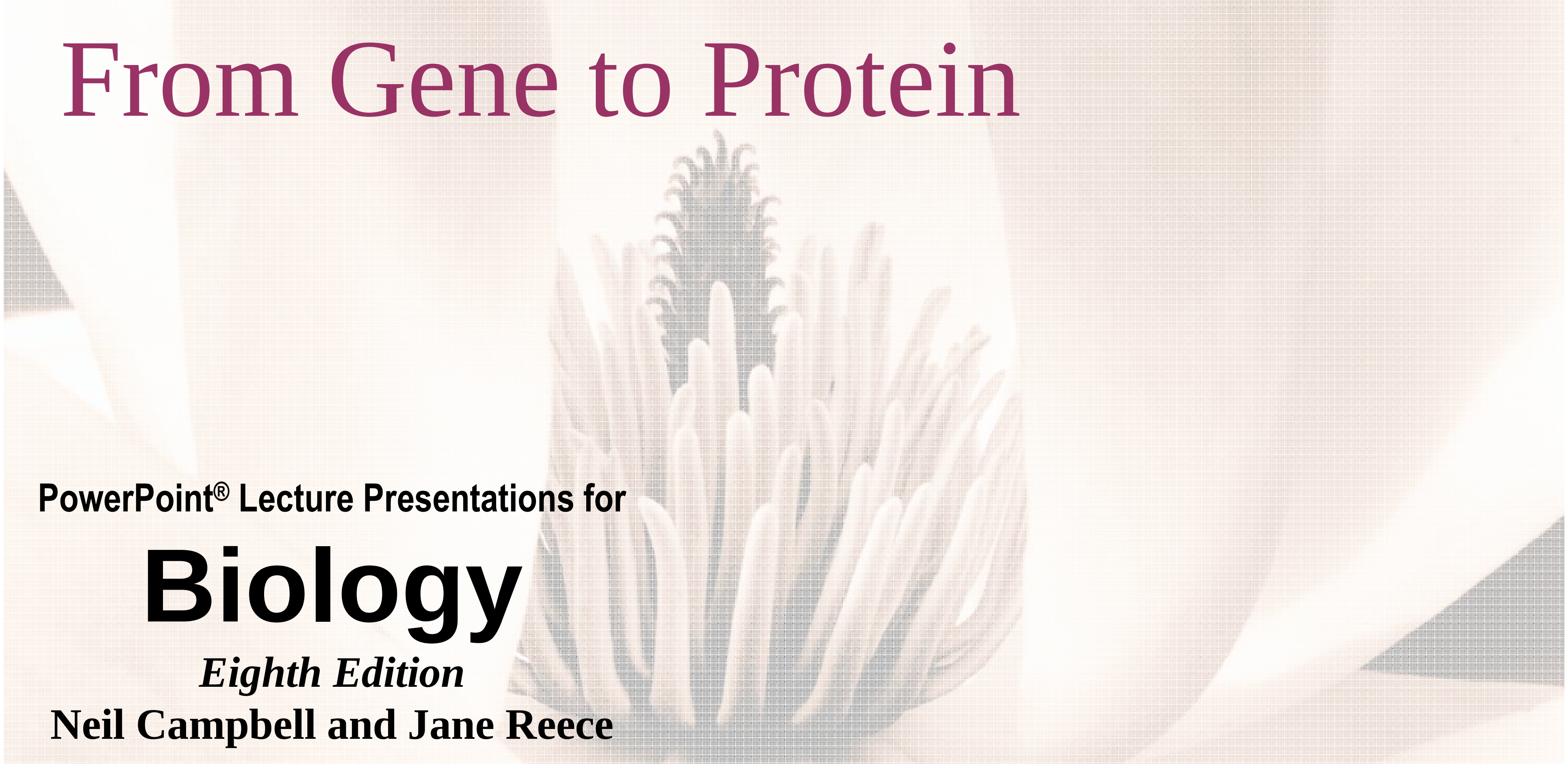
Chapter 17

From Gene to Protein



PowerPoint® Lecture Presentations for

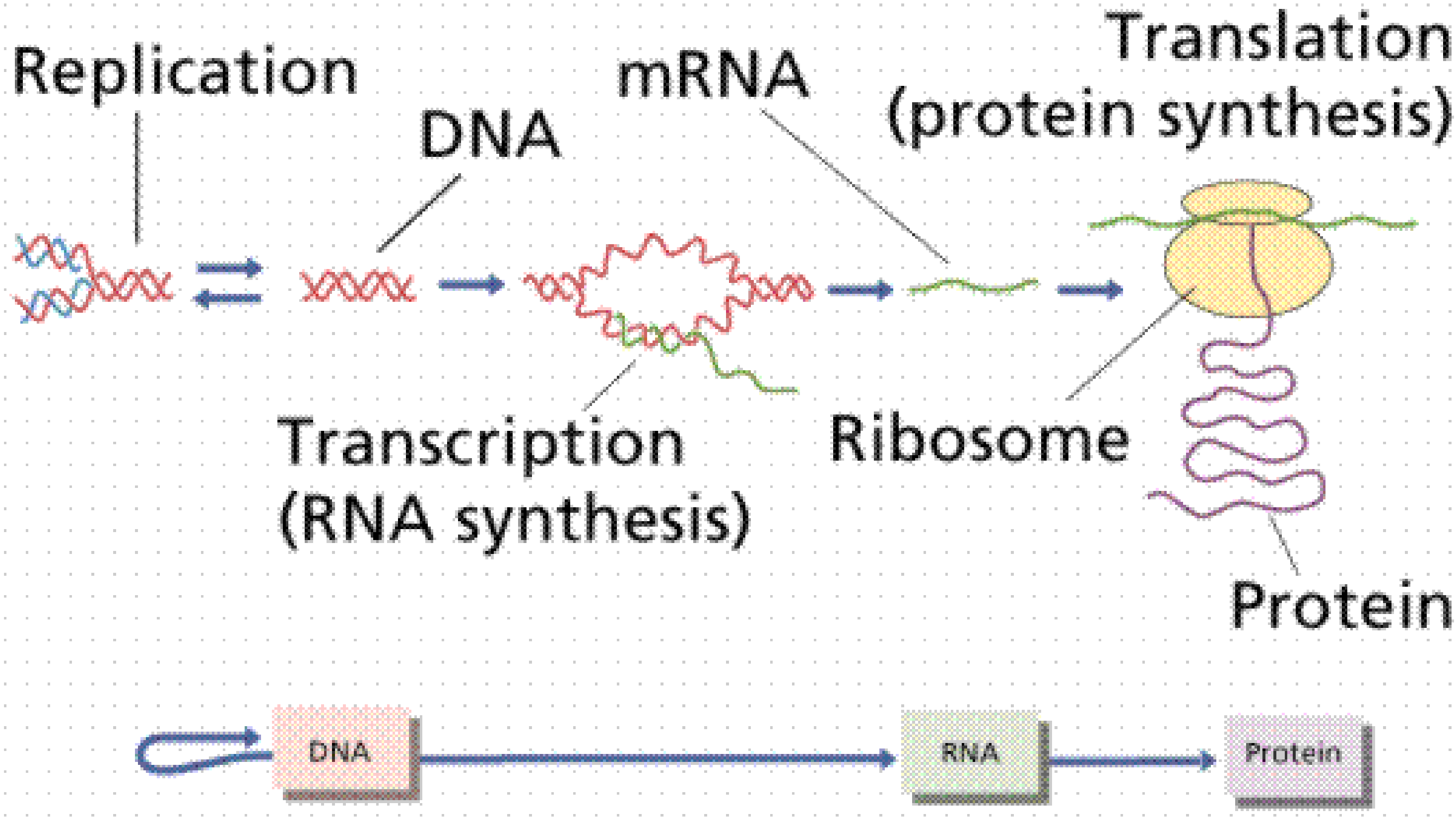
Biology

Eighth Edition

Neil Campbell and Jane Reece

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

Central Dogma of Life

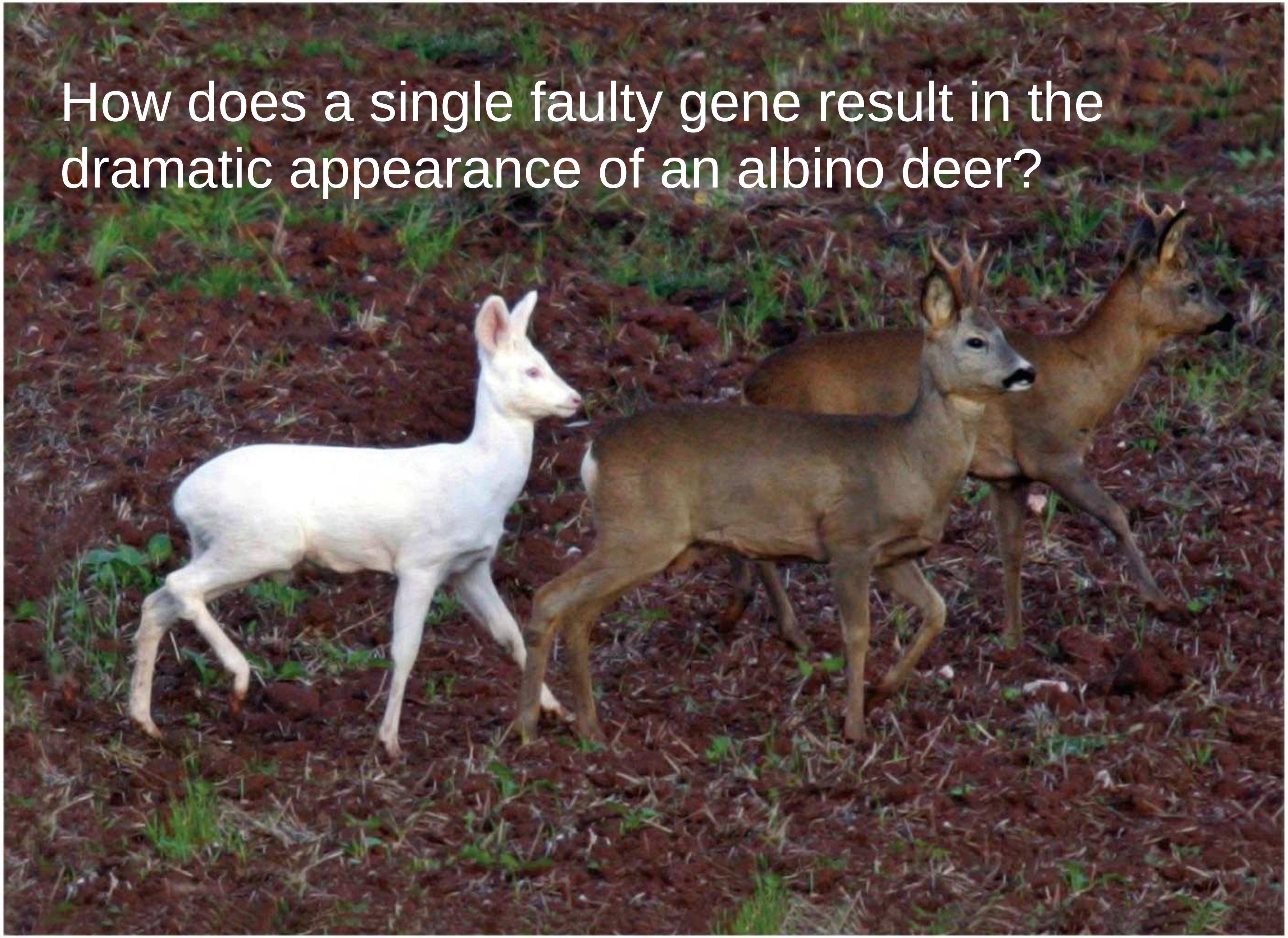


http://www.emc.maricopa.edu/faculty/farabee/BIOBK/BioBookPROTSYn.html

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
- Proteins are the links between genotype and phenotype
- **Gene expression**, the process by which DNA directs protein synthesis, includes two stages: transcription and translation

Fig. 17-1



How does a single faulty gene result in the dramatic appearance of an albino deer?

Concept 17.1: Genes specify proteins via transcription and translation

- How was the fundamental relationship between genes and proteins discovered?

Evidence from the Study of Metabolic Defects

- In 1909, British physician Archibald Garrod first suggested that **genes** dictate phenotypes through **enzymes** that catalyze specific chemical reactions
- He thought symptoms of an inherited disease reflect an inability to synthesize a certain enzyme
- Linking genes to enzymes required understanding that cells synthesize and degrade molecules in a series of steps, a metabolic pathway

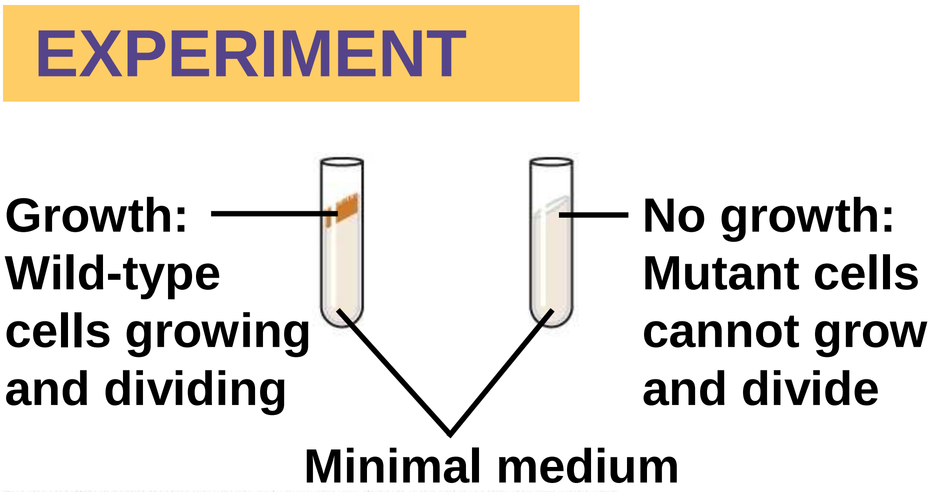
Nutritional Mutants in Neurospora: Scientific Inquiry

- George Beadle and Edward Tatum exposed **bread mold** to X-rays, creating mutants that were unable to survive on minimal medium as a result of inability to synthesize certain molecules

- Using crosses, they identified three classes of arginine-deficient mutants, each lacking a different enzyme necessary for synthesizing arginine

- They developed a *one gene–one enzyme hypothesis*, which states that each gene dictates production of a specific enzyme

Fig. 17-2a



EXPERIMENT

Growth:
Wild-type cells growing and dividing

No growth:
Mutant cells cannot grow and divide

Minimal medium

Fig. 17-2b

RESULTS

Classes of *Neurospora crassa*

Wild type

Class I mutants

Class II mutants

Class III mutants

Condition

Minimal medium (MM) (control)

MM + ornithine

MM + citrulline

MM + arginine (control)

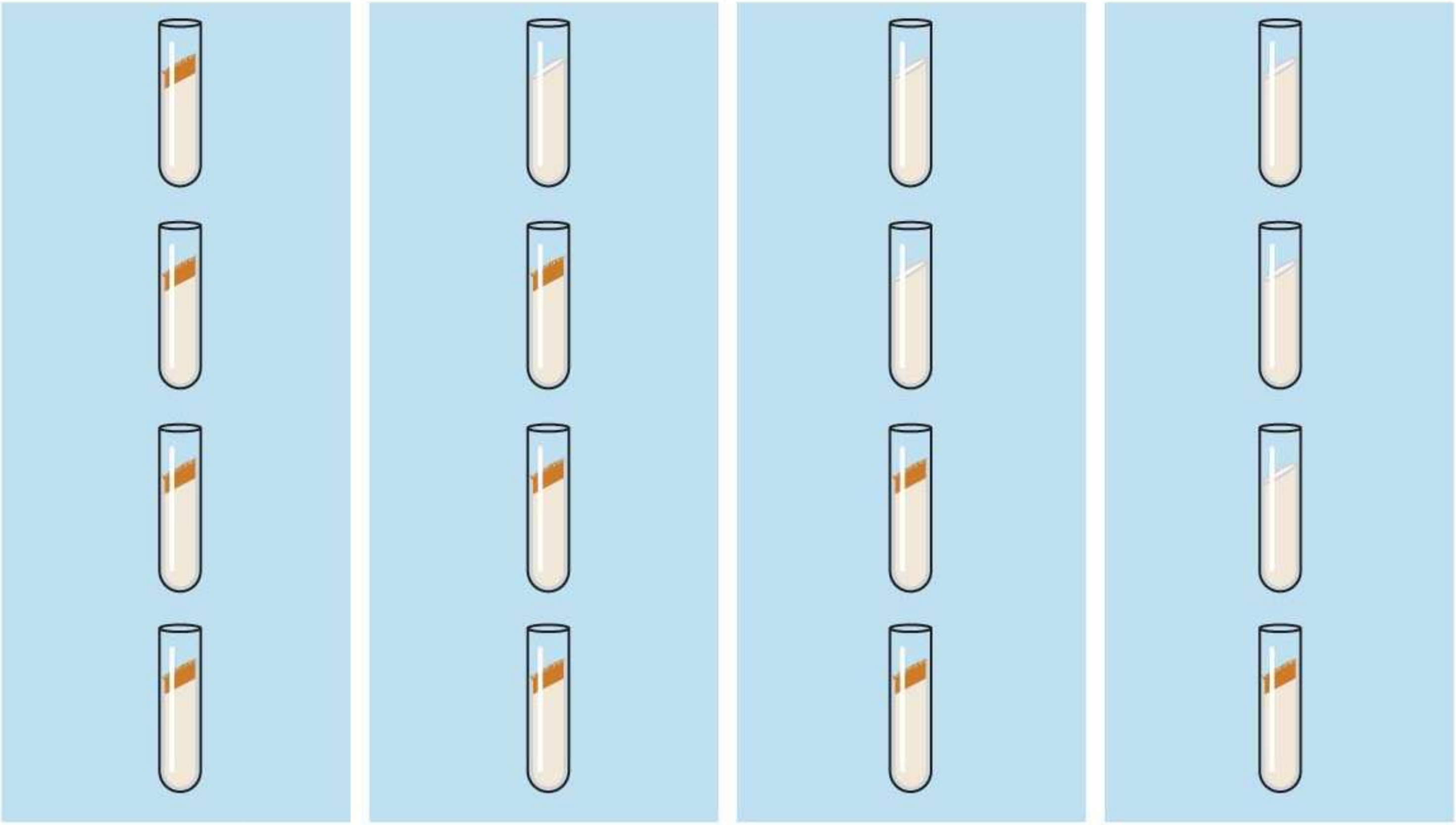
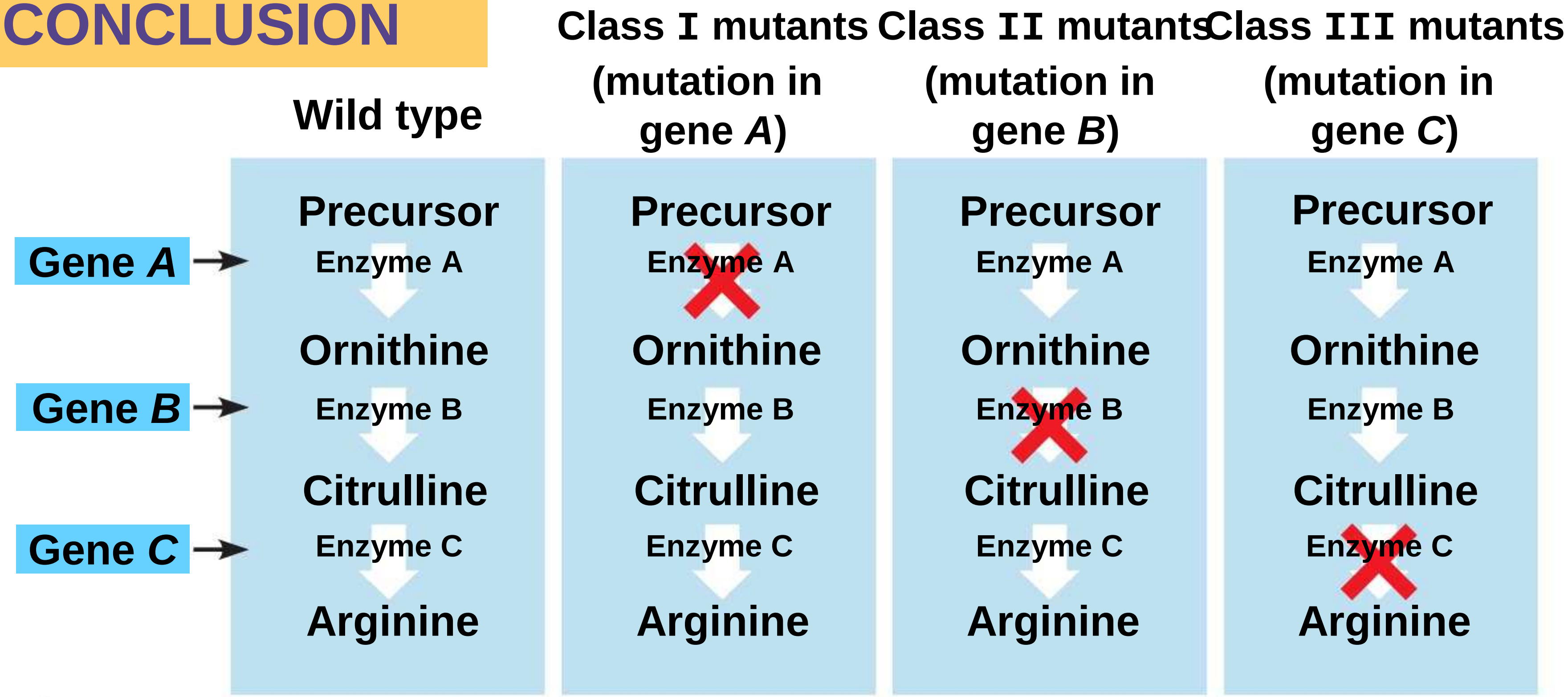


Fig. 17-2c

CONCLUSION



Wild type

Class I mutants (mutation in gene *A*)

Class II mutants (mutation in gene *B*)

Class III mutants (mutation in gene *C*)

Gene *A*

Gene *B*

Gene *C*

Precursor

Enzyme A

Ornithine

Enzyme B

Citrulline

Enzyme C

Arginine

The Products of Gene Expression: A Developing Story

- Some proteins aren’t enzymes, so researchers later revised the hypothesis: *one gene–one protein*
- Many proteins are composed of several polypeptides, each of which has its own gene
- Therefore, Beadle and Tatum’s hypothesis is now restated as the *one gene–one polypeptide hypothesis*
- Note that it is common to refer to gene products as proteins rather than polypeptides

Basic Principles of Transcription and Translation

- **RNA** is the intermediate between genes and the proteins for which they code
- **Transcription** is the synthesis of RNA under the direction of DNA
- Transcription produces **messenger RNA (mRNA)**
- **Translation** is the synthesis of a polypeptide, which occurs under the direction of mRNA
- **Ribosomes** are the sites of translation

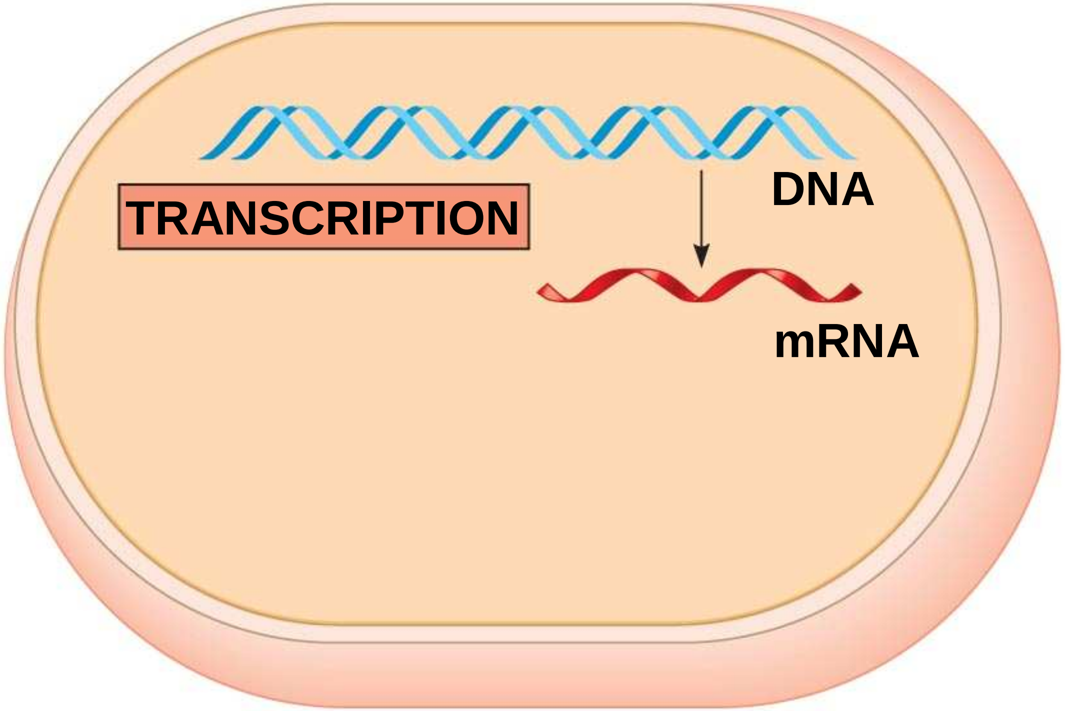
- In prokaryotes, mRNA produced by transcription is immediately translated without more processing
- In a eukaryotic cell, the nuclear envelope separates transcription from translation
- Eukaryotic RNA transcripts are modified through **RNA processing** to yield finished mRNA

- A **primary transcript** is the initial RNA transcript from any gene
- The central dogma is the concept that cells are governed by a cellular chain of command:

