Chapter 4

# Expression of Genetic Information

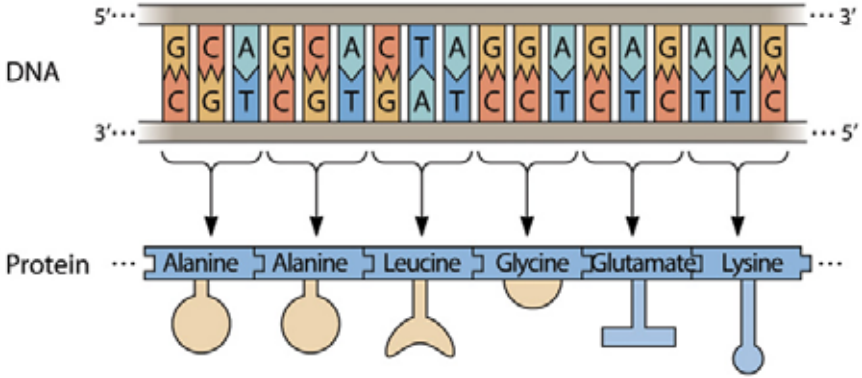
# Genetic Code

- Information in DNA → amino acid sequence in protein

**mRNA**

**5’ GCA GCA CUA GGA GAG AAG 3’**

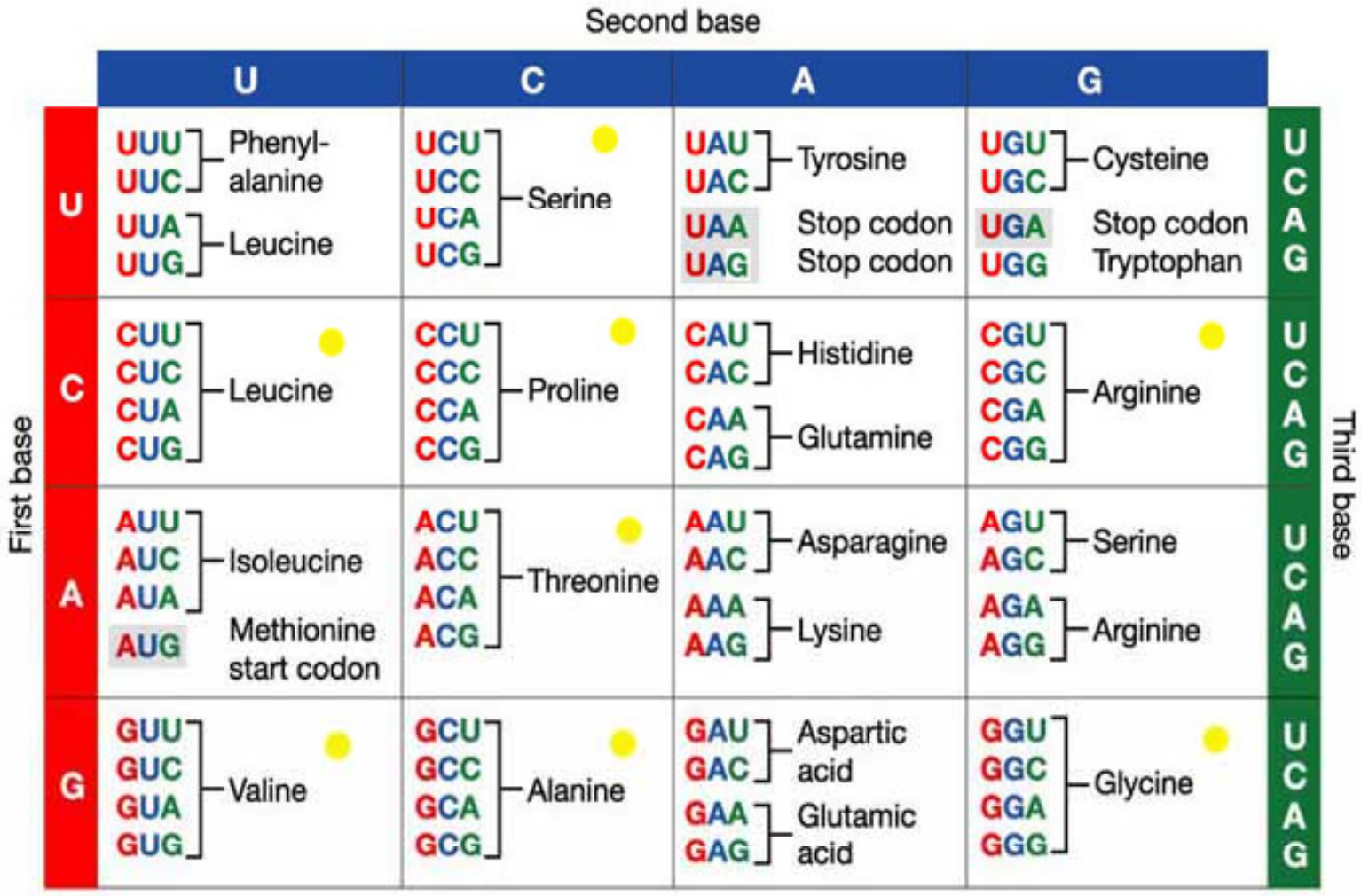
- Codon: ..




