

## Chapter 4

# Expression of Genetic Information



# Genetic Code

- Information in DNA → amino acid sequence in protein

## mRNA

**5' GCA GCA CUA GGA GAG AAG 3'**

- Codon: triplet of RNA bases

20 amino acids in nature

Codon (nt)

1

2

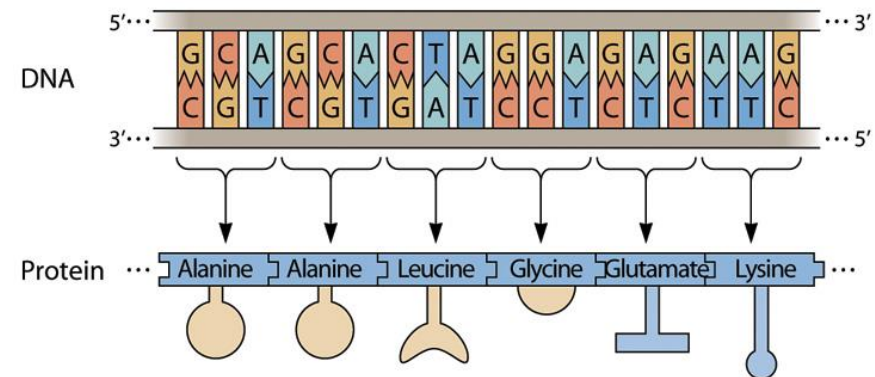
3

Amino acid

4

16

64



# Genetic Code

		Second base				
		U	C	A	G	
First base	U	<b>UUU</b> } Phenyl- alanine <b>UUC</b> } <b>UUA</b> } Leucine <b>UUG</b> }	<b>UCU</b> } <b>UCC</b> } Serine <b>UCA</b> } <b>UCG</b> }	<b>UAU</b> } Tyrosine <b>UAC</b> } <b>UAA</b> } Stop codon <b>UAG</b> } Stop codon	<b>UGU</b> } Cysteine <b>UGC</b> } <b>UGA</b> } Stop codon <b>UGG</b> } Tryptophan	U C A G
	C	<b>CUU</b> } <b>CUC</b> } Leucine <b>CUA</b> } <b>CUG</b> }	<b>CCU</b> } <b>CCC</b> } Proline <b>CCA</b> } <b>CCG</b> }	<b>CAU</b> } Histidine <b>CAC</b> } <b>CAA</b> } Glutamine <b>CAG</b> }	<b>CGU</b> } <b>CGC</b> } Arginine <b>CGA</b> } <b>CGG</b> }	U C A G
	A	<b>AUU</b> } <b>AUC</b> } Isoleucine <b>AUA</b> } <b>AUG</b> } Methionine start codon	<b>ACU</b> } <b>ACC</b> } Threonine <b>ACA</b> } <b>ACG</b> }	<b>AAU</b> } Asparagine <b>AAC</b> } <b>AAA</b> } Lysine <b>AAG</b> }	<b>AGU</b> } Serine <b>AGC</b> } <b>AGA</b> } Arginine <b>AGG</b> }	U C A G
	G	<b>GUU</b> } <b>GUC</b> } Valine <b>GUA</b> } <b>GUG</b> }	<b>GCU</b> } <b>GCC</b> } Alanine <b>GCA</b> } <b>GCG</b> }	<b>GAU</b> } Aspartic acid <b>GAC</b> } <b>GAA</b> } Glutamic acid <b>GAG</b> }	<b>GGU</b> } <b>GGC</b> } Glycine <b>GGA</b> } <b>GGG</b> }	U C A G

# Genome

## ■ Gene

- The complete stretch of DNA needed to determine the amino acid sequence of a protein