DNA → RNA → protein

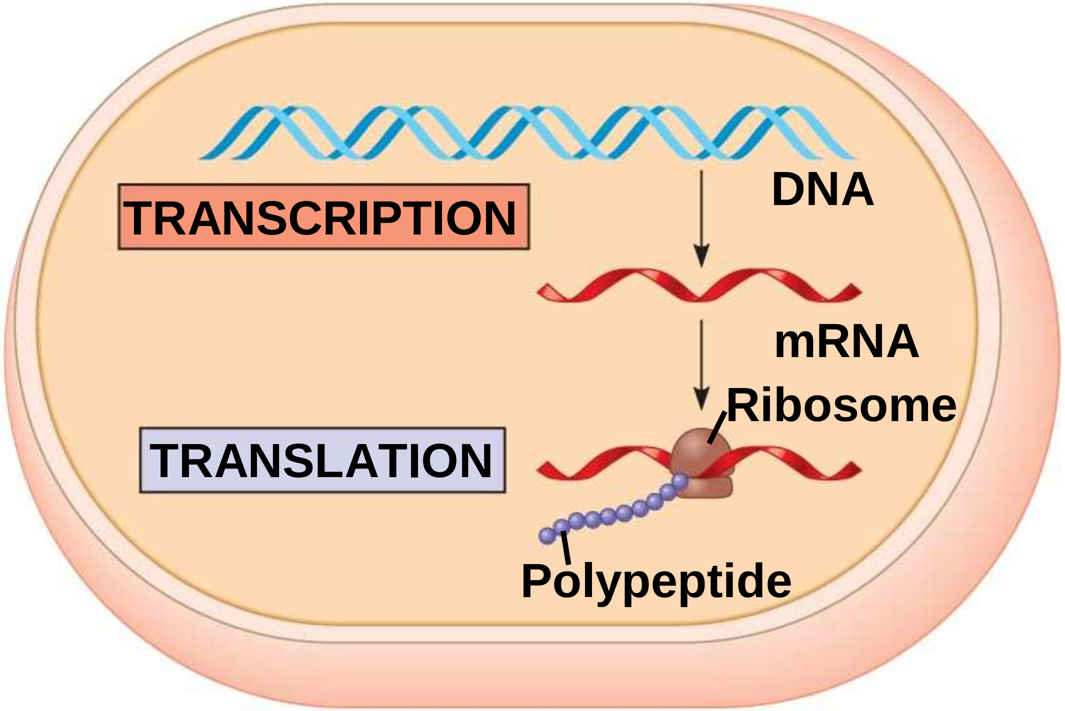
Fig. 17-3a-1

Overview: the roles of transcription and translation in the flow of genetic information

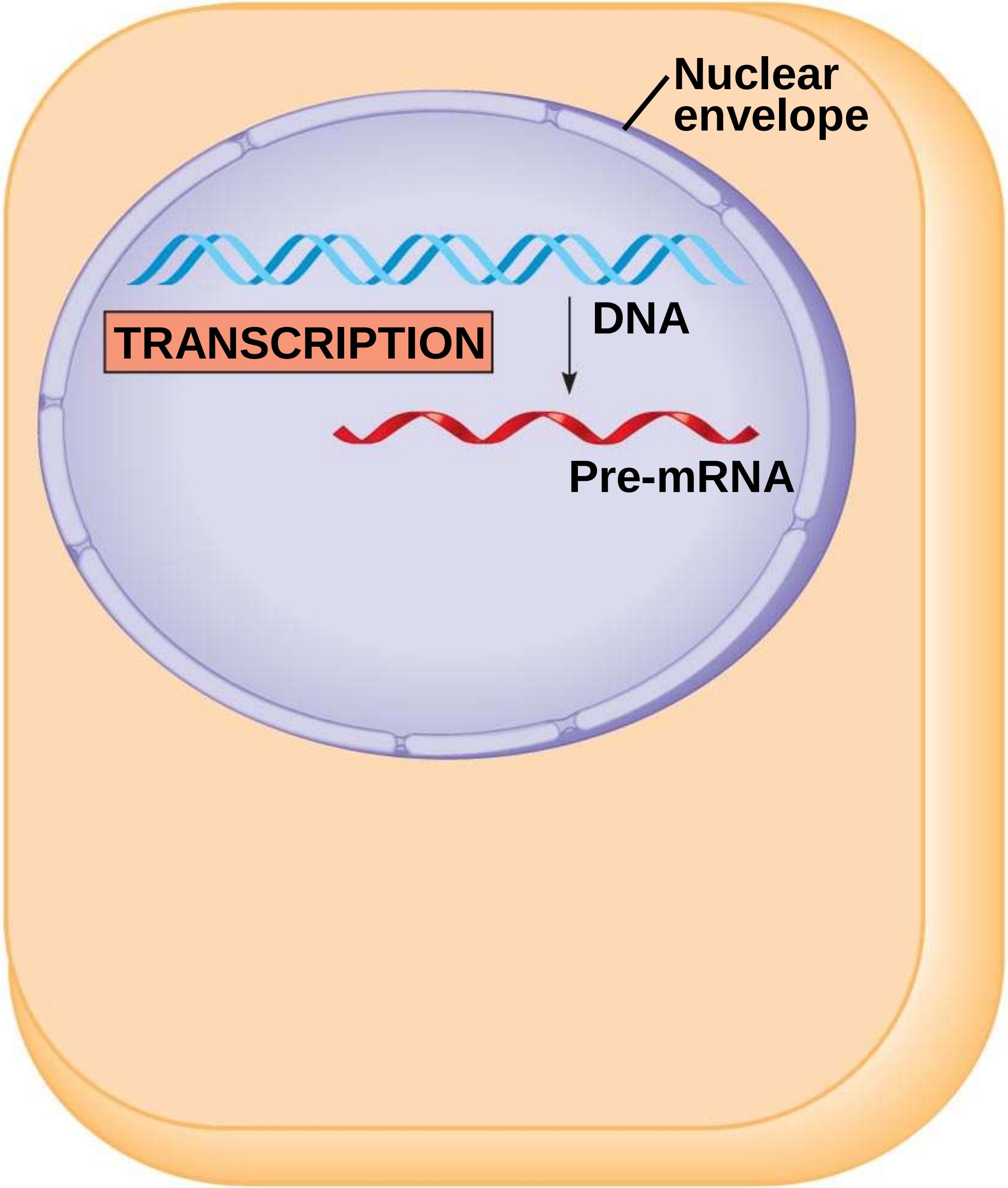


(a) Bacterial cell

Fig. 17-3a-2

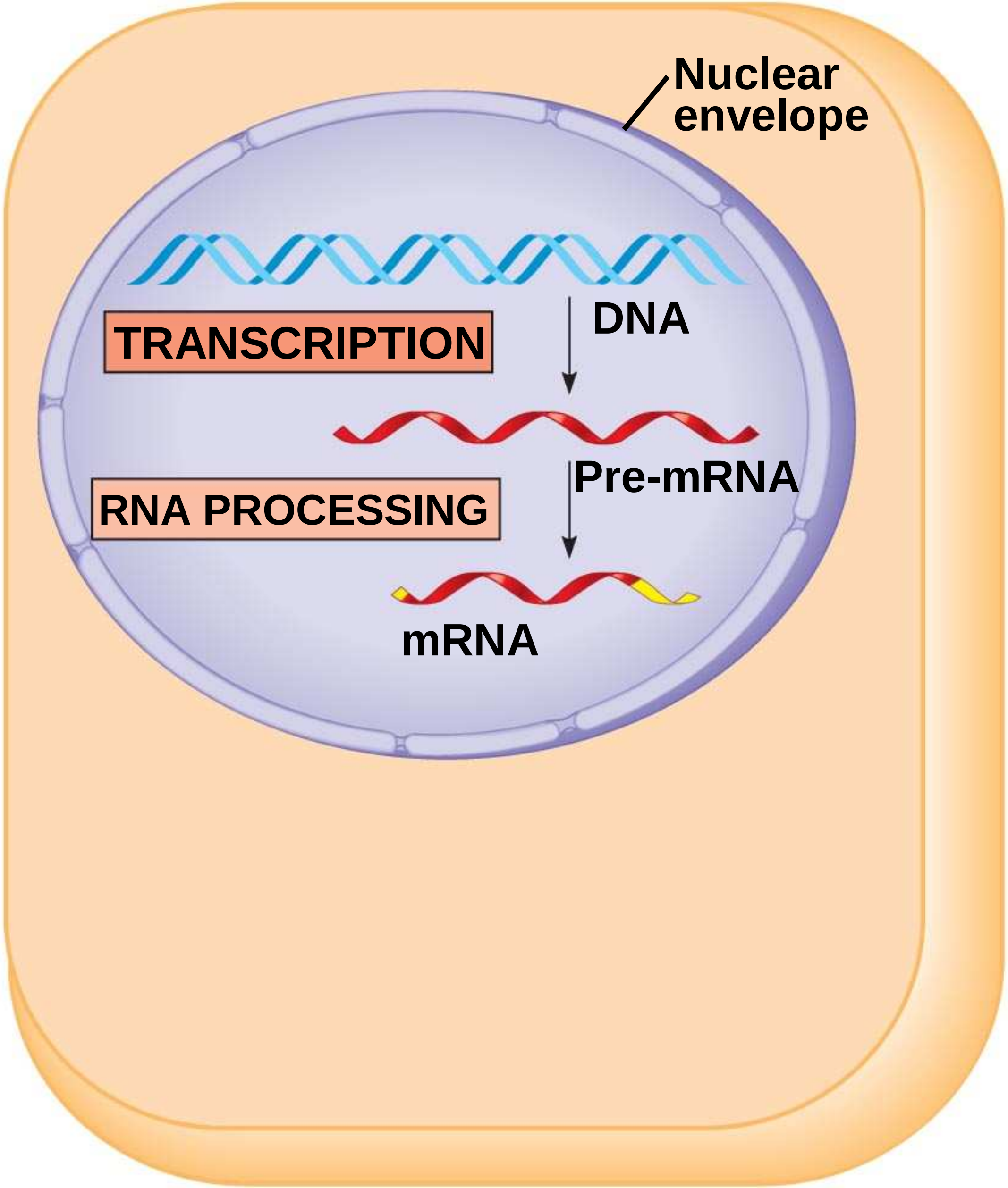


(a) Bacterial cell



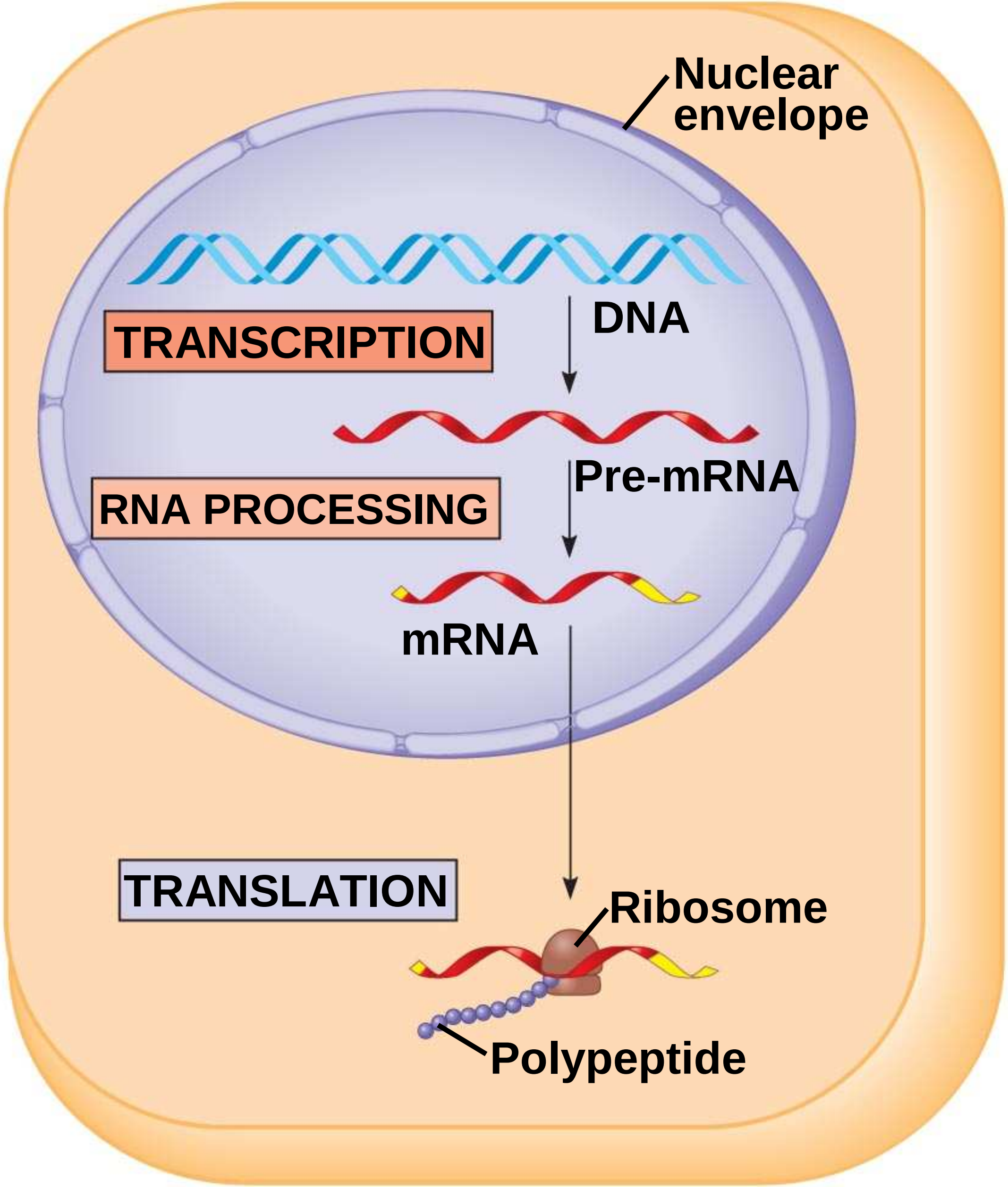
(b) Eukaryotic cell

Fig. 17-3b-2



(b) Eukaryotic cell

Fig. 17-3b-3



(b) Eukaryotic cell

The Genetic Code

- How are the instructions for assembling amino acids into proteins encoded into DNA?
- There are 20 amino acids, but there are only four nucleotide bases in DNA
- How many bases correspond to an amino acid?

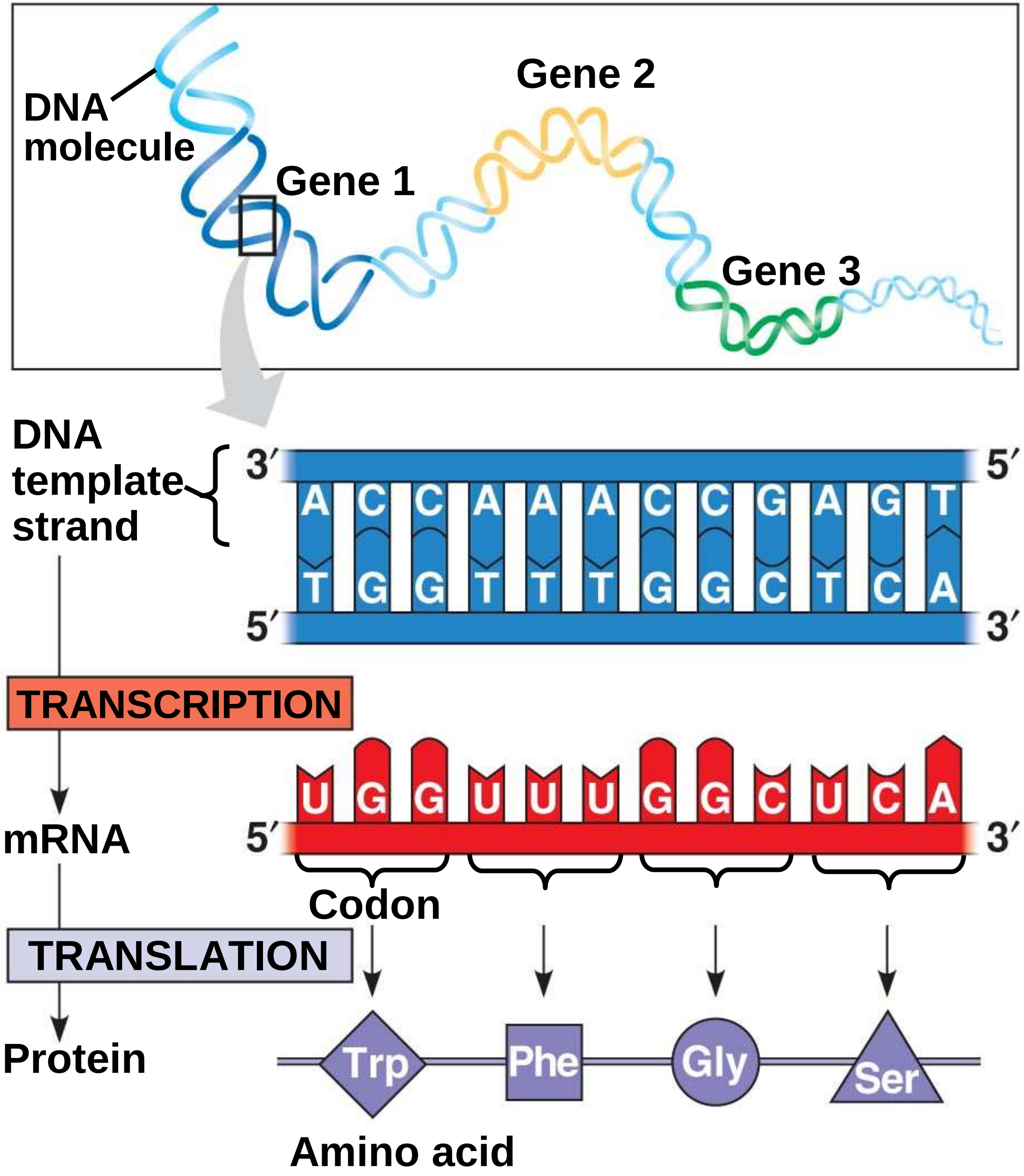
Codons: Triplets of Bases

- 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

- During transcription, one of the two DNA strands called the **template strand** provides a template for ordering the sequence of nucleotides in an RNA transcript
- During translation, the mRNA base triplets, called **codons**, are read in the 5′ to 3′ direction
- Each codon specifies the amino acid to be placed at the corresponding position along a polypeptide

- Codons along an mRNA molecule are read by translation machinery in **the 5′ to 3′ direction**
- Each codon specifies the addition of one of 20 amino acids

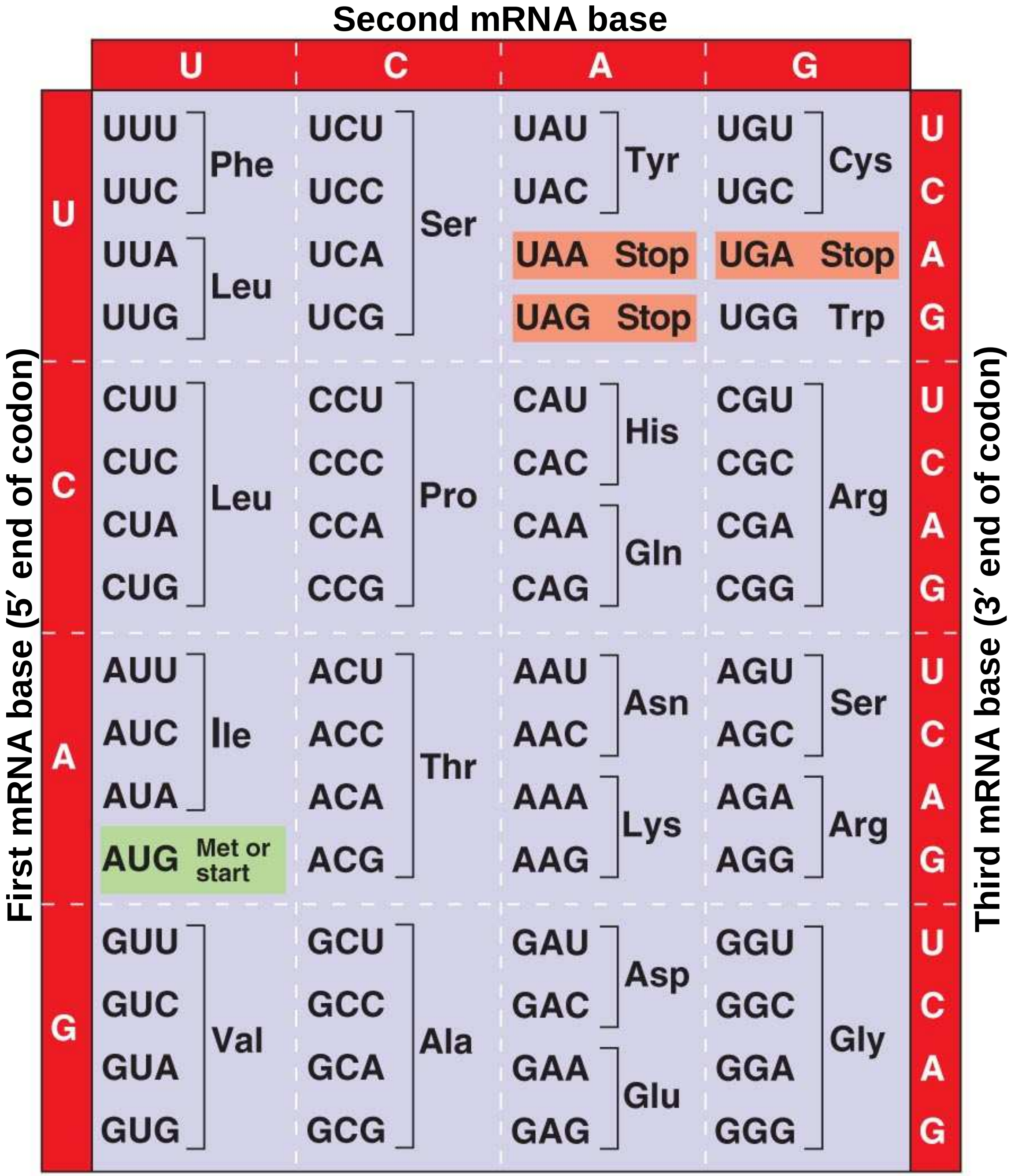
Fig. 17-4


DNA molecule
Gene 1
Gene 2
Gene 3
DNA template strand
3′
5′
A C C A A A C C G A G T
T G G T T T G G C T C A
5′
3′
TRANSCRIPTION
mRNA
5′
U G G U U U G G C U C A
3′
Codon
TRANSLATION
Protein
Trp
Phe
Gly
Ser
Amino acid

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
- Codons must be read in the correct **reading frame** (correct groupings) in order for the specified polypeptide to be produced

Fig. 17-5



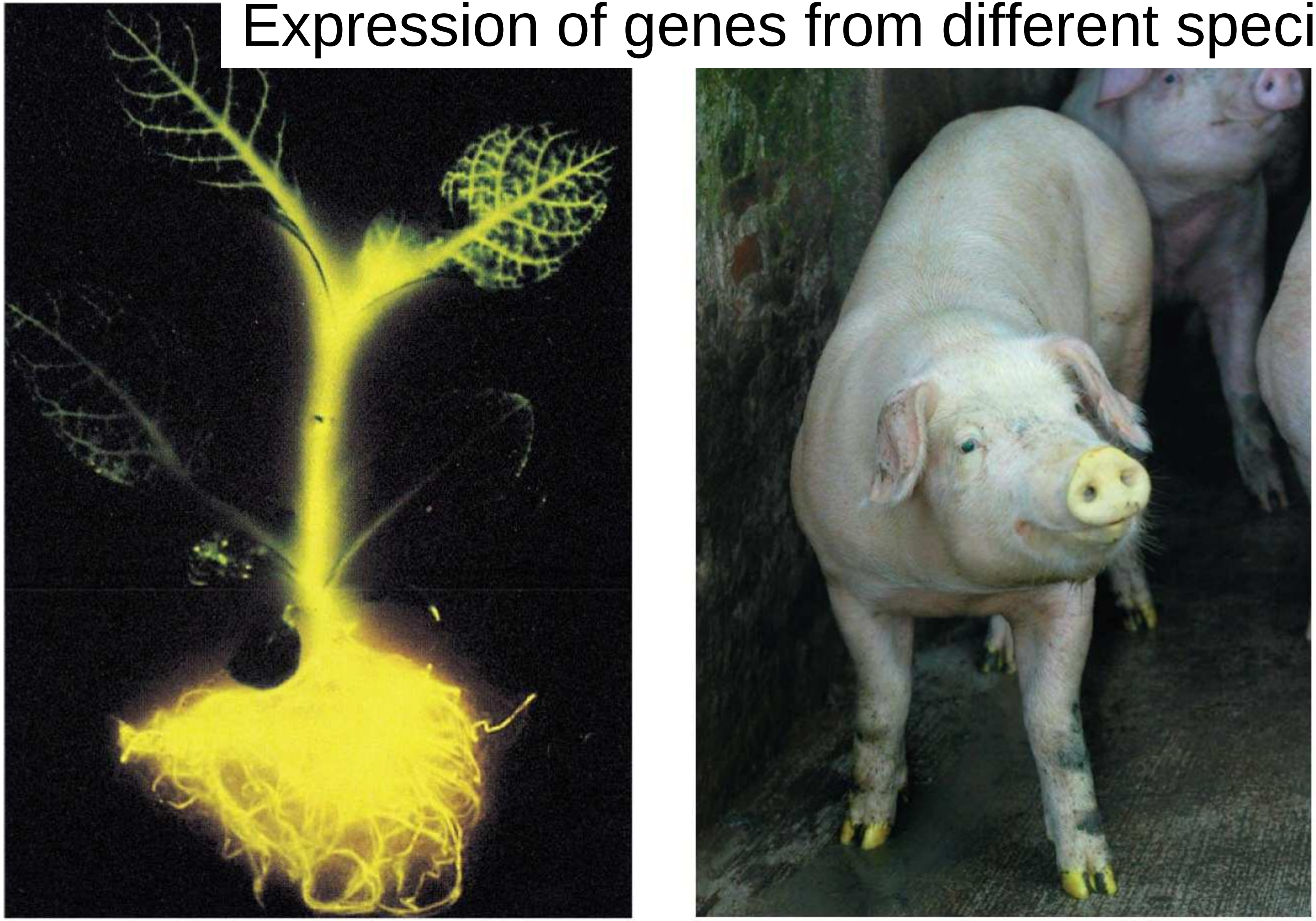
First mRNA base (5′ end of codon)	Second mRNA base				Third mRNA base (3′ end of codon)
	U	C	A	G	
U	UUU Phe	UCU Ser	UAU Tyr	UGU Cys	U
	UUC	UCC	UAC	UGC	C
	UUA Leu	UCA	UAA Stop	UGA Stop	A
	UUG	UCG	UAG Stop	UGG Trp	G
C	CUU Leu	CCU Pro	CAU His	CGU Arg	U
	CUC	CCC	CAC	CGC	C
	CUA	CCA	CAA Gln	CGA	A
	CUG	CCG	CAG	CGG	G
A	AUU Ile	ACU Thr	AAU Asn	AGU Ser	U
	AUC	ACC	AAC	AGC	C
	AUA	ACA	AAA Lys	AGA Arg	A
	AUG Met or start	ACG	AAG	AGG	G
G	GUU Val	GCU Ala	GAU Asp	GGU Gly	U
	GUC	GCC	GAC	GGC	C
	GUA	GCA	GAA Glu	GGA	A
	GUG	GCG	GAG	GGG	G