20 amino acids in nature

| Codon (nt) | Amino acid |
|---|---|
| 1 | |
| 2 | |
| 3 | |

# Genetic Code



# Genome

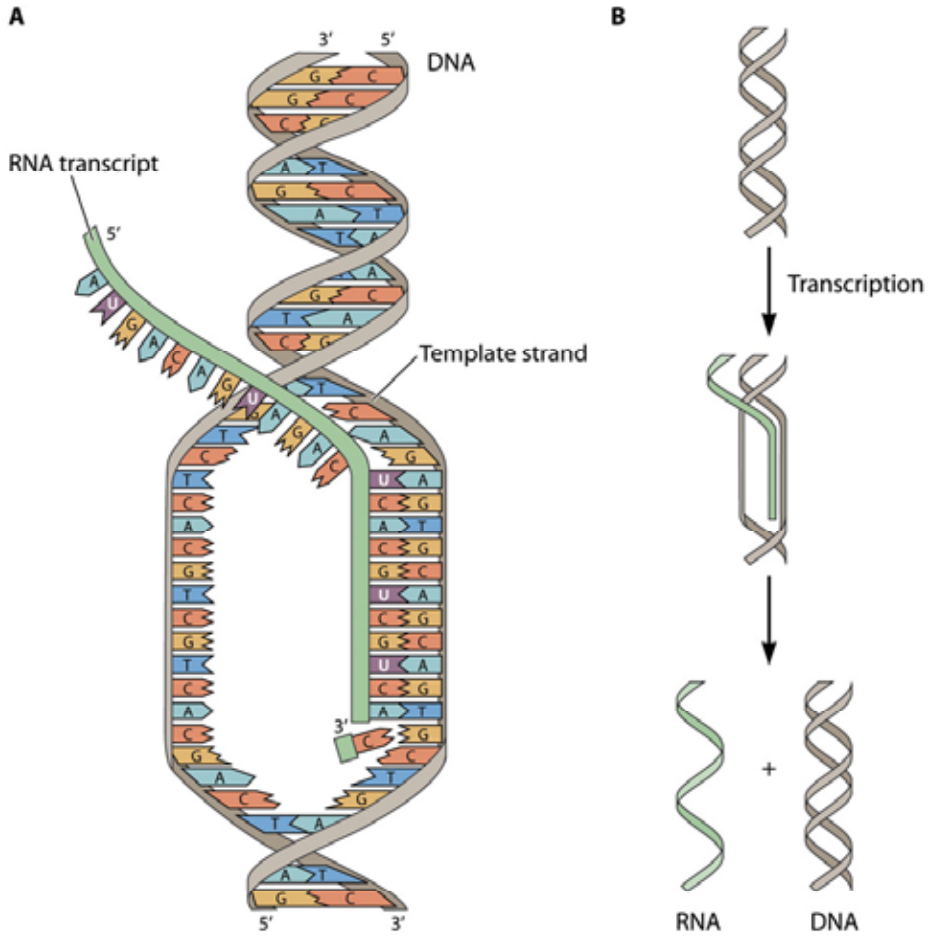
- Gene
  - The complete stretch of DNA needed to determine the amino acid sequence of a protein
- Genome
  - The complete set of genetic material in an organism
  - Human genome project
    - 1990-2003
    - U.S. department of energy and the National Institute of Health
    - $2.8 \times 10^9$ bp, ~30,000 genes
    - 90% is noncoding DNA

# Protein Synthesis

- Transcription
  - From ..  to ..
- Translation
  - From ..  to ..
  - tRNA (transfer RNA) matches ..
  - Ribosome (made of ..  )
    - Protein synthesis