## ■ Genome

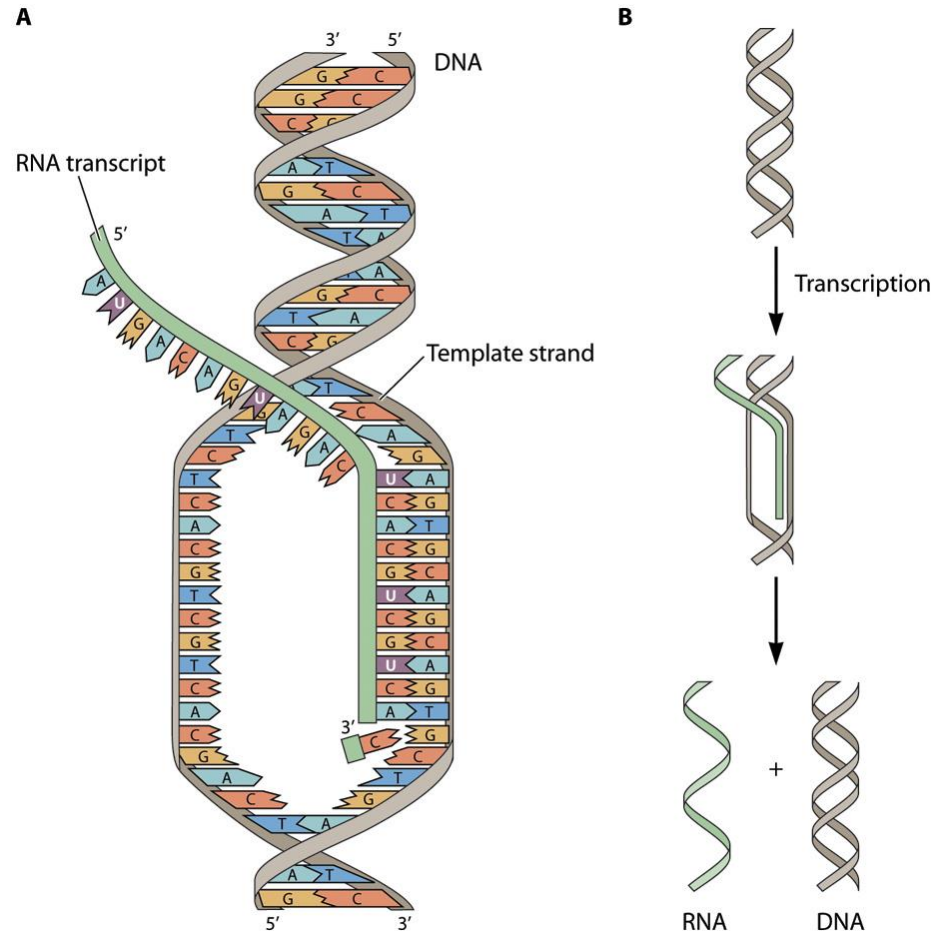
- The complete set of genetic material in an organism
- Human genome project
  - 1990-2003
  - U.S. department of energy and the National Institute of Health
  - $3.2 \times 10^9$  bp, 19,000~20,000 genes
  - 98% is noncoding DNA

# Protein Synthesis

- Transcription
  - From DNA to mRNA (messenger RNA)
- Translation
  - From mRNA to protein
  - tRNA (transfer RNA) matches the codon to amino acid
  - Ribosome (made of Proteins and RNAs)
    - Protein synthesis

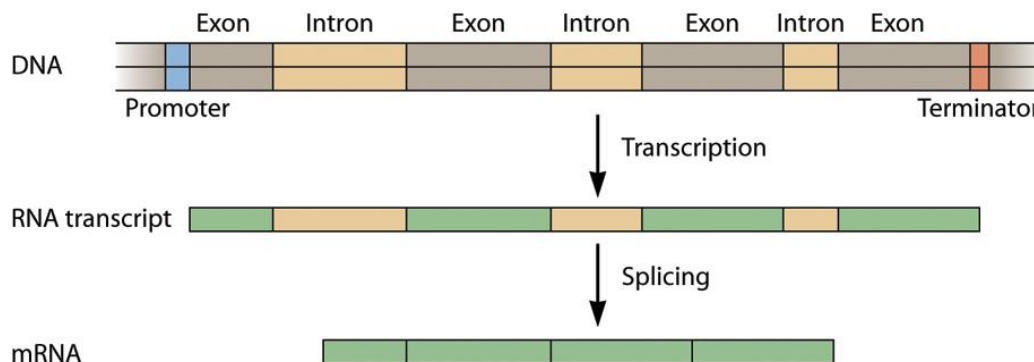
# Transcription

- RNA synthesis using only one strand as a template
  - mRNA → encode protein
  - Ribosomal RNA (rRNA) and tRNA → no translation
- RNA polymerase
  - mRNA synthesis

