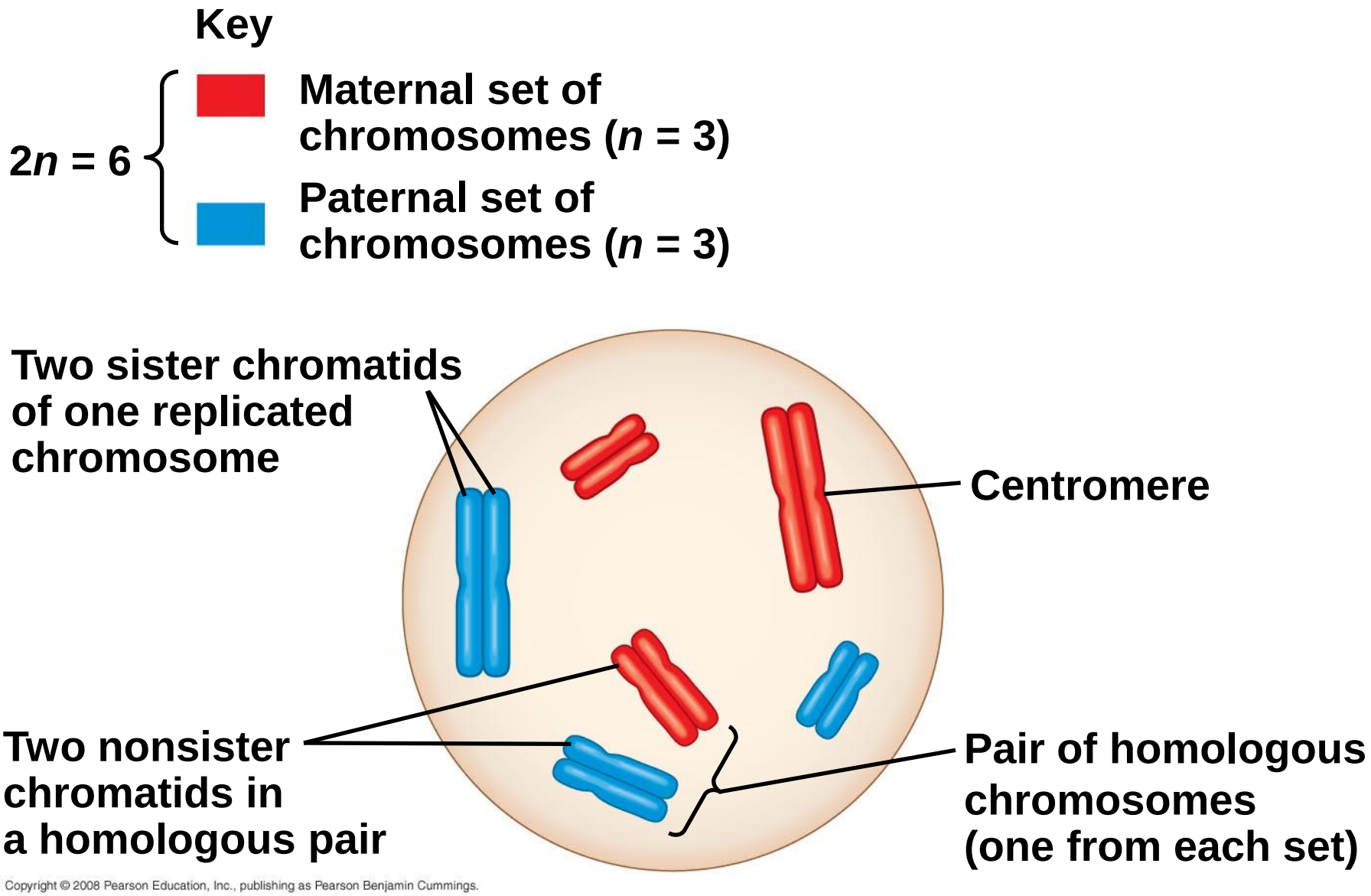


Fig. 13-4



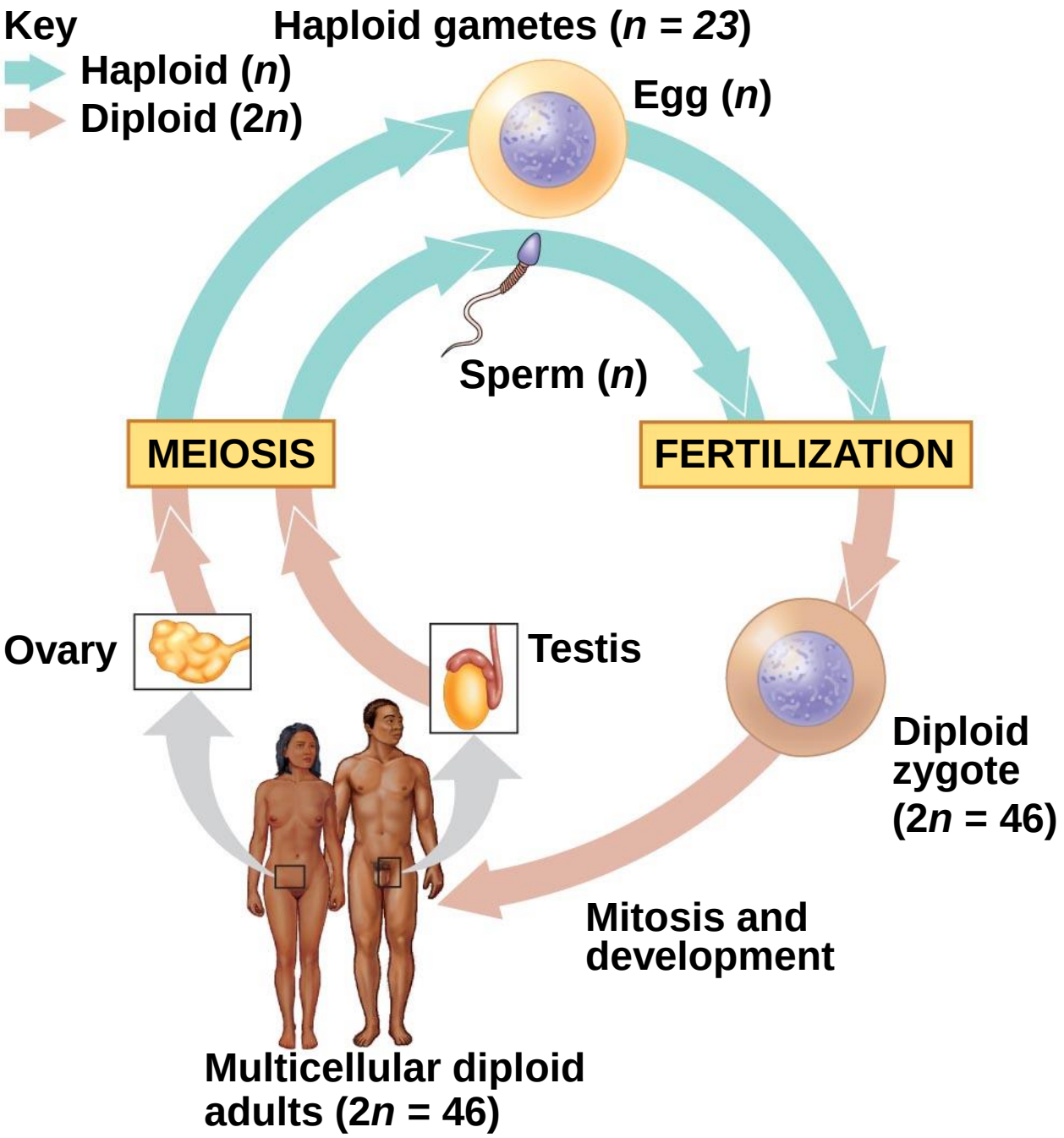
Sex Chromosomes v. Autosomes

- The **sex chromosomes** are called X and Y:
  - Human females have a homologous pair of X chromosomes (XX).
  - Human males have one X and one Y chromosome (XY).
  - The 22 pairs of chromosomes that do not determine sex are called **autosomes.**

Haploid

- A gamete (sperm or egg) contains a single set of chromosomes, and is **haploid** (1*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.

Fig. 13-5



Meiosis is REDUCTION DIVISION

- **Meiosis reduces the number of chromosome sets from diploid to haploid.**
- Like mitosis, meiosis is preceded by the replication of chromosomes.
- Meiosis 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.**

The Stages of Meiosis

- In the first cell division (meiosis I), homologous chromosomes separate:
  - Meiosis I results in two haploid daughter cells with replicated chromosomes; it is called the ***reduction division.***
- In the second cell division (meiosis II), sister chromatids separate:
  - Meiosis II results in four haploid daughter cells with unreplicated chromosomes; it is called the ***equational division.***