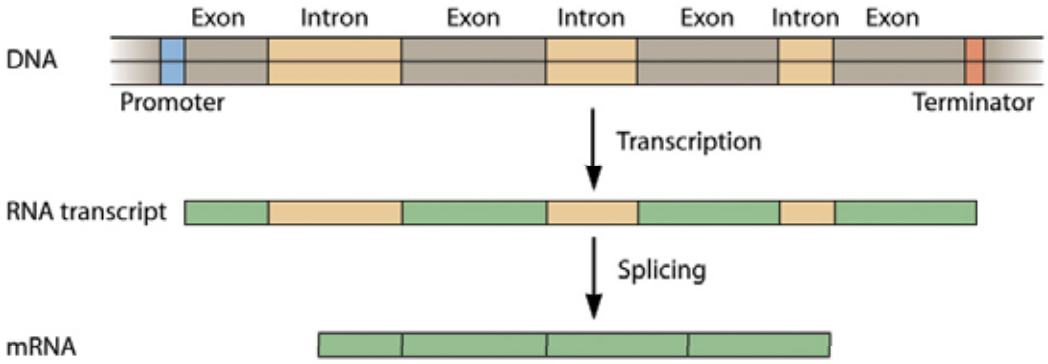
# Transcription

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

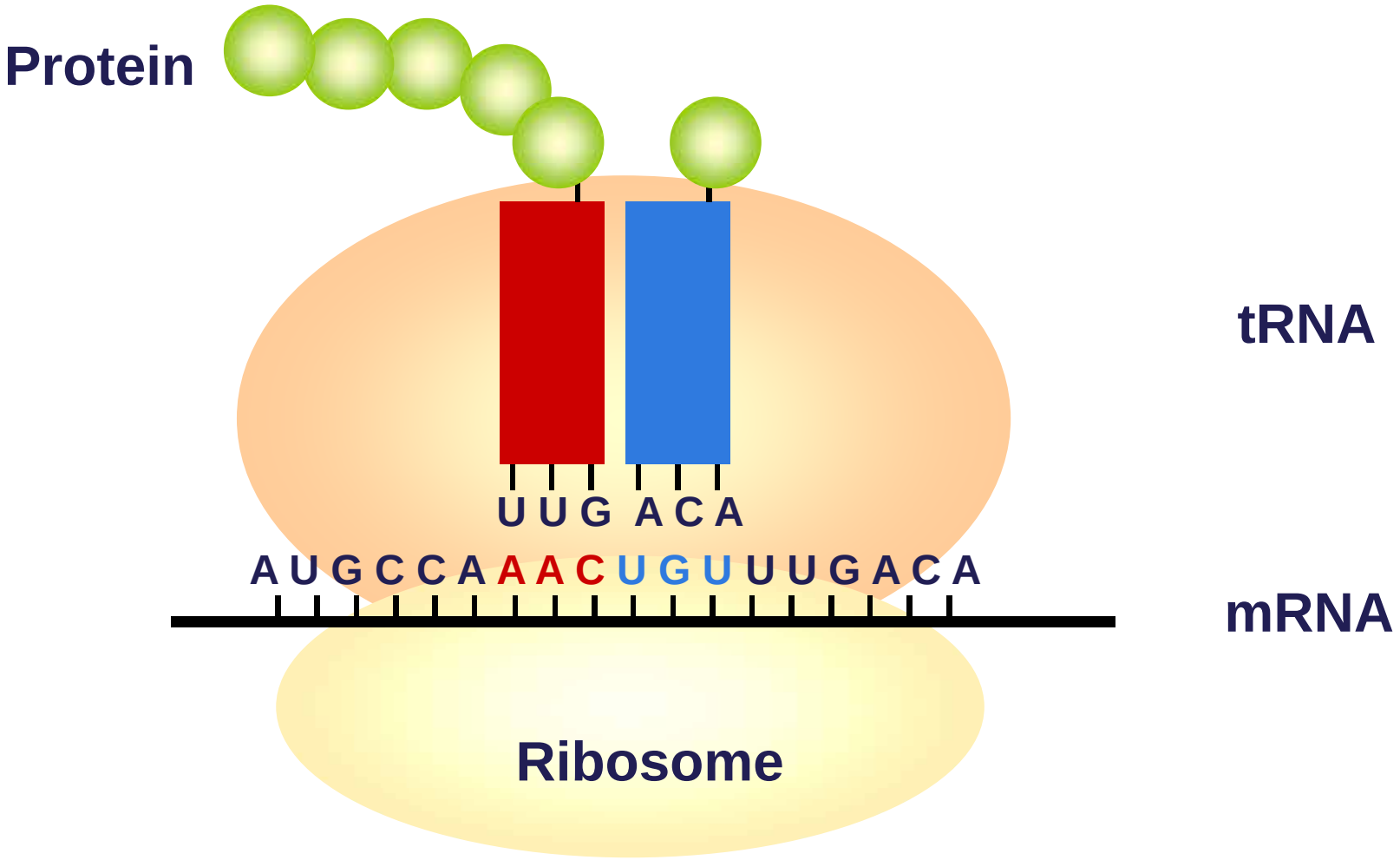


# Regulation of Transcription

- Promoter
  - Binding site of ..
    (transcriptional regulator: )
- Terminator
  - The site where ..
- Processing of eukaryotic RNA
  - Splicing : joining of ..

