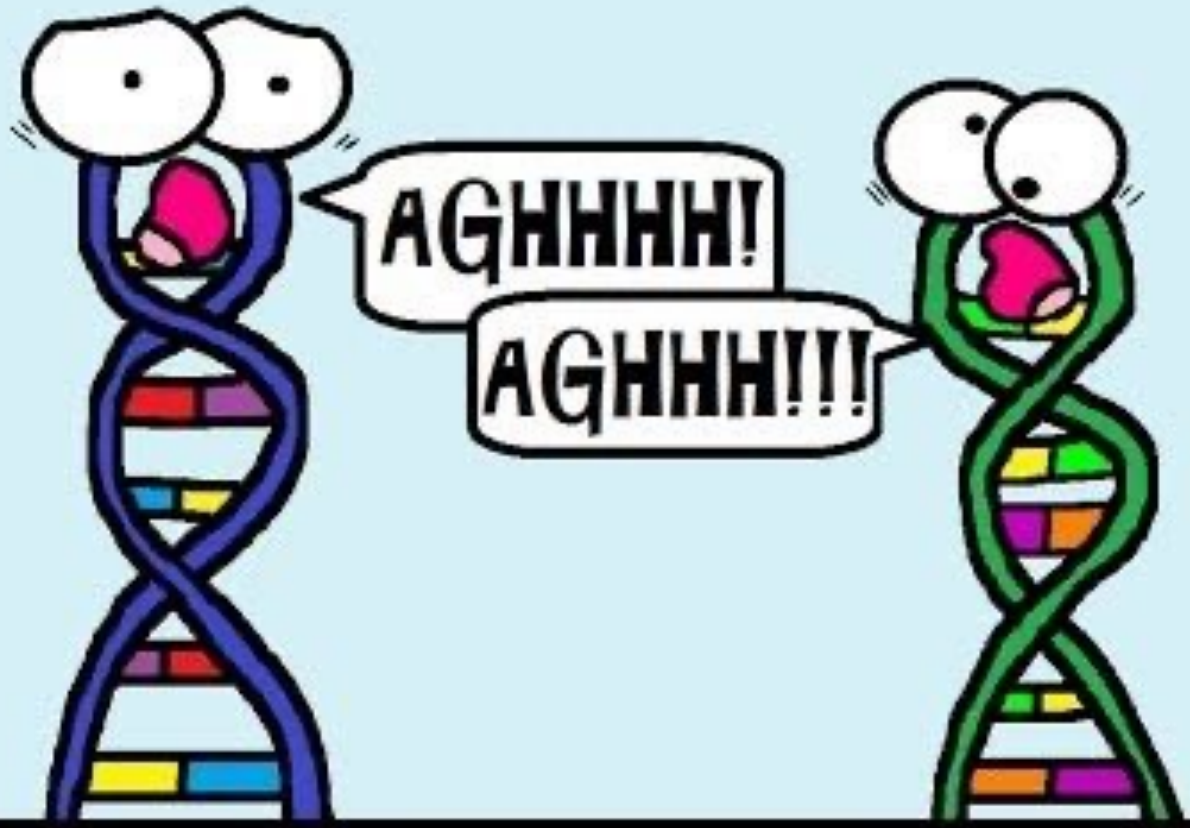
Mutations



Concept of Mutation

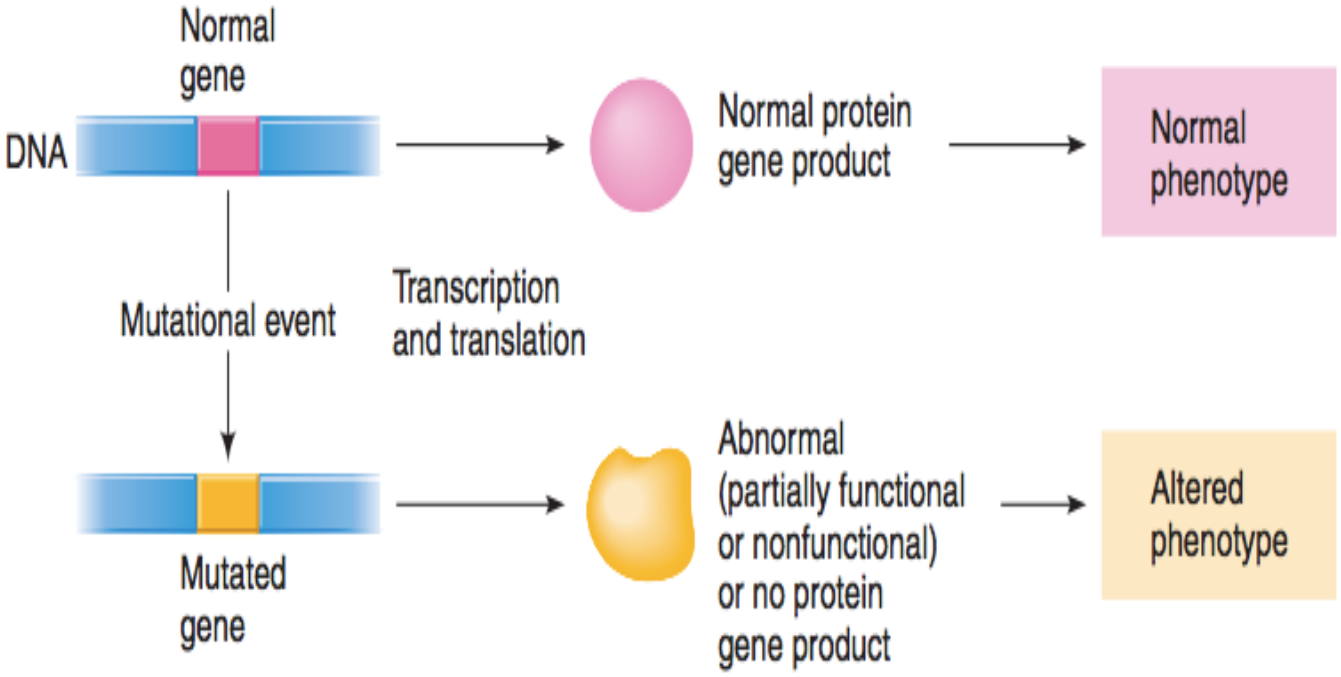
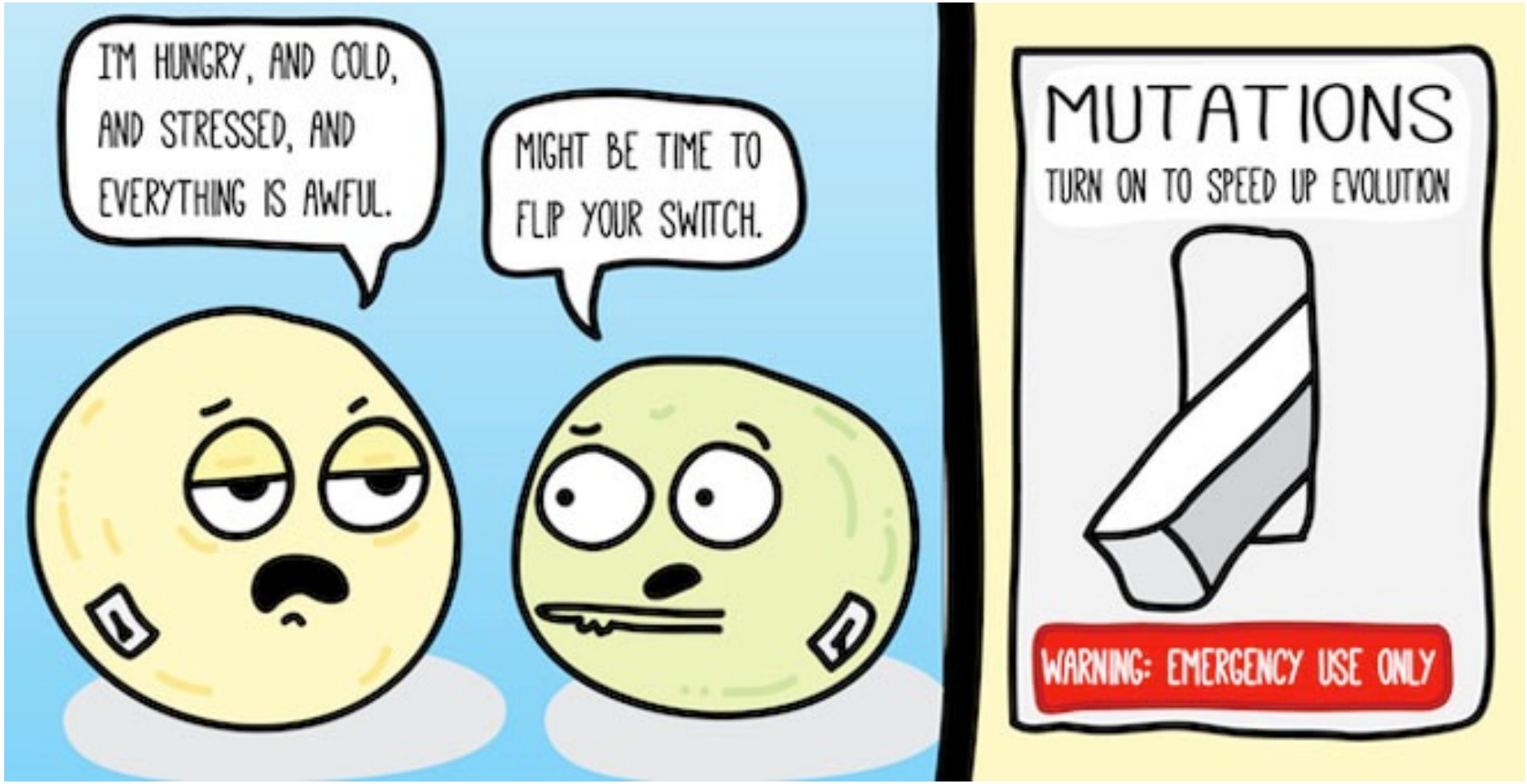
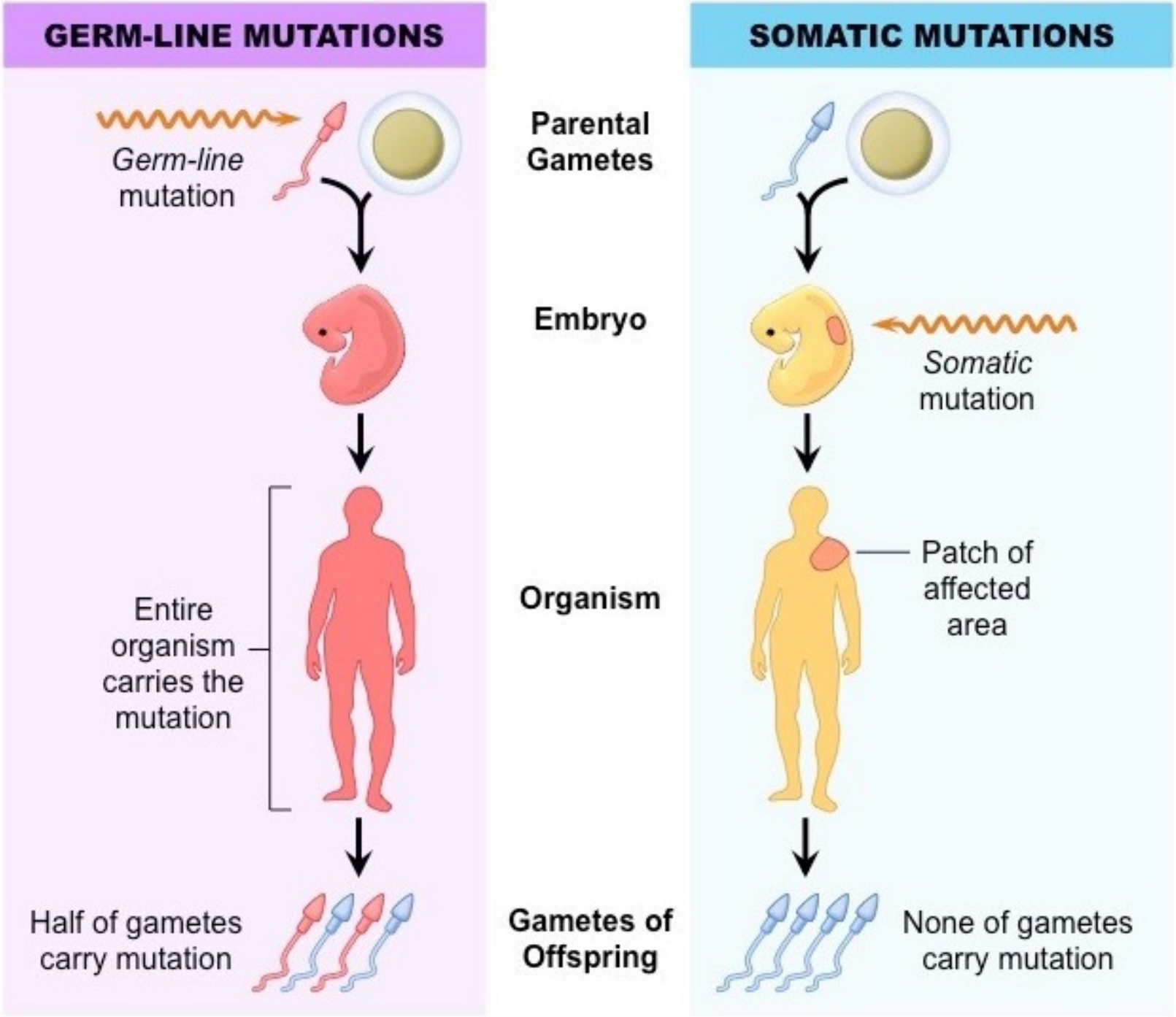