Fig. 13-7-3

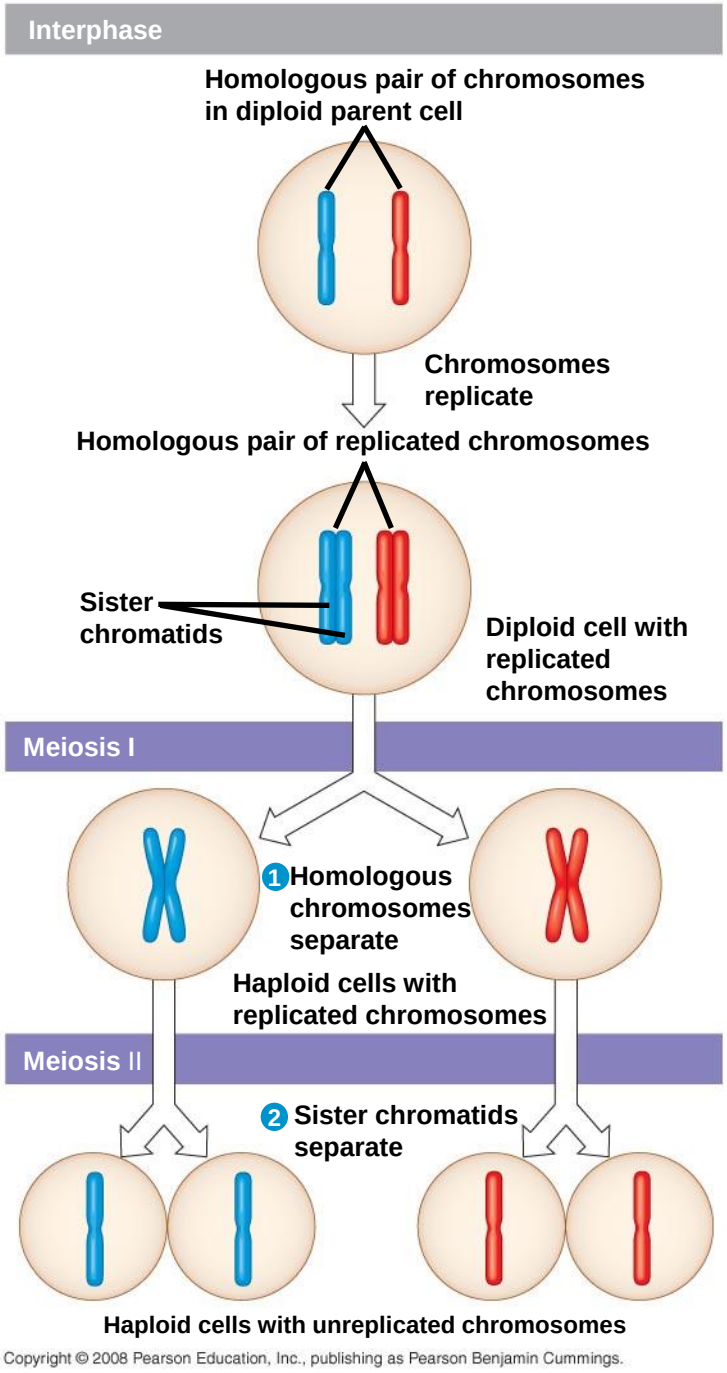


Figure 13.7 The stages of meiotic cell division: Meiosis I

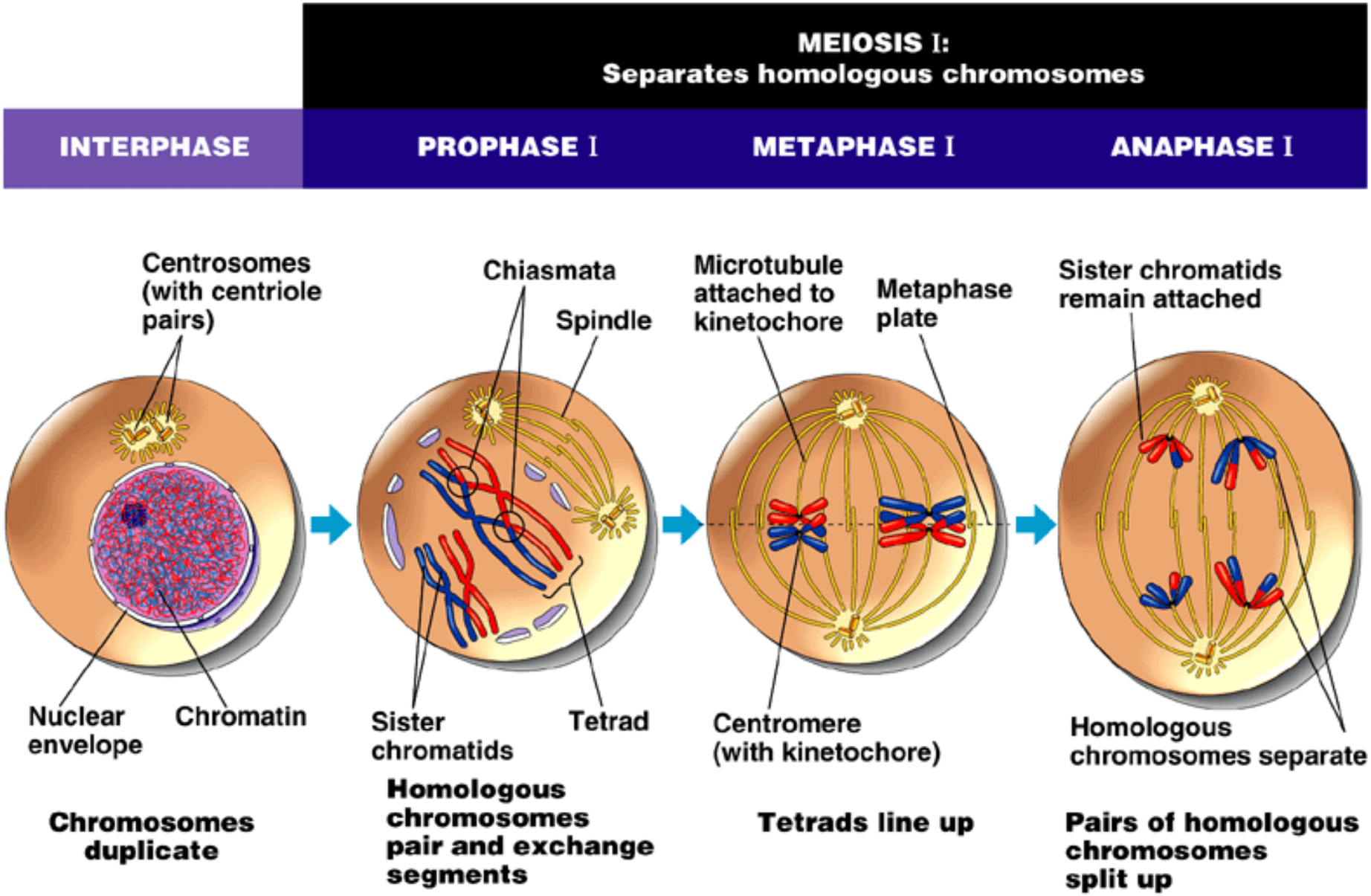
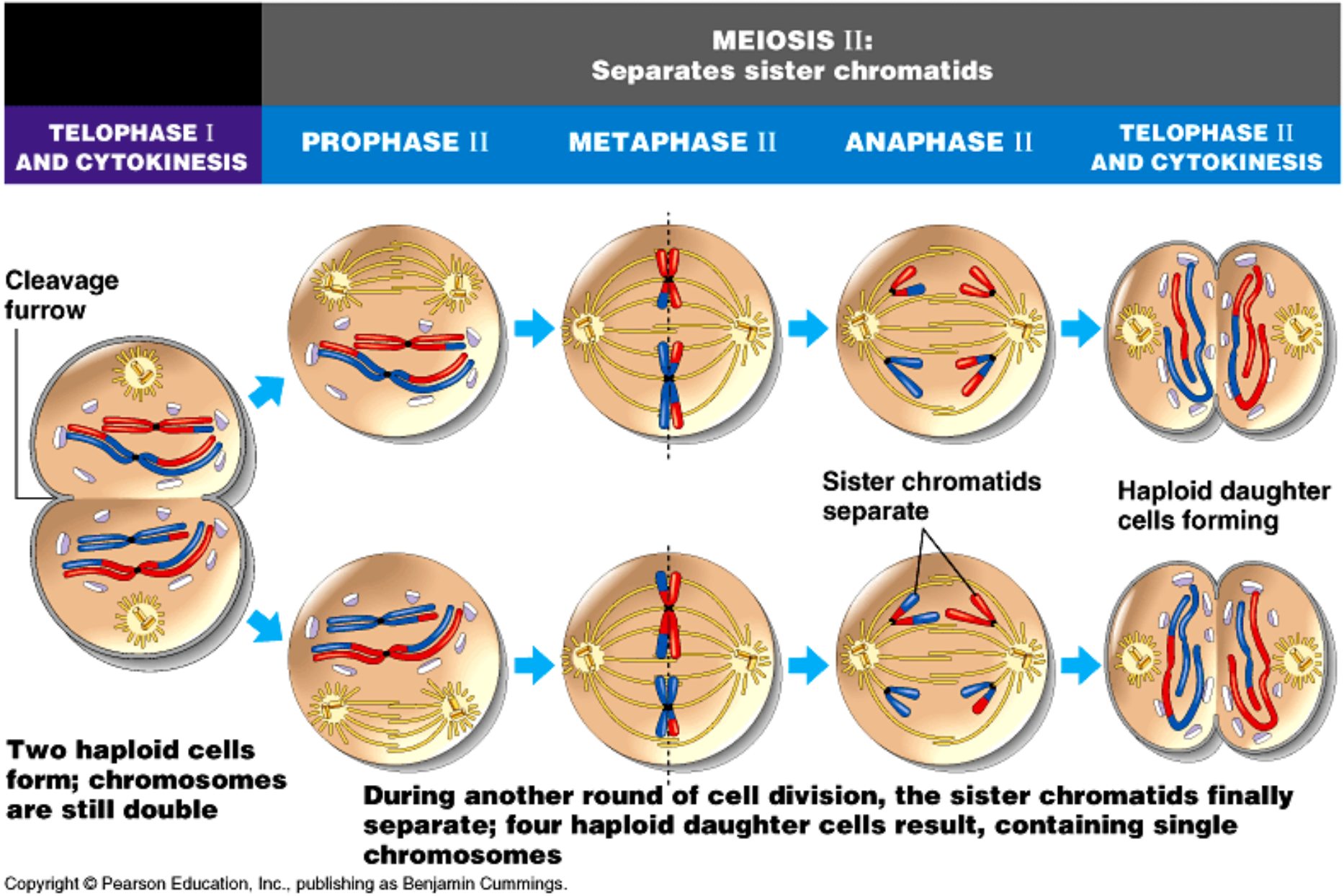


Figure 13.7 The stages of meiotic cell division: Meiosis II



Crossing Over During Meiosis I

http://highered.mcgraw-hill.com/sites/dl/free/0072835125/126997/animation5.html

- As homologous chromosomes pair up and form tetrads in meiosis I, they may exchange portions of their chromatids.
  - This event is called **CROSSING OVER**.
- This results in an exchange of genetic material that gives new gene combinations**:**
  - The site at which the exchange occurs is called the **chiasma**.

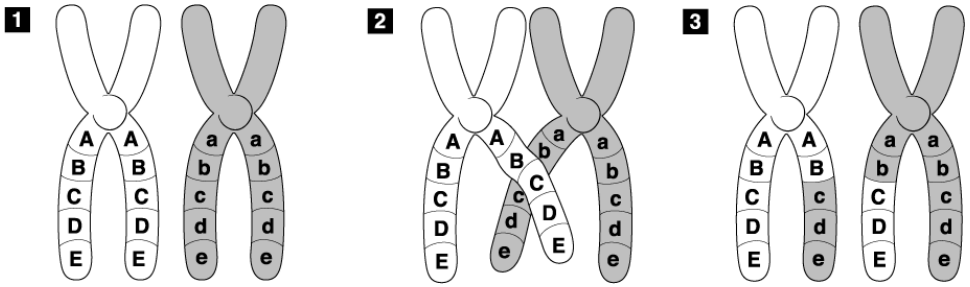
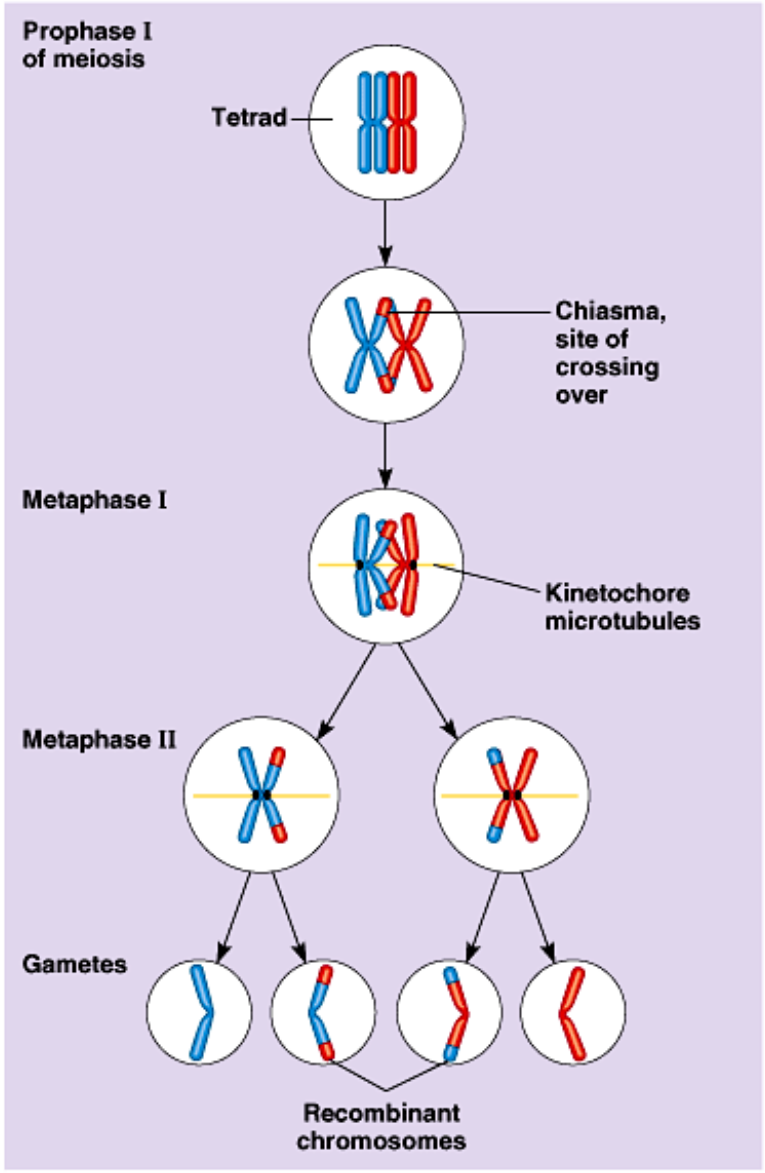


Figure 13.10 The results of crossing over during meiosis.



During prophase I of meiosis, the duplicated chromosomes pair with their homologues, in a process called **synapsis**.

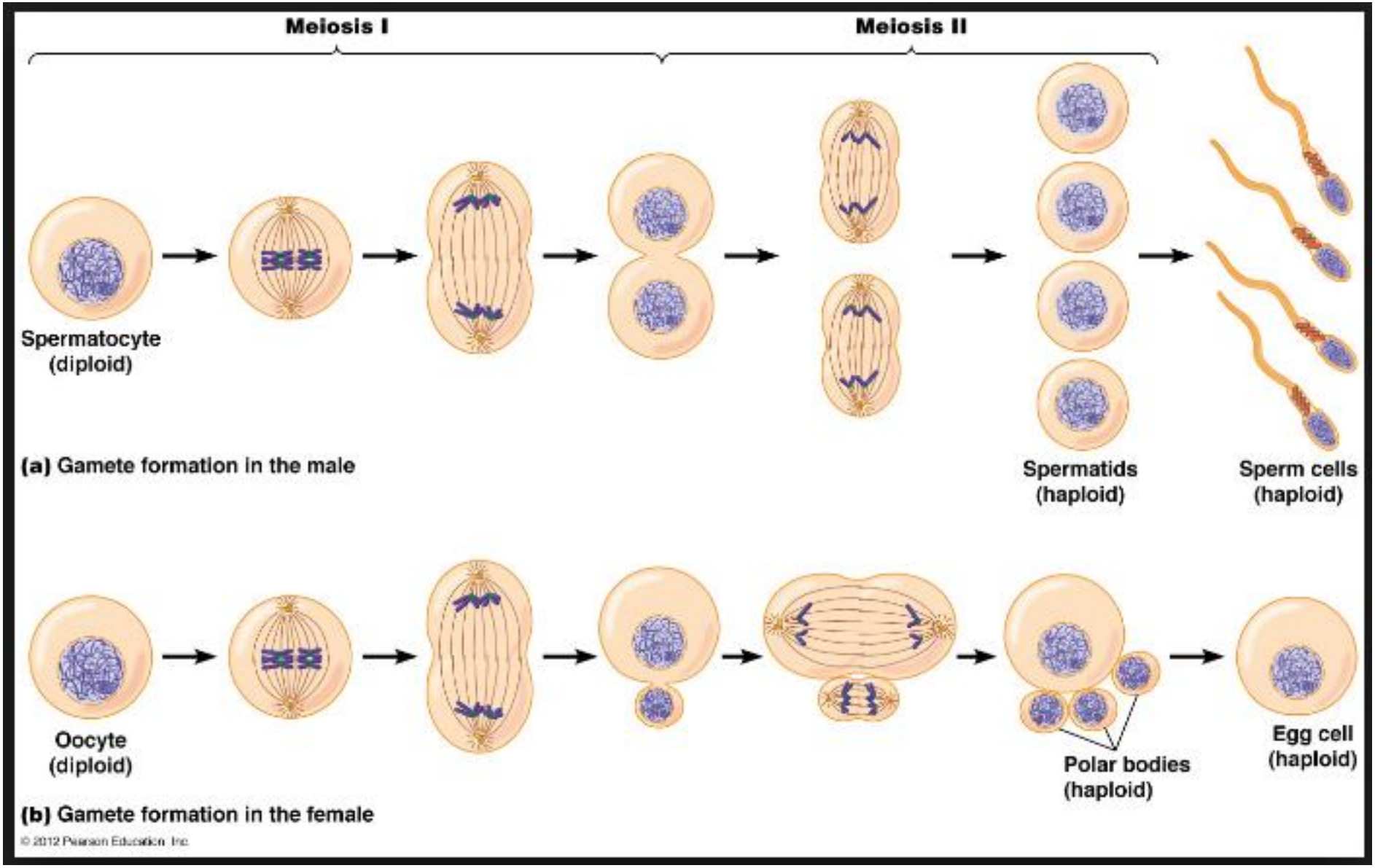
During this process, a protein "zipper" called the **synaptonemal complex** holds the homologous chromosomes tightly together all along their lengths.

When this complex disappears in late prophase, the four closely associated chromatids of a homologous pair are visible as a **tetrad**.

Crossing Over (exchanging portions of homologous chromosomes) occurs during PROPHASE I OF MEIOSIS I.

The site at which the crossing over exchange occurs is called the **chiasma**.

Crossing over gives rise to **recombinant chromosomes**, individual chromosomes that have some combination of DNA originally derived from 2 different parents.