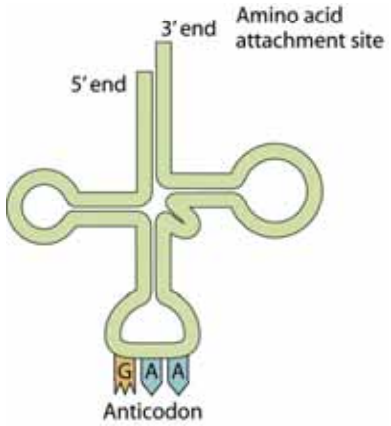


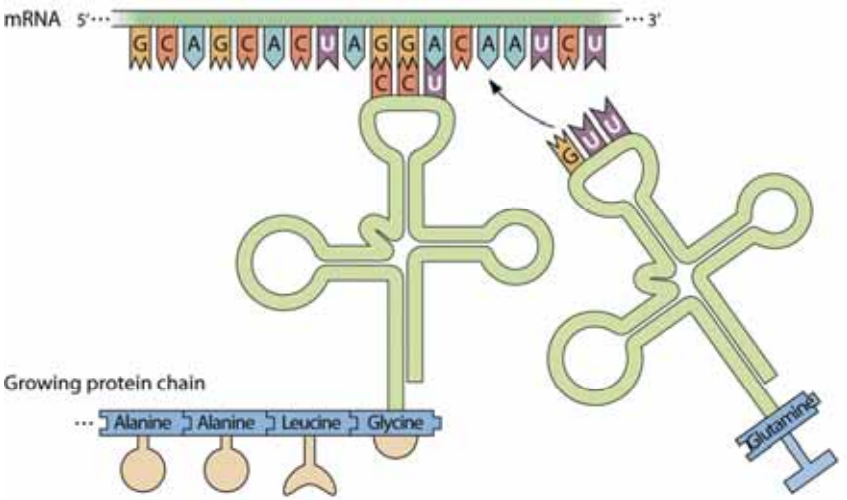
# Translation



# Translation

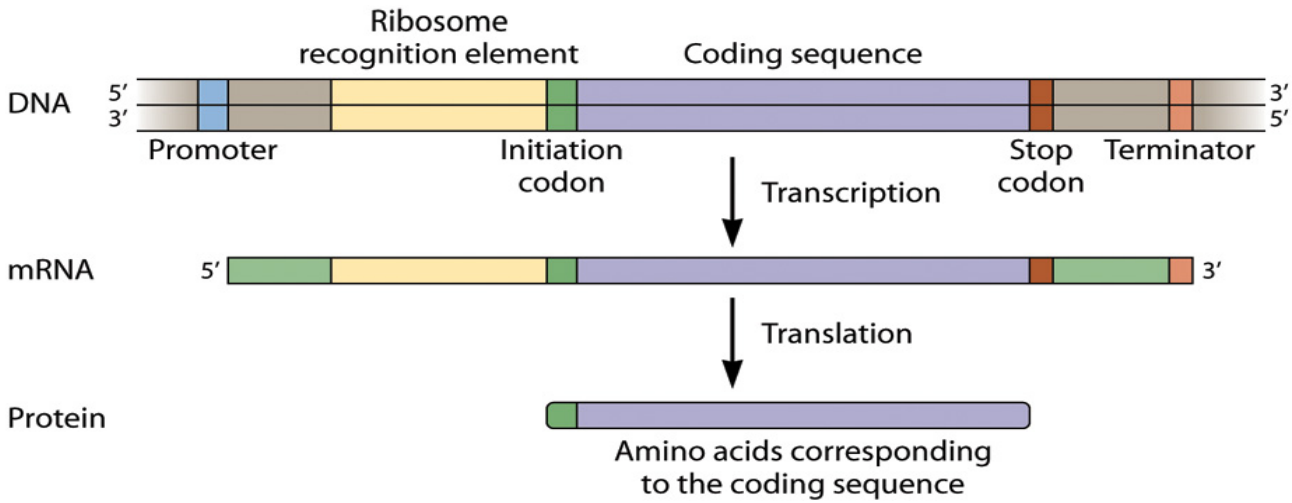
- tRNA
  - Cloverleaves-shaped folding
  - Anticodon:
  - 3’ end:
- Translation
  - Assembly of ..
  - Binding of ..
  - ..
  - Release of ..