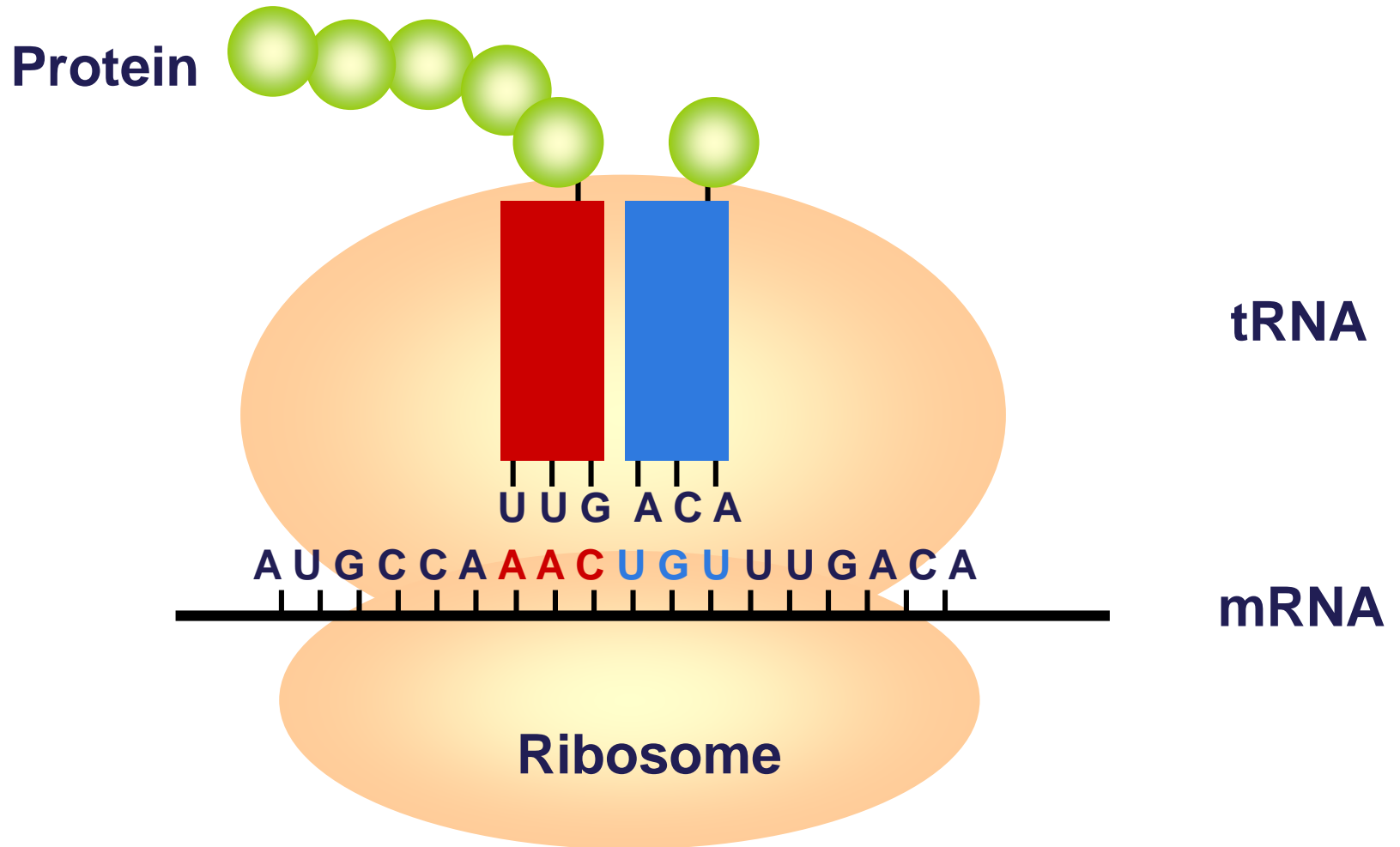


# Regulation of Transcription

- Promoter
  - Binding site of RNA polymerase and regulatory proteins (transcriptional regulator; activator or repressor)
- Terminator
  - The site where transcription stops
- Processing of eukaryotic RNA
  - Splicing : joining of exons