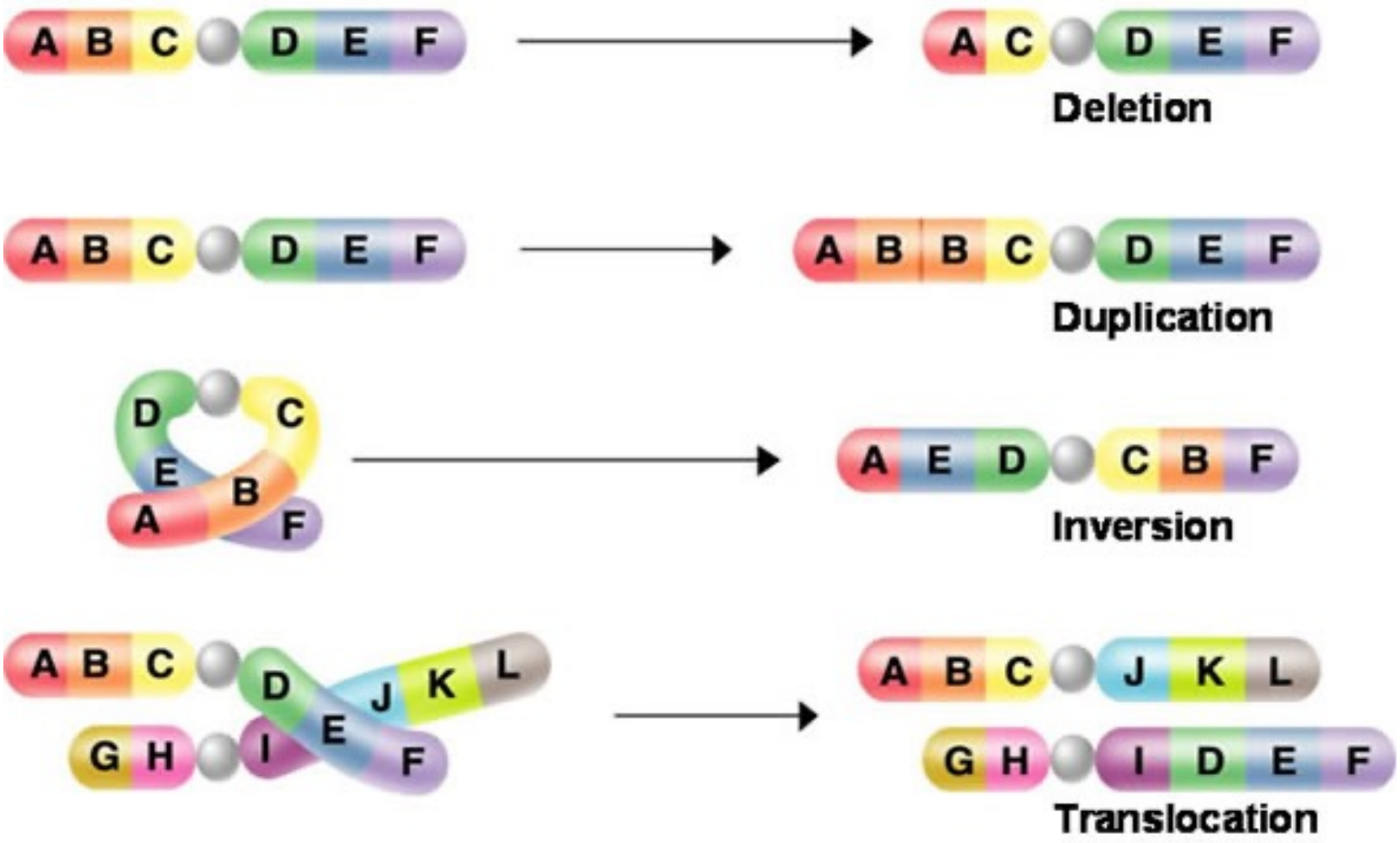
Figure 7.1

Concept of a mutation in the protein-coding region of a gene. (Note that not all mutations lead to altered proteins and that not all mutations are in protein-coding regions.)

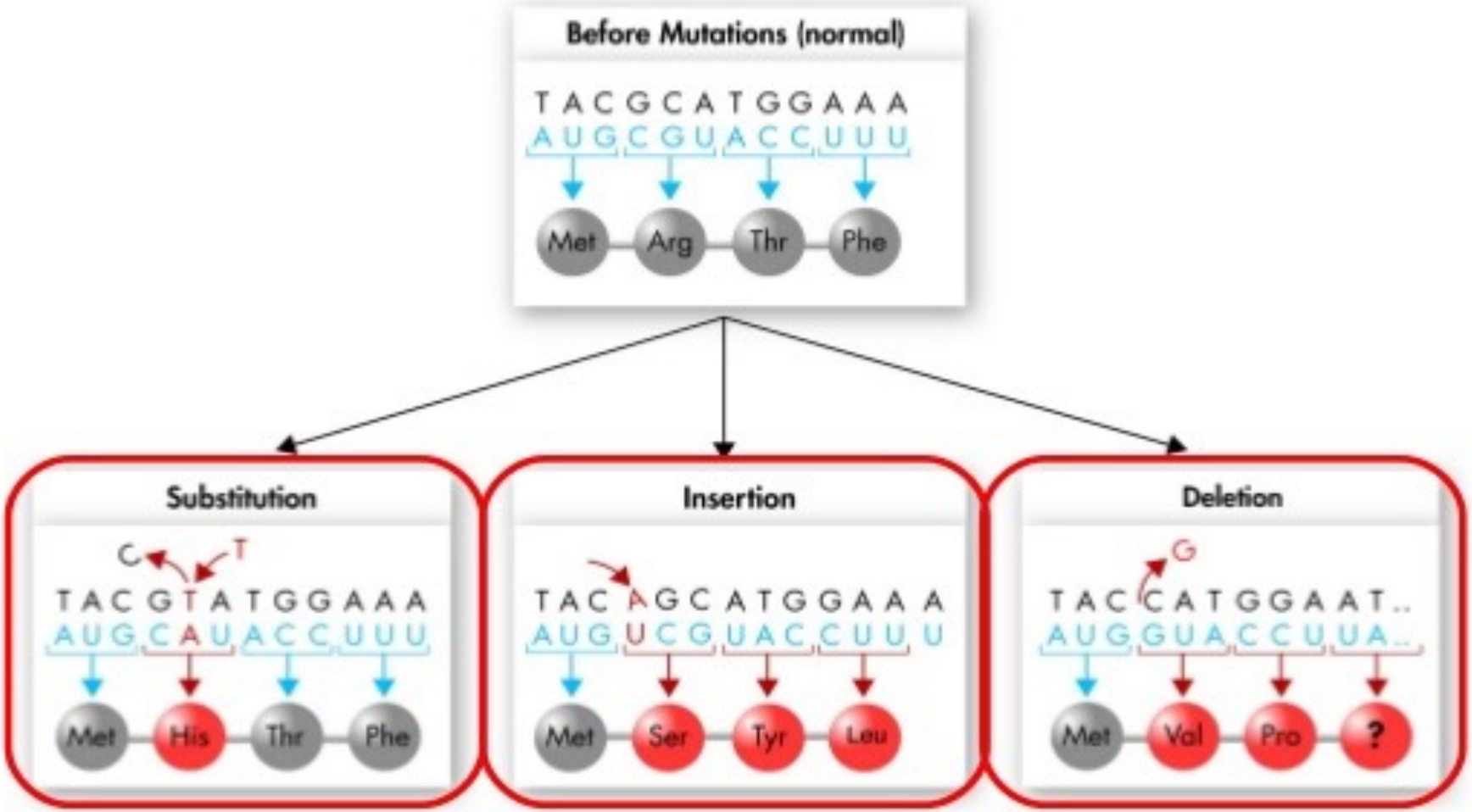

I'M HUNGRY, AND COLD, AND STRESSED, AND EVERYTHING IS AWFUL.
MIGHT BE TIME TO FLIP YOUR SWITCH.
MUTATIONS
TURN ON TO SPEED UP EVOLUTION
WARNING: EMERGENCY USE ONLY