Evolution of the Genetic Code

- The genetic code is nearly universal, shared by the simplest bacteria to the most complex animals
- Genes can be transcribed and translated after being transplanted from one species to another (基因轉殖)

Fig. 17-6

Expression of genes from different species



(a) Tobacco plant expressing a firefly gene

(b) Pig expressing a jellyfish gene

Concept 17.2: Transcription is the DNA-directed synthesis of RNA: *a closer look*

- **Transcription**, the **first stage of gene expression**, can be examined in more detail

DNAi video clip

Molecular Components of Transcription

- RNA synthesis is catalyzed by **RNA polymerase**, which pries the DNA strands apart and hooks together the RNA nucleotides
- RNA synthesis follows the same base-pairing rules as DNA, except uracil substitutes for thymine

- The DNA sequence where RNA polymerase attaches is called the **promoter**; in bacteria, the sequence signaling the end of transcription is called the **terminator**
- The stretch of DNA that is transcribed is called a **transcription unit**

PLAY **Animation: Transcription**

Fig. 17-7

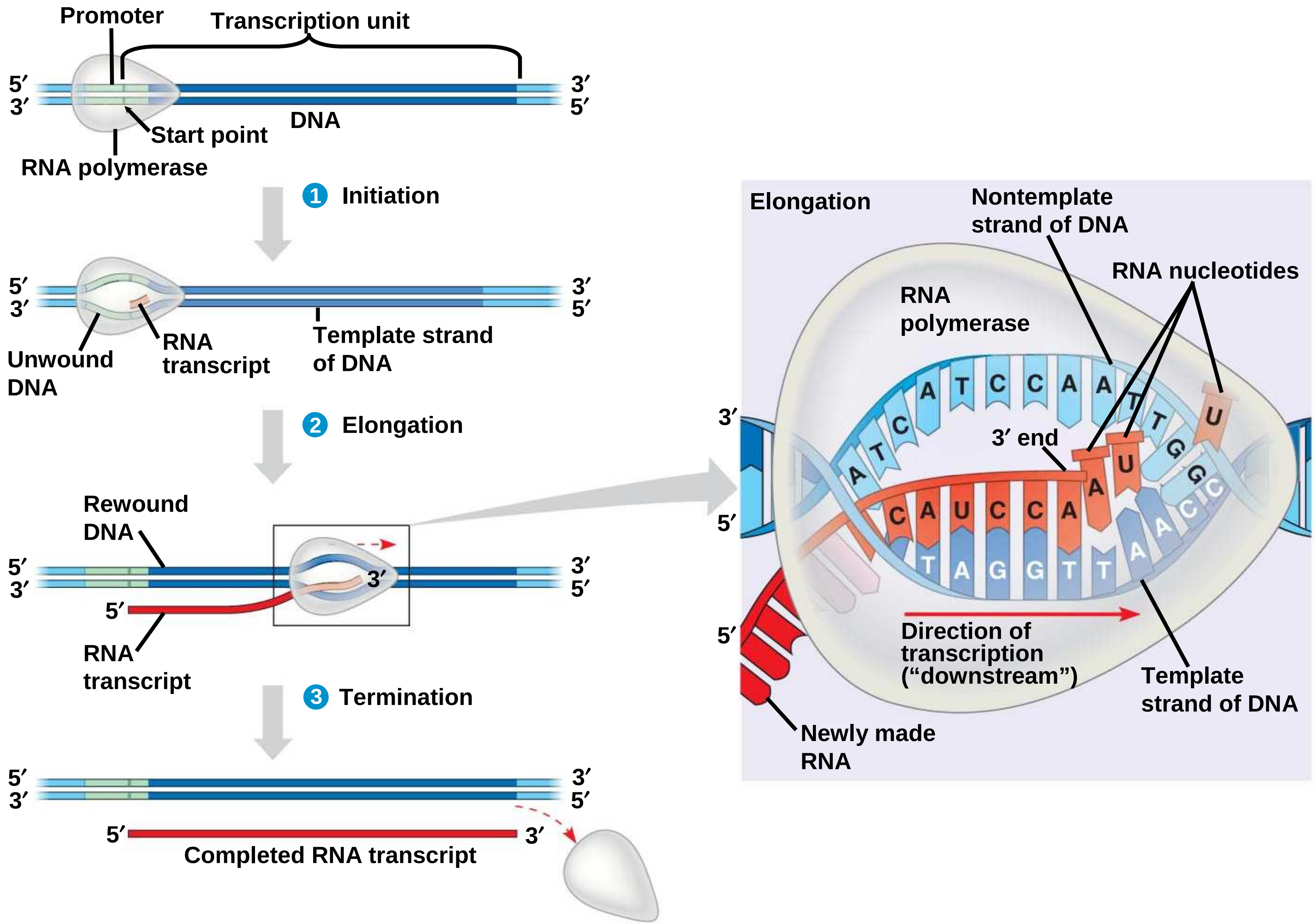

Promoter
Transcription unit
5′
3′
3′
5′
Start point
DNA
RNA polymerase
1 Initiation
5′
3′
3′
5′
Unwound DNA
RNA transcript
Template strand of DNA
2 Elongation
Rewound DNA
5′
3′
3′
3′
5′
5′
RNA transcript
3 Termination
5′
3′
3′
5′
5′
3′
Completed RNA transcript
Elongation
Nontemplate strand of DNA
RNA nucleotides
RNA polymerase
3′
A T C A T C C A A T T G G
U
3′ end
5′
C A U C C A A U
T A G G T T A A C C
5′
Direction of transcription (“downstream”)
Template strand of DNA
Newly made RNA

Fig. 17-7a-1

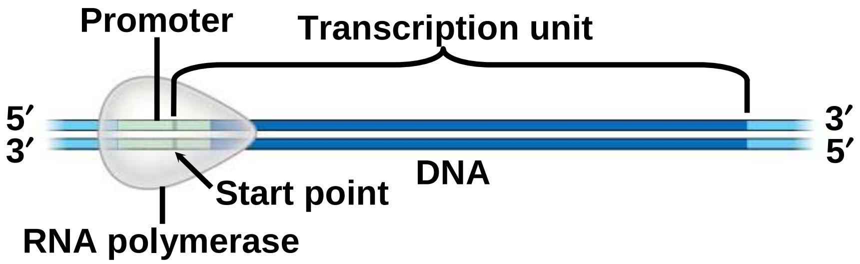

Promoter
Transcription unit
5′
3′
3′
5′
DNA
Start point
RNA polymerase

Fig. 17-7a-2

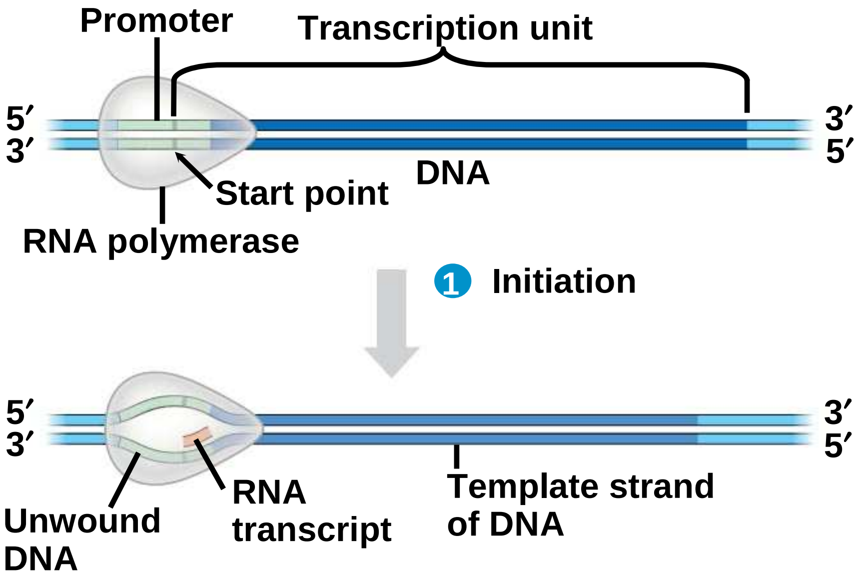

Promoter
Transcription unit
5′
3′
3′
5′
Start point
DNA
RNA polymerase
1 Initiation
5′
3′
3′
5′
Unwound DNA
RNA transcript
Template strand of DNA

Fig. 17-7a-3

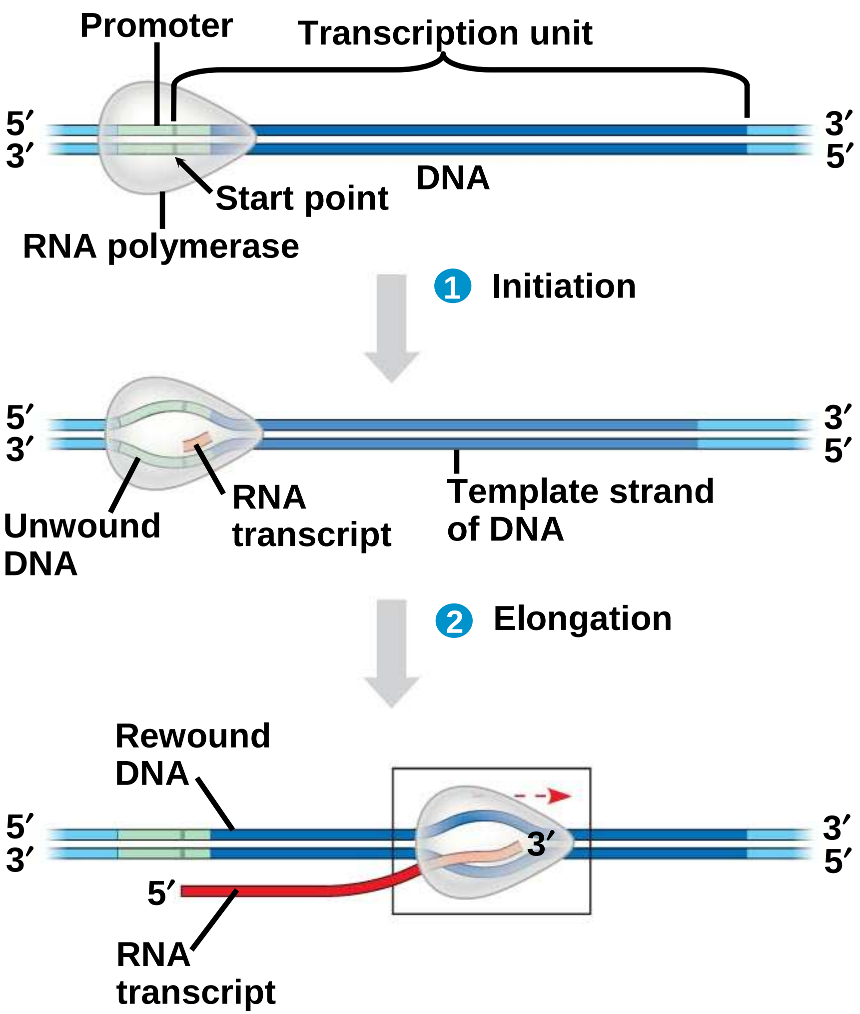

Promoter
Transcription unit
5′
3′
3′
5′
DNA
Start point
RNA polymerase
1 Initiation
5′
3′
3′
5′
Unwound DNA
RNA transcript
Template strand of DNA
2 Elongation
Rewound DNA
5′
3′
3′
5′
3′
5′
RNA transcript

Fig. 17-7a-4

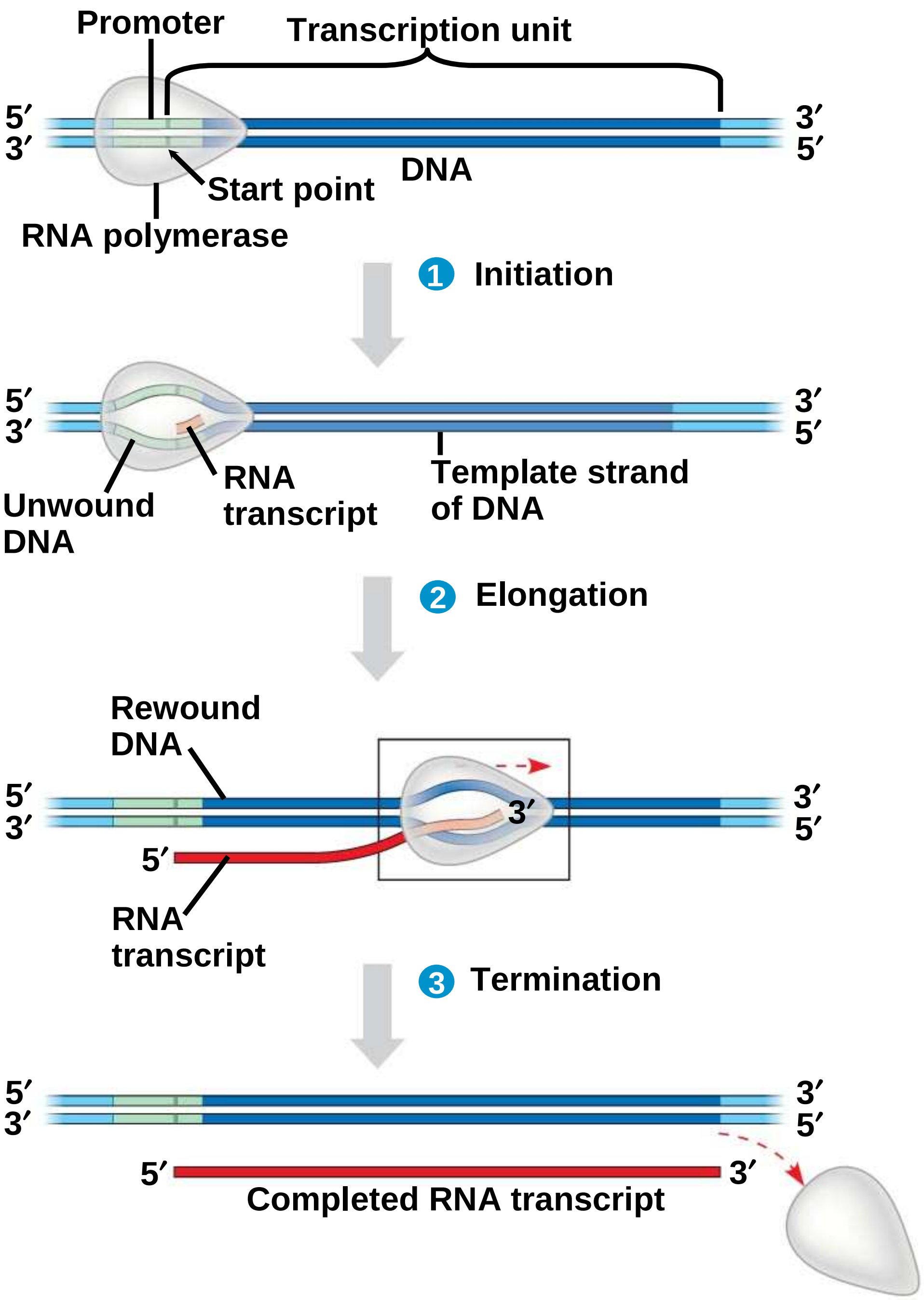
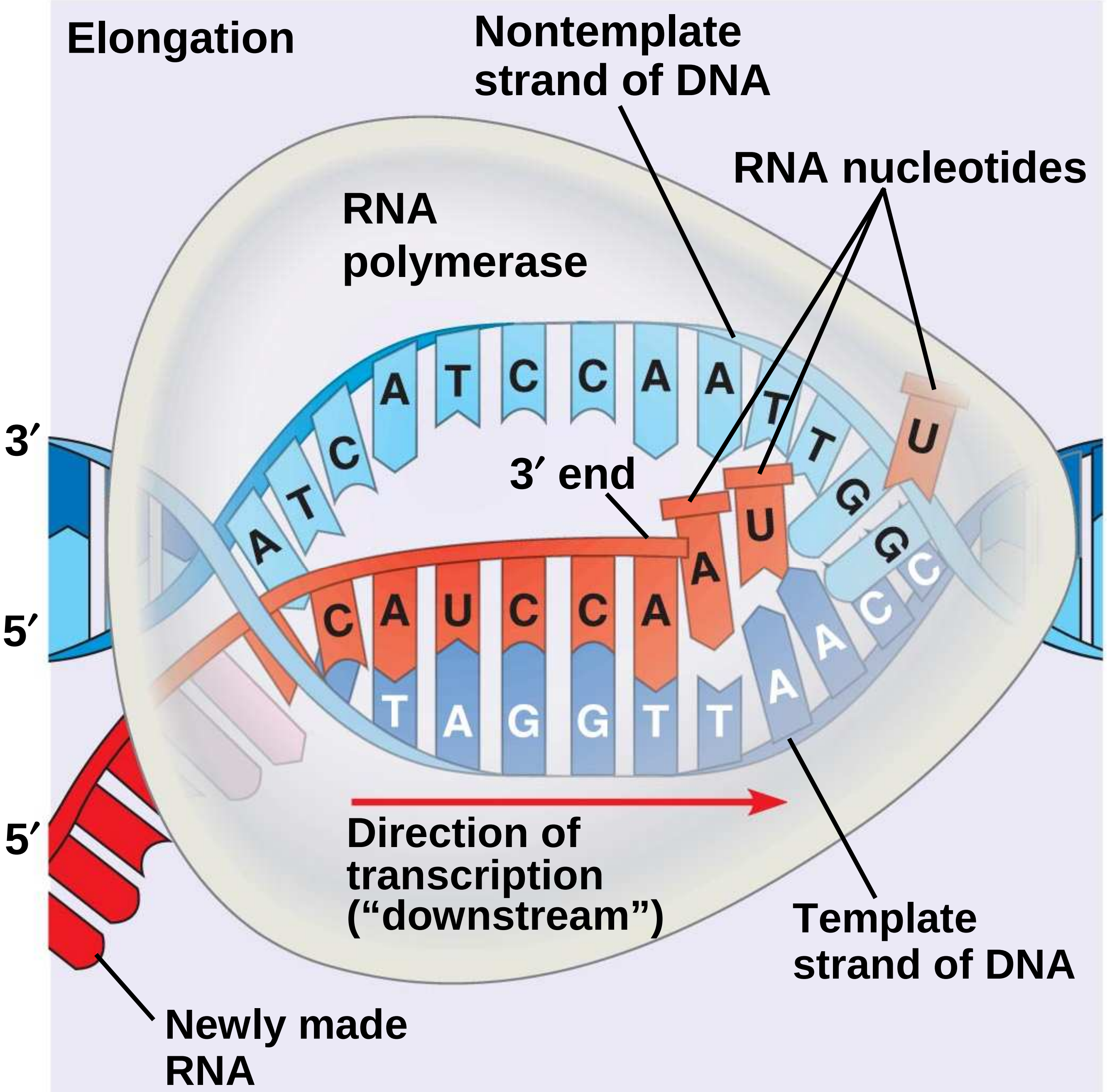

Promoter
Transcription unit
5′
3′
3′
5′
Start point
DNA
RNA polymerase
1 Initiation
5′
3′
3′
5′
Unwound DNA
RNA transcript
Template strand of DNA
2 Elongation
Rewound DNA
5′
3′
3′
3′
5′
5′
RNA transcript
3 Termination
5′
3′
3′
5′
5′
3′
Completed RNA transcript

Fig. 17-7b


Elongation
Nontemplate strand of DNA
RNA nucleotides
RNA polymerase
3′
3′ end
A T C A T C C A A T T G G
U
A U
C A U C C A
T A G G T T A A C C
5′
5′
Direction of transcription (“downstream”)
Template strand of DNA
Newly made RNA

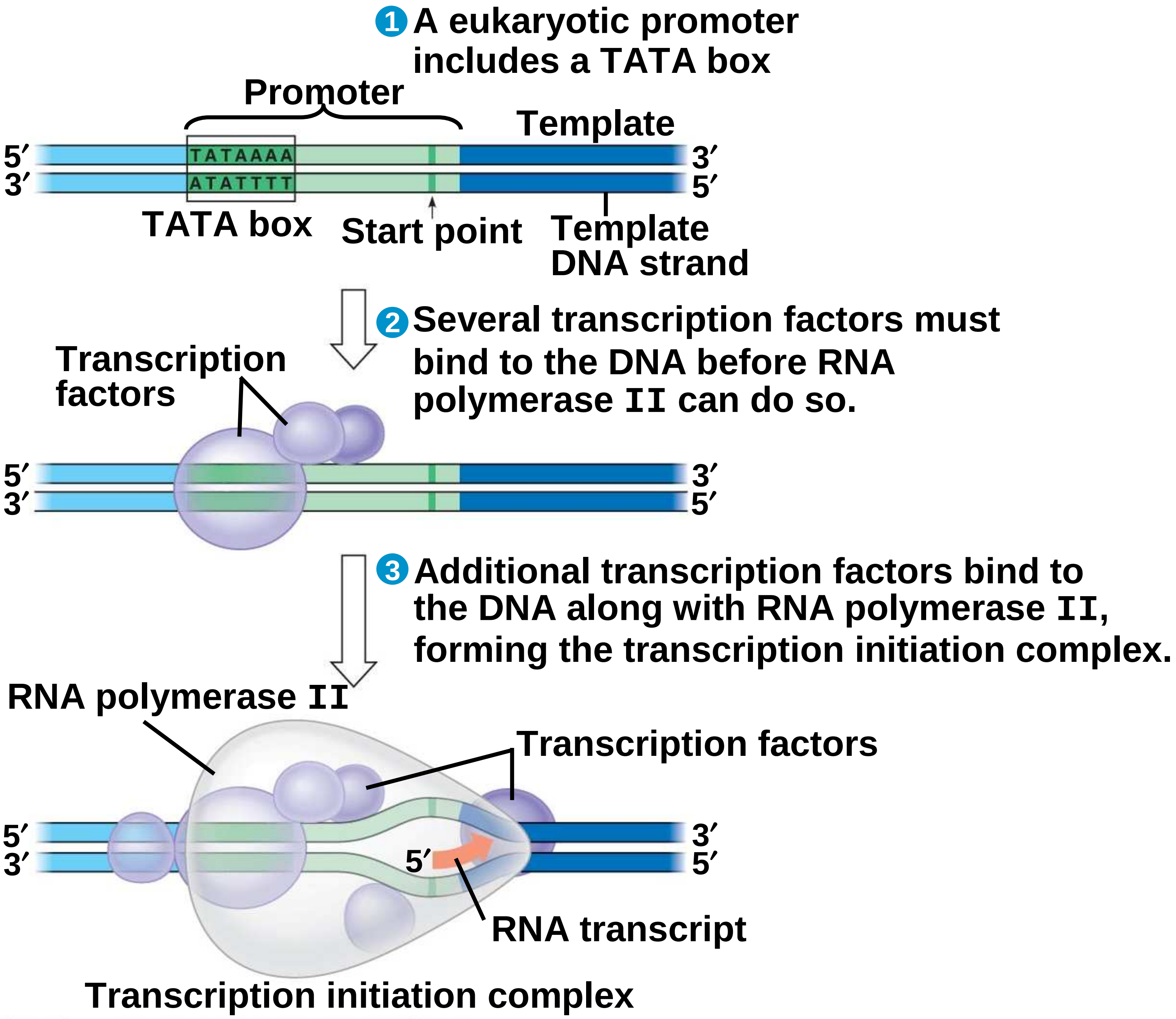
Synthesis of an RNA Transcript

- The three stages of transcription:
  - **Initiation**
  - **Elongation**
  - **Termination**

RNA Polymerase Binding and Initiation of Transcription

- Promoters signal the initiation of RNA synthesis
- **Transcription factors** mediate the binding of RNA polymerase and the initiation of transcription
- The completed assembly of transcription factors and RNA polymerase II bound to a promoter is called a **transcription initiation complex**
- A promoter called a **TATA box** is crucial in forming the initiation complex in eukaryotes

Fig. 17-8



① A eukaryotic promoter includes a TATA box

② Several transcription factors must bind to the DNA before RNA polymerase II can do so.

③ Additional transcription factors bind to the DNA along with RNA polymerase II, forming the transcription initiation complex.