# Translation

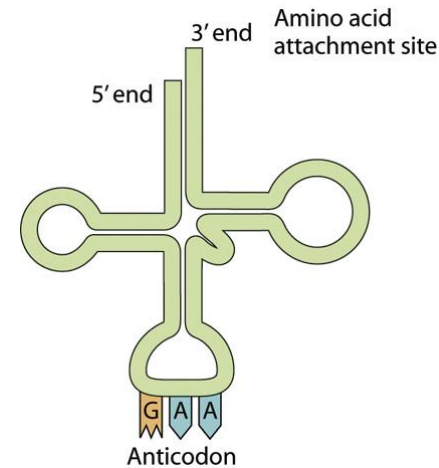




# Translation

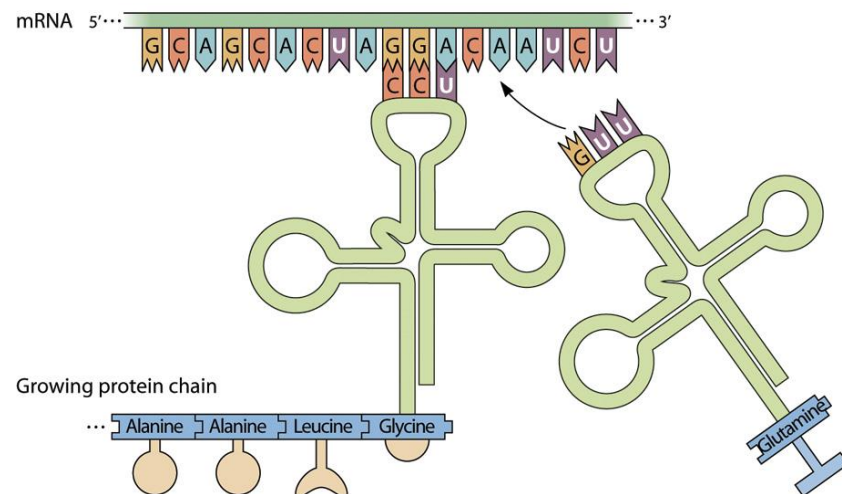
## ■ tRNA

- Cloverleaves shaped folding
- Anticodon: complementary to codon
- 3' end: amino acid attachment



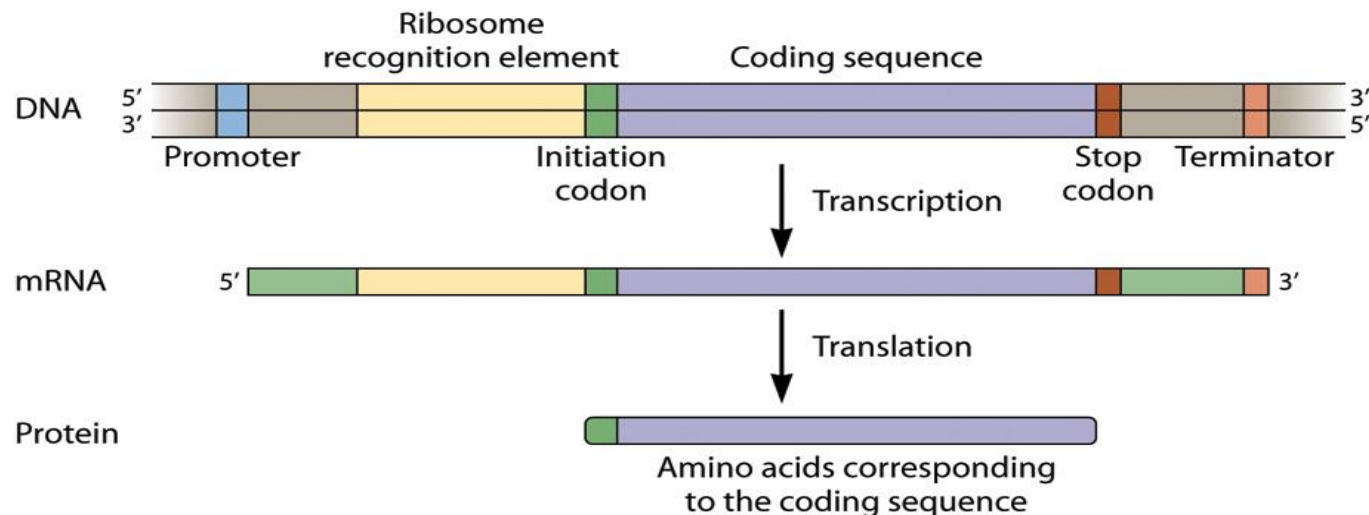
## ■ Translation

- Assembly of ribosome and mRNA
- Binding of tRNA
- Peptide bond formation
- Release of tRNA