GERM-LINE MUTATIONS
SOMATIC MUTATIONS
Parental Gametes
Embryo
Organism
Gametes of Offspring
Germ-line mutation
Somatic mutation
Entire organism carries the mutation
Patch of affected area
Half of gametes carry mutation
None of gametes carry mutation

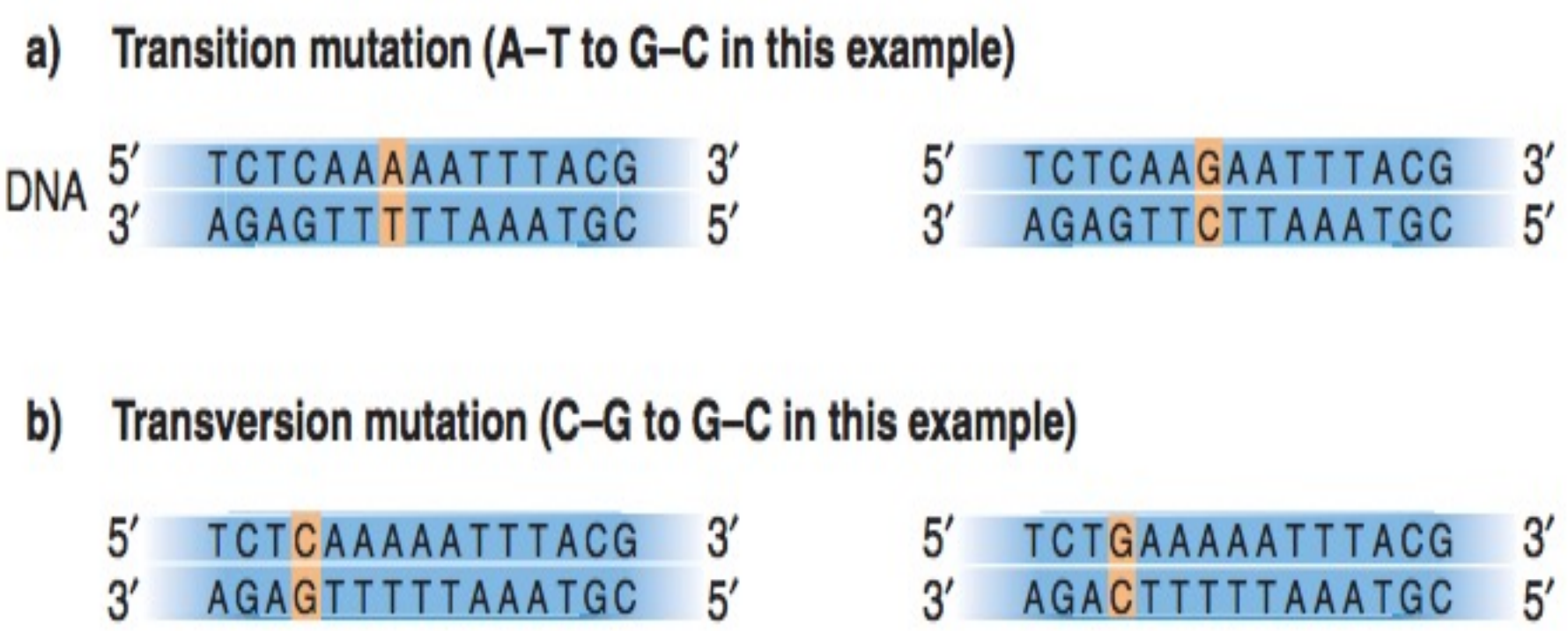
Chromosomal Mutations

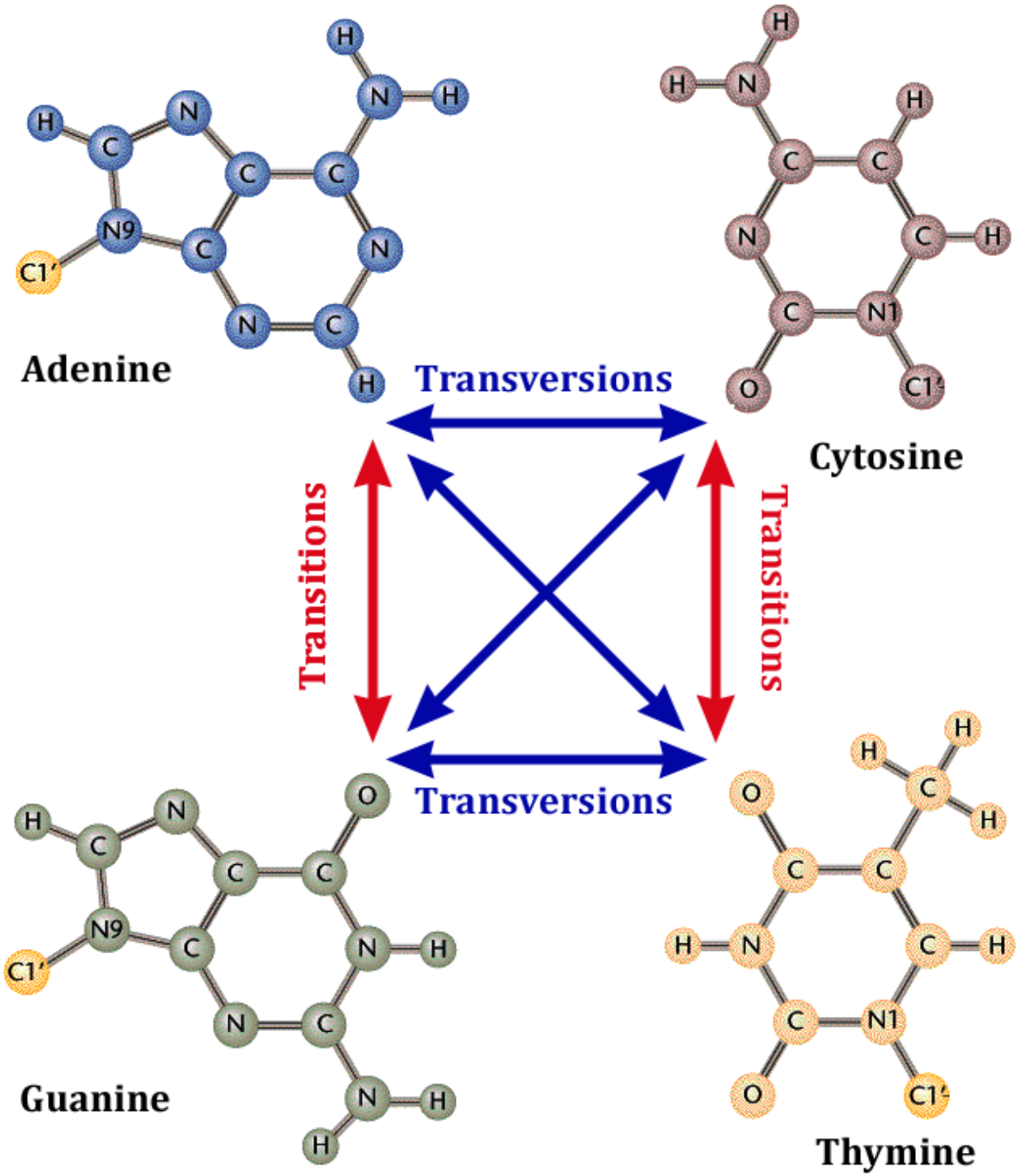


Point Mutations

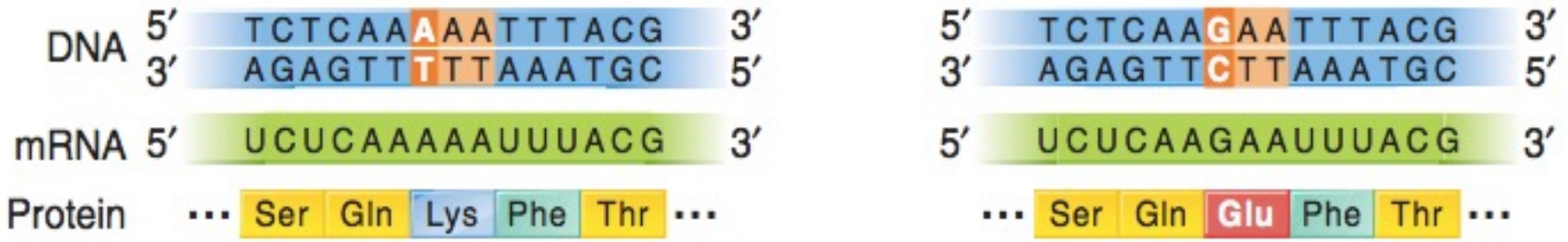


1. Base-Pair Substitution




Adenine
Cytosine
Guanine
Thymine
Transversions
Transversions
Transitions
Transitions

c) Missense mutation (change from one amino acid to another; here, an AT-to-GC transition mutation changes the codon from lysine to glutamic acid)



d) Nonsense mutation (change from an amino acid to a stop codon; here, an AT-to-TA transversion mutation changes the codon from lysine to UAA stop codon)