Meiosis I
Meiosis II
Spermatocyte
(diploid)
(a) Gamete formation in the male
Spermatids
(haploid)
Sperm cells
(haploid)
Oocyte
(diploid)
Polar bodies
(haploid)
Egg cell
(haploid)
(b) Gamete formation in the female
© 2012 Pearson Education, Inc.

A Comparison of Mitosis and Meiosis

- Mitosis **conserves** the number of chromosome sets, producing cells that are genetically identical to the parent cell.
- Meiosis **reduces** the number of chromosomes sets from two (diploid) to one (haploid), producing cells that differ genetically from each other and from the parent cell.
- The mechanism for separating sister chromatids is virtually identical in meiosis II and mitosis.

Events Unique to Meiosis

- Three events are unique to meiosis, and all three occur in meiosis I:
  - ***Synapsis and crossing over in prophase I***: Homologous chromosomes physically connect and exchange genetic information.
  - At the metaphase plate, there are paired homologous chromosomes (***tetrads***), instead of individual replicated chromosomes.
  - At anaphase I, it is ***homologous chromosomes***, instead of sister chromatids, that ***separate.***

Figure 13.8 A comparison of mitosis and meiosis

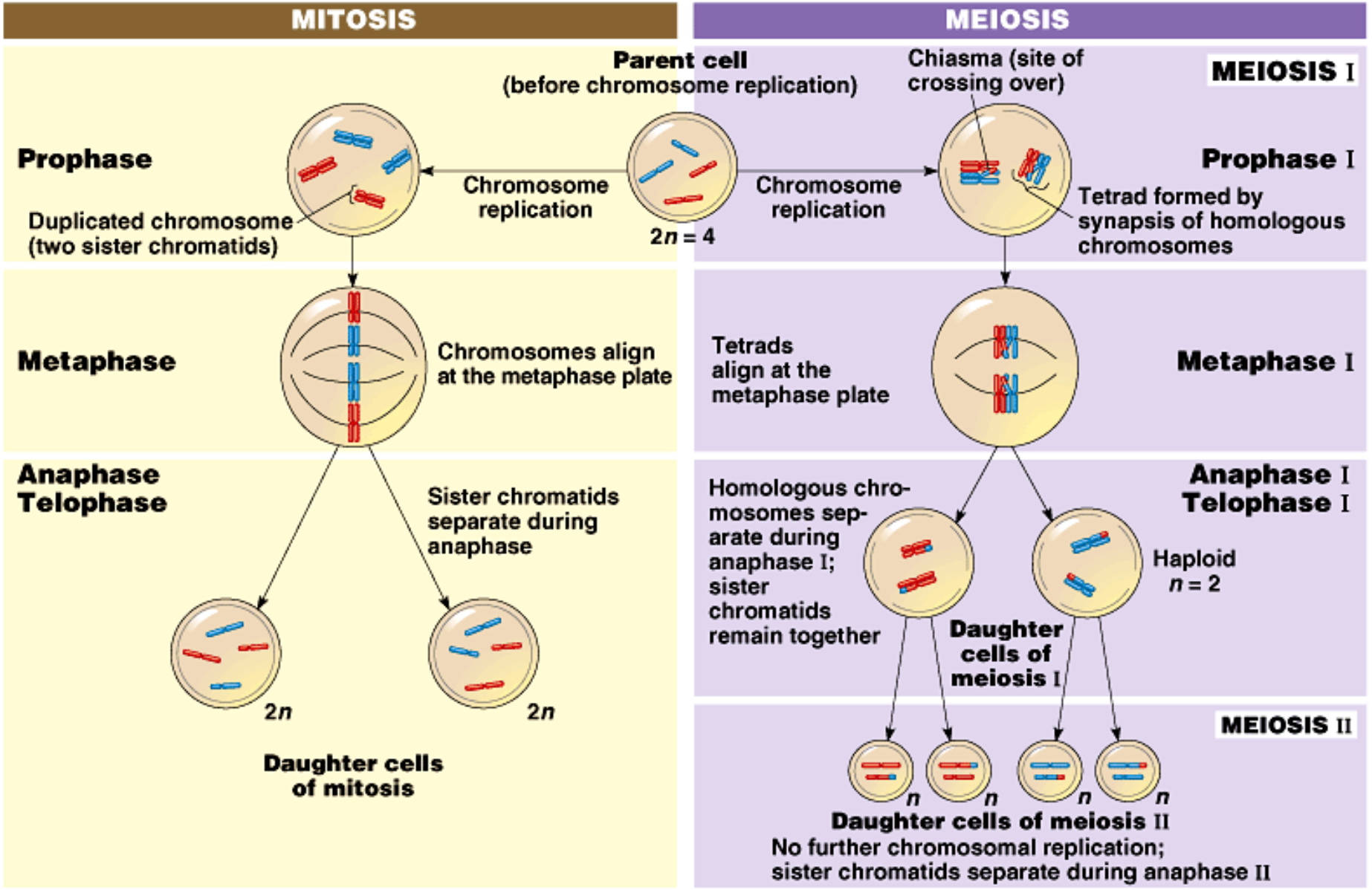


Figure 13.8 A comparison of mitosis and meiosis: summary

SUMMARY

Event	Mitosis	Meiosis
DNA replication	Occurs during interphase before nuclear division begins	Occurs once, during the interphase before meiosis I begins
Number of divisions	One, including prophase, metaphase, anaphase, and telophase	Two, each including prophase, metaphase, anaphase, and telophase
Synapsis of homologous chromosomes	Does not occur	Synapsis is unique to meiosis: During prophase I, the homologous chromosomes join along their length, forming tetrads (groups of four chromatids); synapsis is associated with crossing over between nonsister chromatids
Number of daughter cells and genetic composition	Two, each diploid ($2n$) and genetically identical to the parent cell	Four, each haploid (n), containing half as many chromosomes as the parent cell; genetically nonidentical to the parent cell and to each other
Role in the animal body	Enables multicellular adult to arise from zygote; produces cells for growth and tissue repair	Produces gametes; reduces chromosome number by half and introduces genetic variability among the gametes

IPMAT + cytokinesis

IPMAT + PMAT + cytokinesis

Genetic Variation and Evolution

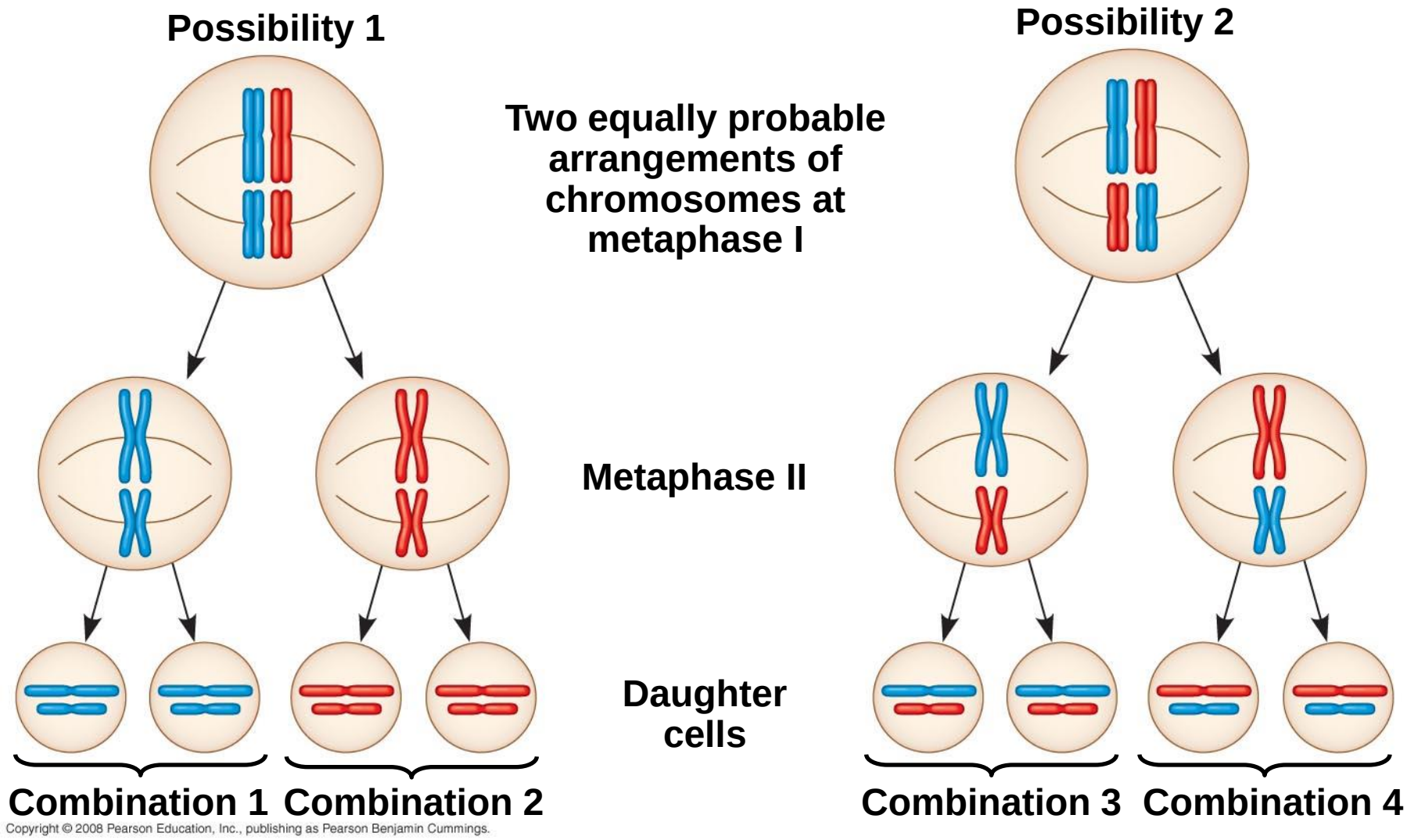
- Genetic variation produced in sexual life cycles contributes to evolution.
  - Mutations (changes in an organism's DNA) are the original source of genetic diversity.
  - Mutations create different versions of genes called *alleles*.
  - Reshuffling of alleles during sexual reproduction produces genetic 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

Fig. 13-11-3 **Independent Assortment of Chromosomes**

http://www.sumanasinc.com/webcontent/animations/content/independentassortment.html



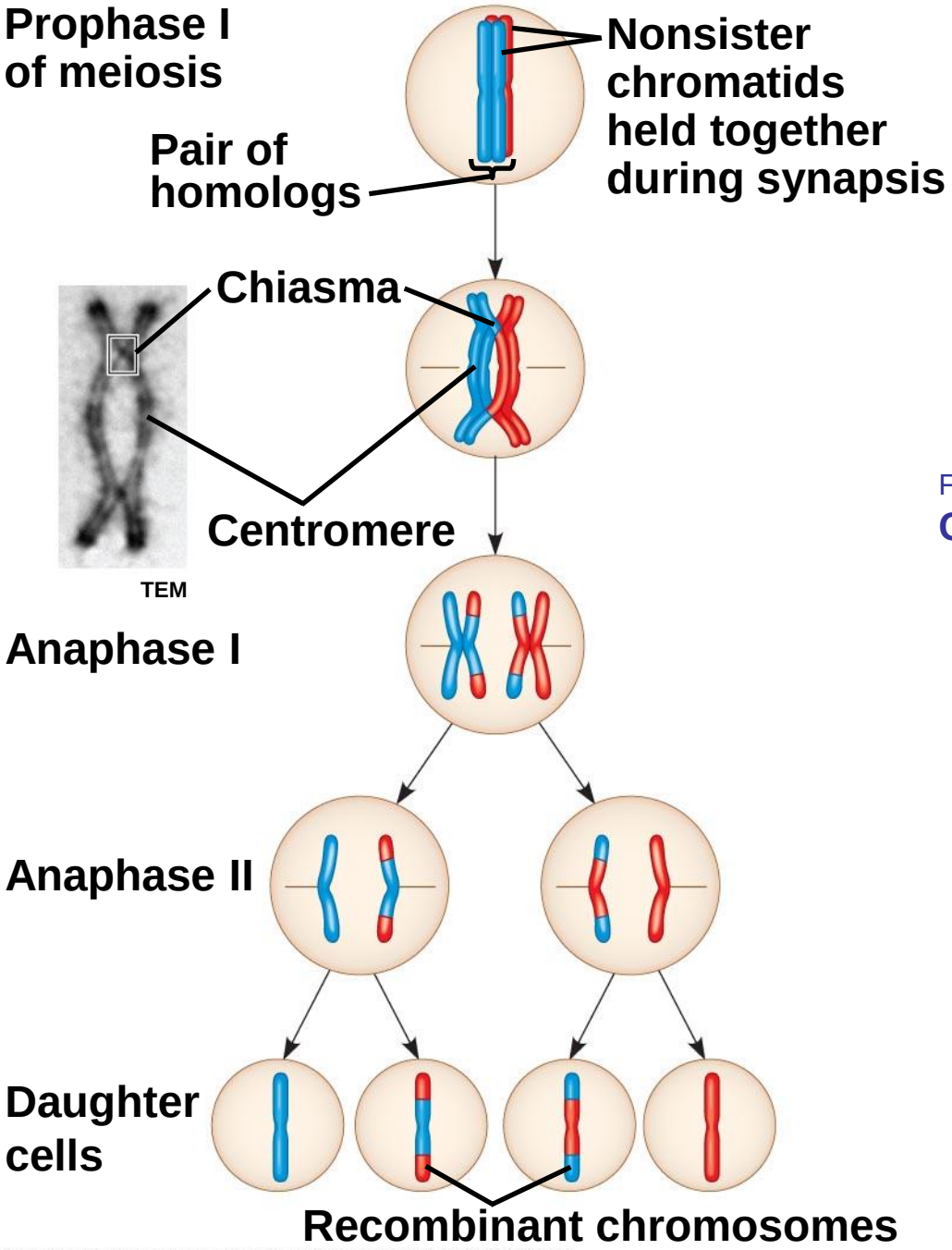
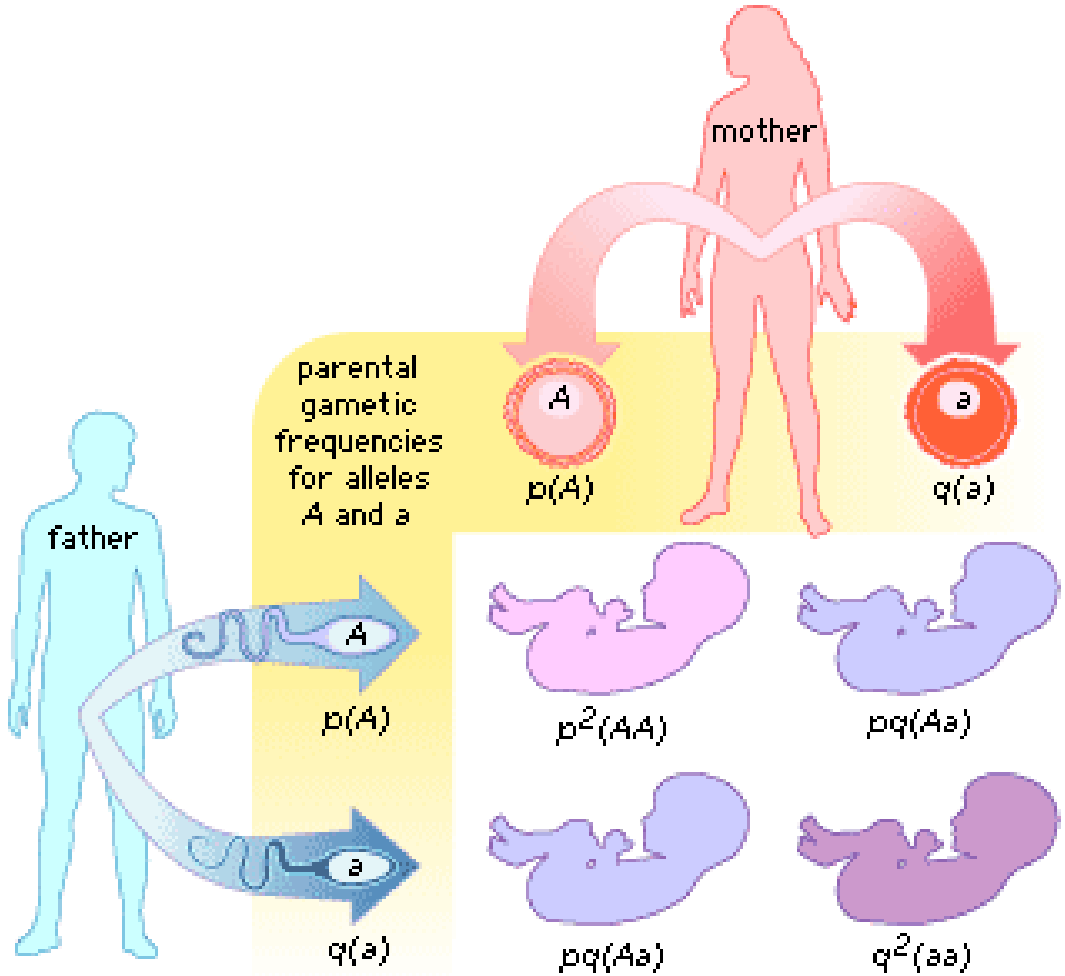