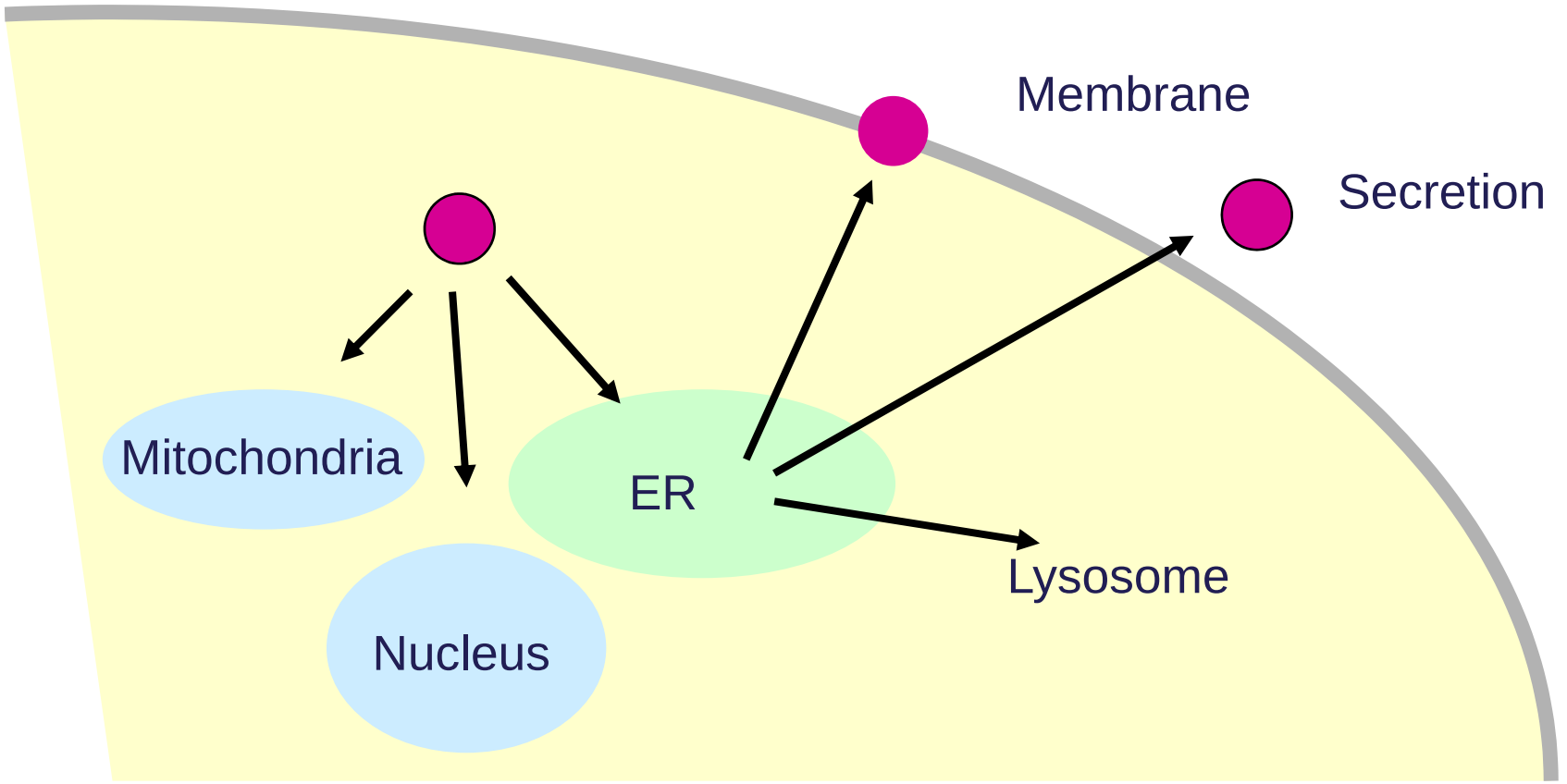
# Signals for Transcription and Translation

- Ribosome binding site in mRNA
  - ..
- Initiation codon
  - ..
- Stop codon
  - UGA, UAA, UAG :