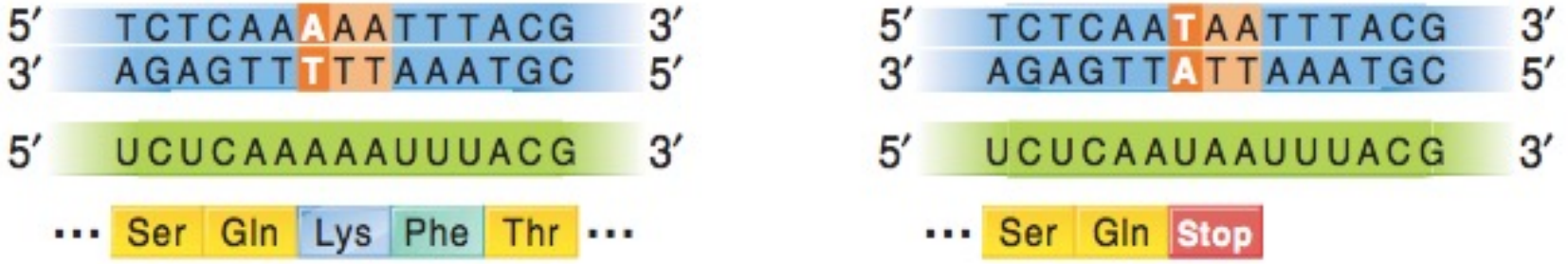
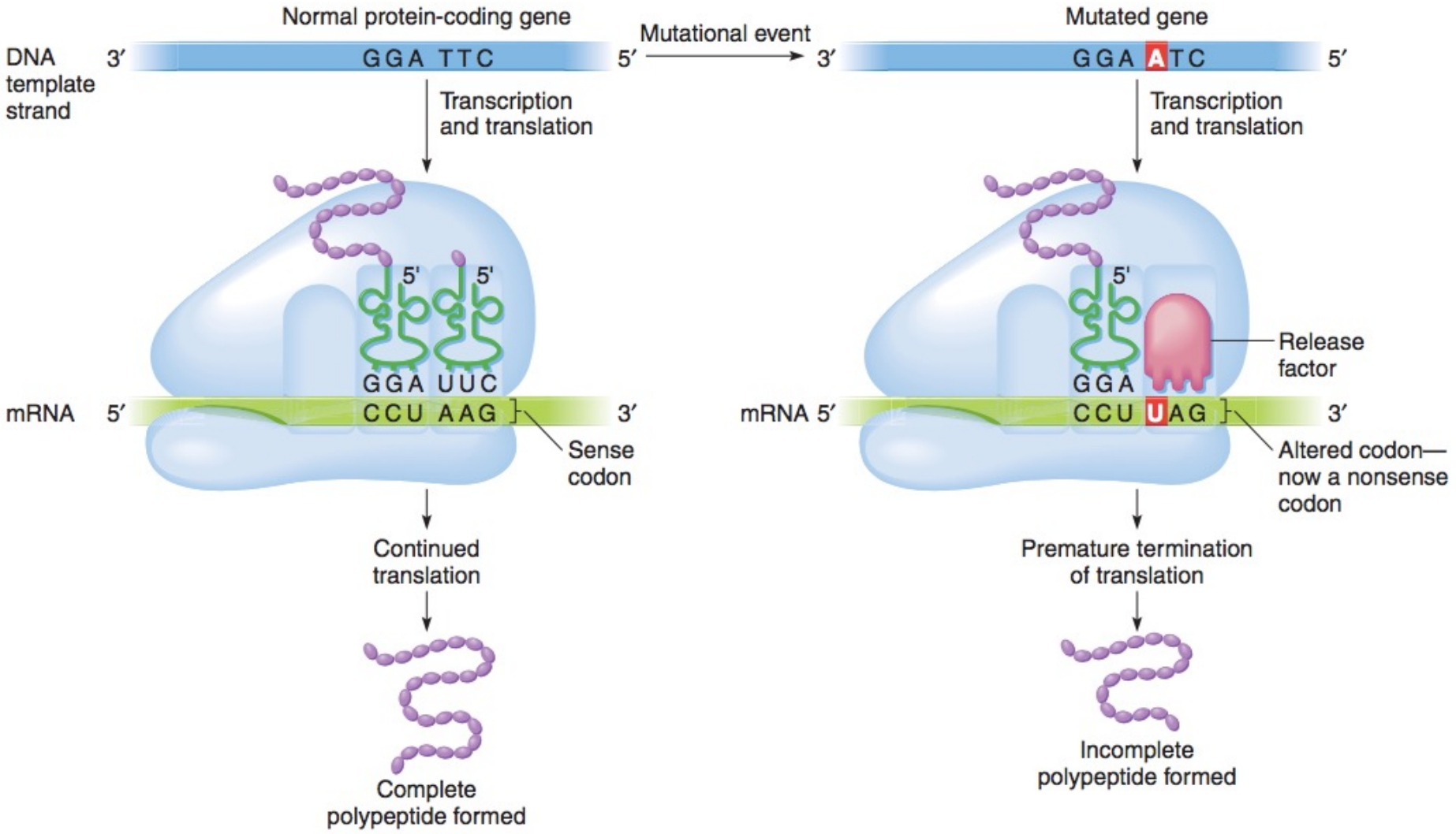
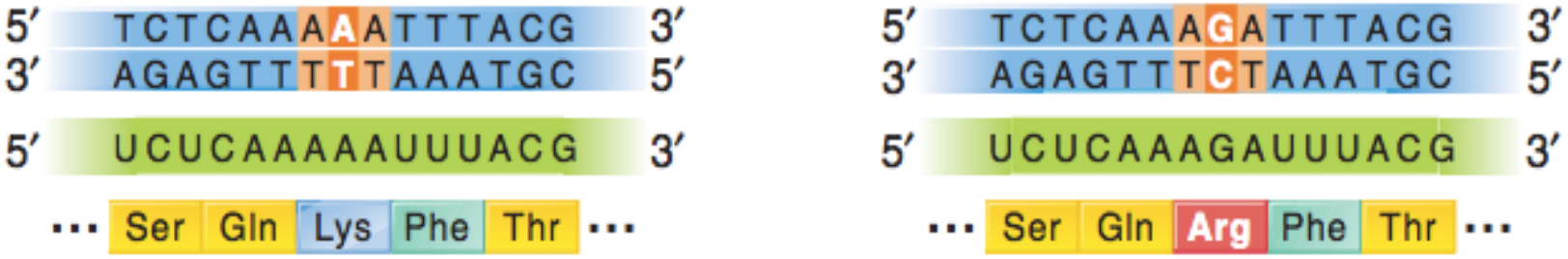


Figure 7.4

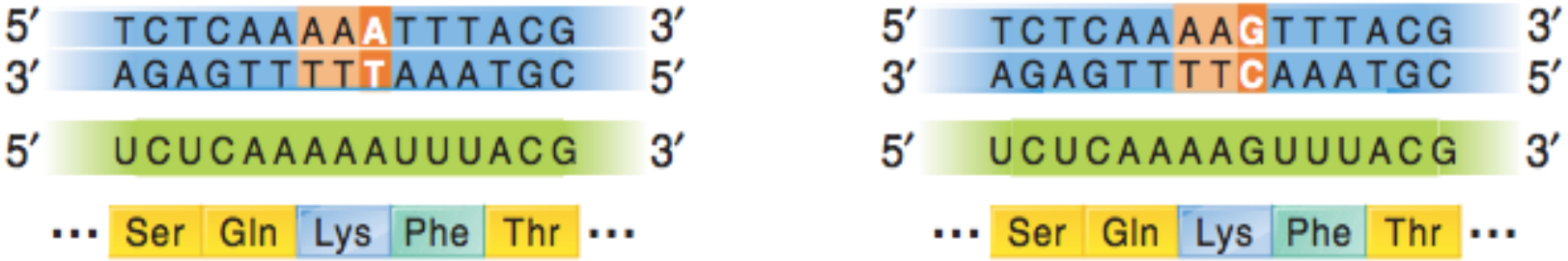
A nonsense mutation and its effect on translation.



e) **Neutral mutation (change from an amino acid to another amino acid with similar chemical properties; here, an AT-to-GC transition mutation changes the codon from lysine to arginine)**

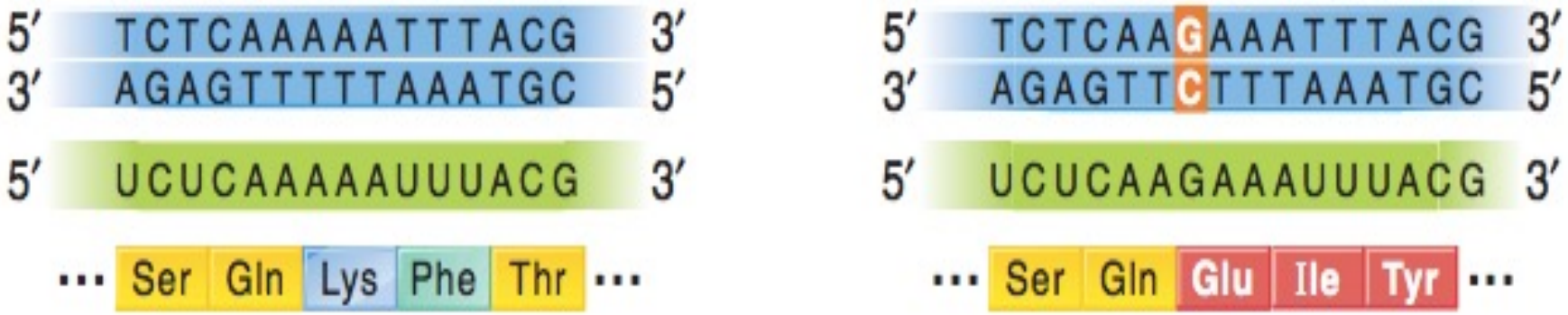


f) **Silent mutation (change in codon such that the same amino acid is specified; here, an AT-to-GC transition in the third position of the codon gives a codon that still encodes lysine)**