Fig. 13-12-5
Crossing Over

Random Fertilization



Copyright © 2008 Pearson Education, Inc., publishing as Pearson Benjamin Cummings

BIG IDEA III

Living systems store, retrieve, transmit and respond to information essential to life processes.

Enduring Understanding 3.A

Heritable information provides for continuity of life.

Essential Knowledge 3.A.3

The chromosomal basis of inheritance provides an understanding of the pattern of passage (transmission) of genes from parent to offspring.

PowerPoint® Lecture Presentations for

Biology

Eighth Edition

Neil Campbell and Jane Reece

Lectures by Chris Romero, updated by Erin Barley with contributions from Joan Sharp

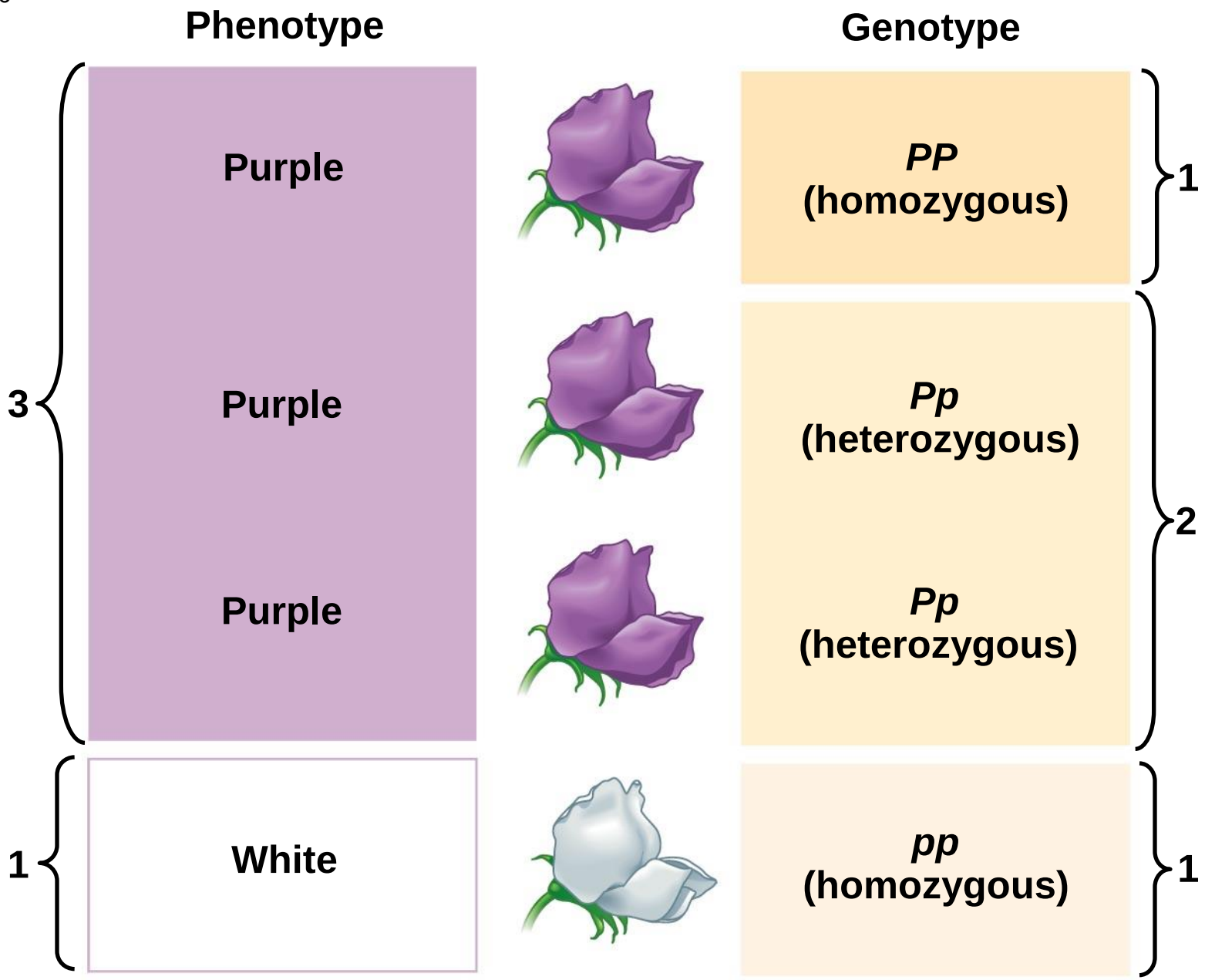
Essential Knowledge 3.A.3: *The chromosomal basis of inheritance provides an understanding of the pattern of passage (transmission) of genes from parent to offspring.*

- **Learning Objectives:**
  - **(3.12)** The student is able to **construct a representation that connects** the process of meiosis to the passage of traits from parent to offspring.
  - **(3.13)** The student is able to **pose questions** about ethical, social or medical issues surrounding human genetic disorders.
  - **(3.14)** The student is able to **apply mathematical routines** to determine Mendelian patterns of inheritance provided by data sets.

Useful Genetics Vocabulary

- An organism with two identical alleles for a character is said to be **homozygous** for the gene controlling that character:
  - Example: TT or tt for tall trait
- An organism that has two different alleles for a gene is said to be **heterozygous** for the gene controlling that character (i.e. a ***carrier***):
  - Example: Tt for tall trait
- Unlike homozygotes, heterozygotes are not true-breeding…they are HYBRIDS!

Fig. 14-6



Rules of probability can be applied to analyze passage of single gene traits from parent to offspring.

- The laws of probability govern Mendelian inheritance:
  - Mendel's laws of segregation and independent assortment reflect the rules of probability.
  - When tossing a coin, the outcome of one 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.

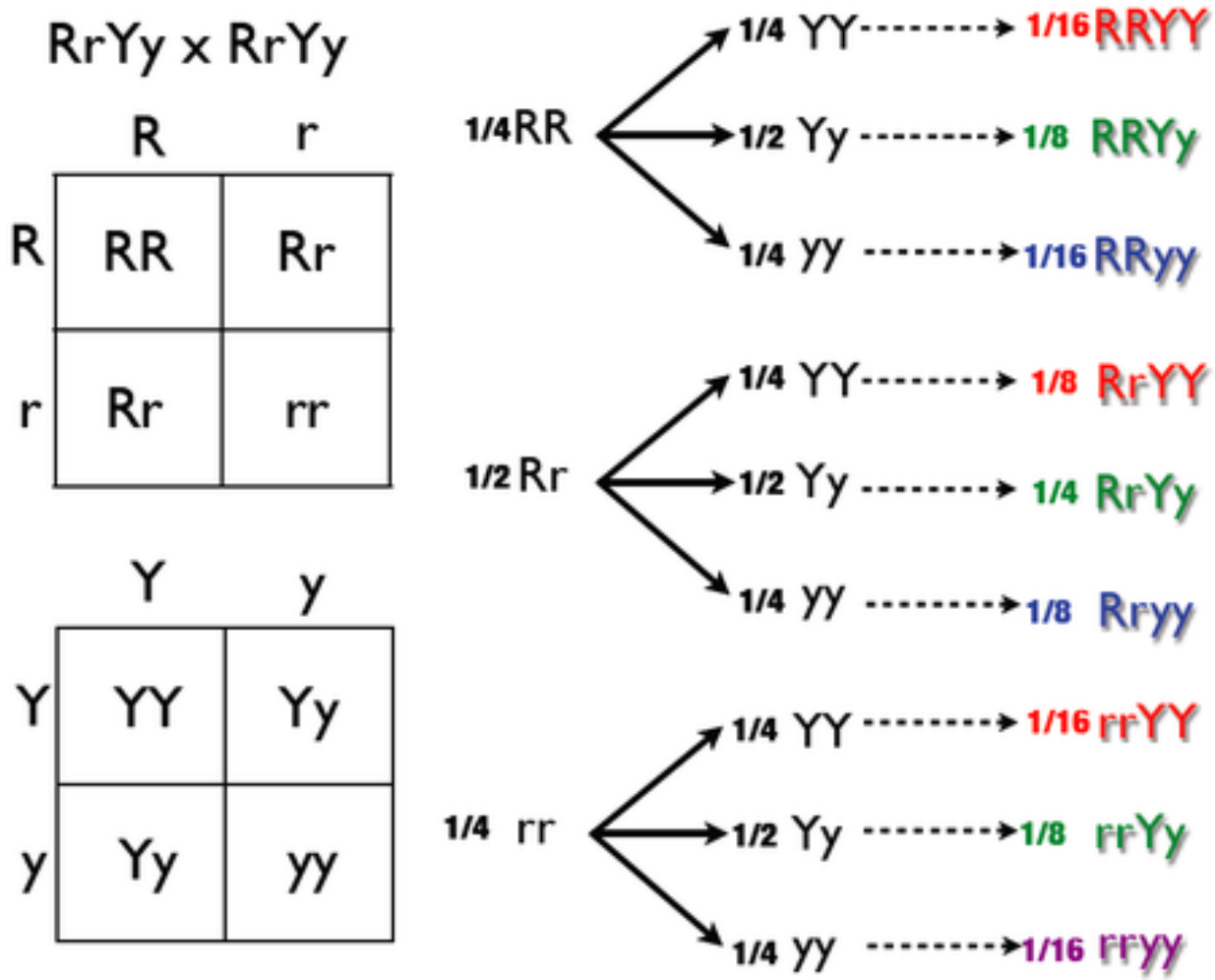
The Multiplication & Addition Rule

- The *multiplication rule*
  - states that the probability that two or more **independent events** will occur together is the **product** of their individual probabilities
- The *rule of addition*
  - states that the probability that **any one of two or more exclusive events** will occur is calculated by **adding together** their individual probabilities

Rule of Multiplication

- When calculating the probability that two or more *independent events* will occur together in a specific combination, *multiply the probabilities of each of the two events*.
- ***Example:*** the probability of a coin landing face up two times in two flips is:
  - ½ x ½ = ¼
- ***Example:*** If you cross two organisms with the genotypes AABbCc and AaBbCc, the probability of an offspring having the genotype AaBbcc is:
  - ½ x ½ x ¼ = 1/16

Solving Complex Genetics Problems with the Rules of Probability



Rule of Addition

- When calculating the probability that any of two or more mutually exclusive events will occur, you need to add together their individual probabilities.
- ***Example:*** if you are tossing a die, what is the probability that it will land on either the side with 4 spots or the side with 5 spots?
  - 1/6 + 1/6 = 1/3

- The multiplication and addition rules can be combined to solve complex genetic problems.