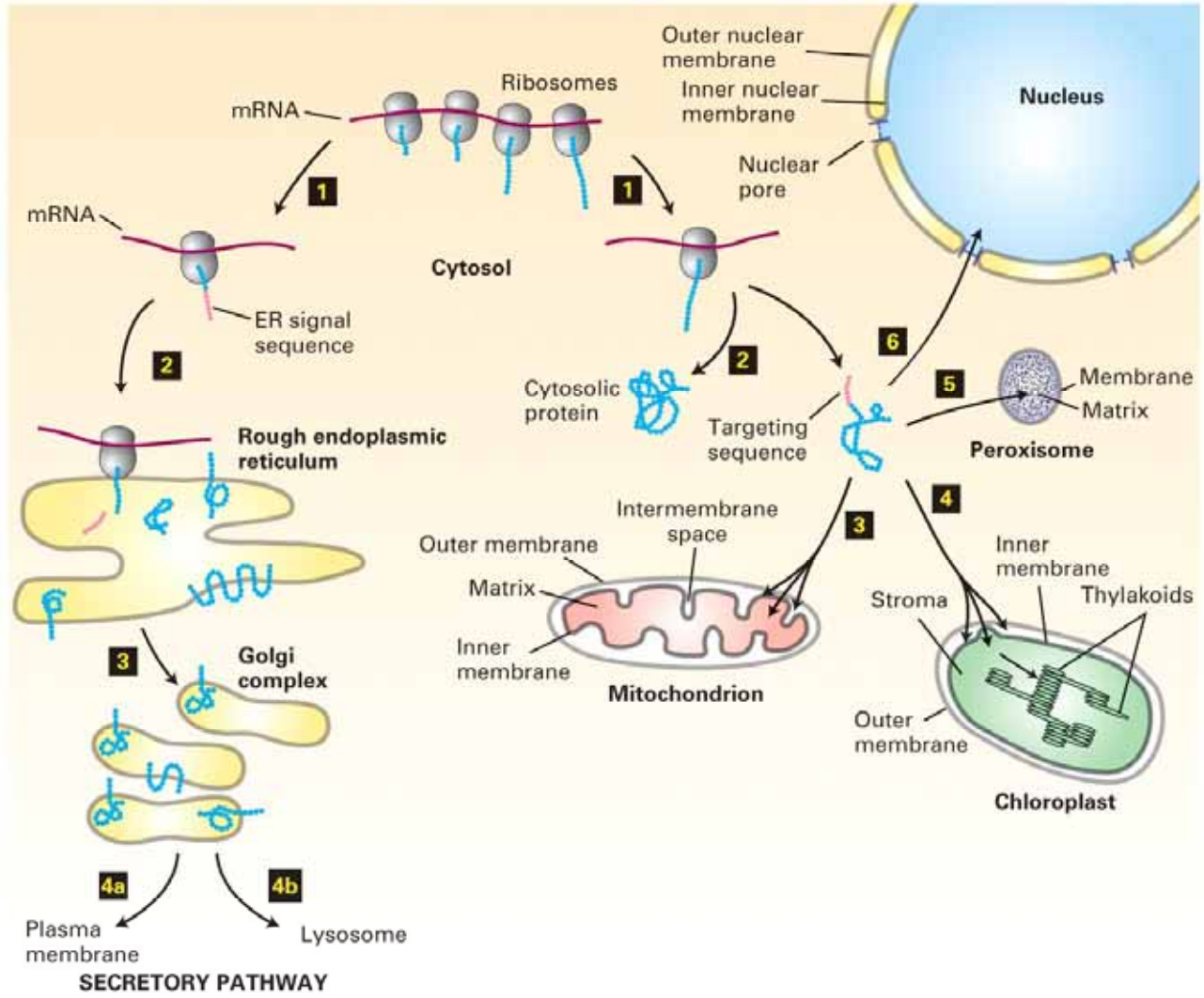


# Cellular Fate of Proteins

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

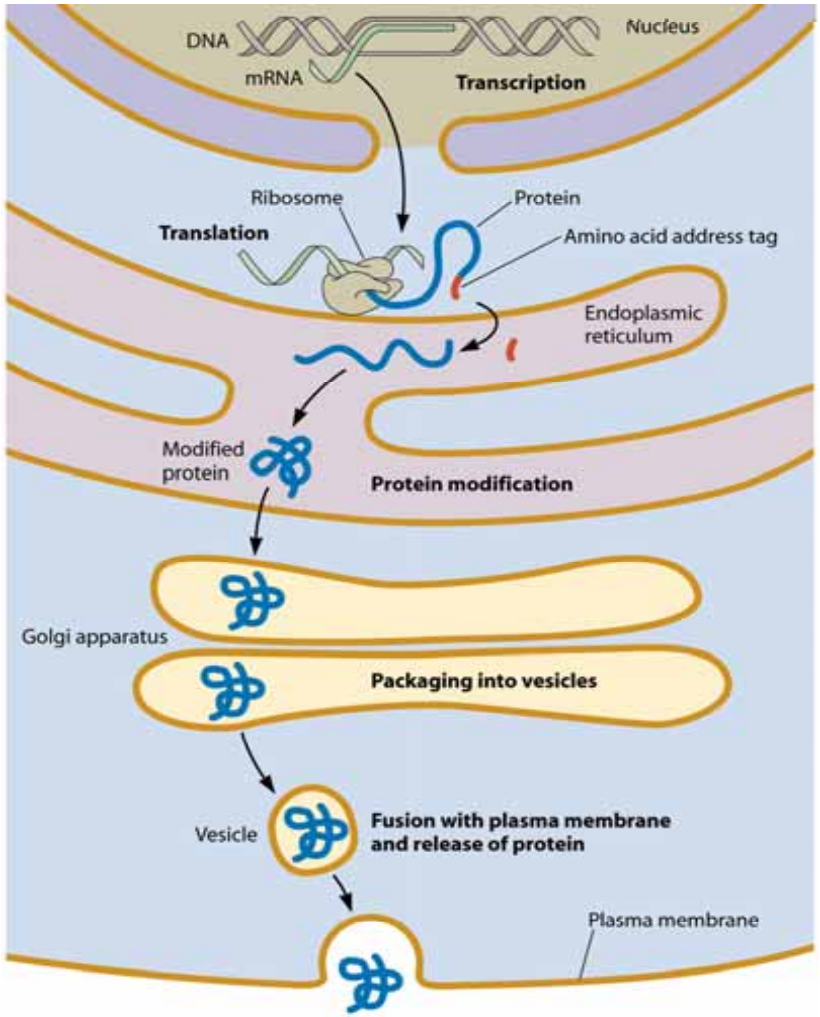


# Overview of Protein Sorting Pathway



# 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

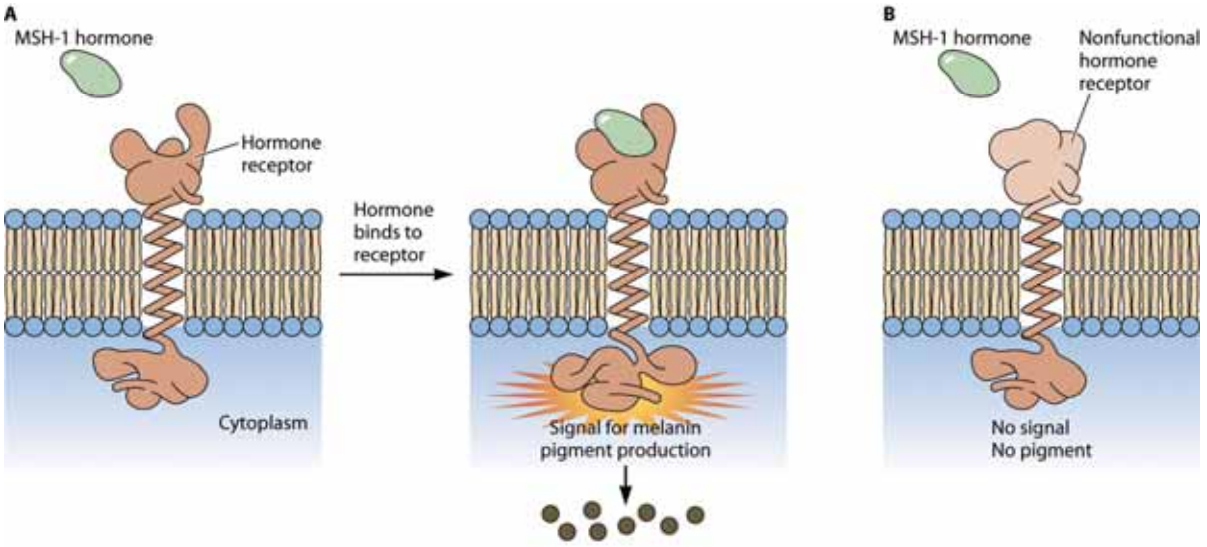


# Mutations

- 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: nt change with 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