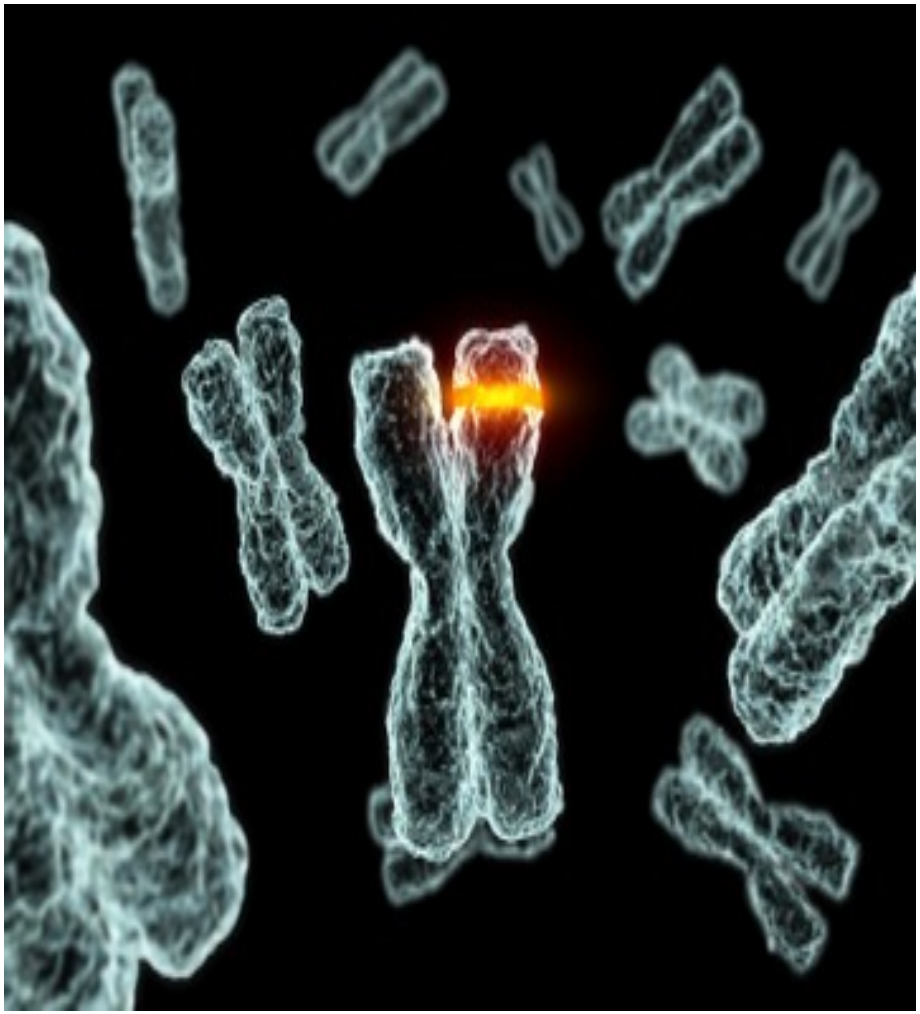


2. Frameshift Mutation

Frameshift mutation (addition or deletion of one or a few base pairs leads to a change in reading frame; here, the insertion of a G–C base pair scrambles the message after glutamine)



Spontaneous Mutations



1. Error during DNA replication
(a) Tautomeric shift
(b) Wobble pairing
(c) DNA looping-out errors

2. Spontaneous chemical changes
(a) Depurination
(b) Deamination

Figure 7.6

Normal Watson-Crick and non–Watson-Crick base pairing in DNA.

Tautomeric shift

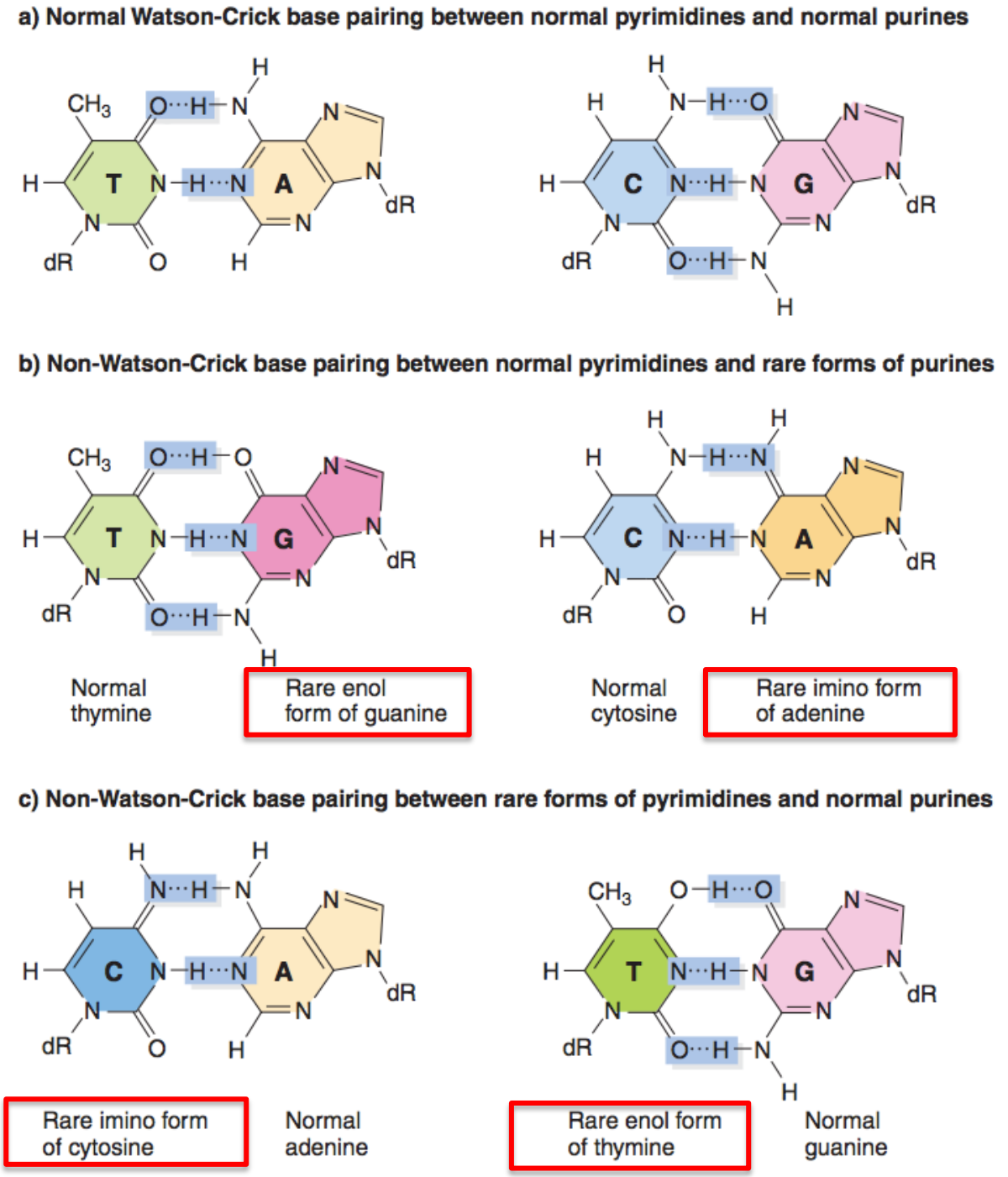
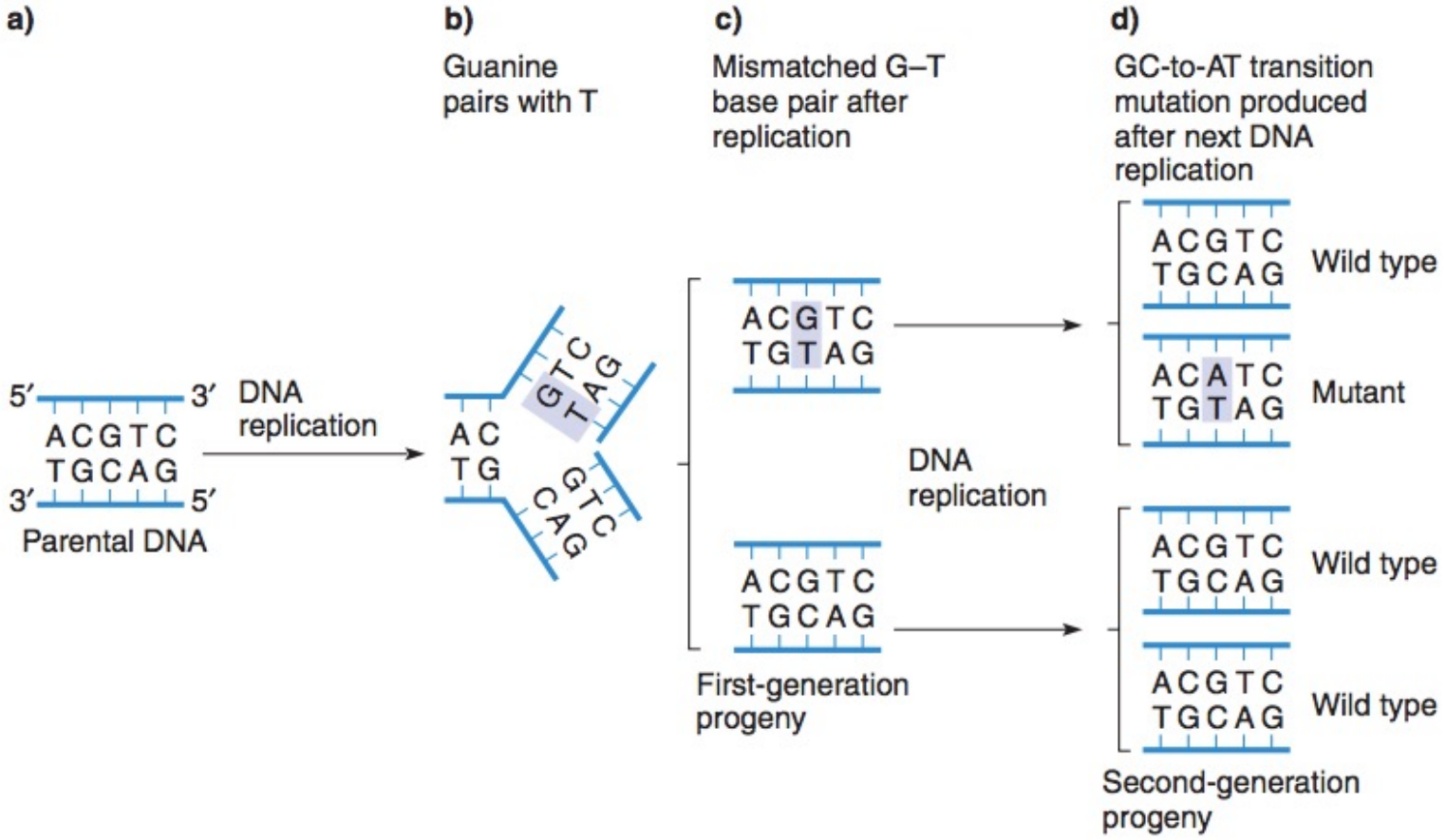


Figure 7.7

Production of a mutation as a result of a mismatch caused by non–Watson-Crick base pairing. The details are explained in the text.