- Let's examine the following cross: *PpYyRr x Ppyyrr*

- What fraction of offspring from this cross would be predicted to exhibit the recessive phenotypes for *at least two* of the three characters?

ppyyRr	¼ (probability of *pp*) × ½ (*yy*) × ½ (*Rr*)	= 1/16
ppYyrr	¼ × ½ × ½	= 1/16
Ppyyrr	½ × ½ × ½	= 2/16
PPyyrr	¼ × ½ × ½	= 1/16
ppyyrr	¼ × ½ × ½	= 1/16
Chance of *at least two* recessive traits		**= 6/16 or 3/8**

Study Tips: Laws of Probability

- What are the chances of event 1 **and** event 2?
  - Multiply
- What are the chances of event 1 **or** event 2?
  - Add

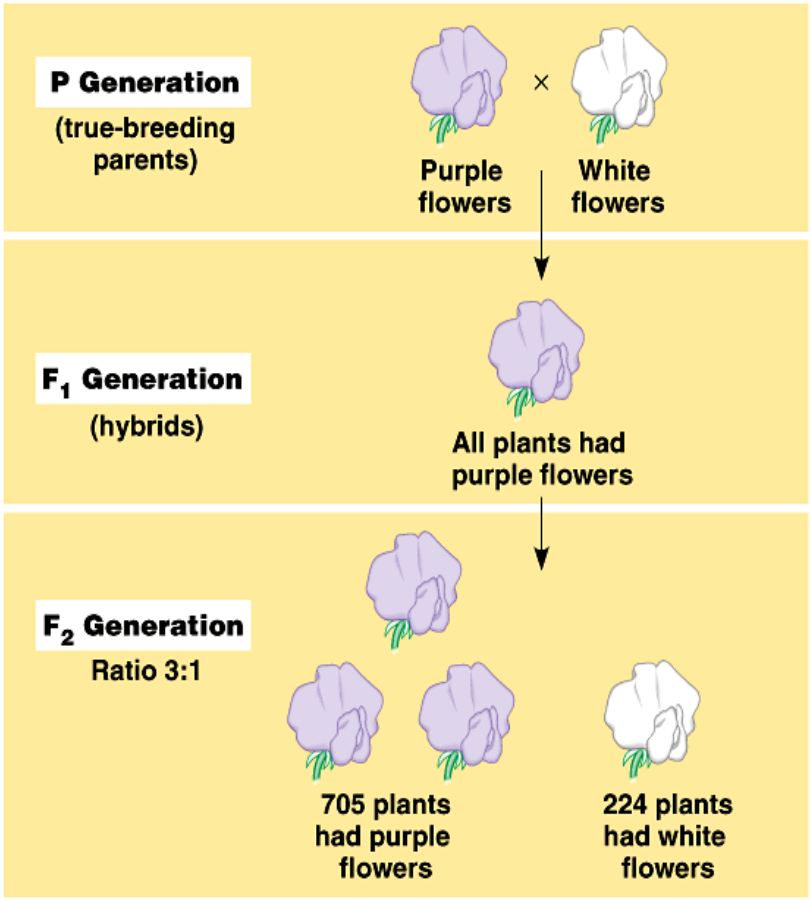
Calculating Gamete Formations

- How many possible gametes can be formed from an individual with the genotype AABBCcDdeeFf?
  - AA = 1
  - BB = 1
  - Cc = 2
  - Dd = 2
  - ee = 1
  - Ff = 2

Mendel's Model: 5 Big Ideas

1. Alternative versions **(different alleles)** of genes account for variations in inherited characters.
2. For each character, an organism inherits two alleles, **one from each parent**.
3. If the two alleles differ, the dominant allele is expressed in the organism's appearance, and the other, a recessive allele is masked.
   - **Law of Dominance**
4. Allele pairs from homologous chromosomes separate during gamete formation (i.e. T separates from t). This separation corresponds to the distribution of homologous chromosomes to different gametes in meiosis.
   - **Law of Segregation**
5. Allele pairs of different genes segregate *independently* during gamete formation - applies when genes for two characteristics are located on *different pairs of homologous chromosomes* (i.e. a person having brown hair does not affect their chance of having brown eyes if these genes are located on DIFFERENT chromosomes).
   - **Principle of Independent Assortment**

Mendel's Method

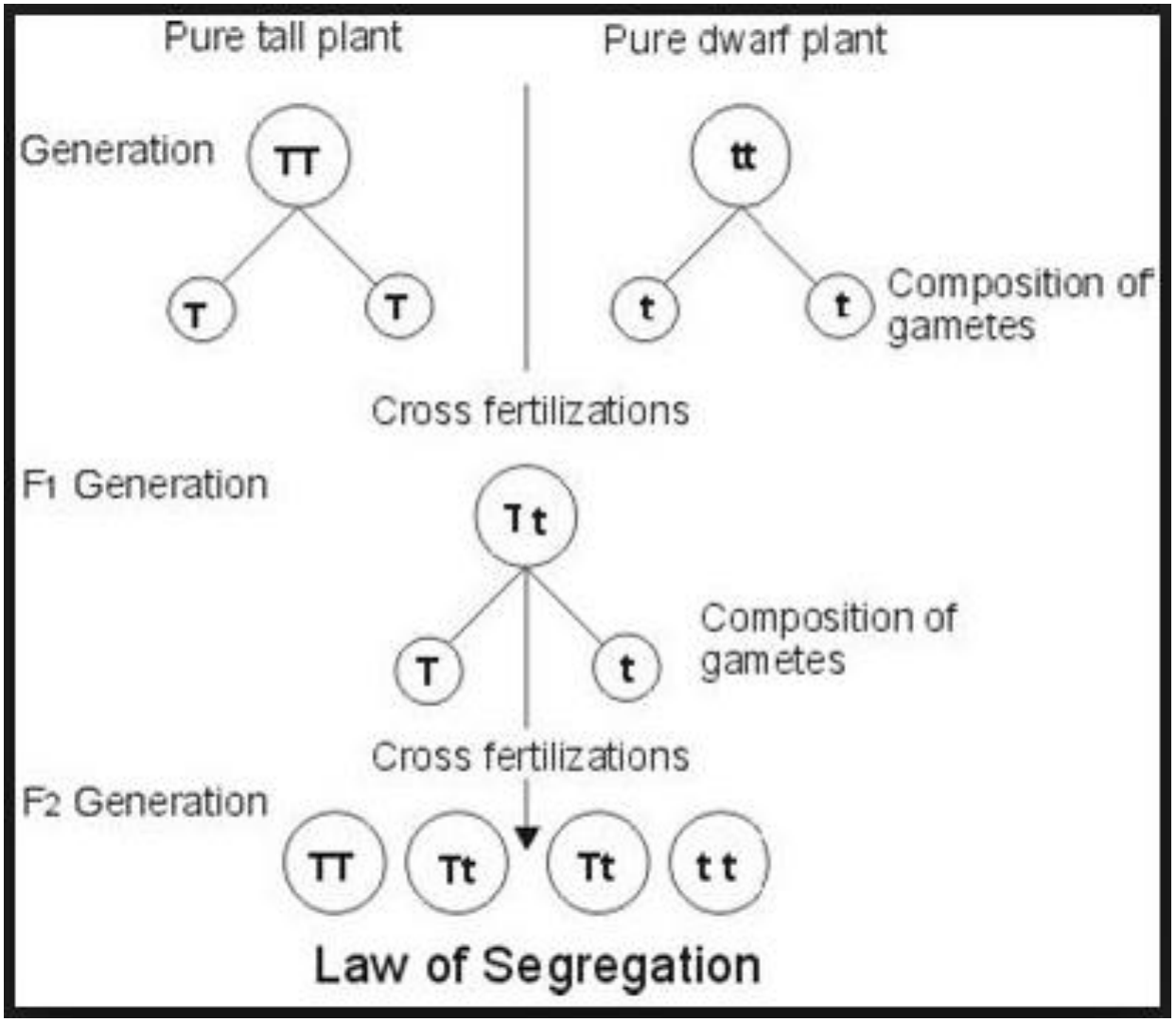


- For his experiments, Mendel chose to CROSS POLLINATE (mate different plants to each other) plants that were TRUE BREEDING (meaning if the plants were allowed to self-pollinate, all their offspring would be of the same variety).
- P generation – parentals; true-breeding parents that were cross-pollinated
- F_1 generation – (first filial) - hybrid offspring of parentals that were allowed to self-pollinate
- F_2 generation – (second filial) - offspring of F_1's

Segregation and independent assortment of chromosomes result in genetic variation.

- Segregation and independent assortment can be applied to genes that are on *different* chromosomes.
  - ***Law of Segregation***: The two alleles for each character separate during gamete production. ***This means that P (purple) will separate from (p) white during meiosis and the production of gametes.***
  - ***Law of Independent Assortment***: Each pair of alleles will separate independently during gamete formation. ***This means that in metaphase I, when homologous chromosomes are lined up on the metaphase plate, they can pair up in any combination – there is a 50% chance that a particular daughter cell will get a maternal or paternal chromosome from the pair.***

Law of Segregation



Law of Independent Assortment

http://www.sumanasinc.com/webcontent/animations/content/independentassortment.html

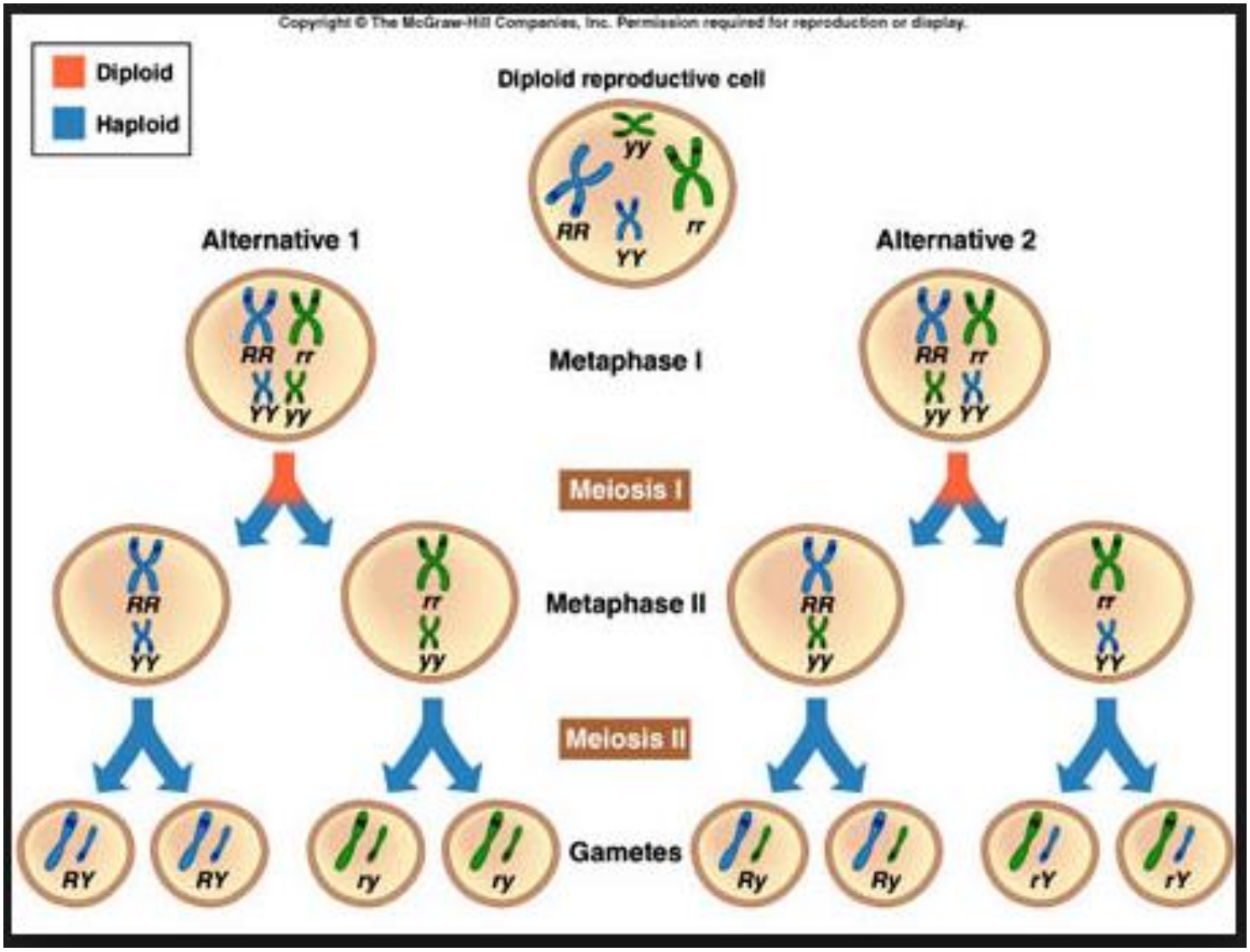
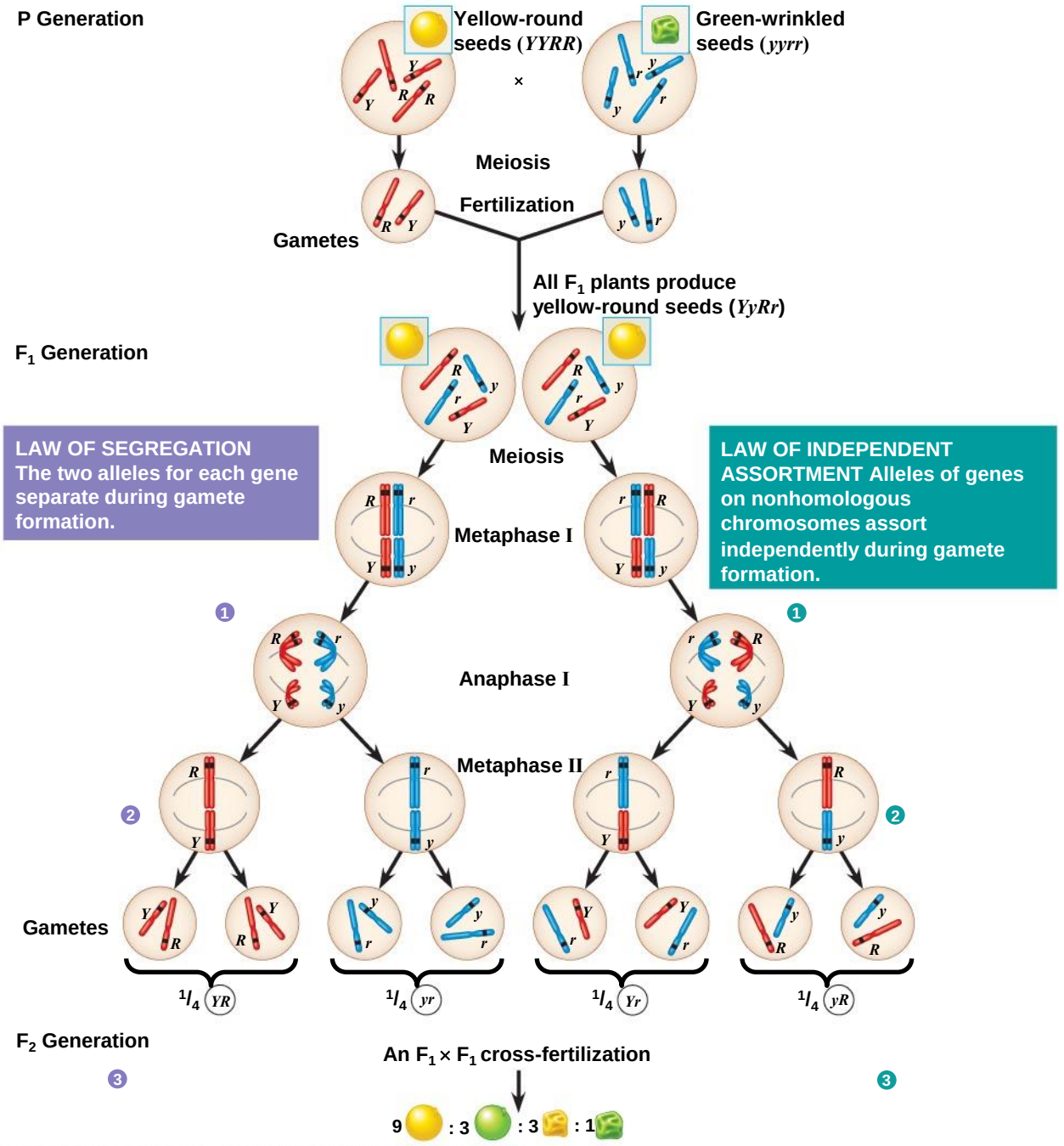