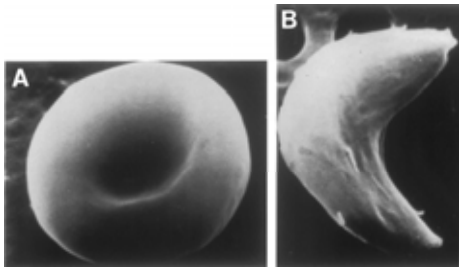
# Mutations

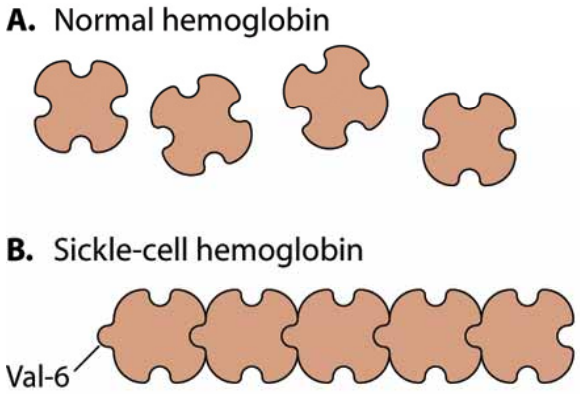
- No effect on survival
  - e.g. hair color




# Mutations

- Harmful
  - e.g. sickle-cell anemia
    - A to T mutation of hemoglobin
      - → 6th amino acid change from glu to val
      - → hydrophobic aggregation of hemoglobin



# Mutations

- Benign erythrocytosis
  - Elevated levels of RBC
  - Mutation in erythropoetin receptor