Elongation of the RNA Strand

- As RNA polymerase moves along the DNA, it untwists the double helix, 10 to 20 bases at a time
- Transcription progresses at a rate of 40 nucleotides per second in eukaryotes
- A gene can be transcribed simultaneously by several RNA polymerases

Termination of Transcription

- The mechanisms of termination are different in bacteria and eukaryotes
- In bacteria, the polymerase stops transcription at the end of the terminator
- In eukaryotes, the polymerase continues transcription after the pre-mRNA is cleaved from the growing RNA chain; the polymerase eventually falls off the DNA

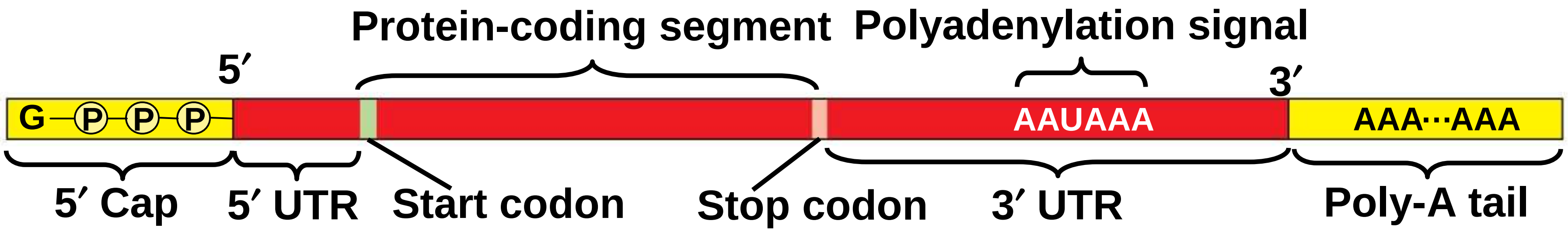
Concept 17.3: Eukaryotic cells modify RNA after transcription

- 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

Alteration of mRNA Ends

- Each end of a pre-mRNA molecule is modified in a particular way:
  - The 5′ end receives a modified nucleotide **5′ cap**
  - The 3′ end gets a **poly-A tail**
- These modifications share several functions:
  - They seem to facilitate the export of mRNA
  - They protect mRNA from hydrolytic enzymes
  - They help ribosomes attach to the 5′ end

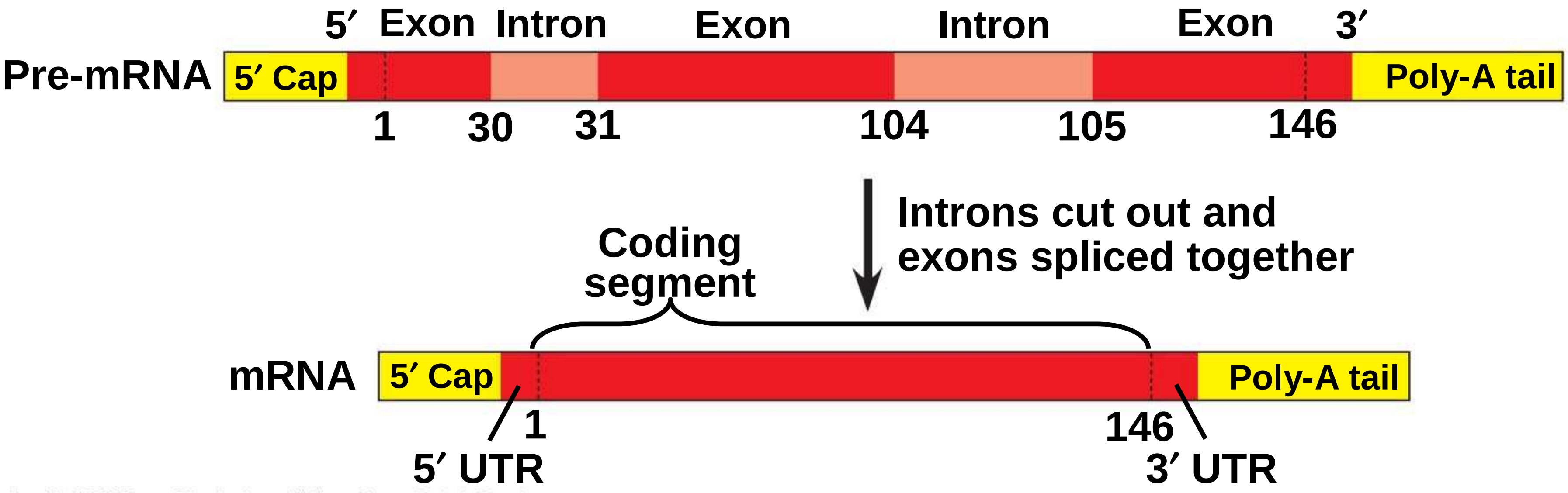
Fig. 17-9


Protein-coding segment
Polyadenylation signal
5′
3′
G
P
P
P
AAUAAA
AAA···AAA
5′ Cap
5′ UTR
Start codon
Stop codon
3′ UTR
Poly-A tail

Split Genes and RNA Splicing

- Most eukaryotic genes and their RNA transcripts have long noncoding stretches of nucleotides that lie between coding regions
- These noncoding regions are called intervening sequences, or **introns**
- The other regions are called **exons** because they are eventually expressed, usually translated into amino acid sequences
- **RNA splicing** removes introns and joins exons, creating an mRNA molecule with a continuous coding sequence

Fig. 17-10


5′
Exon
Intron
Exon
Intron
Exon
3′
Pre-mRNA
5′ Cap
Poly-A tail
1
30
31
104
105
146
Introns cut out and exons spliced together
Coding segment
mRNA
5′ Cap
Poly-A tail
1
146
5′ UTR
3′ UTR

- In some cases, RNA splicing is carried out by spliceosomes
- **Spliceosomes** consist of a variety of proteins and several small nuclear ribonucleoproteins (snRNPs) that recognize the splice sites

Fig. 17-11-1

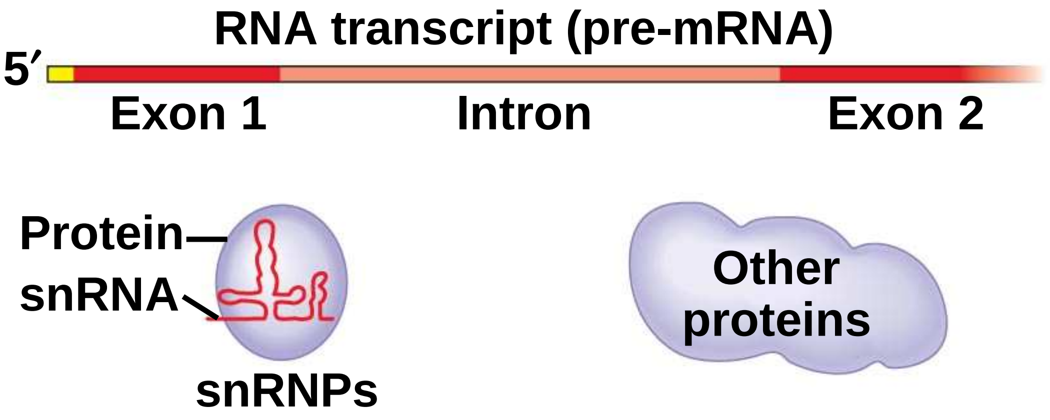

RNA transcript (pre-mRNA)
5′
Exon 1
Intron
Exon 2
Protein
snRNA
snRNPs
Other proteins

Fig. 17-11-2

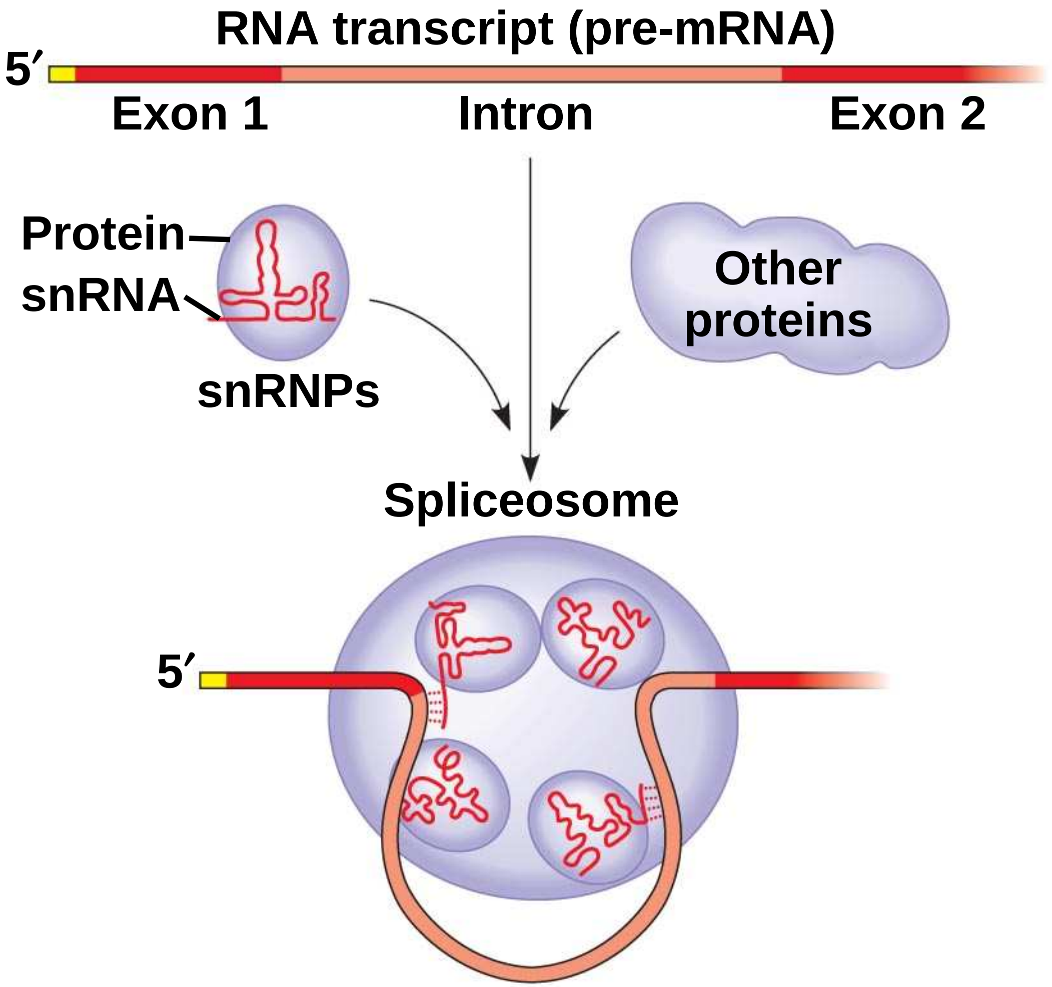
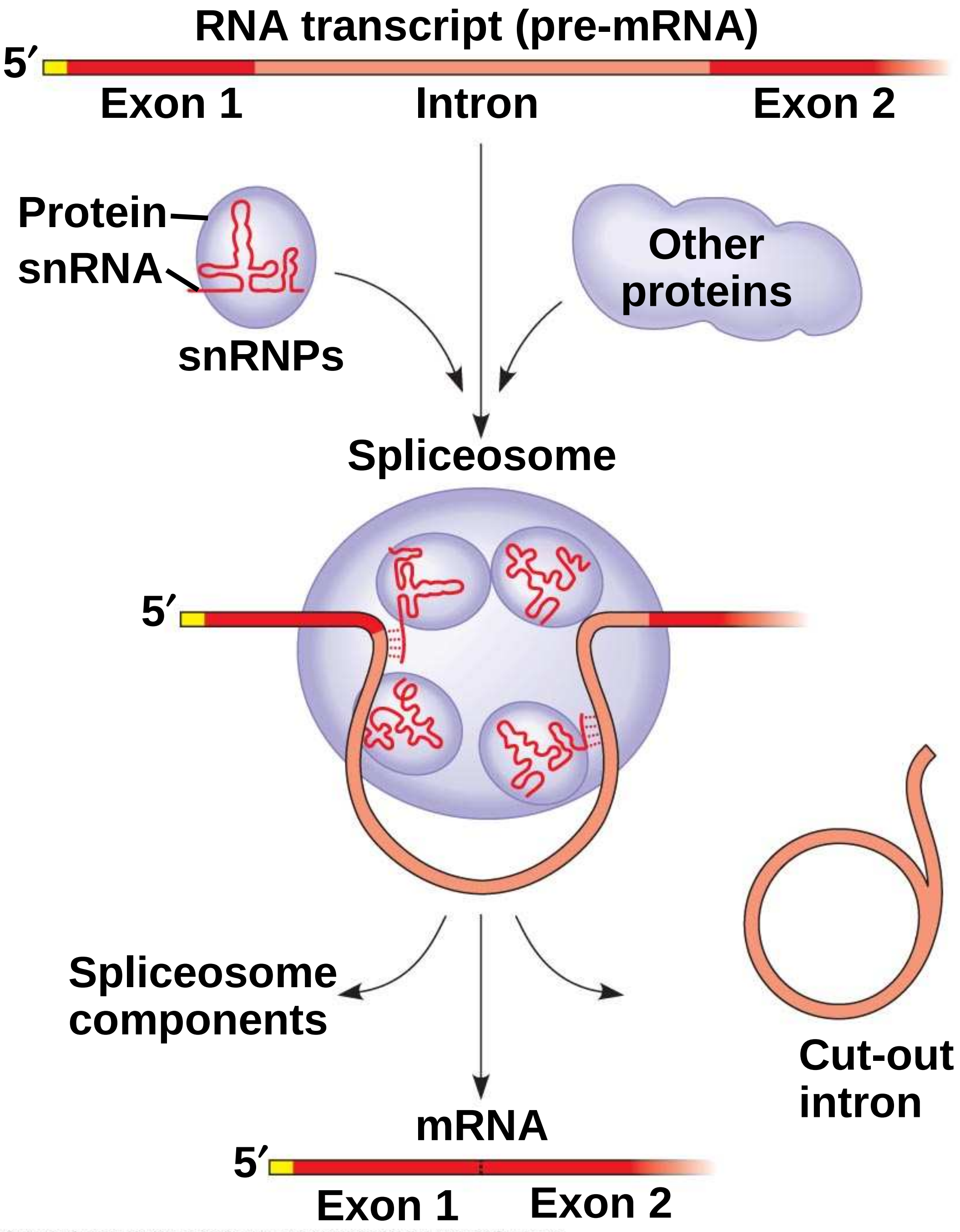

RNA transcript (pre-mRNA)
5′
Exon 1
Intron
Exon 2
Protein
snRNA
snRNPs
Other proteins
Spliceosome
5′

Fig. 17-11-3


RNA transcript (pre-mRNA)
5′
Exon 1
Intron
Exon 2
Protein
snRNA
snRNPs
Other proteins
Spliceosome
5′
Spliceosome components
Cut-out intron
mRNA
5′
Exon 1
Exon 2

Ribozymes

- **Ribozymes** are catalytic RNA molecules that function as enzymes and can splice RNA
- The discovery of ribozymes rendered obsolete the belief that all biological catalysts were proteins

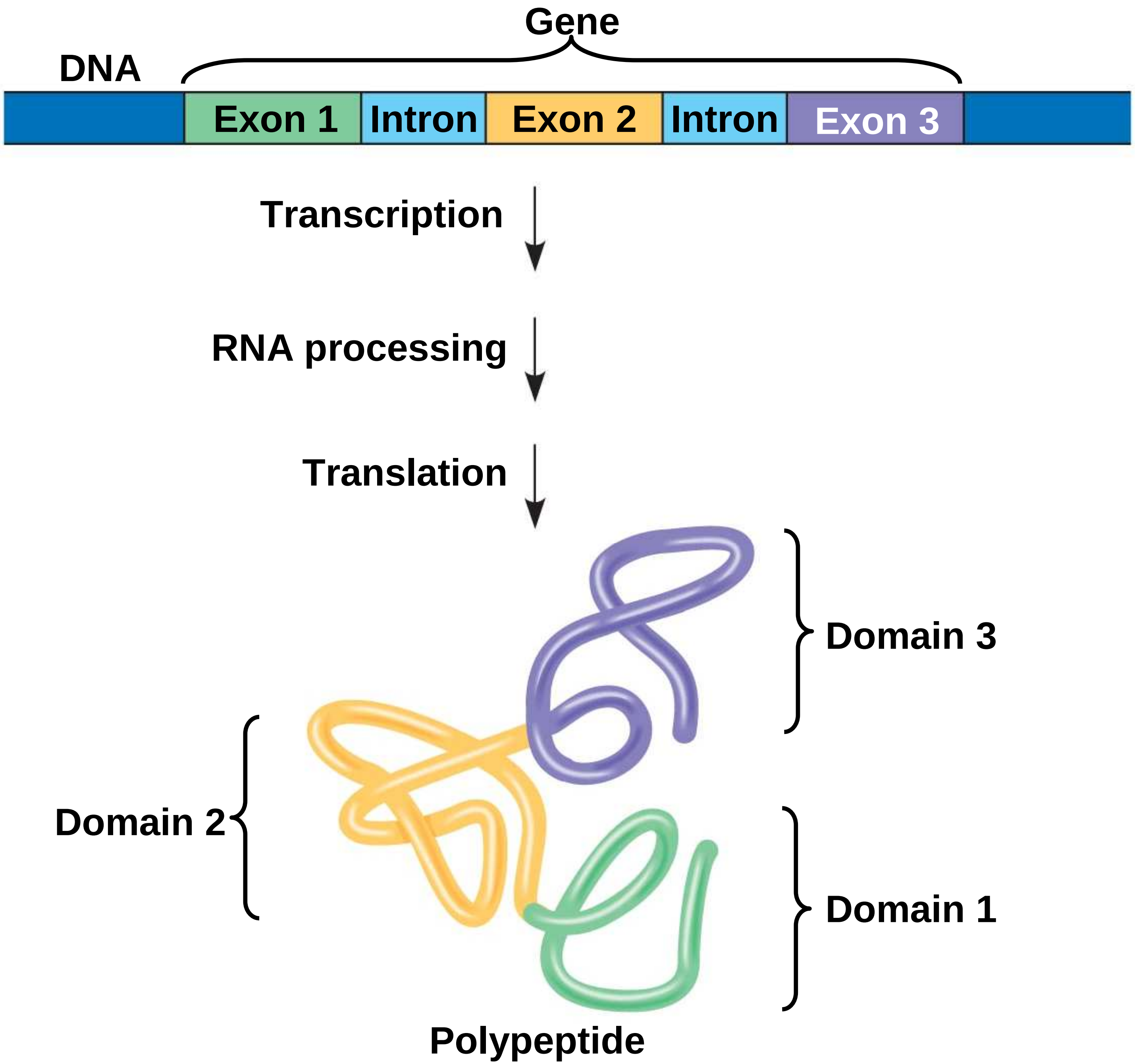
- Three properties of RNA enable it to function as an enzyme
  - It can form a three-dimensional structure because of its ability to base pair with itself
  - Some bases in RNA contain functional groups
  - RNA may hydrogen-bond with other nucleic acid molecules

The Functional and Evolutionary Importance of Introns

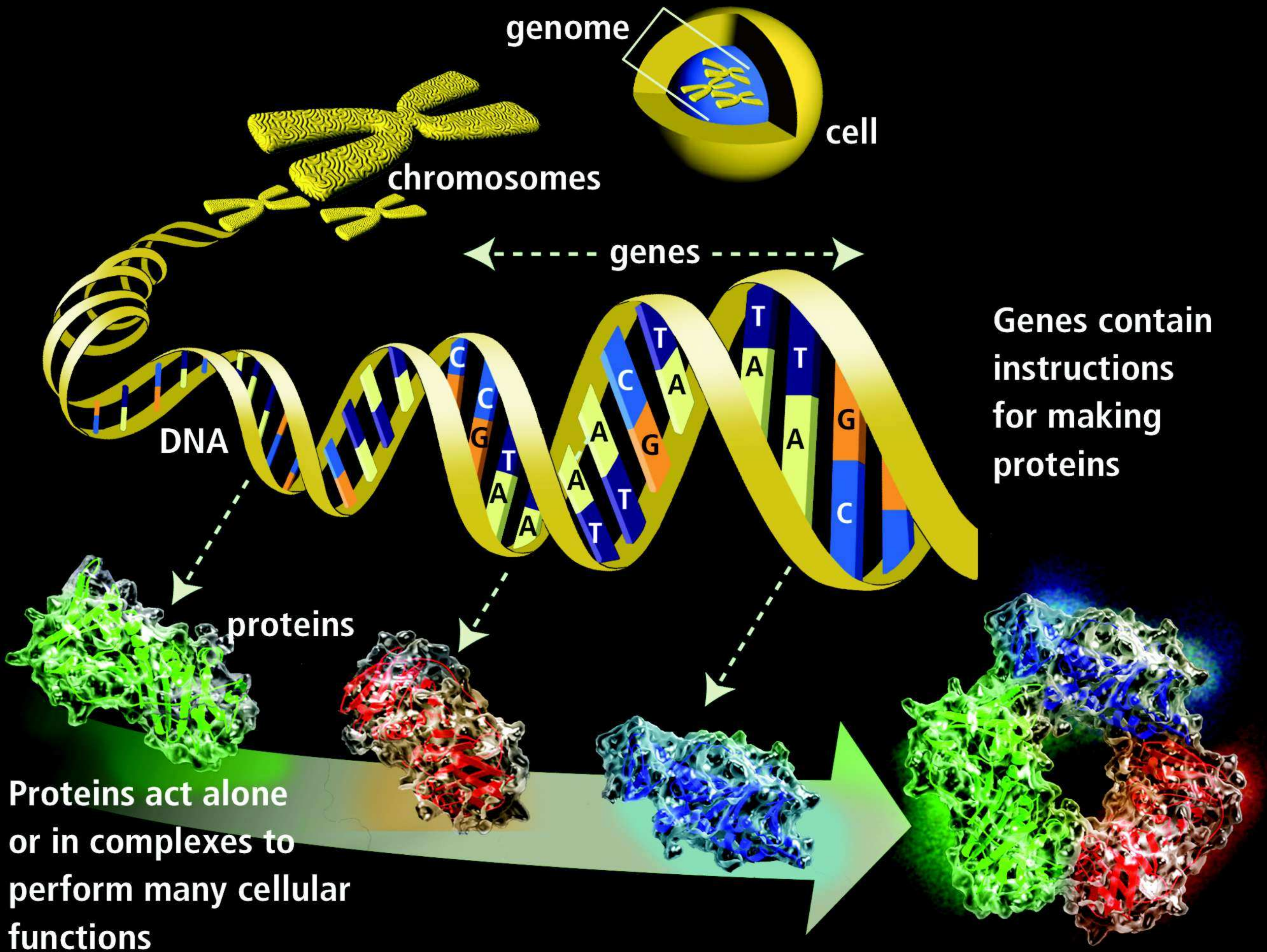
- Some genes can encode more than one kind of polypeptide, depending on which segments are treated as exons during RNA splicing
- Such variations are called **alternative RNA splicing**
- Because of alternative splicing, the number of different proteins an organism can produce is much greater than its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

- Proteins often have a modular architecture consisting of discrete regions called **domains**
- In many cases, different exons code for the different domains in a protein
- Exon shuffling may result in the evolution of new proteins

Fig. 17-12


Gene
DNA
Exon 1
Intron
Exon 2
Intron
Exon 3
Transcription
RNA processing
Translation
Domain 3
Domain 2
Domain 1
Polypeptide

Paused



Genes contain instructions for making proteins

Proteins act alone or in complexes to perform many cellular functions

U.S. DEPARTMENT OF ENERGY

Concept 17.4: Translation is the RNA-directed synthesis of a polypeptide: *a closer look*

- The **translation** of **mRNA to protein** can be examined in more detail

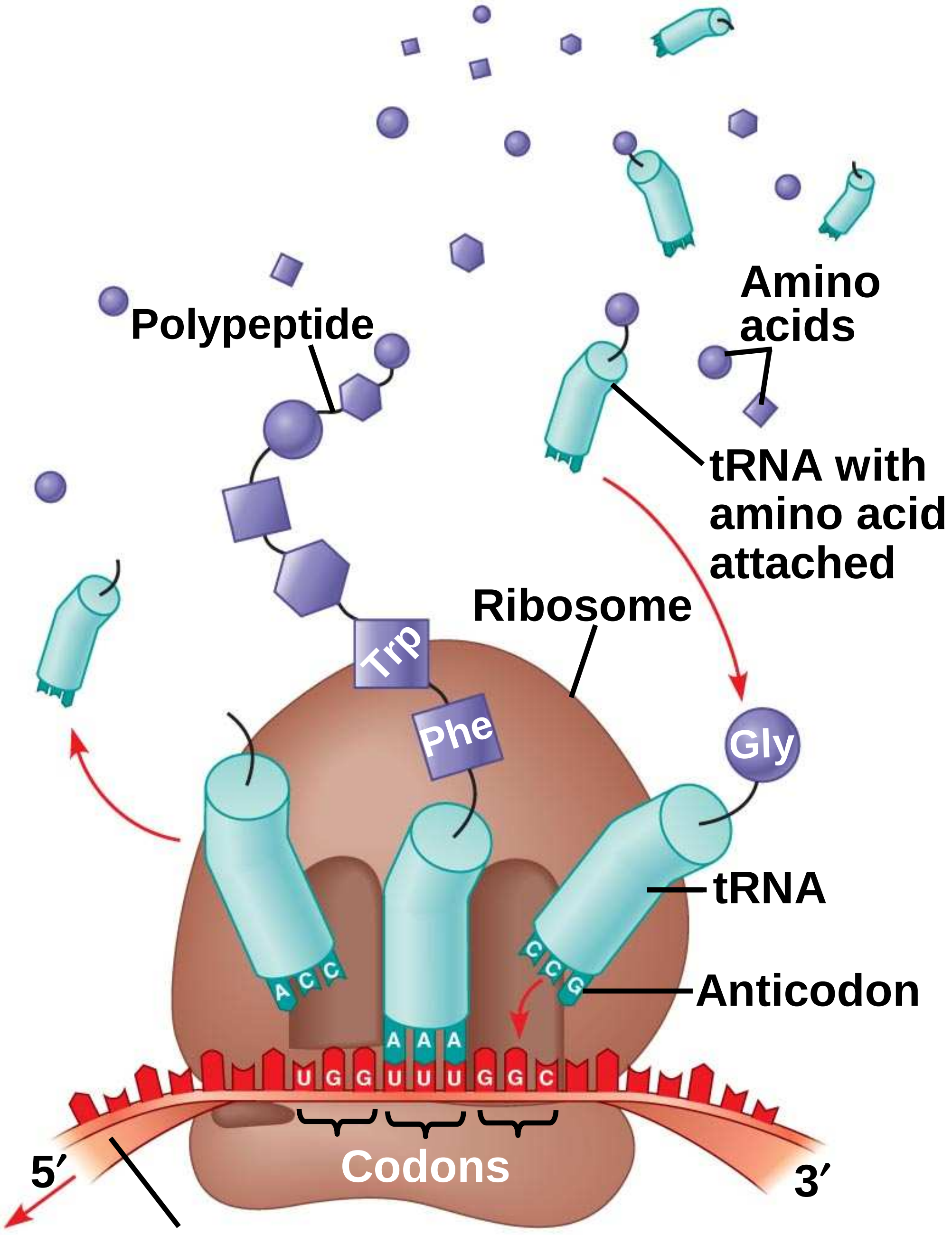
Molecular Components of Translation

- A cell translates an mRNA message into protein with the help of **transfer RNA (tRNA)**
- Molecules of tRNA are not identical:
  - Each carries a specific amino acid on one end
  - Each has an **anticodon** on the other end; the anticodon base-pairs with a complementary codon on mRNA