Fig. 15-2

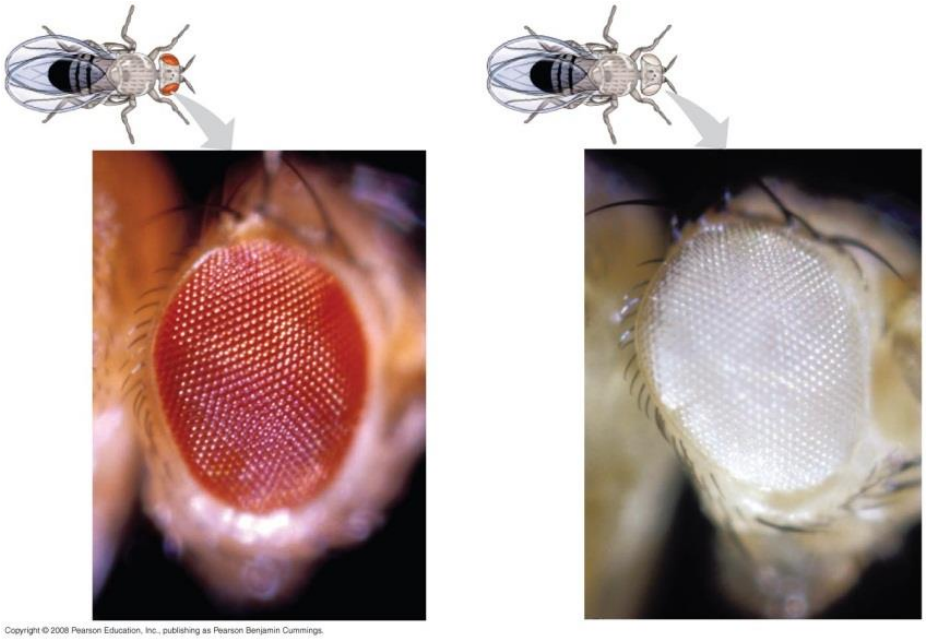


Morgan's Experimental Evidence: Scientific Inquiry

- The first solid evidence associating a specific gene with a specific chromosome came from ***Thomas Hunt Morgan***, an embryologist.
- Morgan's experiments with fruit flies provided convincing evidence that chromosomes are the location of Mendel's heritable factors.
- Several characteristics make fruit flies a convenient organism for genetic studies:
  - They breed at a high rate
  - A generation can be bred every two weeks
  - They have only four pairs of chromosomes

Wild Type v. Mutant

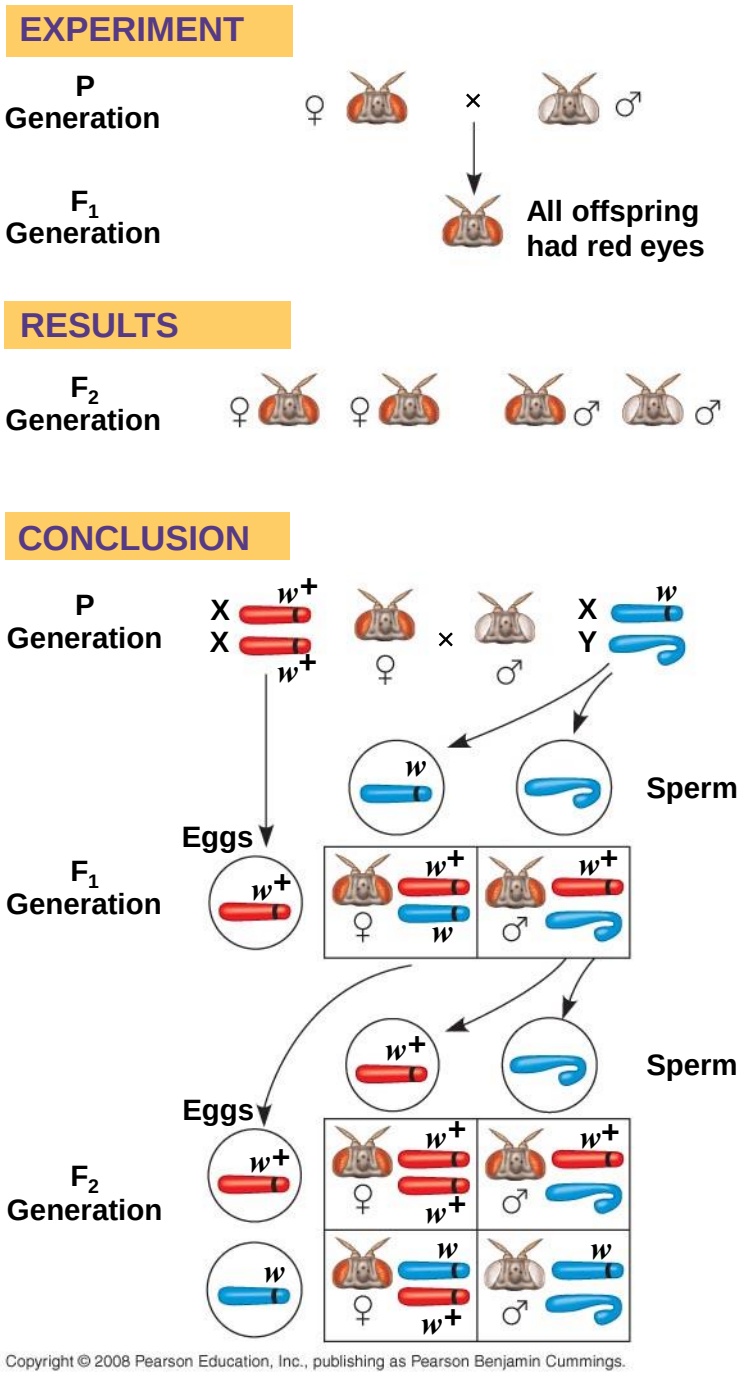
- Morgan noted **wild type**, or normal, phenotypes that were common in the fly populations
- Traits alternative to the wild type are called ***mutant*** *phenotypes*



Correlating Behavior of a Gene's Alleles with Behavior of a Chromosome Pair

- In one experiment, Morgan mated male flies with white eyes (mutant) with female flies with red eyes (wild type):
  - The F_1 generation all had red eyes
  - The F_2 generation showed the 3:1 red:white eye ratio, but only males had white eyes
- Morgan determined that the white-eyed mutant allele must be located on the X chromosome.
- Morgan's finding supported the chromosome theory of inheritance.

Fig. 15-4



Inheritance of Sex-Linked Genes

- The sex chromosomes have genes for many characters unrelated to sex.
- A gene located on either sex chromosome is called a **sex-linked gene.**
- In humans, sex-linked usually refers to a gene on the larger X chromosome.

Inheritance of Sex-Linked Genes

- Sex-linked genes follow specific patterns of inheritance.
- For a recessive sex-linked trait to be expressed:
  - A female needs two copies of the allele
  - A male needs only one copy of the allele
- ***Sex-linked recessive disorders are much more common in males than in females…why?***

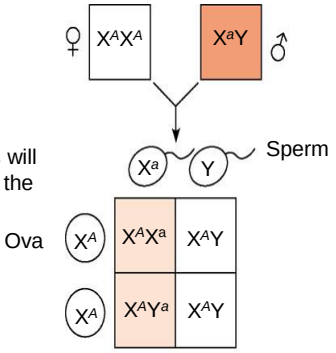
Inheritance of Sex-Linked Genes

- Some disorders caused by recessive alleles on the X chromosome in humans:
  - Color blindness
  - **Duchenne muscular dystrophy**
  - **Hemophilia**

Inheritance of Sex-Linked Genes

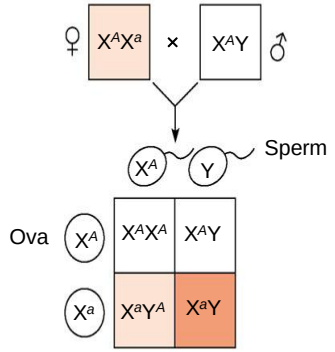
(a)

A father with the disorder will transmit the mutant allele to all daughters but to no sons. When the mother is a dominant homozygote, the daughters will have the normal phenotype but will be carriers of the mutation.



(b)

If a carrier mates with a male of normal phenotype, there is a 50% chance that each daughter will be a carrier like her mother, and a 50% chance that each son will have the disorder.



(c)

If a carrier mates with a male who has the disorder, there is a 50% chance that each child born to them will have the disorder, regardless of sex. Daughters who do not have the disorder will be carriers, where as males without the disorder will be completely free of the recessive allele.

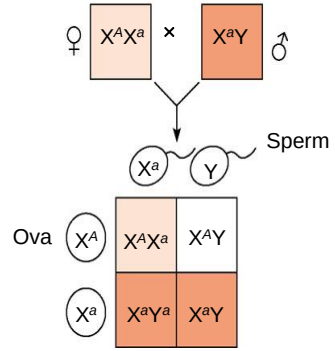
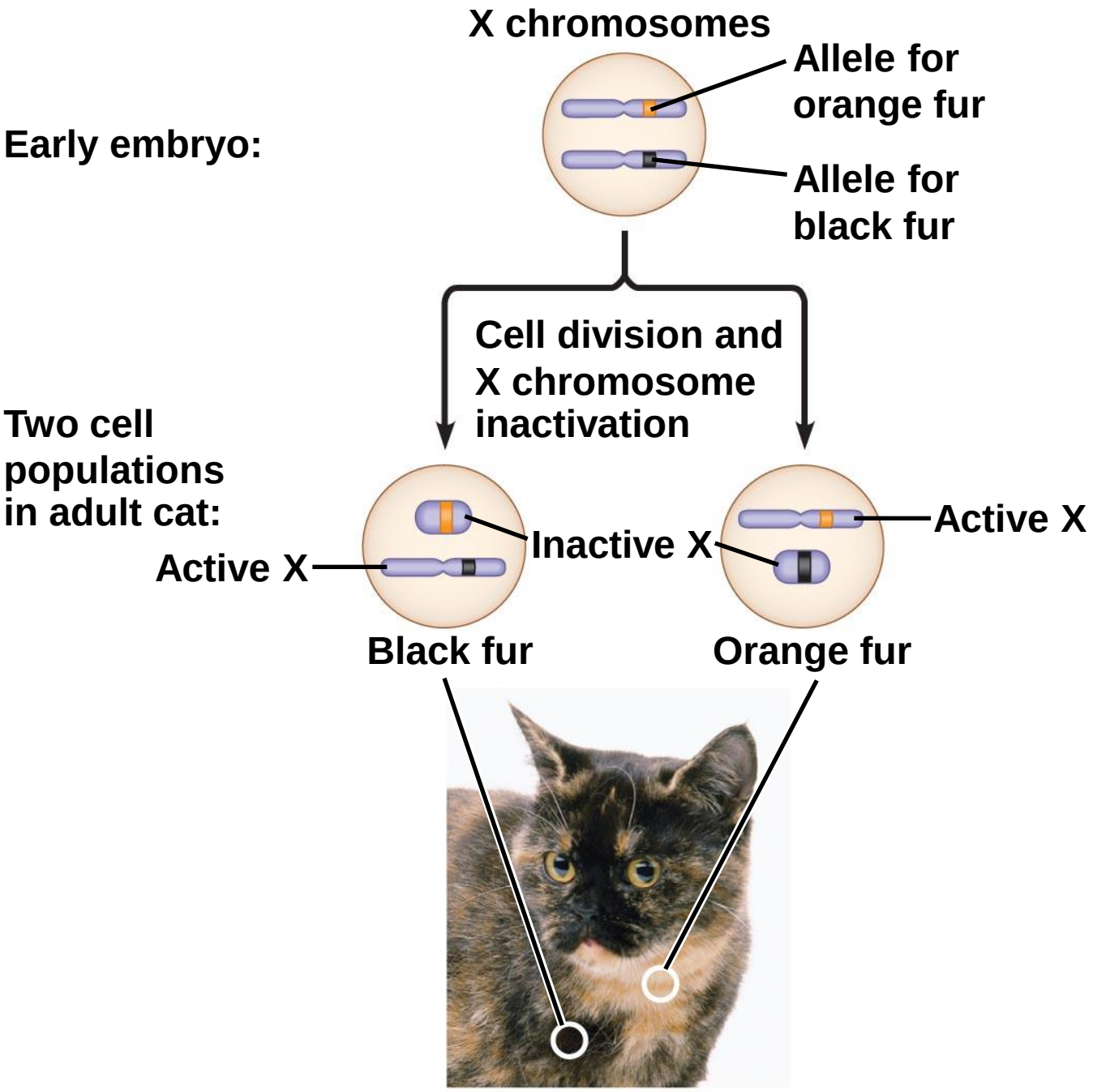


Figure 15.10a–c

X Inactivation in Female Mammals

- In mammalian females, one of the two X chromosomes in each cell is randomly inactivated during embryonic development.
- Otherwise – females would have twice as many proteins produced by DNA on the X chromosome than males!
  - The inactive X condenses into a **Barr body**
  - ***Effectively – males and females have the same "dose" of the X chromosome***
  - In ovaries – the Barr body chromosomes are reactivated in the cells that give rise to eggs – so every female gamete has an active X chromosome.