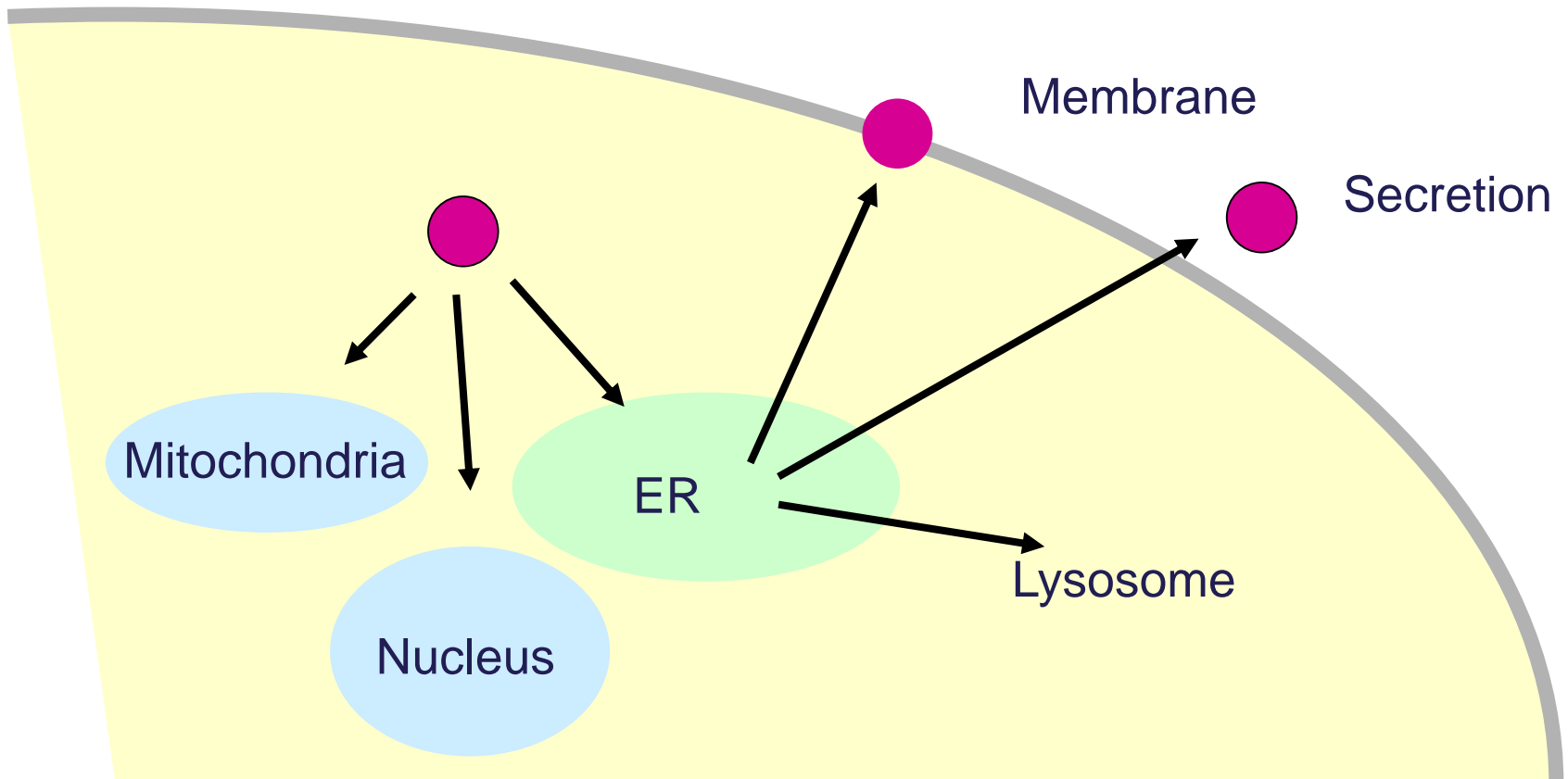
# Signals for Transcription and Translation

- Ribosome binding site in mRNA
  - Shine-Dalgarno Sequence
- Initiation codon
  - AUG for methionine
- Stop codon
  - UGA, UAA, UAG : no corresponding tRNA