“Wobble” T-G pairing

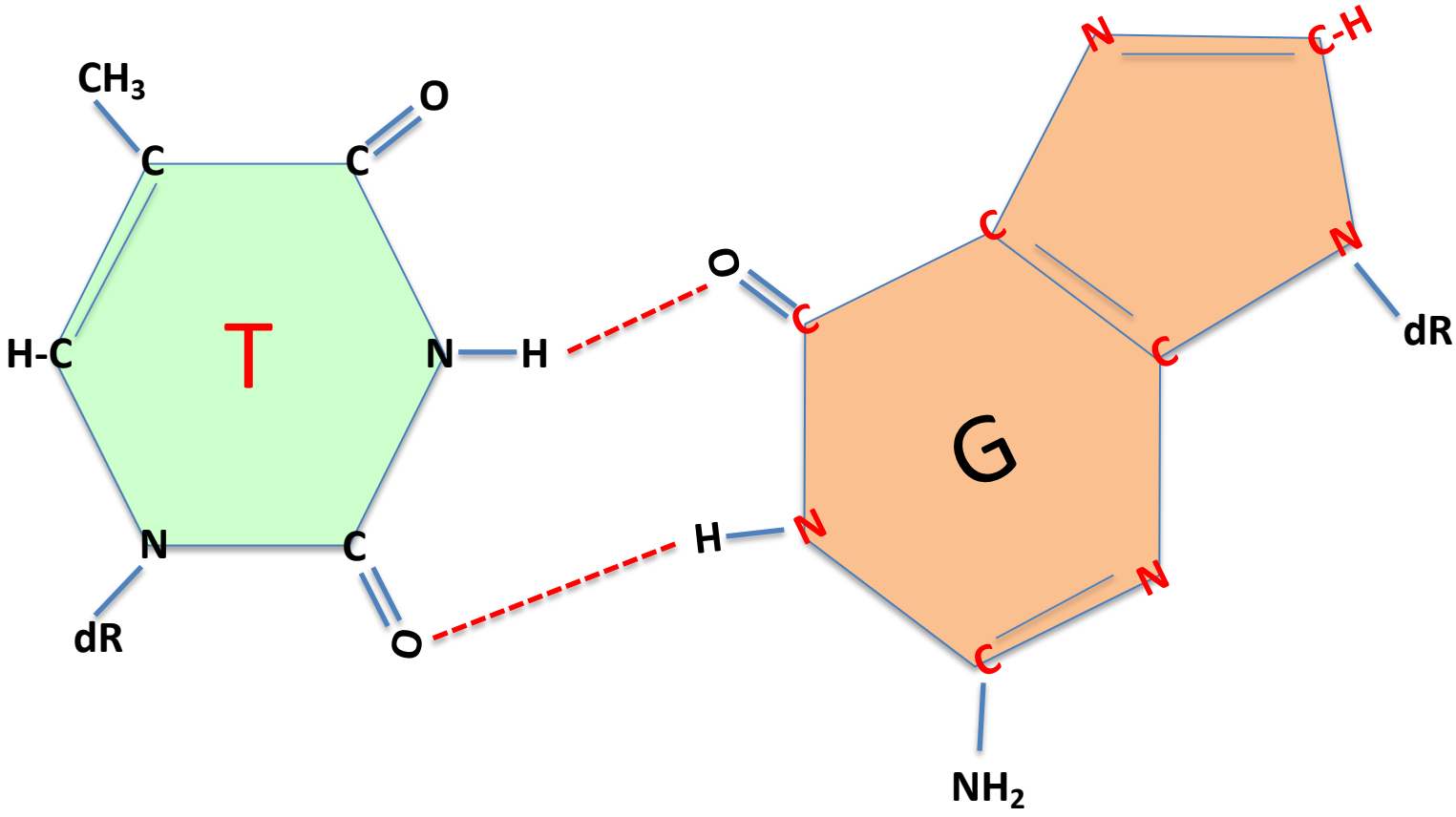
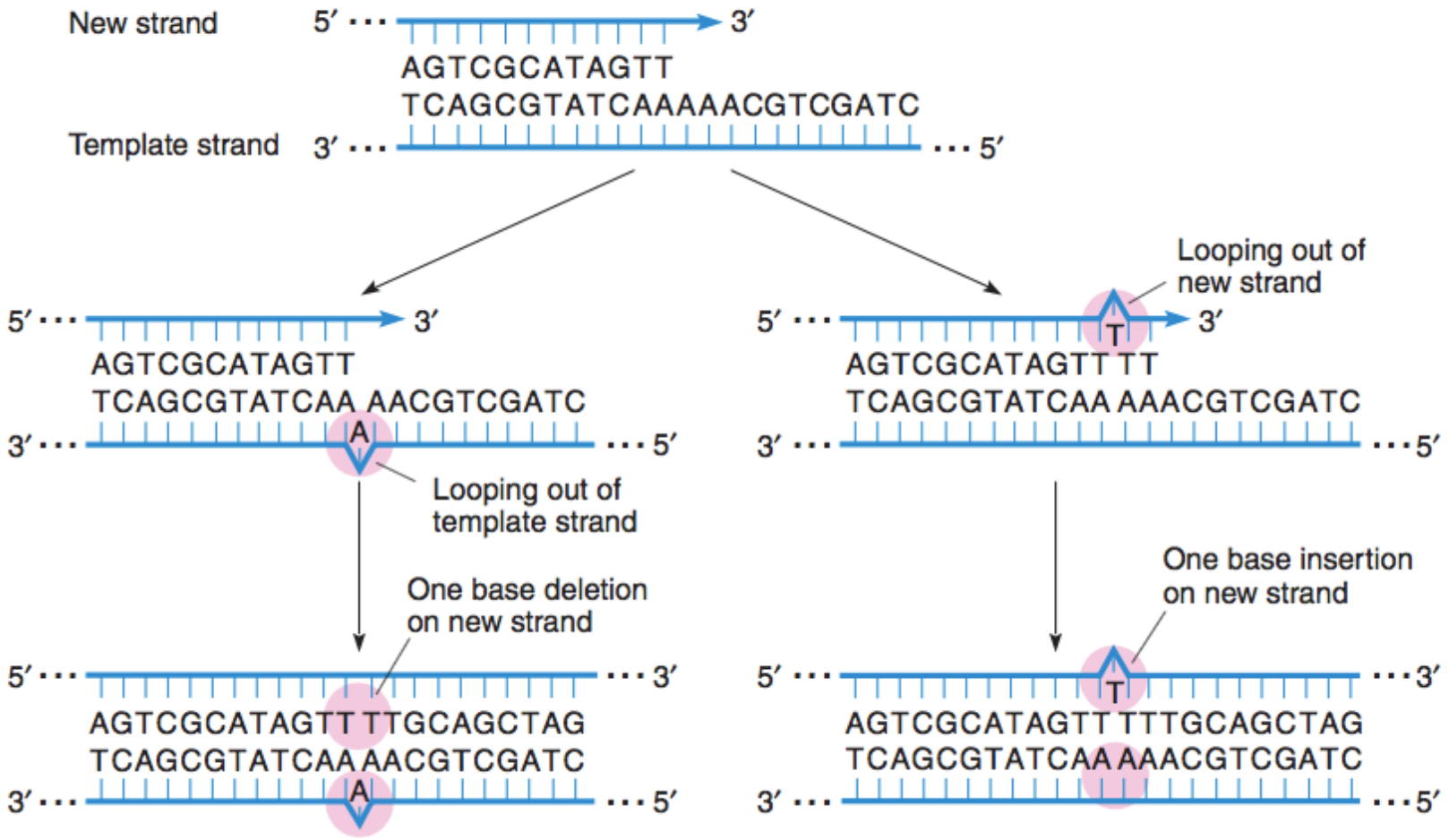
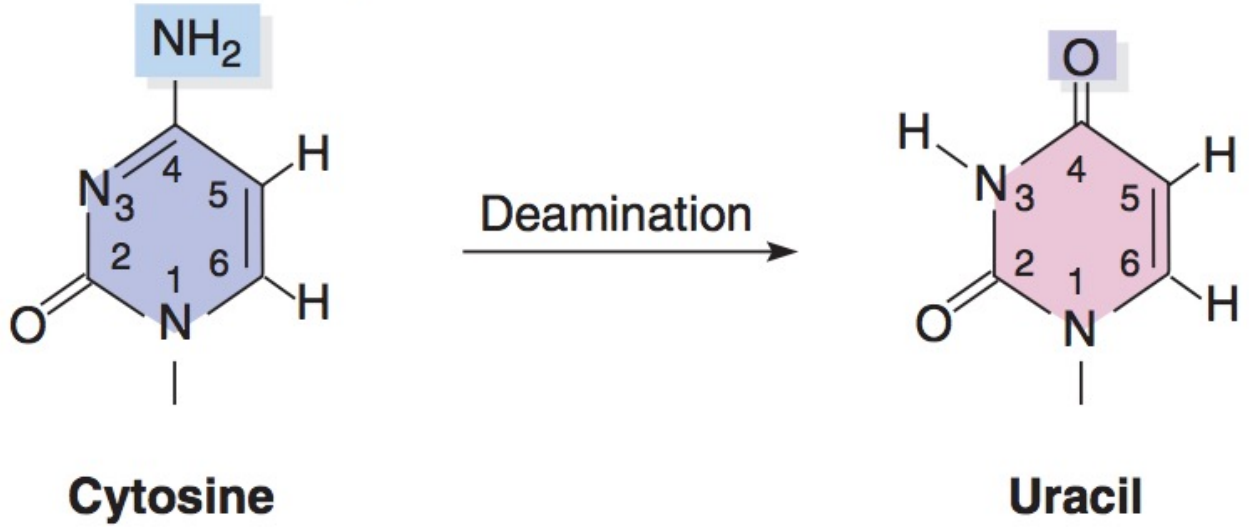


Figure 7.8

Spontaneous generation of addition and deletion mutants by DNA looping-out errors during replication.