PLAY

BioFlix: Protein Synthesis

Fig. 17-13

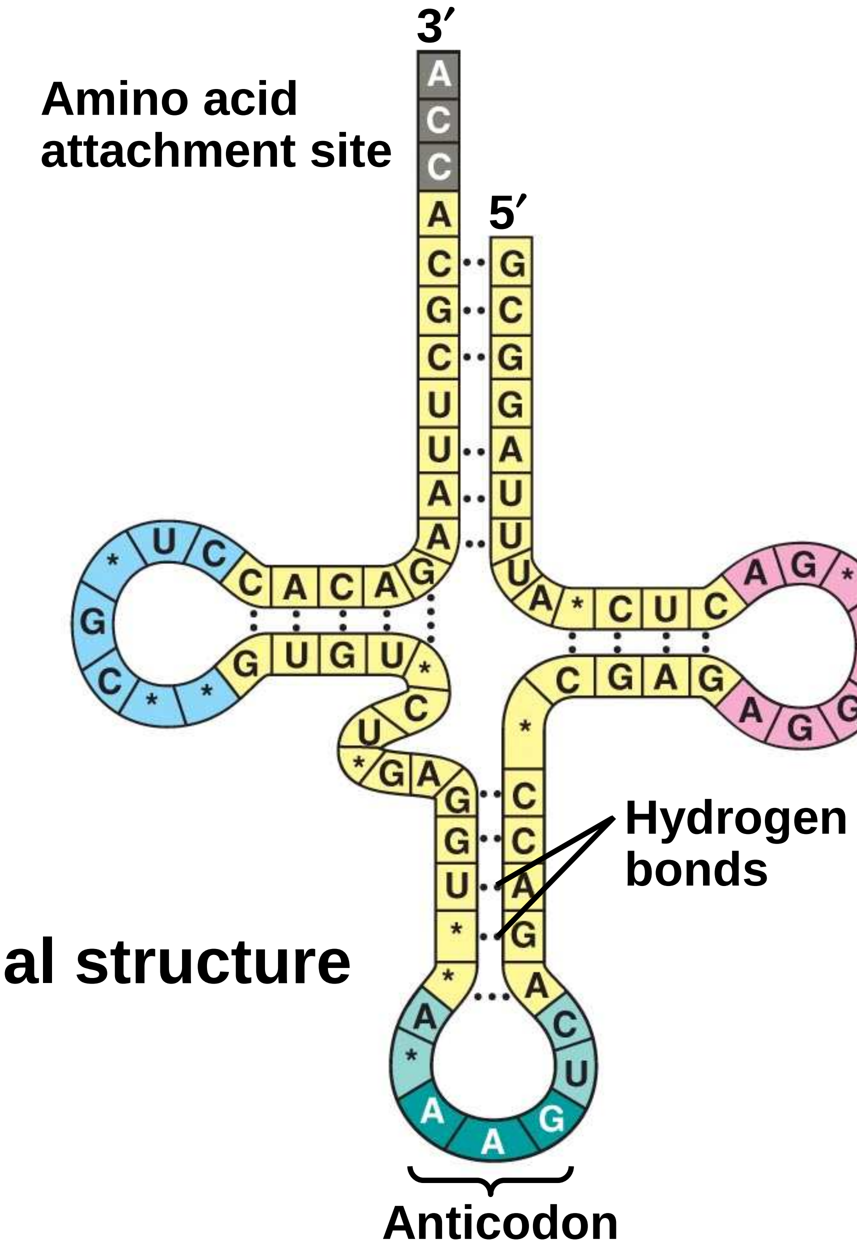

Amino acids
Polypeptide
tRNA with amino acid attached
Ribosome
Trp
Phe
Gly
tRNA
Anticodon
A C C
A A A
C C G
U G G U U U G G C
Codons
5′
3′
mRNA

The Structure and Function of Transfer RNA

- A tRNA molecule consists of a single RNA strand that is only about 80 nucleotides long

- Flattened into one plane to reveal its base pairing, a tRNA molecule looks like a cloverleaf

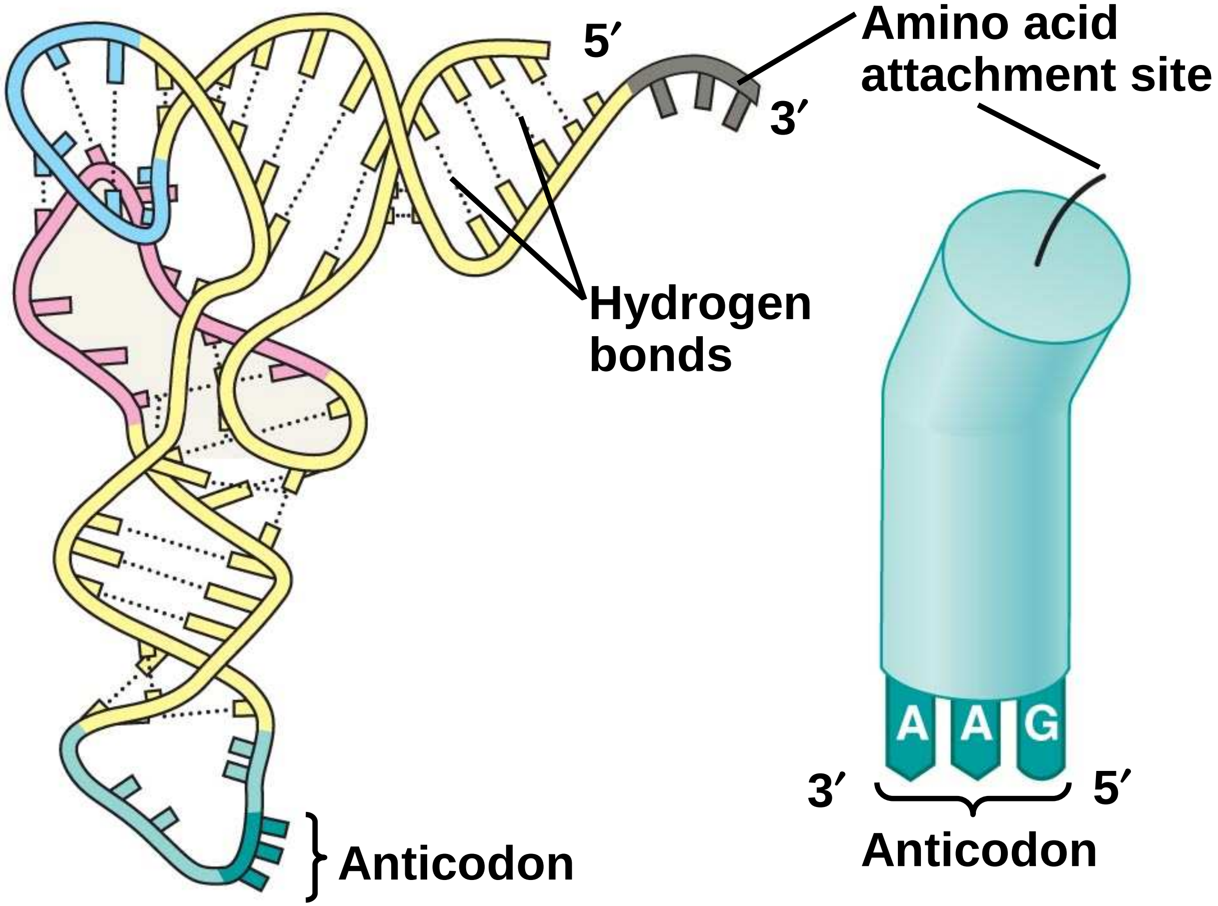
Fig. 17-14a



(a) Two-dimensional structure

Fig. 17-14b

(b) Three-dimensional structure



- Because of hydrogen bonds, tRNA actually twists and folds into a three-dimensional molecule
- tRNA is roughly L-shaped

- Accurate translation requires two steps:
  - **1.** a correct match between a tRNA and an amino acid, done by the enzyme **aminoacyl-tRNA synthetase**
  - **2.** a correct match between the tRNA anticodon and an mRNA codon
- Flexible pairing at the third base of a codon is called **wobble** and allows some tRNAs to bind to more than one codon

Fig. 17-15-1

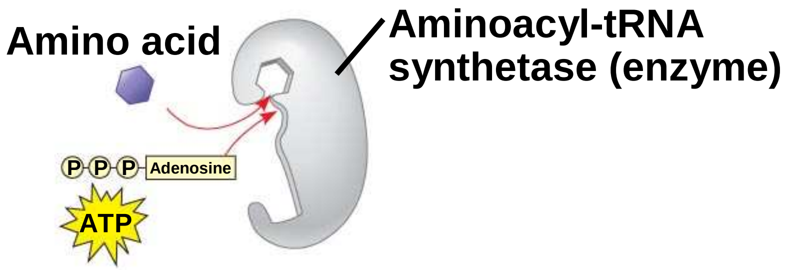

Amino acid
Aminoacyl-tRNA synthetase (enzyme)
P
P
P
Adenosine
ATP

Fig. 17-15-2

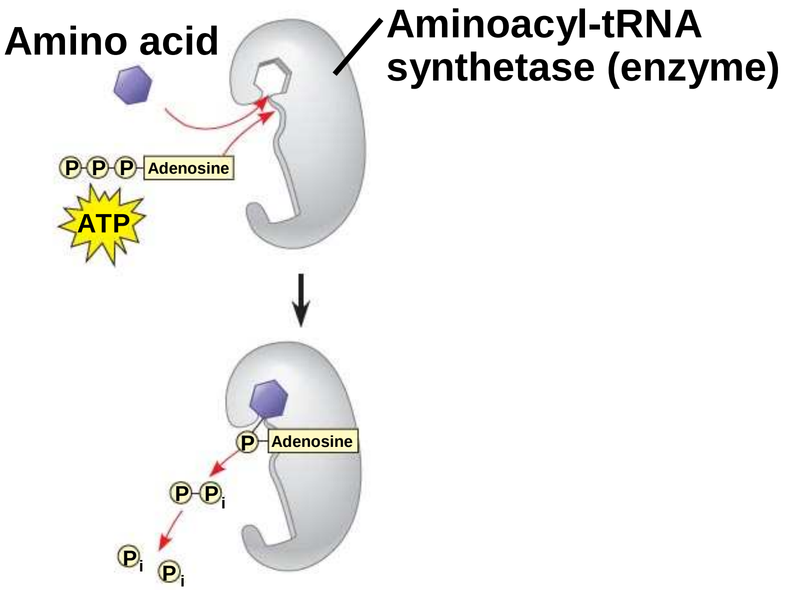

Amino acid
Aminoacyl-tRNA synthetase (enzyme)
P P P Adenosine
ATP
P Adenosine
P P$_i$
P$_i$ P$_i$

Fig. 17-15-3

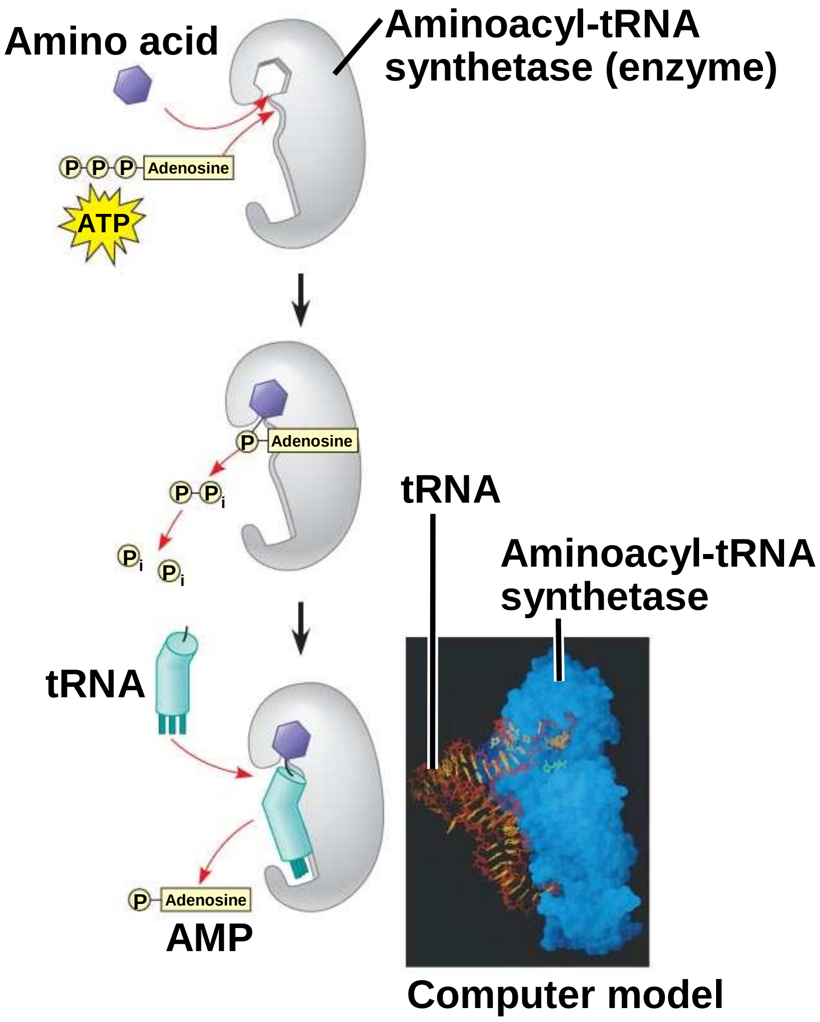
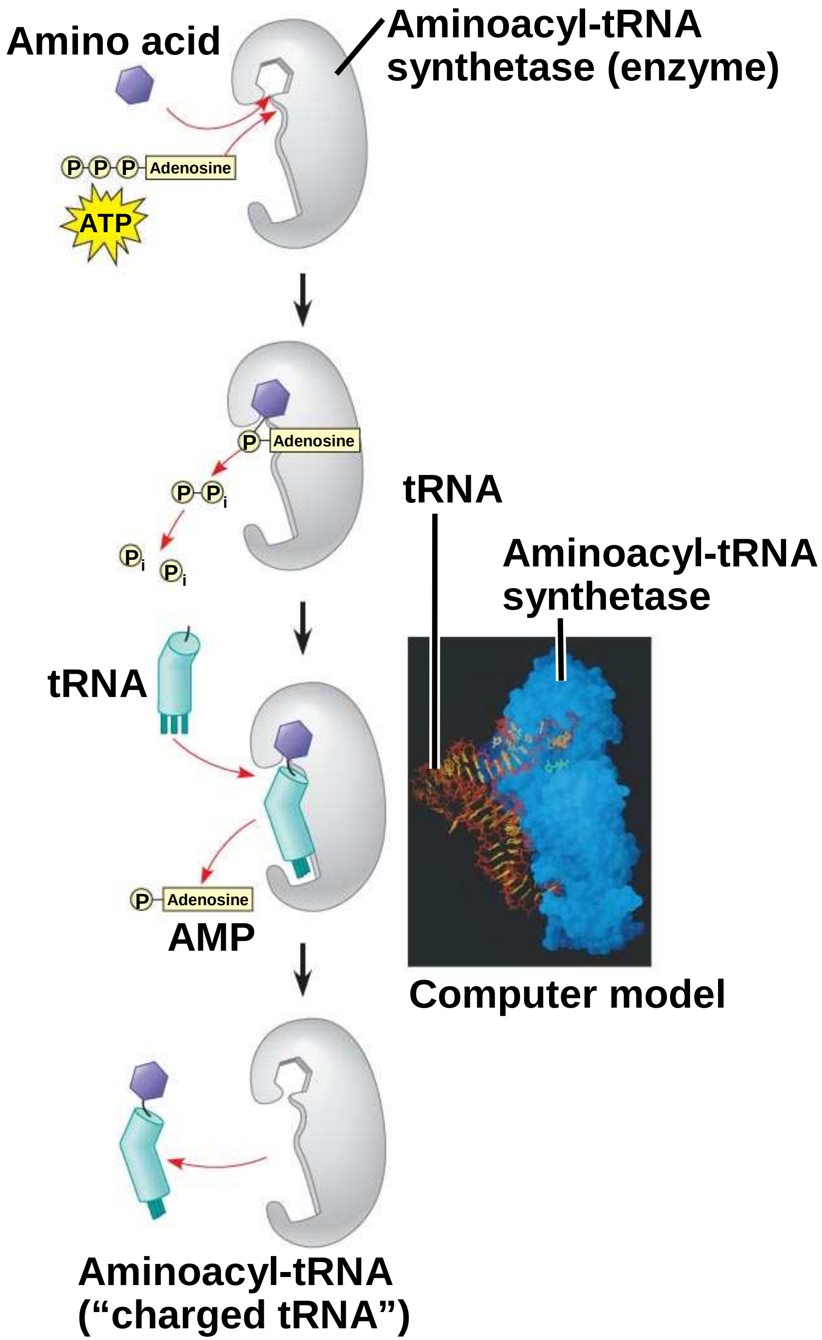

Amino acid
Aminoacyl-tRNA synthetase (enzyme)
P P P Adenosine
ATP
P Adenosine
P P$_i$
P$_i$
P$_i$
tRNA
P Adenosine
AMP
tRNA
Aminoacyl-tRNA synthetase
Computer model

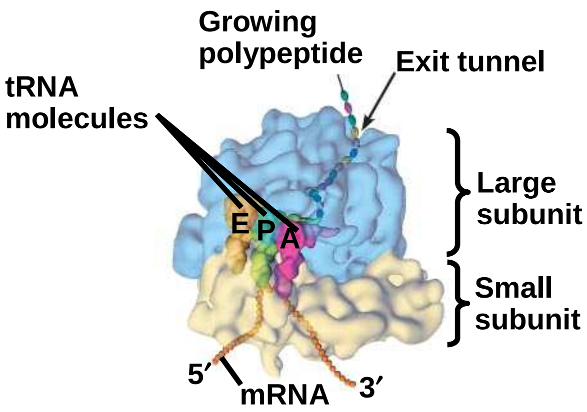
Fig. 17-15-4


Amino acid
Aminoacyl-tRNA synthetase (enzyme)
P P P Adenosine
ATP
P Adenosine
P P$_i$
P$_i$ P$_i$
tRNA
P Adenosine
AMP
Aminoacyl-tRNA
("charged tRNA")
tRNA
Aminoacyl-tRNA synthetase
Computer model

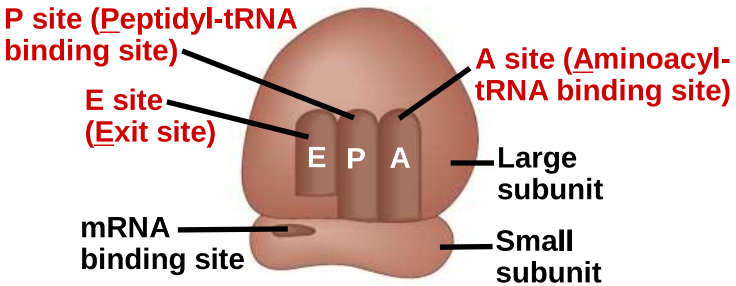
Ribosomes

- Ribosomes facilitate specific coupling of tRNA anticodons with mRNA codons in protein synthesis
- The two ribosomal subunits (large and small) are made of proteins and **ribosomal RNA (rRNA)**

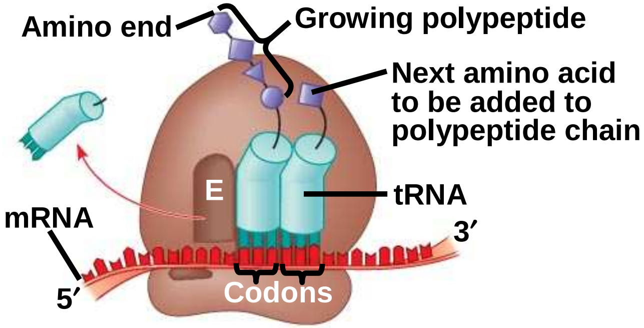
Fig. 17-16



(a) Computer model of functioning ribosome

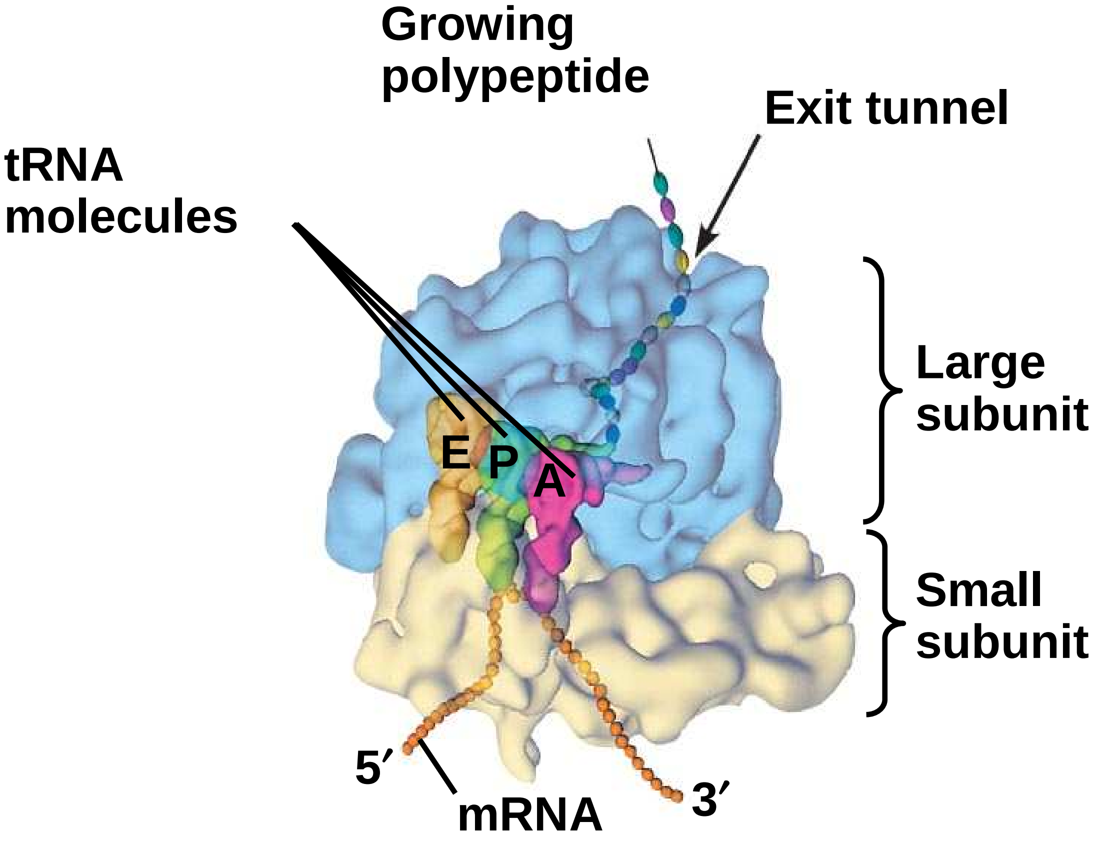


(b) Schematic model showing binding sites



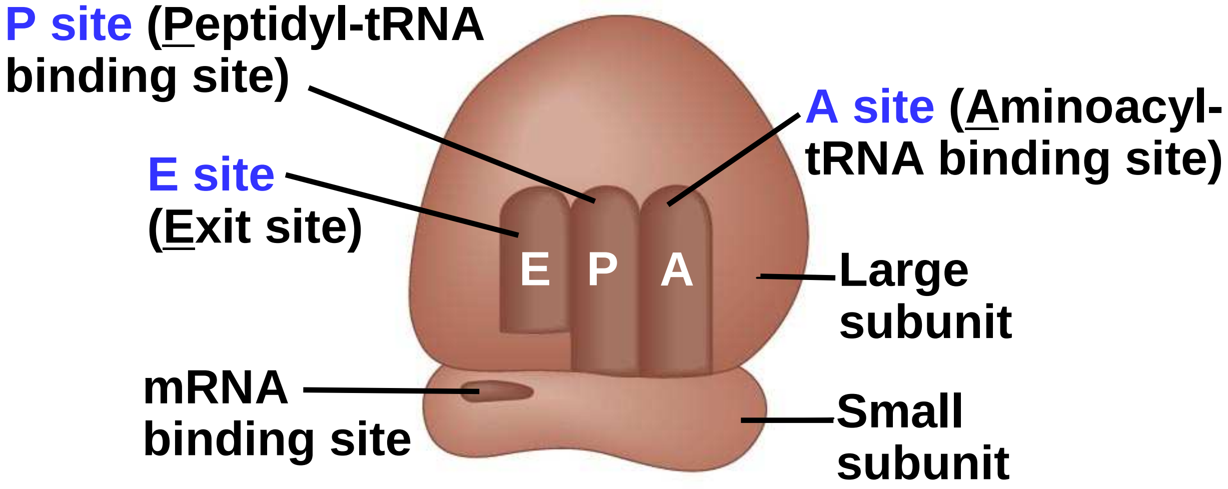
(c) Schematic model with mRNA and tRNA

Fig. 17-16a



(a) Computer model of functioning ribosome

Fig. 17-16b



P site (Peptidyl-tRNA binding site)

A site (Aminoacyl-tRNA binding site)

E site (Exit site)