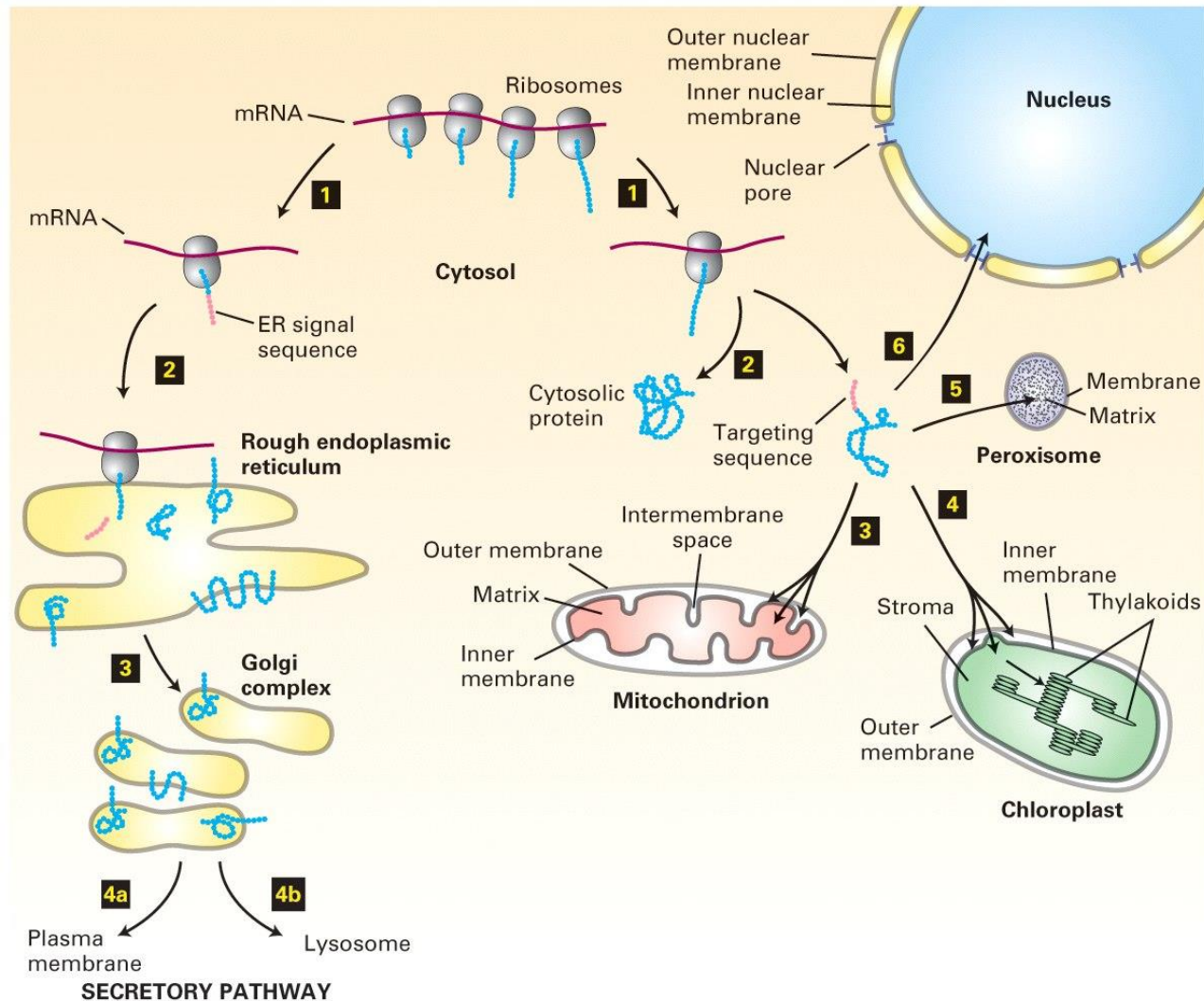


# Cellular Fate of Proteins

- Protein Targeting to specific compartment (ER, Nucleus, Mitochondria ) is guided by **signal peptide (tags)**