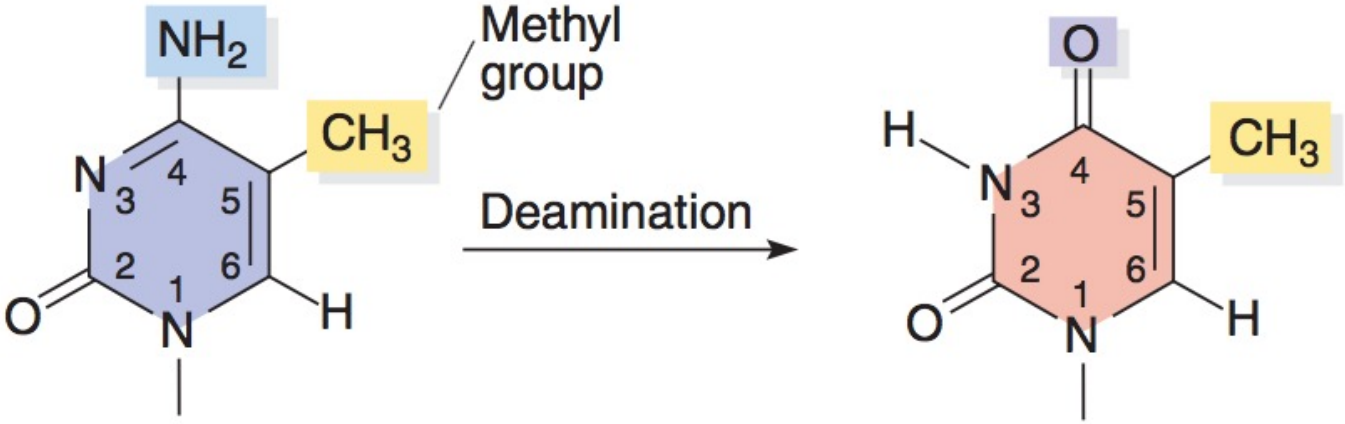


a) Deamination of cytosine to uracil



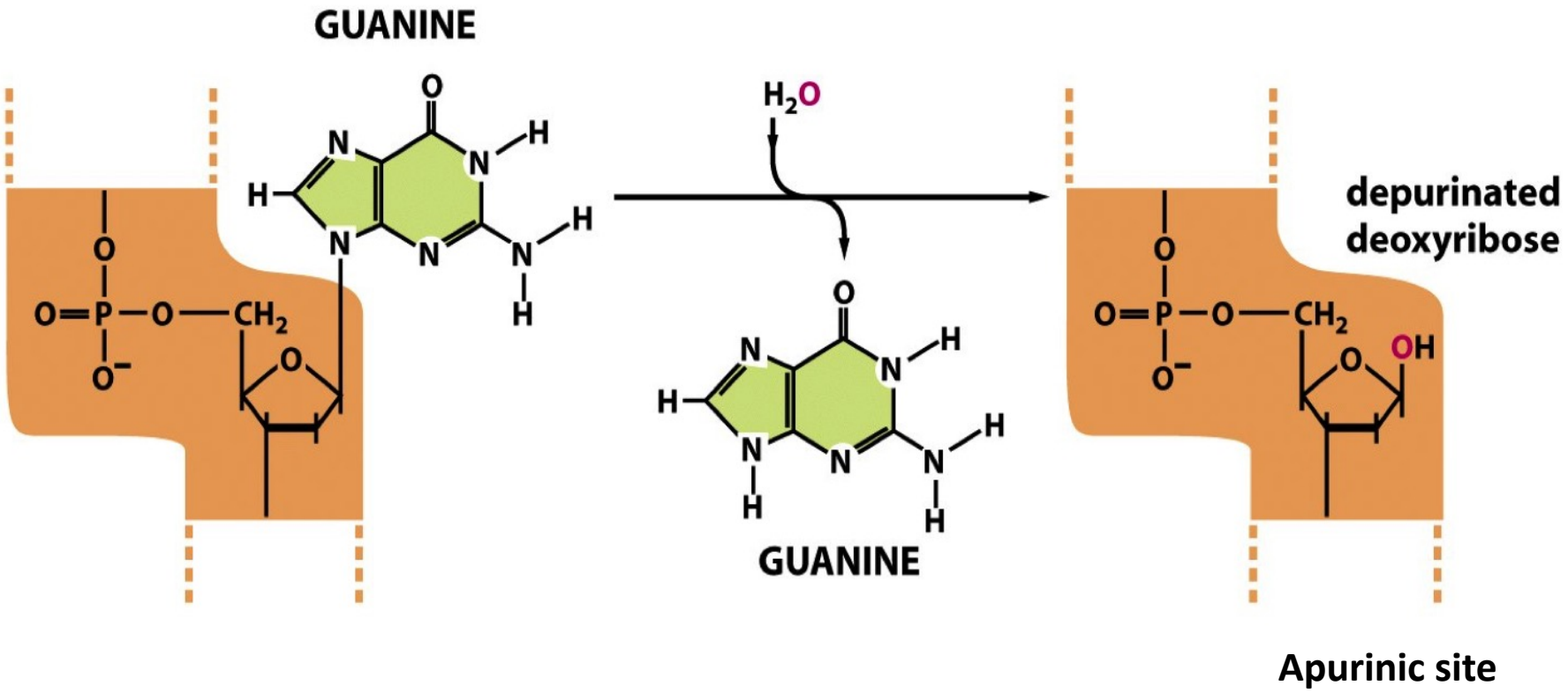
Cytosine **Uracil**

b) Deamination of 5-methylcytosine ($5^{m}C$) to thymine

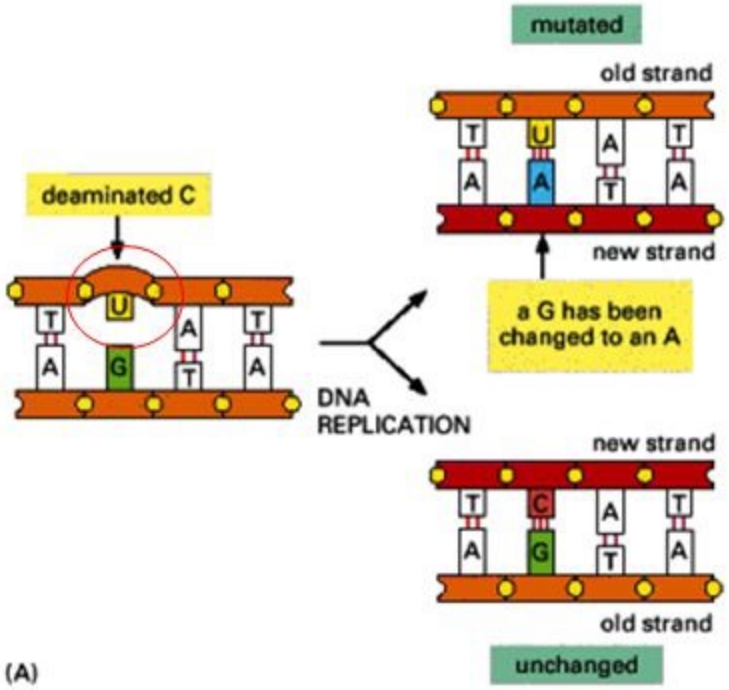


5-methylcytosine ($5^{m}C$) **Thymine (T)**

Depurination

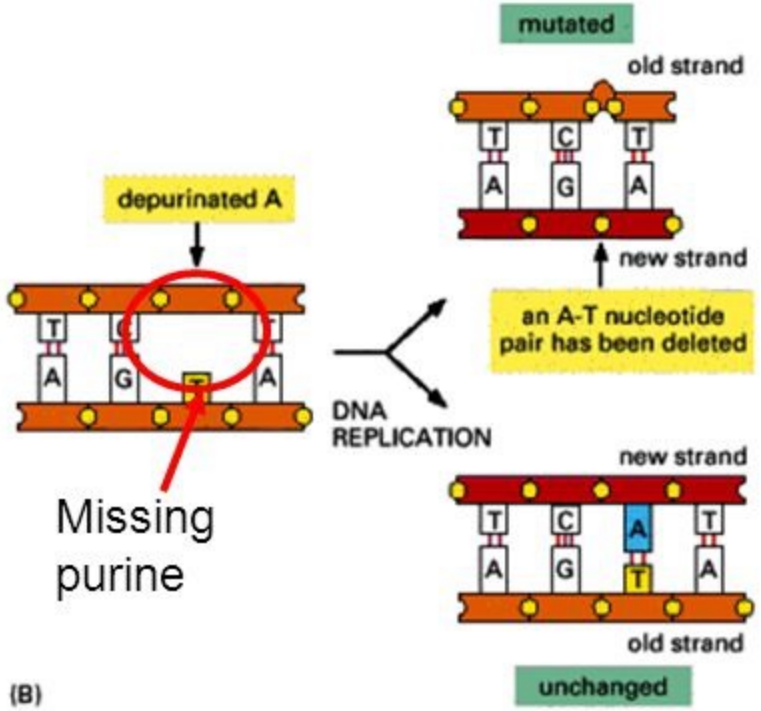


A. Deamination of cytosine produces uracil



Results in the substitution of one base for another when the DNA is replicated

B. Depurination



If uncorrected, can lead to either the substitution or the loss of a nucleotide pair.