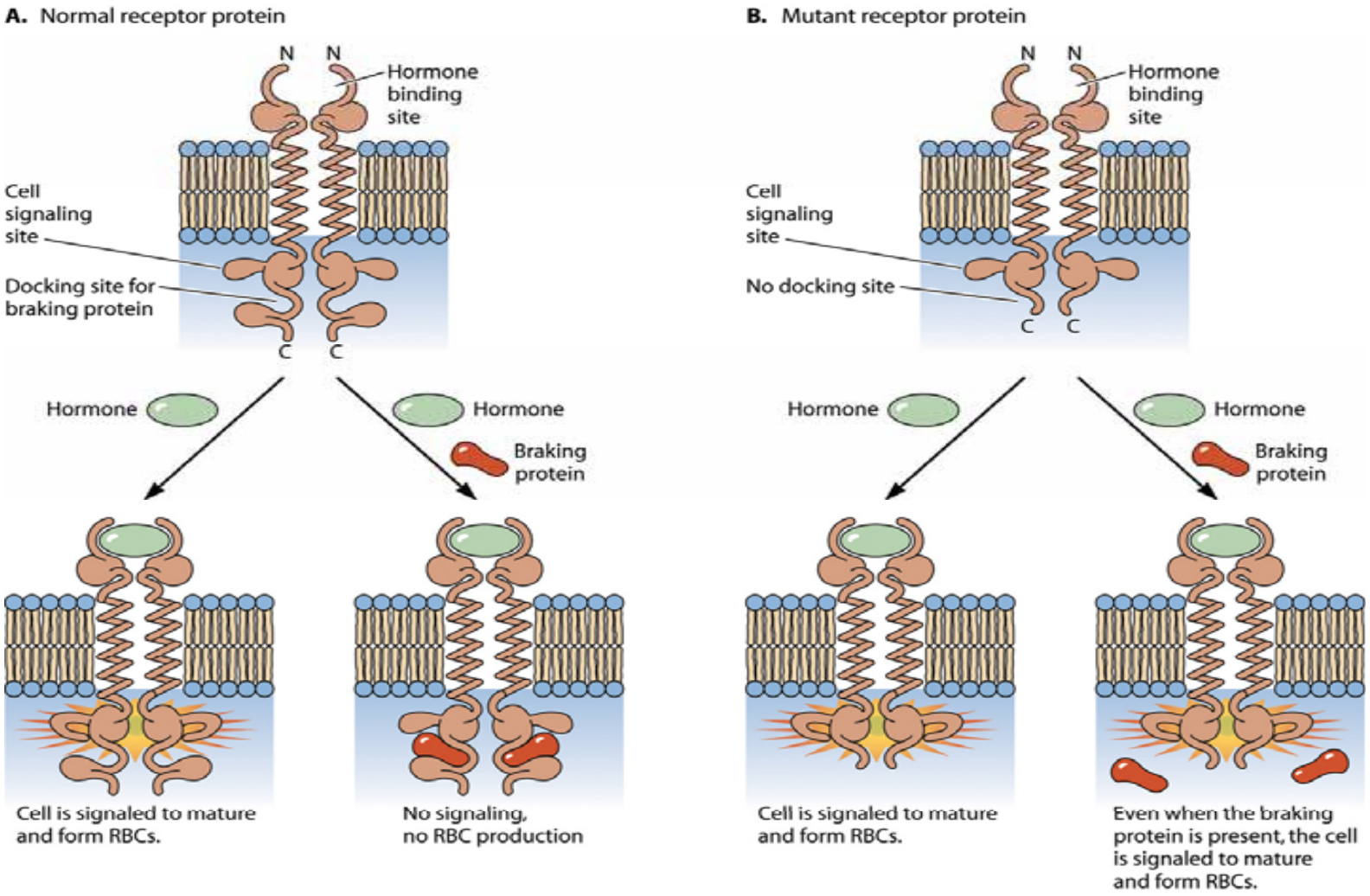
    -- 481 TGG to TAG (stop codon)
  - Deletion of 70 amino acid for repression of signal transduction

    →More RBC production from bone marrow stem cells

    →Greatly enhanced stamina

    (Finnish athlete Eero Mantyranta won three gold medals for cross-country skiing in the 1964 Winter Olympics)

# Mutations


A. Normal receptor protein
N
N
Hormone binding site
Cell signaling site
Docking site for braking protein
C
C
Hormone
Hormone
Braking protein
Cell is signaled to mature and form RBCs.
No signaling, no RBC production
B. Mutant receptor protein
N
N
Hormone binding site
Cell signaling site
No docking site
C
C
Hormone
Hormone
Braking protein
Cell is signaled to mature and form RBCs.
Even when the braking protein is present, the cell is signaled to mature and form RBCs.