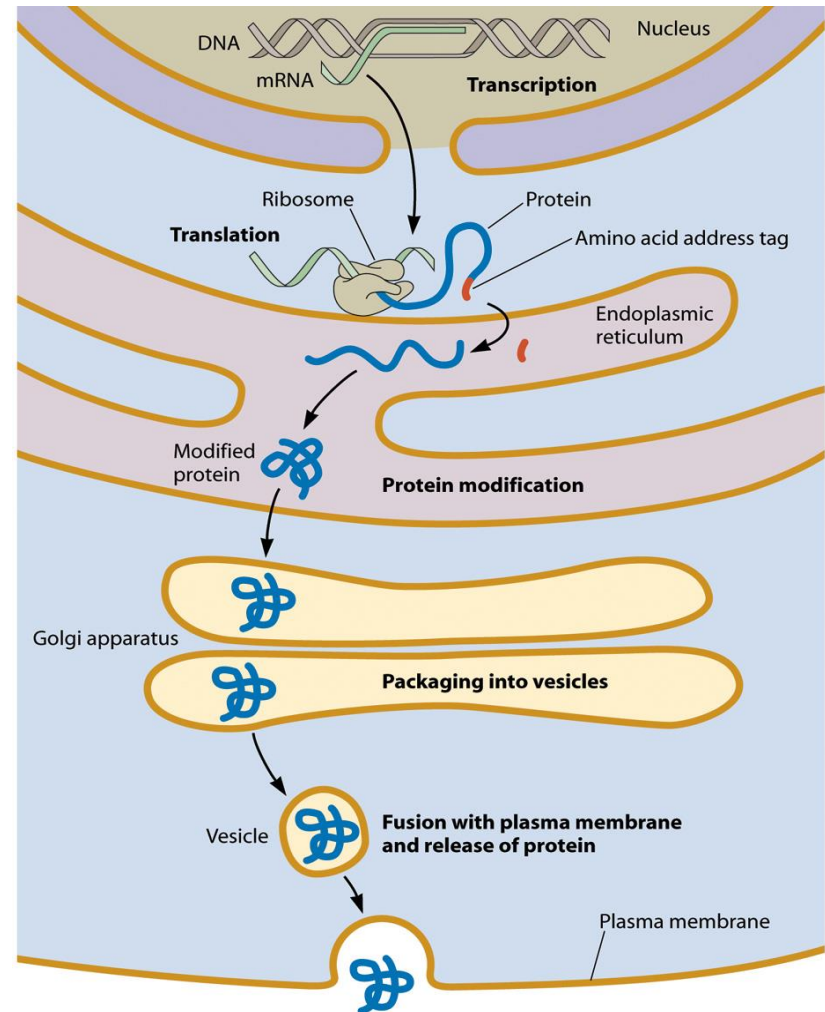


# Overview of Protein Sorting Pathway



# Protein Targeting to ER

- Proteins with signal peptide
  - Secretory proteins
  - Membrane proteins
  - Proteins in ER, Golgi, and lysosome
- Modification during transport from ER to Golgi apparatus
  - Glycosylation



# Mutations

## ■ Mutation

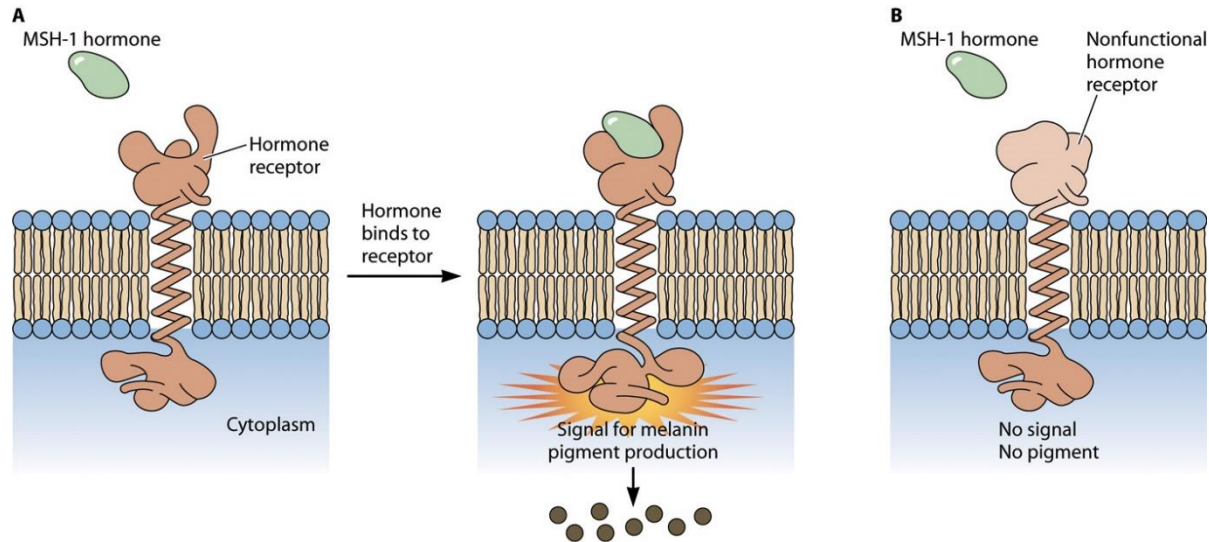
- Any change in a DNA sequence
  - During normal cellular processes
    - Error of DNA polymerase
    - Transposition (Chapter 13)
  - Environmental factors
    - DNA damage by UV or chemicals
- Source of genetic variation and evolution