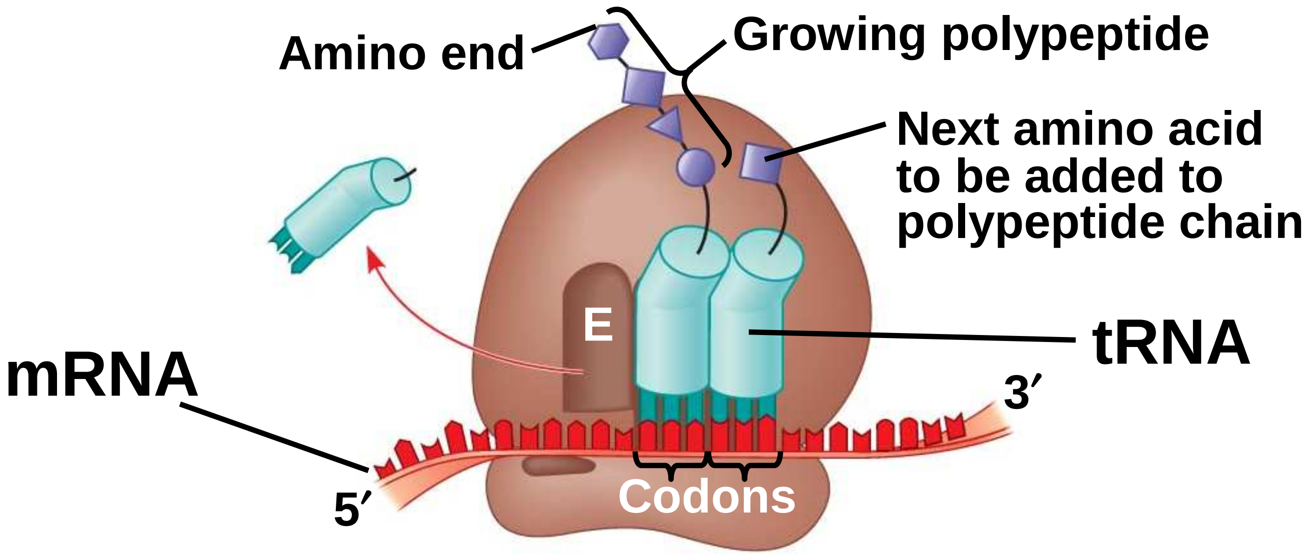
E P A

Large subunit

mRNA binding site

Small subunit

(b) Schematic model showing binding sites



Amino end

Growing polypeptide

Next amino acid to be added to polypeptide chain

E

tRNA

mRNA

3′

5′

Codons

(c) Schematic model with mRNA and tRNA

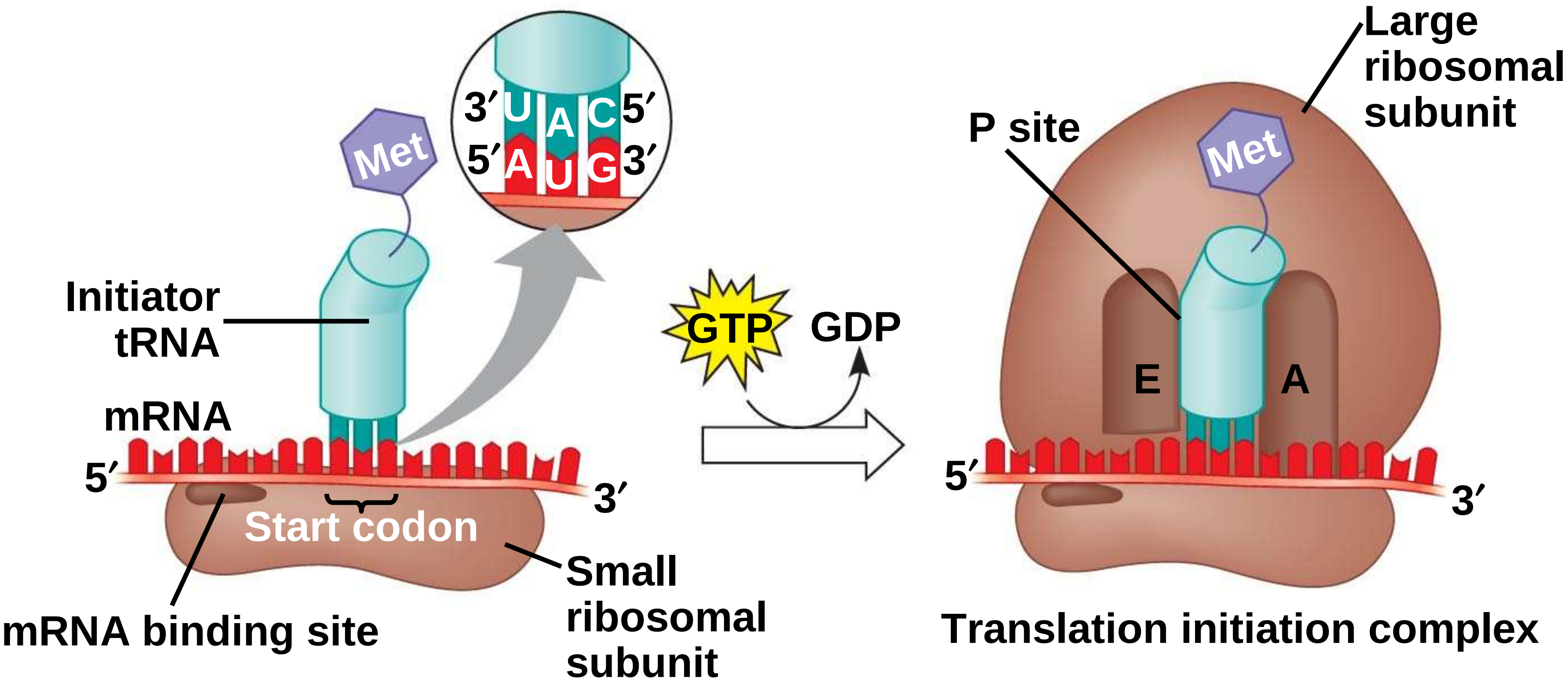
- A ribosome has three binding sites for tRNA:
  - The **P site** holds the tRNA that carries the growing polypeptide chain
  - The **A site** holds the tRNA that carries the next amino acid to be added to the chain
  - The **E site** is the exit site, where discharged tRNAs leave the ribosome

Building a Polypeptide

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

Fig. 17-17

The initiation of translation



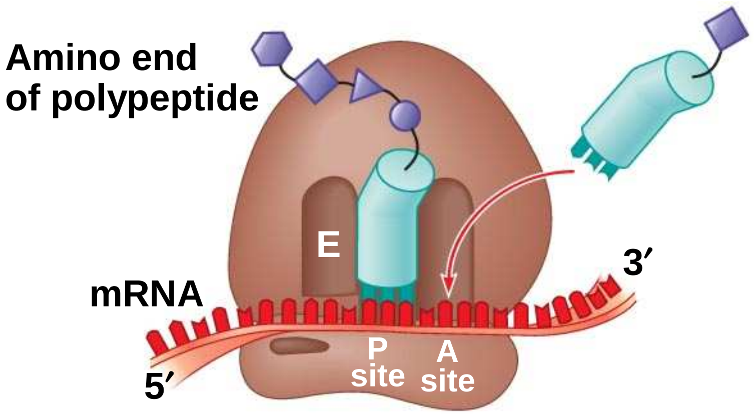
Ribosome Association and Initiation of Translation

- The initiation stage of translation brings together mRNA, a tRNA with the first amino acid, and the two ribosomal subunits
  - First, a small ribosomal subunit binds with mRNA and a special initiator tRNA
  - Then the small subunit moves along the mRNA until it reaches the start codon (AUG)
  - Proteins called initiation factors bring in the large subunit that completes the translation initiation complex

Elongation of the Polypeptide Chain

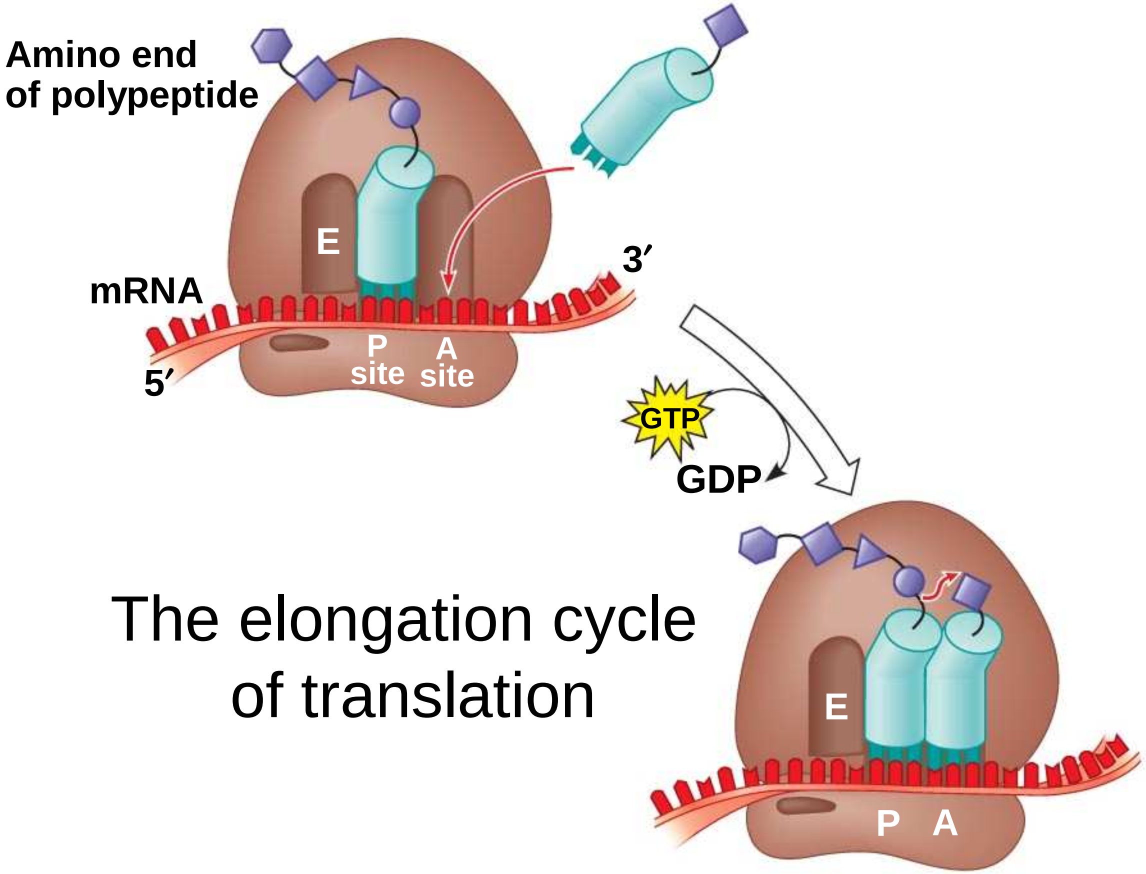
- During the elongation stage, amino acids are added one by one to the preceding amino acid
  - Each addition involves proteins called elongation factors and occurs in three steps:
    - Codon recognition,
    - Peptide bond formation, and
    - Translocation

Fig. 17-18-1



The elongation cycle of translation

Fig. 17-18-2



The elongation cycle of translation

Fig. 17-18-3

The elongation cycle of translation

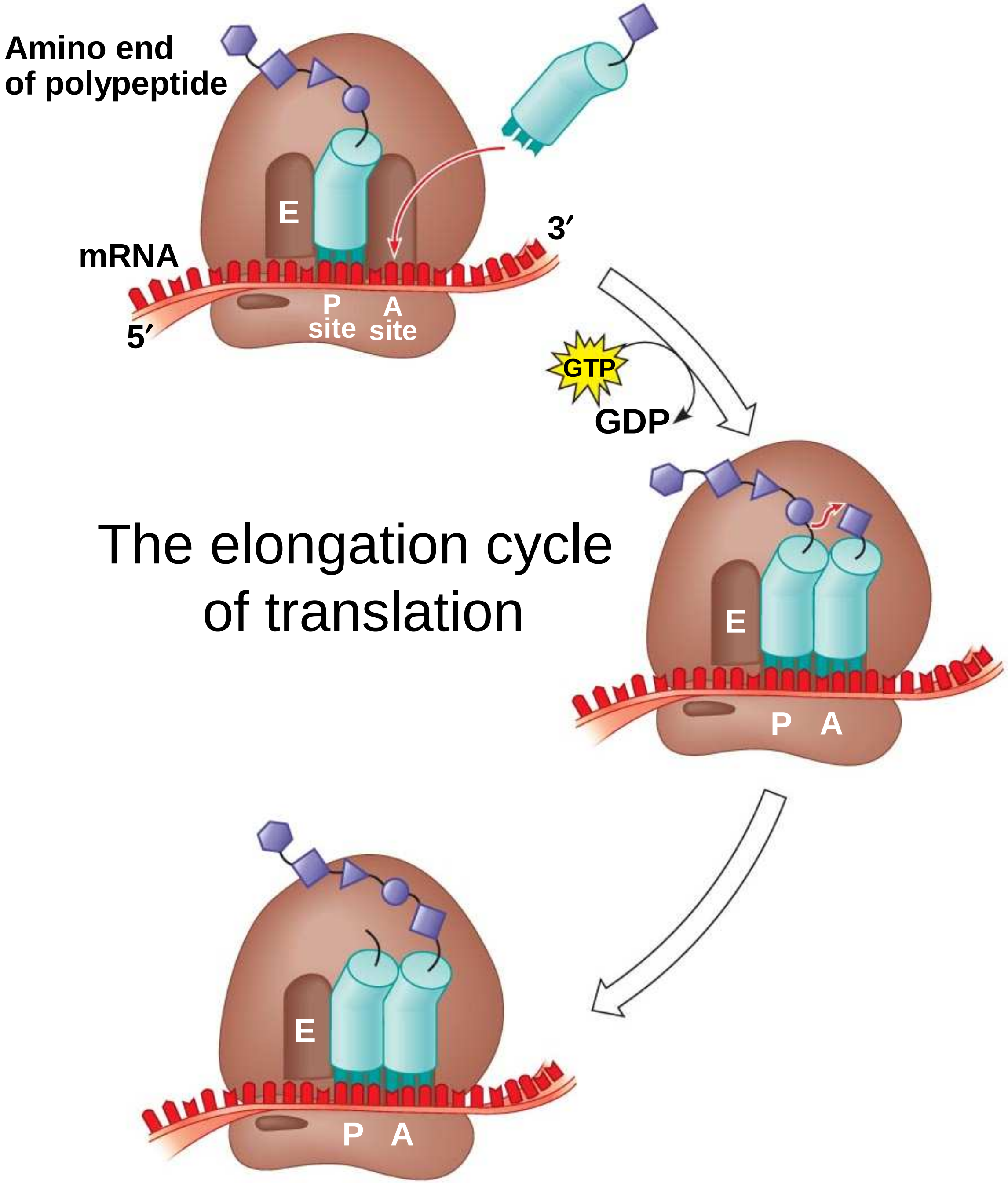
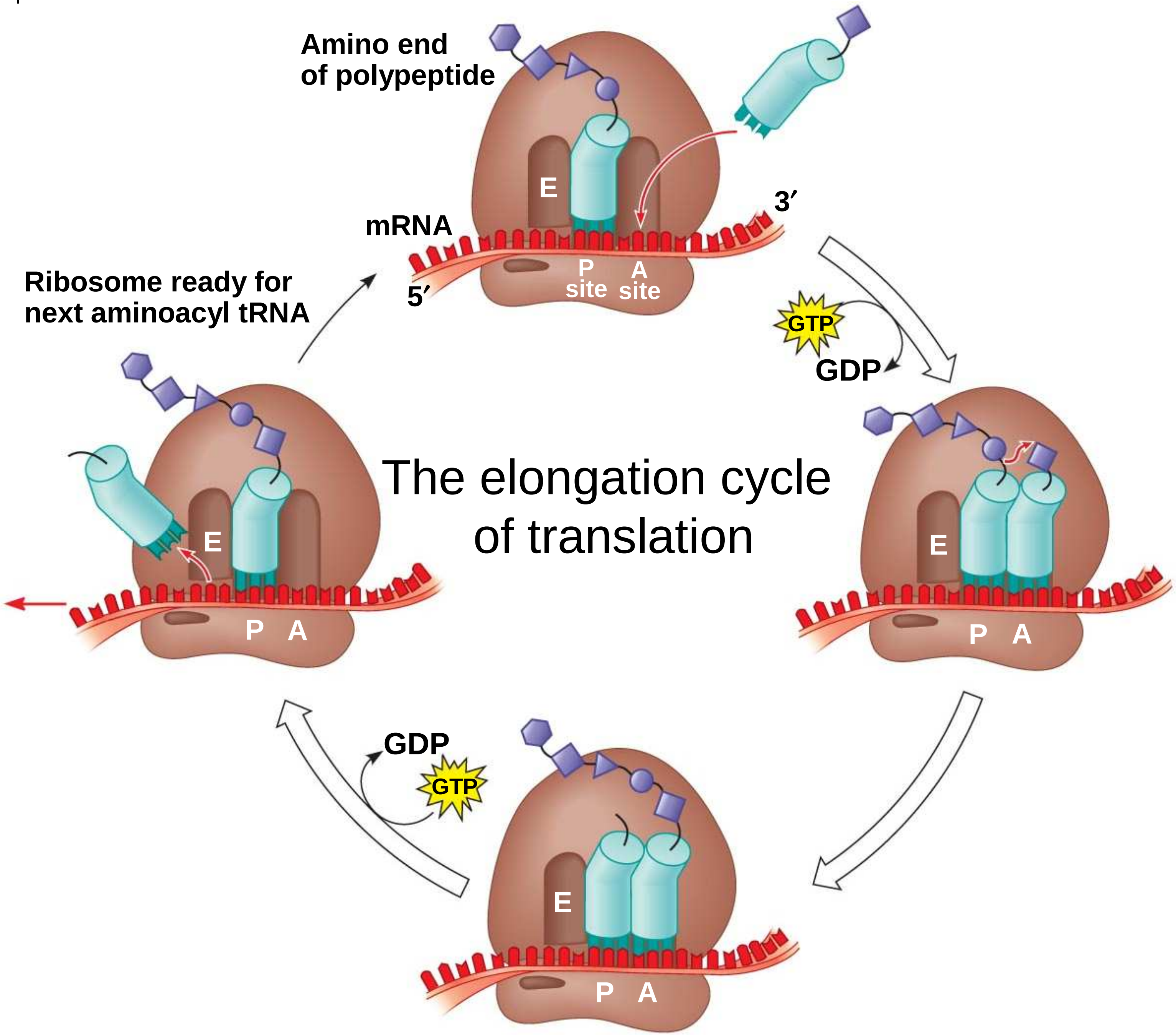


Fig. 17-18-4

The elongation cycle of translation



Termination of Translation

- Termination occurs when a stop codon in the mRNA reaches the A site of the ribosome
  - The A site accepts a protein called a release factor
  - The release factor causes the addition of a water molecule instead of an amino acid
  - This reaction releases the polypeptide, and the translation assembly then comes apart

PLAY Animation: Translation

Fig. 17-19-1

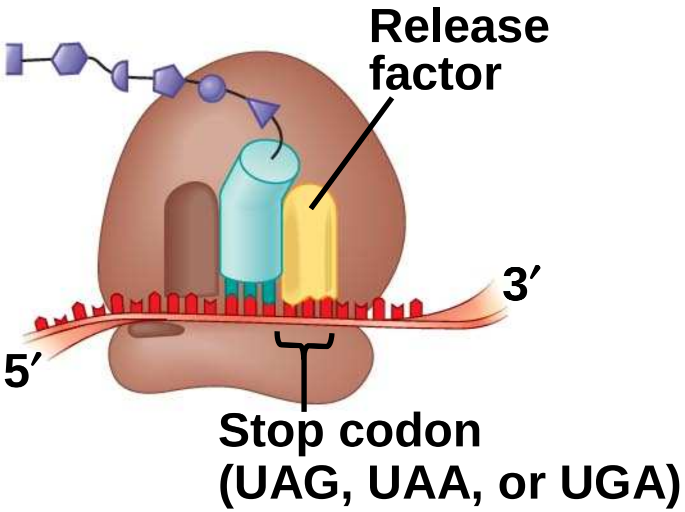


Fig. 17-19-2

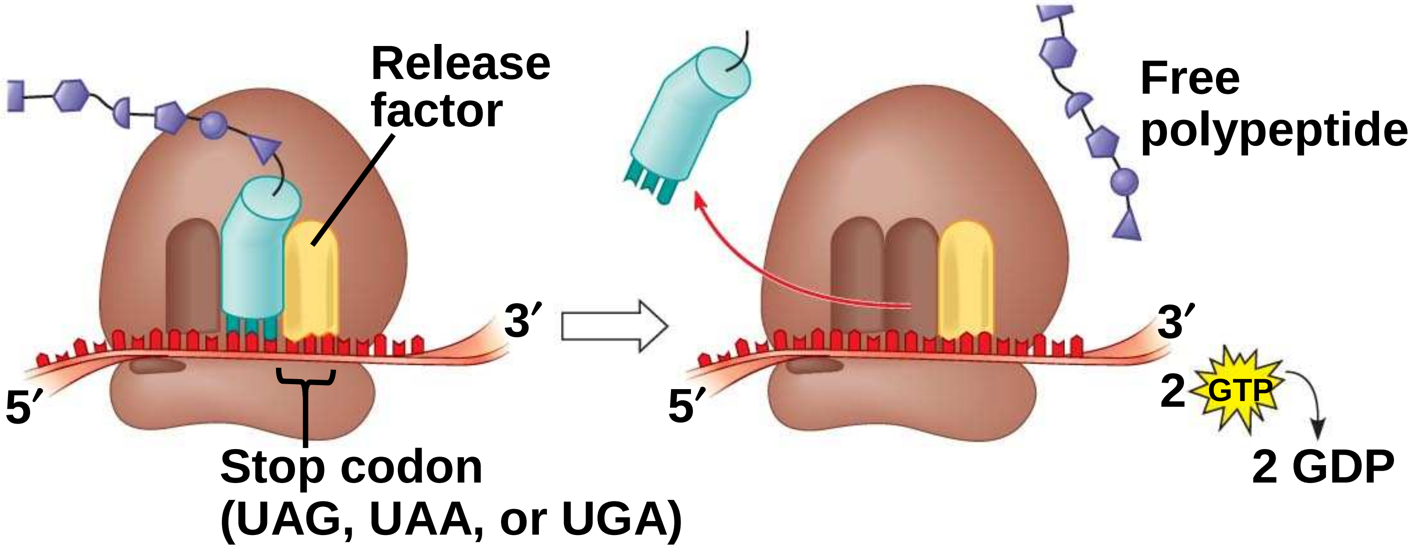
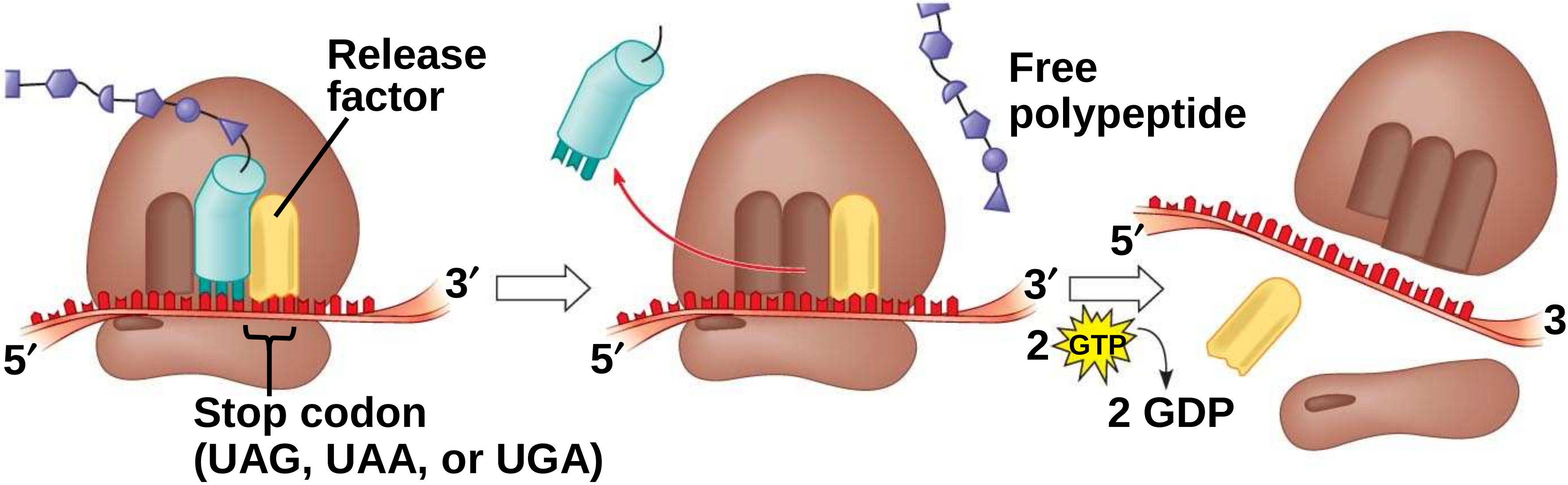

Release factor
Free polypeptide
3′
5′
3′
5′
Stop codon (UAG, UAA, or UGA)
2 GTP
2 GDP