Fig. 15-8

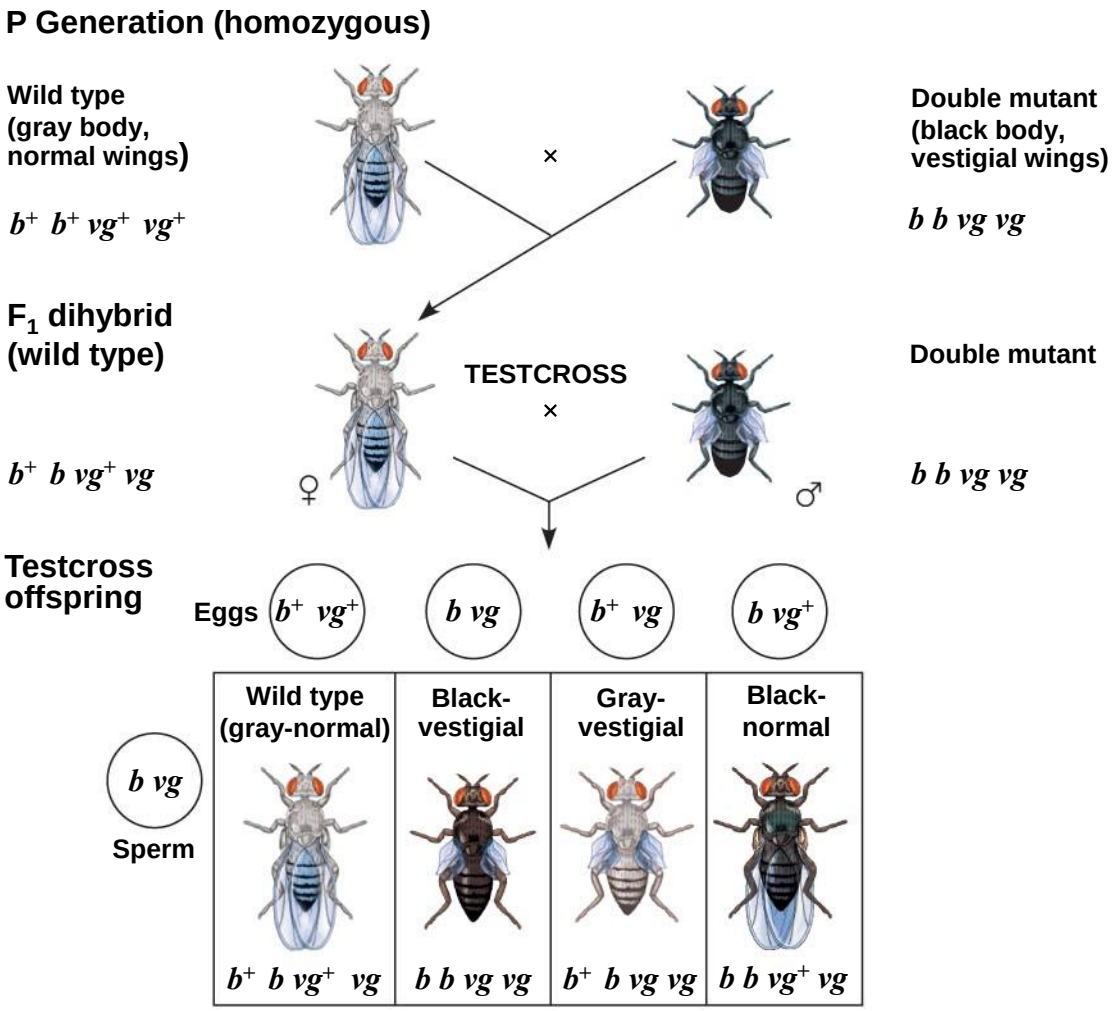


Genes that are adjacent and close to each other on the same chromosome tend to move as a unit.

- The probability that these genes will segregate as a unit is a function of the distance between them.
- Genes located on the same chromosome that tend to be inherited together are called **linked genes.**
  - Linked genes tend to be inherited together because they are located near each other on the same chromosome.
  - Morgan did other experiments with fruit flies to see how linkage affects inheritance of two characters.
  - Morgan crossed flies that differed in traits of body color and wing size.

Fig. 15-9-4

EXPERIMENT



PREDICTED RATIOS

If genes are located on different chromosomes:	1	:	1	:	1	:	1
If genes are located on the same chromosome *and* parental alleles are always inherited together:	1	:	1	:	0	:	0
	965	:	944	:	206	:	185

RESULTS

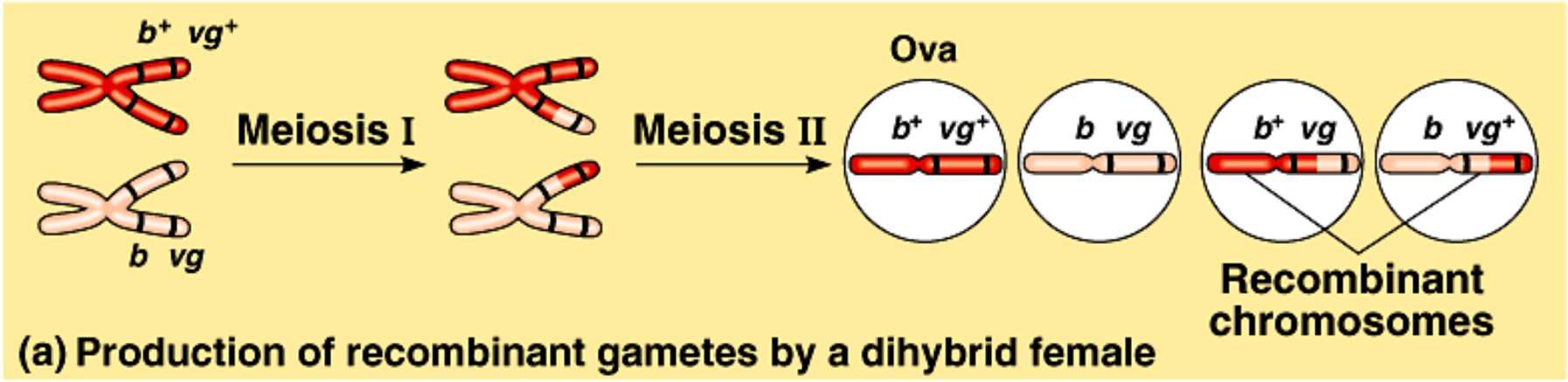
Genetic Recombination and Linkage

- The genetic findings of Mendel and Morgan relate to the chromosomal basis of recombination:
  - Mendel observed that combinations of traits in some offspring differ from either parent.
  - Offspring with a phenotype matching one of the parental phenotypes are called **parental types.**
  - Offspring with non-parental phenotypes (new combinations of traits) are called **recombinant types**, or **recombinants.**
  - A 50% frequency of recombination is observed for any two genes on different chromosomes (i.e. NOT LINKED) – 50% of offspring look like either parent; 50% look like a recombinant because of crossing over.

Recombination of Linked Genes: Crossing Over

- Morgan discovered that genes can be linked, but the linkage was incomplete, as evident from recombinant phenotypes.
  - Morgan proposed that some process must sometimes break the physical connection between genes on the same chromosome.
  - That mechanism was the **crossing over** of homologous chromosomes.

Figure 15.5a Recombination due to crossing over



In crossing over during prophase of meiosis I, chromatids of paired homologous chromosomes break, and homologous chromatid fragments switch places…crossing over. This creates recombinant chromosomes.

NOTICE the parental types and the recombinants created during Meiosis I in the diagram above.

Fig. 15-10

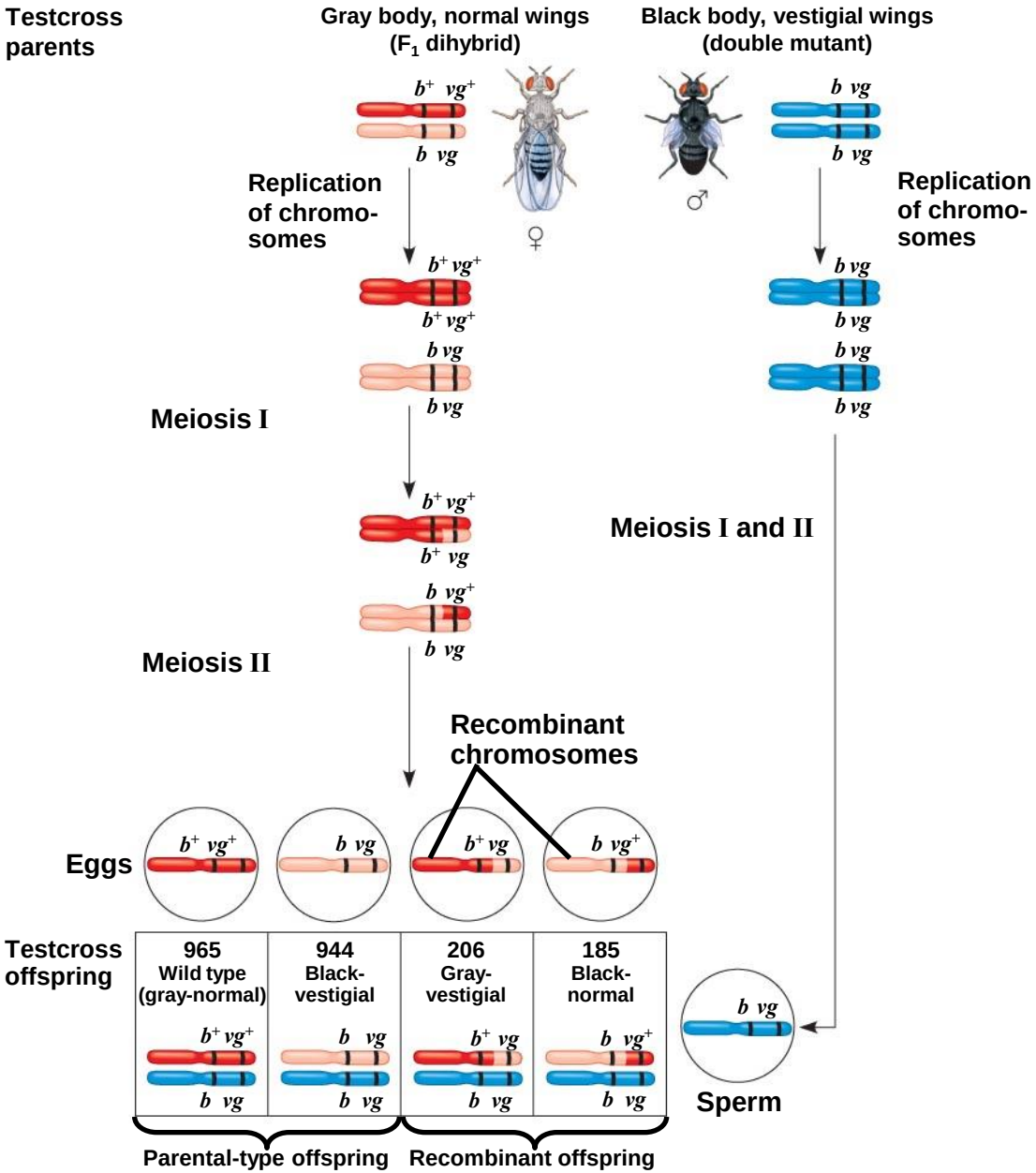
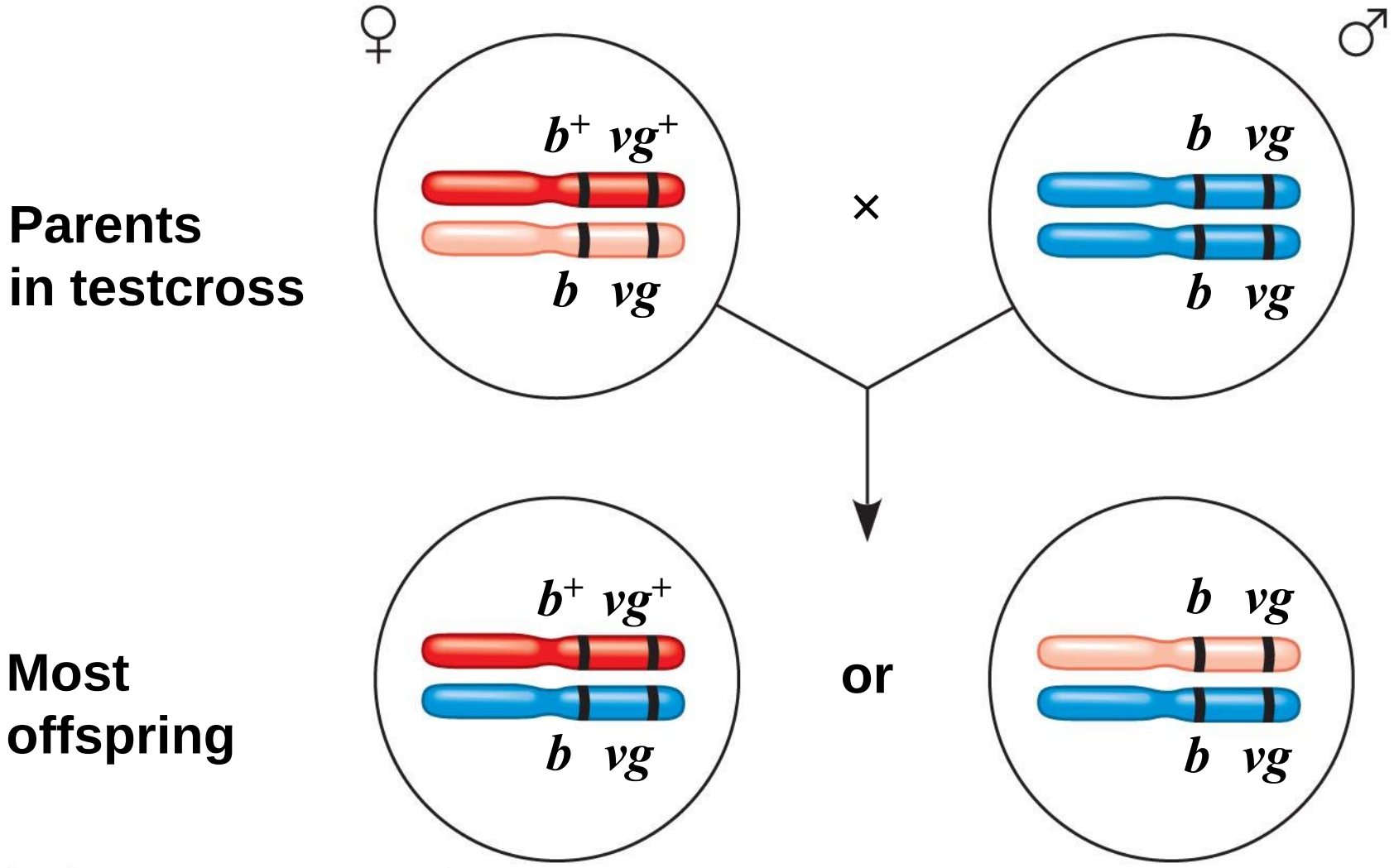