## ■ Types of mutation

- Silent mutation: : mutation with no effect on a protein (the codon encoding the same amino acid)
- Mutations having slight effect : mutation in non-functional domain of a protein
- Mutations affecting protein function
  - Promoter or ribosome binding sequence : no protein synthesis
  - Essential protein sequence

# Mutations

- No effect on survival
  - e.g. hair color





# Mutations

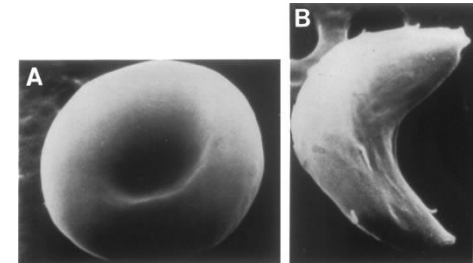
- Harmful

- e.g. sickle-cell anemia

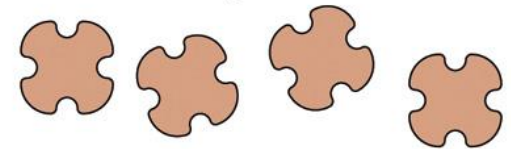
- A to T mutation of hemoglobin

- 6<sup>th</sup> amino acid change from  
glu to val

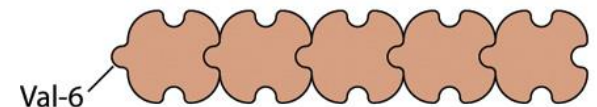
- hydrophobic aggregation of  
hemoglobin



**A.** Normal hemoglobin



**B.** Sickle-cell hemoglobin



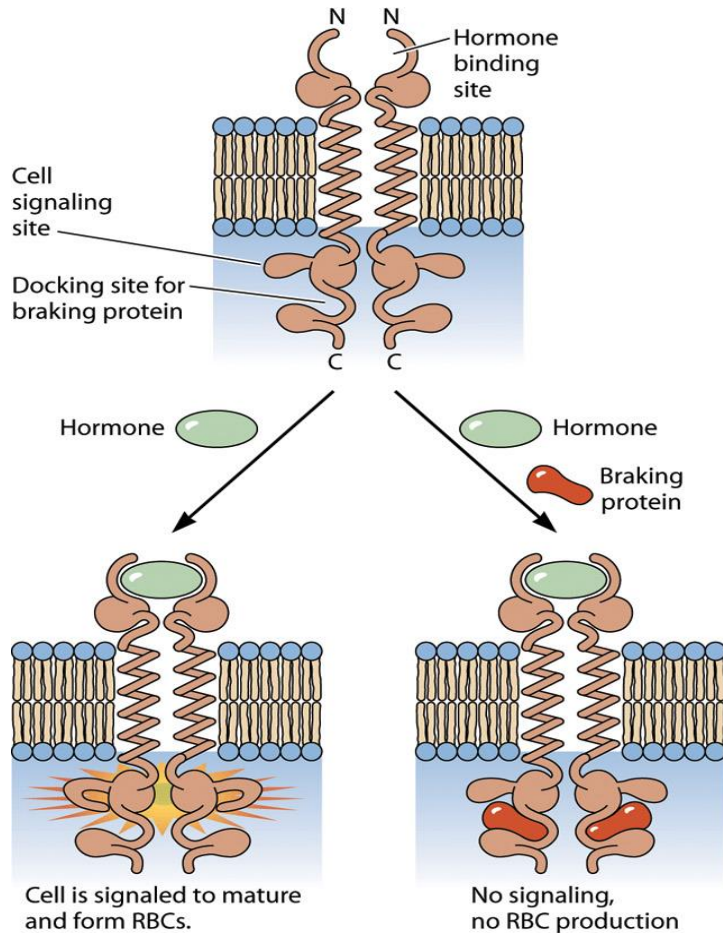


# Mutations

- Benign erythrocytosis
  - Elevated levels of RBC
  - Mutation in erythropoietin receptor
    - 481 TGG to TAG (stop codon)
  - Deletion of 70 amino acids for repression of signal transduction
    - More RBC production from bone marrow stem cells
    - Greatly enhanced stamina
    - (Finnish athlete Eero Mäntyranta won three gold medals for cross-country skiing in the 1964 Winter Olympics)

# Mutations

**A. Normal receptor protein**



**B. Mutant receptor protein**