Fig. 17-19-3

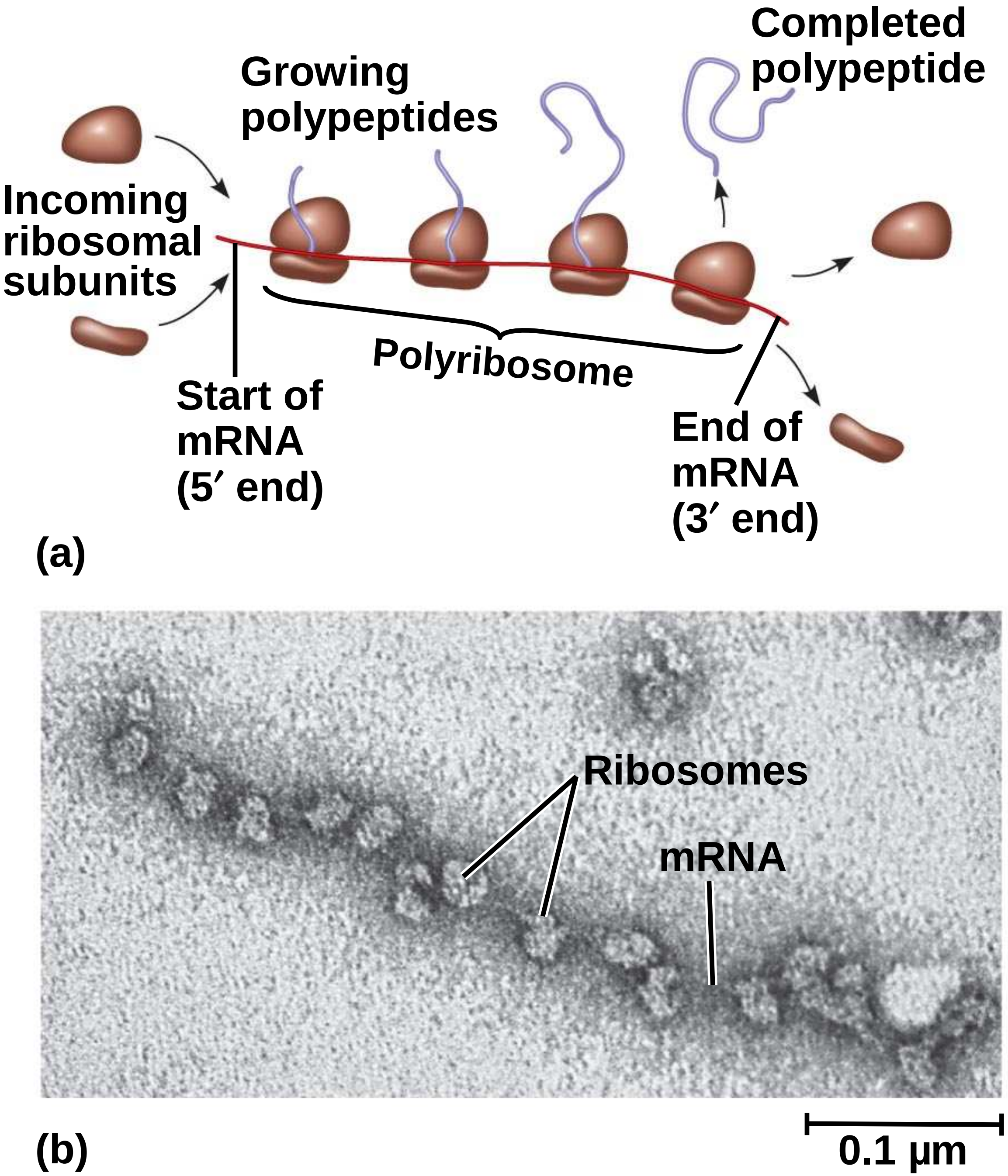
The termination of translation



Polyribosomes

- A number of ribosomes can translate a single mRNA simultaneously, forming a **polyribosome** (or **polysome**)
- Polyribosomes enable a cell to make many copies of a polypeptide very quickly

Fig. 17-20


Completed polypeptide
Growing polypeptides
Incoming ribosomal subunits
Polyribosome
Start of mRNA (5′ end)
End of mRNA (3′ end)
(a)
Ribosomes
mRNA
(b)
0.1 µm

Completing and Targeting the Functional Protein

- Often translation is not sufficient to make a functional protein
- Polypeptide chains are modified after translation
  - Example: cleavage, addition of other entities (i.e. glyco-, lipo-protein)
- Completed proteins are targeted to specific sites in the cell
  - Example: membrane protein, nuclear protein

Protein Folding and Post-Translational Modifications

- 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 ribsomes (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

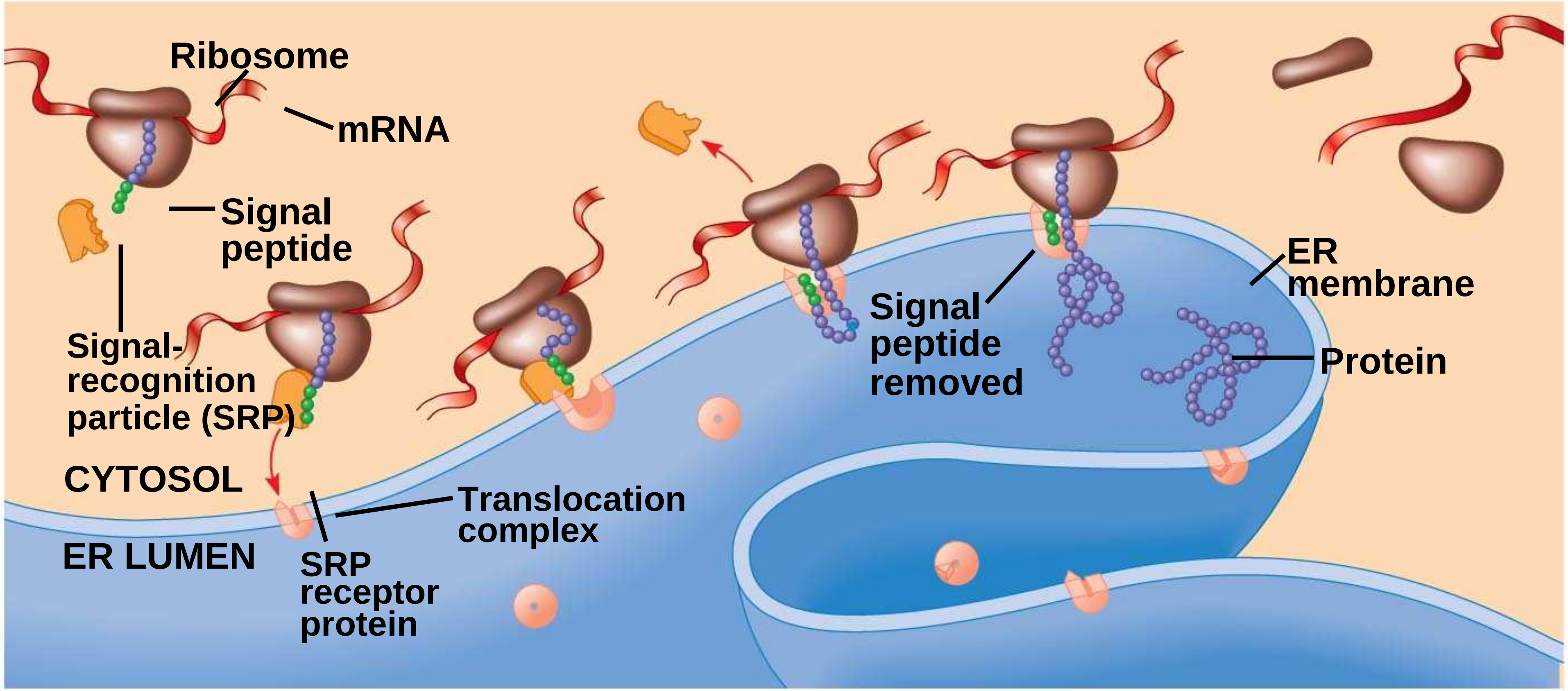
- Polypeptide synthesis always begins in the cytosol

- Synthesis finishes in the cytosol *unless* the polypeptide signals the ribosome to attach to the ER

- Polypeptides destined for the ER or for secretion are marked by a **signal peptide**

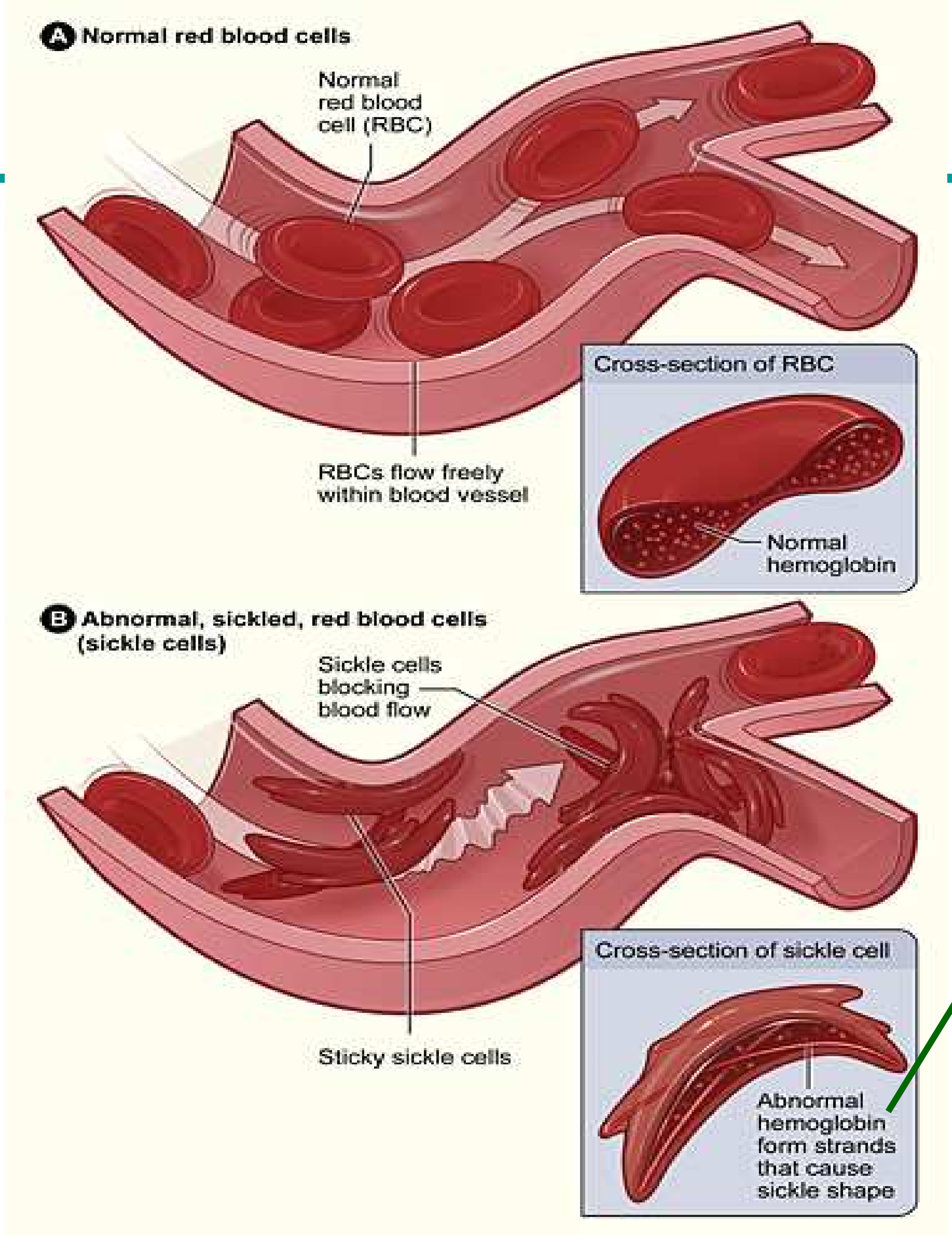
- A **signal-recognition particle (SRP)** binds to the signal peptide
- The SRP brings the signal peptide and its ribosome to the ER

Fig. 17-21


Ribosome
mRNA
Signal peptide
Signal-recognition particle (SRP)
CYTOSOL
ER LUMEN
SRP receptor protein
Translocation complex
Signal peptide removed
ER membrane
Protein

Concept 17.5: Point mutations can affect protein structure and function

- **Mutations** are changes in the genetic material of a cell or virus
- **Point mutations** are chemical changes in just one base pair of a gene
- The change of a single nucleotide in a DNA template strand can lead to the production of an abnormal protein


A Normal red blood cells
Normal red blood cell (RBC)
RBCs flow freely within blood vessel
Cross-section of RBC
Normal hemoglobin
B Abnormal, sickled, red blood cells (sickle cells)
Sickle cells blocking blood flow
Sticky sickle cells
Cross-section of sickle cell
Abnormal hemoglobin form strands that cause sickle shape

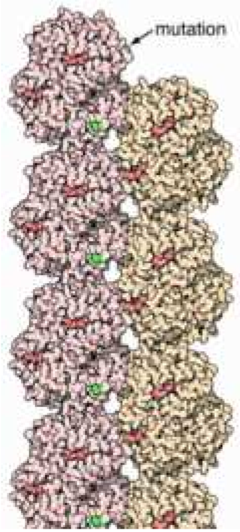
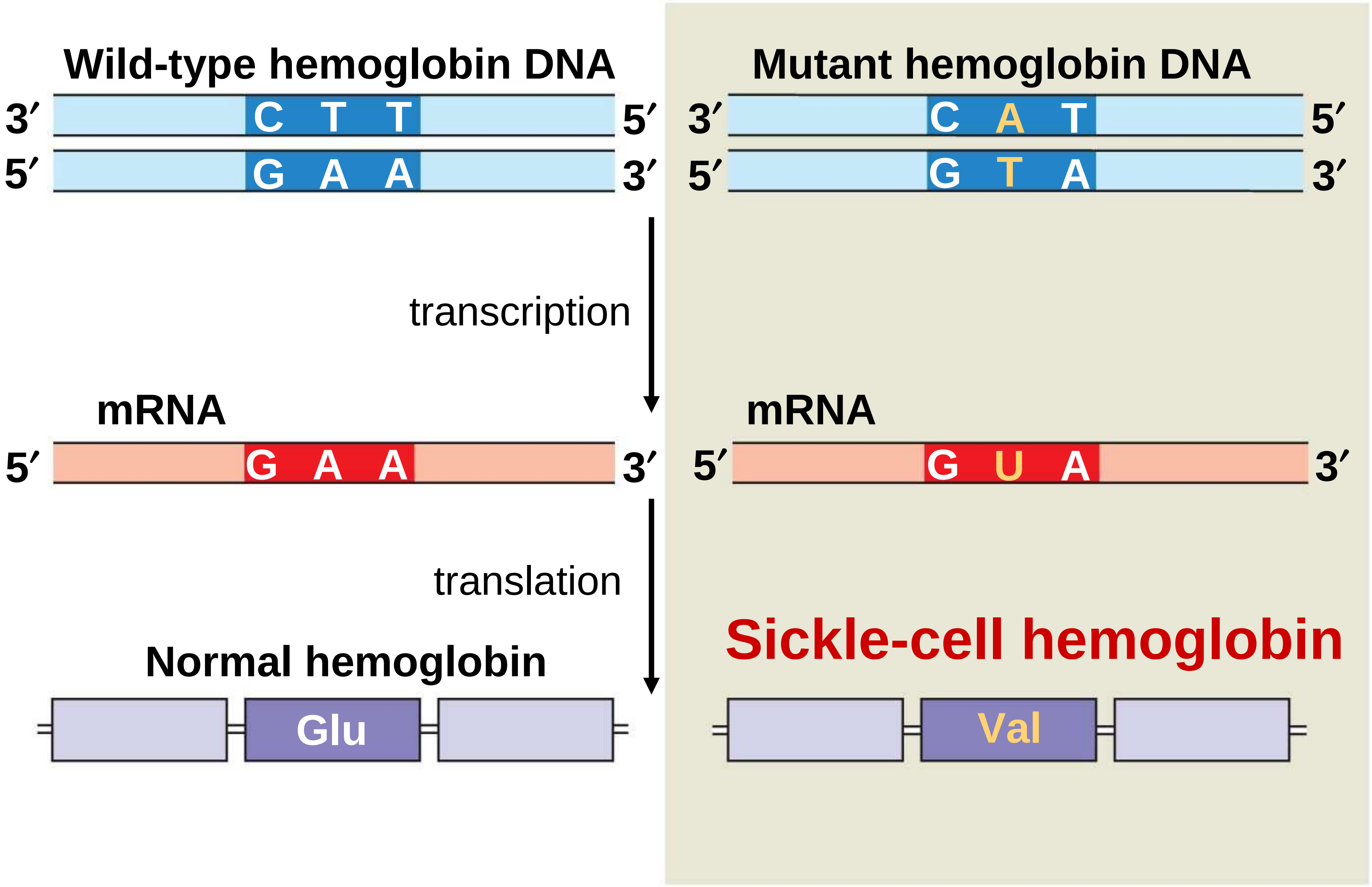

mutation

Fig. 17-22



Wild-type hemoglobin DNA

3′ C T T 5′

5′ G A A 3′

transcription

mRNA

5′ G A A 3′

translation

Normal hemoglobin

Glu

Mutant hemoglobin DNA

3′ C A T 5′

5′ G T A 3′

mRNA

5′ G U A 3′

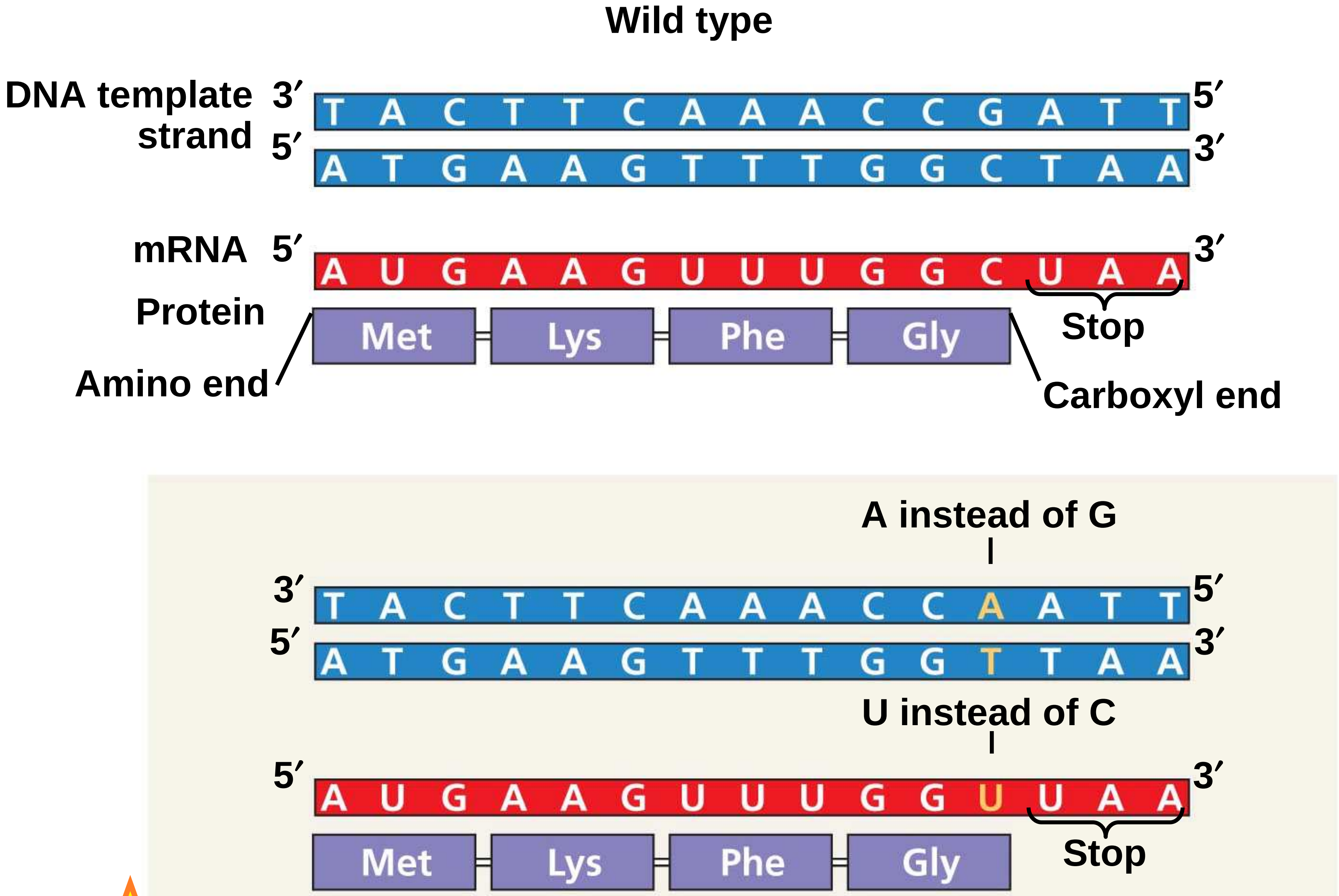
Sickle-cell hemoglobin

Val

Types of Point Mutations

- Point mutations within a gene can be divided into two general categories
  - Base-pair substitutions
  - Base-pair insertions or deletions (indel)

Fig. 17-23a



Wild type

DNA template strand

3′ T A C T T C A A A C C G A T T 5′

5′ A T G A A G T T T G G C T A A 3′

mRNA 5′ A U G A A G U U U G G C U A A 3′

Protein: Met Lys Phe Gly Stop

Amino end

Carboxyl end

A instead of G

3′ T A C T T C A A A C C A A T T 5′

5′ A T G A A G T T T G G T T A A 3′

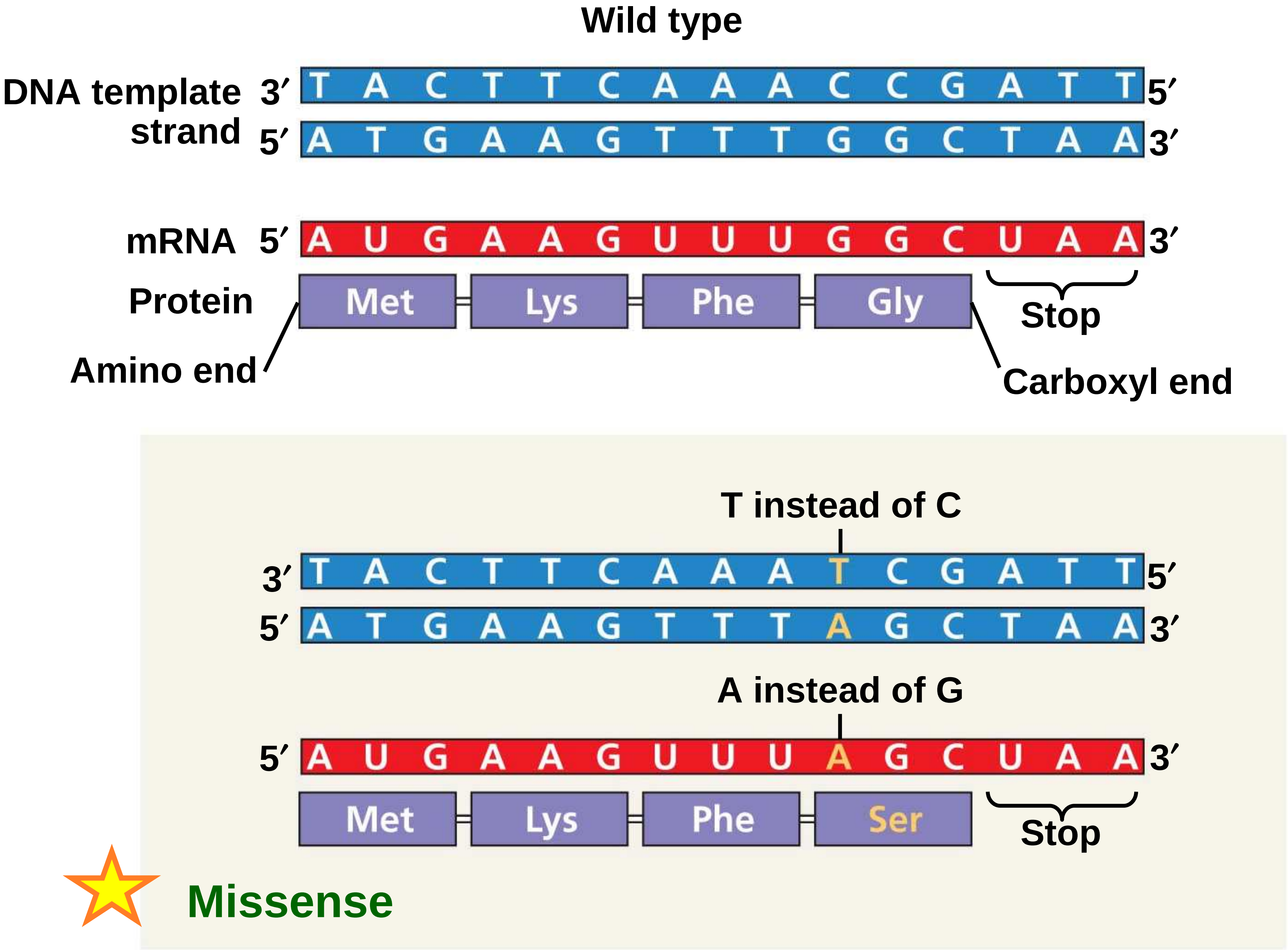
U instead of C

5′ A U G A A G U U U G G U U A A 3′

Met Lys Phe Gly Stop

Silent (no effect on amino acid sequence)