Fig. 15-UN1



Mapping the Distance Between Genes Using Recombination Data: Scientific Inquiry

- Alfred Sturtevant, one of Morgan's students, constructed a **genetic map**, an ordered list of the genetic loci along a particular chromosome.
- Sturtevant predicted that ***the farther apart two genes are, the higher the probability that a crossover will occur between them and therefore the higher the recombination frequency.***

Linkage Maps

- A **linkage map** is a genetic map of a chromosome based on recombination frequencies.
  - Distances between genes can be expressed as **map units**; one map unit, or centimorgan, represents a 1% recombination frequency.
  - Map units indicate relative distance and order, not precise locations of genes.

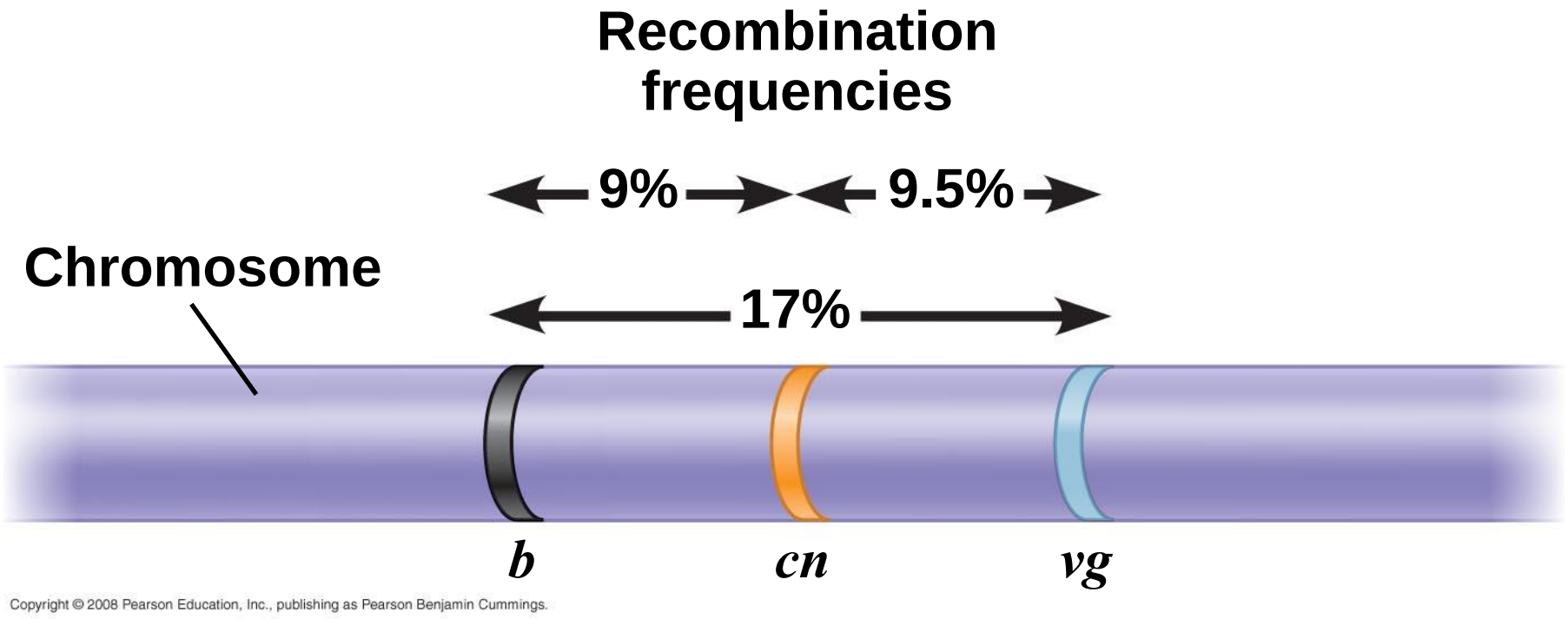
Fig. 15-11

b **= body color**

cn **= cinnabar eyes (brighter red)**

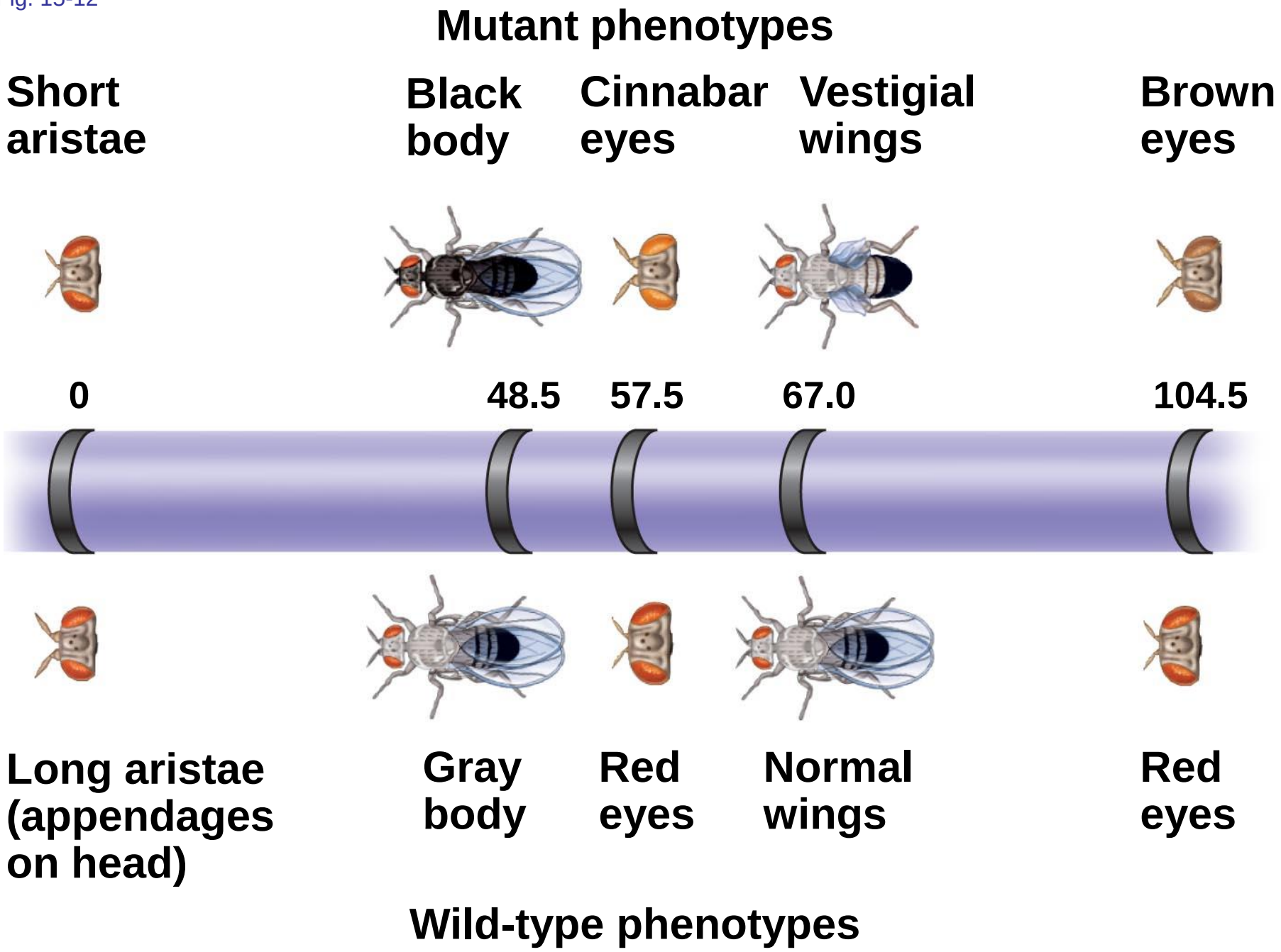
vg **= wing size**

RESULTS



The probability of a crossover between two genetic loci is proportional to the distance separating the loci.

Fig. 15-12



Constructing a Linkage Map

- Determine the sequence of genes along a chromosome based on the following recombination frequencies:
  - A-B = 8%
  - A-C = 28%
  - A-D = 25%
  - B-C = 20%
  - B-D = 33%

Certain human genetic disorders can be attributed to the inheritance of single gene traits or specific chromosomal changes, such as nondisjunction.

- ***Sickle Cell Anemia*** is caused by a point mutation (substitution) that codes for a mutant hemoglobin molecule – codominant disorder.
- ***Tay Sachs disease*** is caused by an allele that codes for a dysfunctional enzyme, which is unable to break down certain lipids in the brain. Lipids accumulate in brain cells – leads to death – recessive disorder.
- **Huntington's disease** is caused by a lethal dominant allele, leads to degeneration of the nervous system around age 40 – dominant disorder.
- **X-linked colorblindness** caused by a recessive allele located on x chromosome – more common in males than females.
- **Down Syndrome/Trisomy 21** is caused by nondisjuction of the 21st chromosome during meiosis – child inherits 3 copies of chromosome #21 – chromosomal disorder.

Many ethical, social and medical issues surround human genetic disorders.

- Reproduction issues – should those with genetic disorders reproduce? Why or why not?
- Civic issues – such as ownership of genetic information, privacy, historical contexts, etc.

BIG IDEA III

Living systems store, retrieve, transmit and respond to information essential to life processes.

Enduring Understanding 3.A

Heritable information provides for continuity of life.

Essential Knowledge 3.A.4

The inheritance pattern of many traits cannot be explained by simple Mendelian genetics.

PowerPoint® Lecture Presentations for

Biology

Eighth Edition

Neil Campbell and Jane Reece

Lectures by Chris Romero, updated by Erin Barley with contributions from Joan Sharp

Essential Knowledge 3.A.4: *The inheritance pattern of many traits cannot be explained by simple Mendelian genetics.*

- **Learning Objectives:**
  - **(3.15)** The student is able to **explain** deviations from Mendel's model of the inheritance of traits.
  - **(3.16)** The student is able to **explain** how the inheritance patterns of many traits cannot be accounted for by Mendelian genetics.
  - **(3.17)** The student is able to **describe representations of an appropriate example** of inheritance patterns that cannot be explained by Mendel's model of the inheritance of traits.

Many traits are the product of multiple genes and/or physiological processes.

- Patterns of inheritance of many traits DO NOT follow ratios predicted by Mendel's laws and can be identified by quantitative analysis, where observed phenotypic ratios statistically differ from the predicted ratios.
- This means that you must be able to:
  - Predict the ***expected ratios*** of a genetic cross.
  - Determine the ***observed ratios*** of a genetic cross.
  - Perform a ***Chi-square analysis*** to determine the probable mode of inheritance of the trait(s) being studied.

Some traits are determined by genes on sex chromosomes.

- Sex-linked genes reside on sex chromosomes (X in humans).
- In mammals and flies, the Y chromosome is very small and carries few genes.
- In mammals and flies, females are XX and males are XY; as such, X-linked recessive traits are always expressed in males (BE SURE YOU CAN EXPLAIN WHY AND SUPPORT YOUR EXPLANATION WITH A PUNNETT SQUARE)!
- Some traits are sex limited, and expression depends on the sex of the individual, such as milk production in female mammals and pattern baldness in males.

Some trains result from nonnuclear inheritance.

- Some inheritance patterns are exceptions to the standard chromosome theory.
- For example, some inheritance patterns result from NONNUCLEAR inheritance (i.e. genes located OUTSIDE the nucleus).
  - Chloroplast DNA
  - Maternal mitochondrial DNA

Inheritance of Organelle Genes

- Chloroplasts and mitochondria are randomly assorted to gametes and daughter cells; thus, traits determined by chloroplast and mitochondrial DNA do not follow simple Mendelian rules.
- In animals, mitochondrial DNA is transmitted by the **egg** and NOT by the sperm; as such, mitochondrial-determined traits are maternally inherited.

Evidence of Maternal Mitochondrial Inheritance in Humans

- The products of mitochondrial genes help to make up the protein complexes of the ETC and ATP synthase in animal cells.
- Defects in one or more of these proteins have been shown to cause a number of rare human disorders.
- Disorders of this nature are MATERNALLY inherited – through the maternal mitochondria.
- Because the parts of the body most susceptible to energy deprivation are the nervous system and the muscles, most mitochondrial diseases primarily affect these systems.
  - *Mitochondrial myopathy* causes weakness, intolerance of exercise, and muscle deterioration.

Determining Inheritance Patterns Using Pedigrees

- A pedigree is a family tree describing the transmission of traits from parents to children across generations.