Fig. 17-23b



Wild type

DNA template strand 3′ T A C T T C A A A C C G A T T 5′

5′ A T G A A G T T T G G C T A A 3′

mRNA 5′ A U G A A G U U U G G C U A A 3′

Protein Met Lys Phe Gly Stop

Amino end

Carboxyl end

T instead of C

3′ T A C T T C A A A T C G A T T 5′

5′ A T G A A G T T T A G C T A A 3′

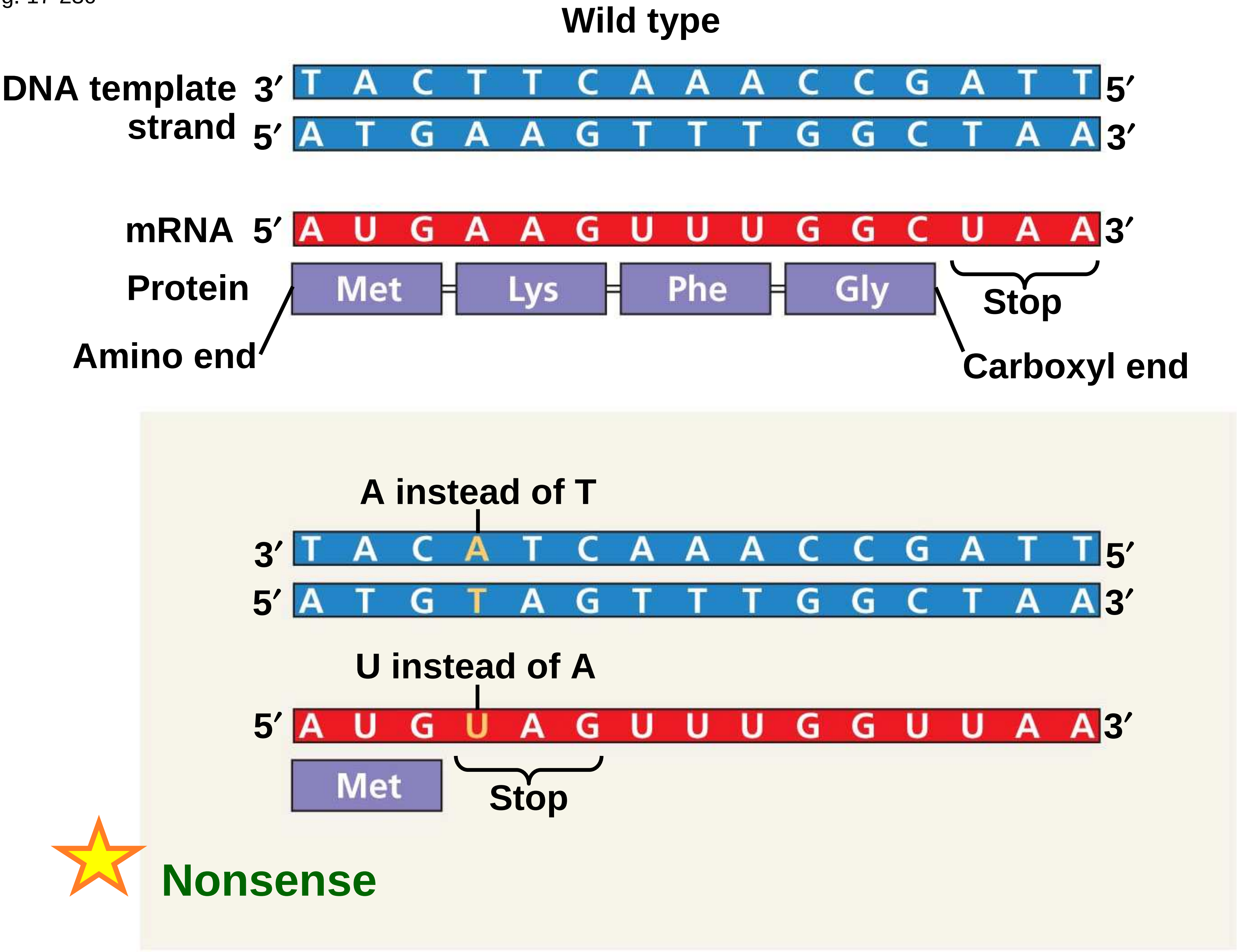
A instead of G

5′ A U G A A G U U U A G C U A A 3′

Met Lys Phe Ser Stop

Missense

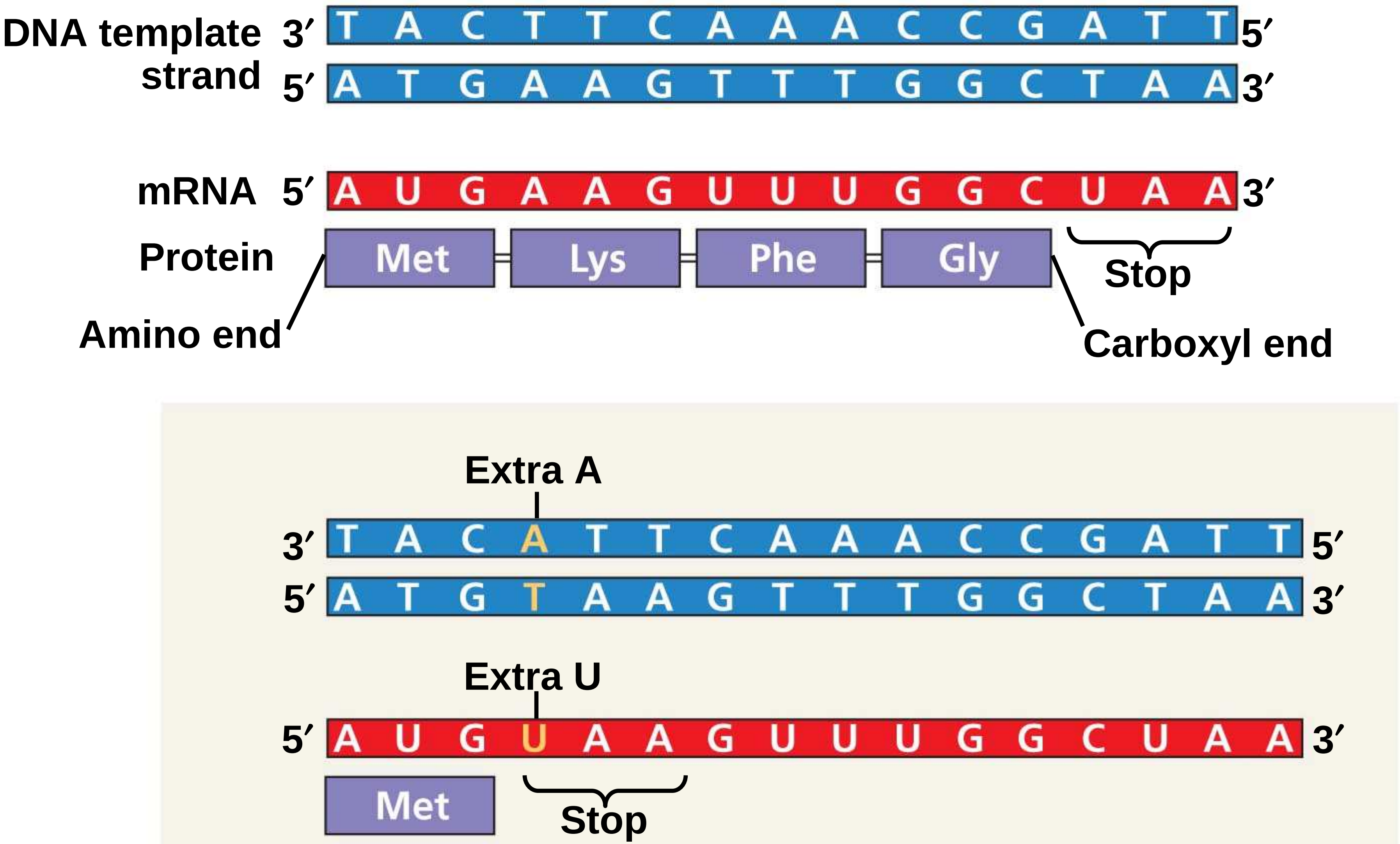
Fig. 17-23c



Nonsense

Fig. 17-23d

Wild type



DNA template strand 3′ T A C T T C A A A C C G A T T 5′

5′ A T G A A G T T T G G C T A A 3′

mRNA 5′ A U G A A G U U U G G C U A A 3′

Protein Met Lys Phe Gly Stop

Amino end

Carboxyl end

Extra A

3′ T A C A T T C A A A C C G A T T 5′

5′ A T G T A A G T T T G G C T A A 3′

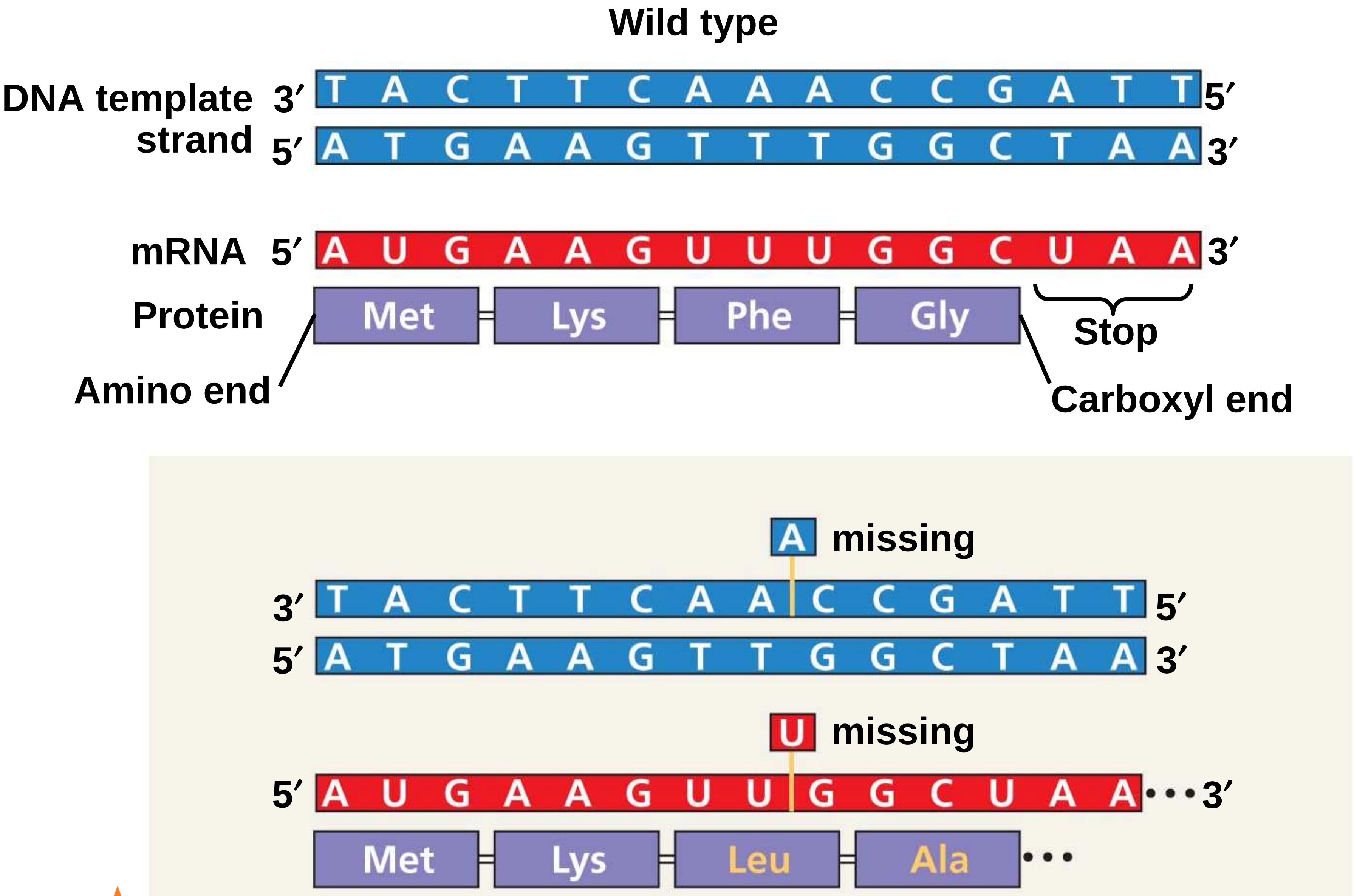
Extra U

5′ A U G U A A G U U U G G C U A A 3′

Met Stop

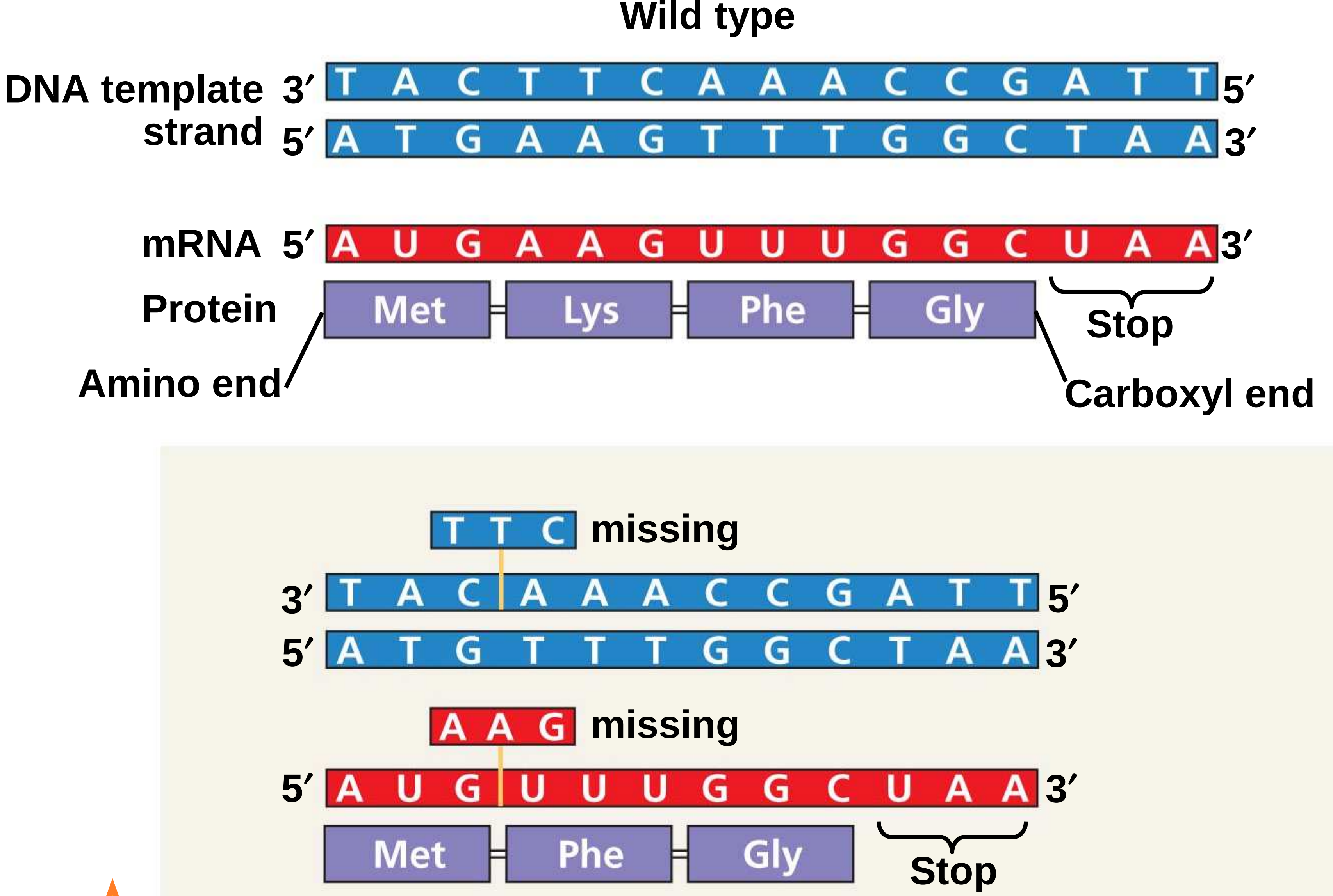
☆ Frameshift causing immediate nonsense (1 base-pair insertion)

Fig. 17-23e



Frameshift causing extensive missense (1 base-pair deletion)

Fig. 17-23f



Wild type

DNA template strand 3′ T A C T T C A A A C C G A T T 5′

5′ A T G A A G T T T G G C T A A 3′

mRNA 5′ A U G A A G U U U G G C U A A 3′

Protein Met Lys Phe Gly Stop

Amino end

Carboxyl end

T T C missing

3′ T A C A A A C C G A T T 5′

5′ A T G T T T G G C T A A 3′

A A G missing

5′ A U G U U U G G C U A A 3′

Met Phe Gly Stop

No frameshift, but one amino acid missing (3 base-pair deletion)

Substitutions

- A **base-pair substitution** replaces one nucleotide and its partner with another pair of nucleotides
  - **Silent mutations** have no effect on the amino acid produced by a codon because of redundancy in the genetic code
  - **Missense mutations** still code for an amino acid, but not necessarily the right amino acid
  - **Nonsense mutations** change an amino acid codon into a stop codon, nearly always leading to a nonfunctional protein

Insertions and Deletions

- **Insertions** and **deletions** are additions or losses of nucleotide pairs in a gene
- These mutations have a disastrous effect on the resulting protein more often than substitutions do
- Insertion or deletion of nucleotides may alter the reading frame, producing a **frameshift mutation**

Mutagens

- Spontaneous mutations can occur during DNA replication, recombination, or repair
- **Mutagens** are physical or chemical agents that can cause mutations

Concept 17.6: While gene expression differs among the domains of life, the concept of a gene is universal

- Archaea **[ahr**-kee-on**]** are prokaryotes, but share many features of gene expression with eukaryotes

Comparing Gene Expression in Bacteria, Archaea, and Eukarya

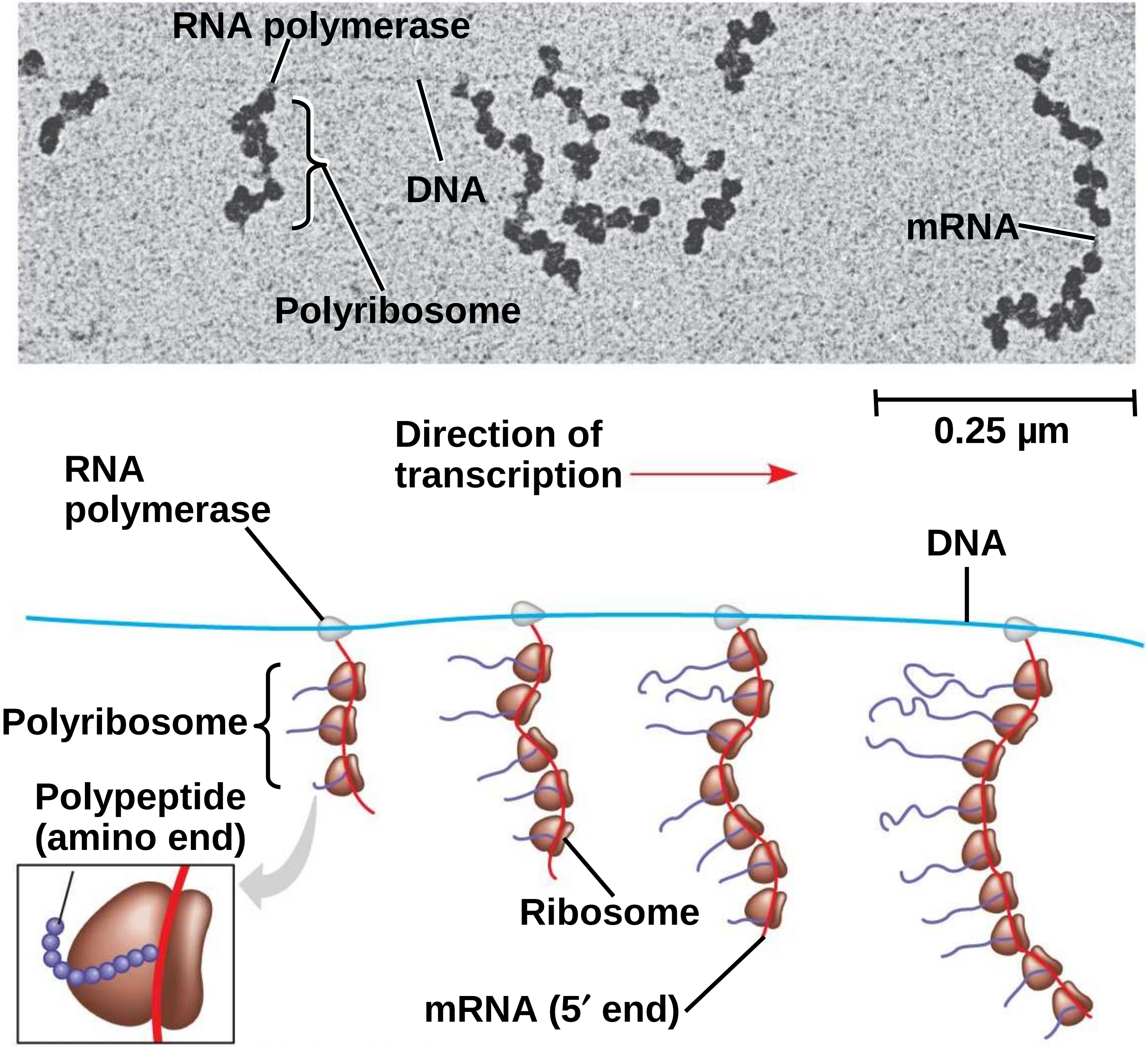
- **Bacteria** and **eukarya** differ in their RNA polymerases, termination of transcription and ribosomes; **archaea** tend to resemble **eukarya** in these respects

- **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

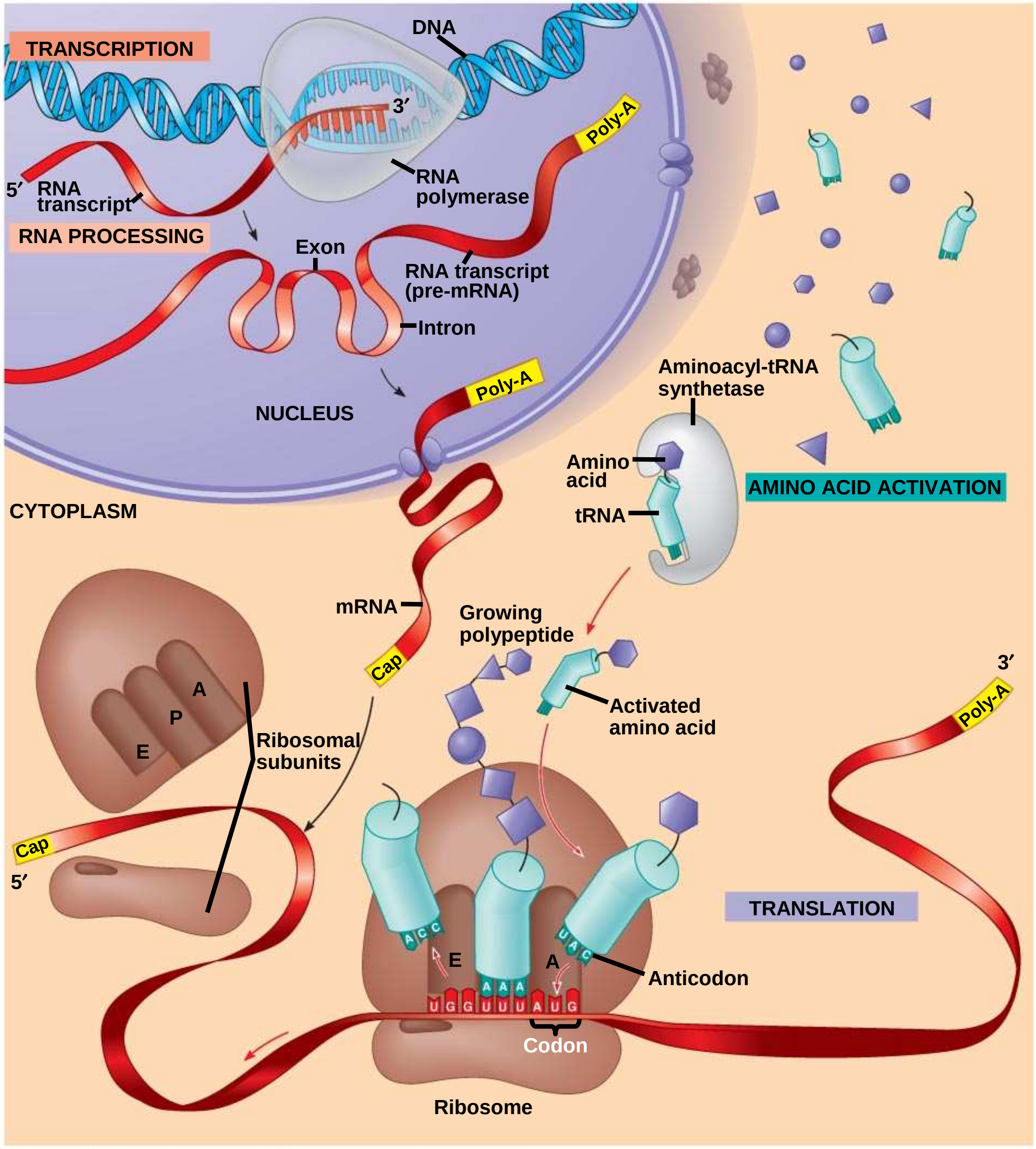
Fig. 17-24


RNA polymerase
DNA
Polyribosome
mRNA
0.25 µm
Direction of transcription
RNA polymerase
DNA
Polyribosome
Polypeptide (amino end)
Ribosome
mRNA (5′ end)

What Is a Gene? *Revisiting the Question*

- The idea of the gene itself is a unifying concept of life
- We have considered a gene as:
  - A discrete unit of inheritance
  - A region of specific nucleotide sequence in a chromosome
  - A DNA sequence that codes for a specific polypeptide chain

Fig. 17-25


TRANSCRIPTION
DNA
3′
RNA polymerase
5′
RNA transcript
RNA PROCESSING
Exon
RNA transcript (pre-mRNA)
Intron
Poly-A
Poly-A
NUCLEUS
Aminoacyl-tRNA synthetase
Amino acid
tRNA
AMINO ACID ACTIVATION
CYTOPLASM
mRNA
Cap
Growing polypeptide
Activated amino acid
3′
Poly-A
A
P
E
Ribosomal subunits
Cap
5′
A C C
E
A A A
A
U A C
Anticodon
U G G U U U A U G
Codon
TRANSLATION
Ribosome

- In summary, a gene can be defined as a region of DNA that can be expressed to produce a final functional product, either a polypeptide or an RNA molecule

You should now be able to:

1. Describe the contributions made by Garrod, Beadle, and Tatum to our understanding of the relationship between genes and enzymes
2. Briefly explain how information flows from gene to protein
3. Compare transcription and translation in bacteria and eukaryotes
4. Explain what it means to say that the genetic code is redundant and unambiguous

5. Include the following terms in a description of transcription: mRNA, RNA polymerase, the promoter, the terminator, the transcription unit, initiation, elongation, termination, and introns
6. Include the following terms in a description of translation: tRNA, wobble, ribosomes, initiation, elongation, and termination