Induced Mutations

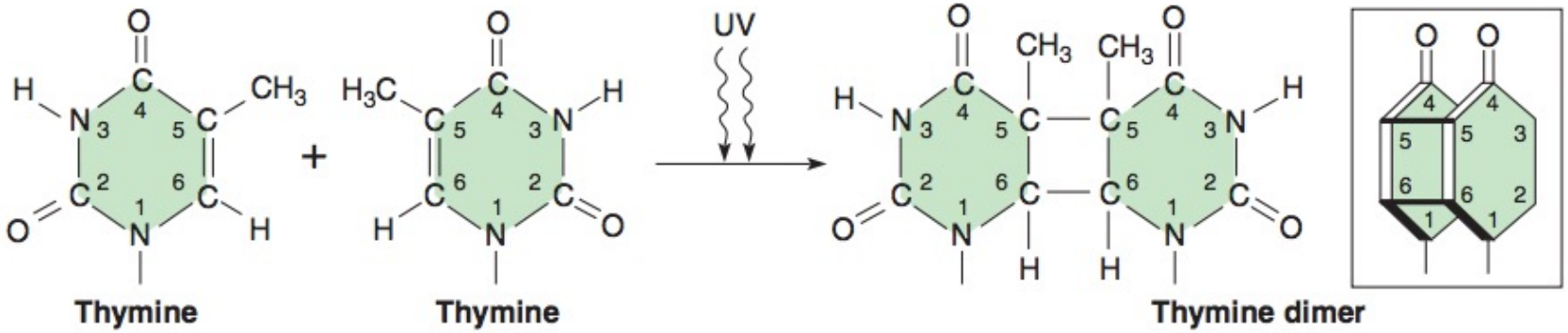
Mutagens

- Physical : Radiations
- Chemical
- Biological

UV Radiation

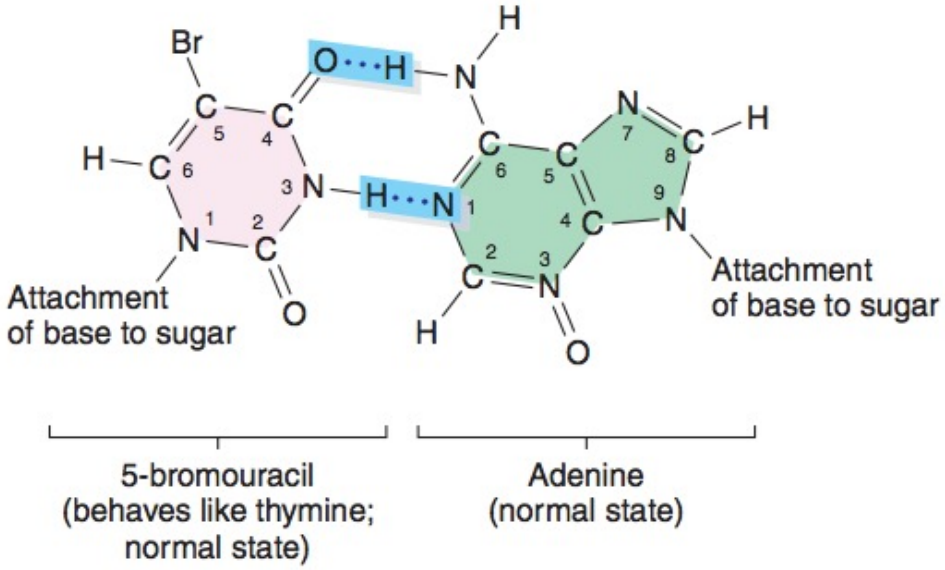
Figure 7.10

Production of thymine dimers by ultraviolet light irradiation. The two components of the dimer are covalently linked in such a way that the DNA double helix is distorted at that position.

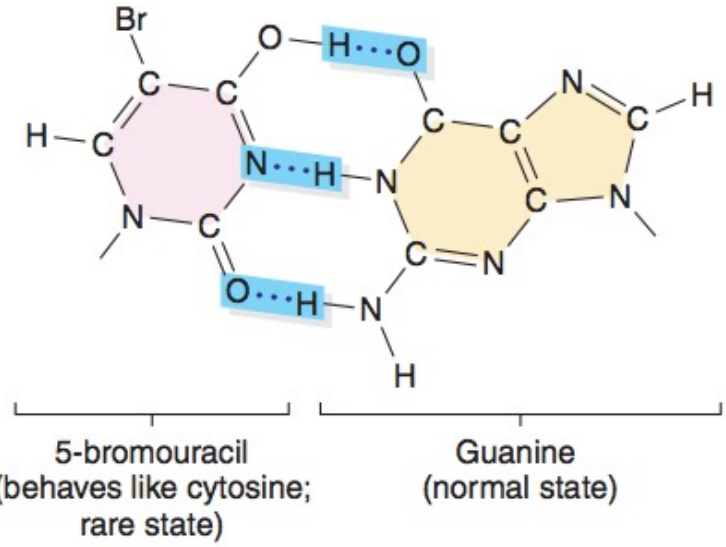


Base Analogs

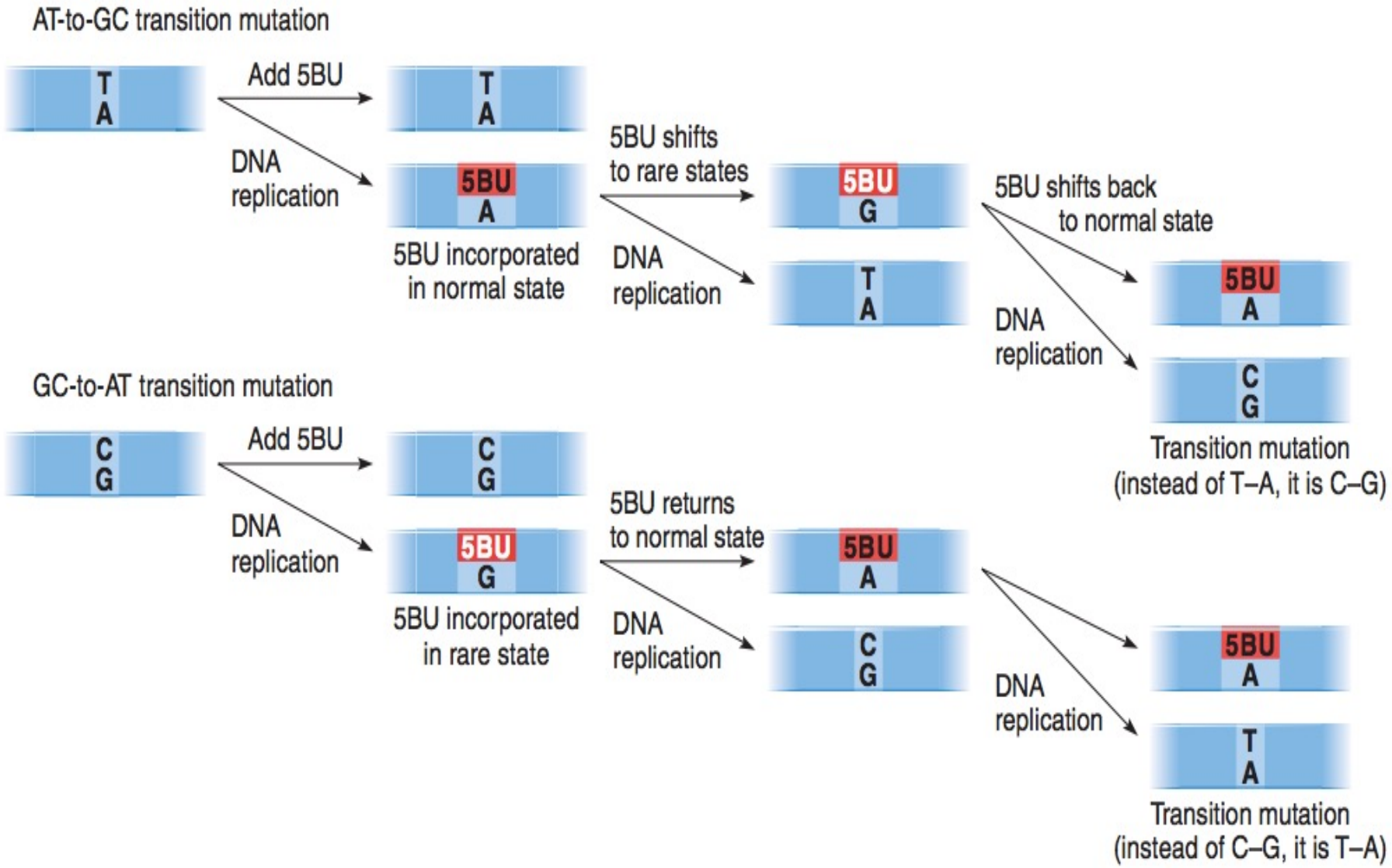
a) Base pairing of 5-bromouracil in its normal state



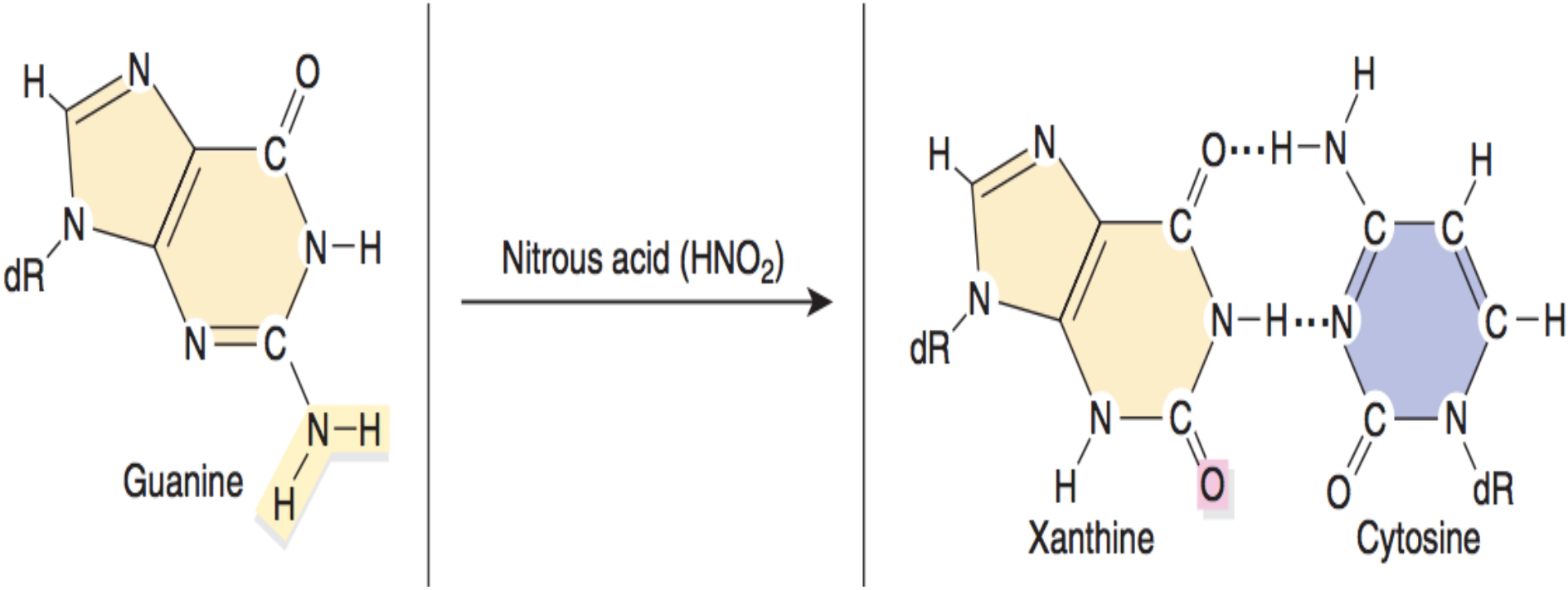
b) Base pairing of 5-bromouracil in its rare state

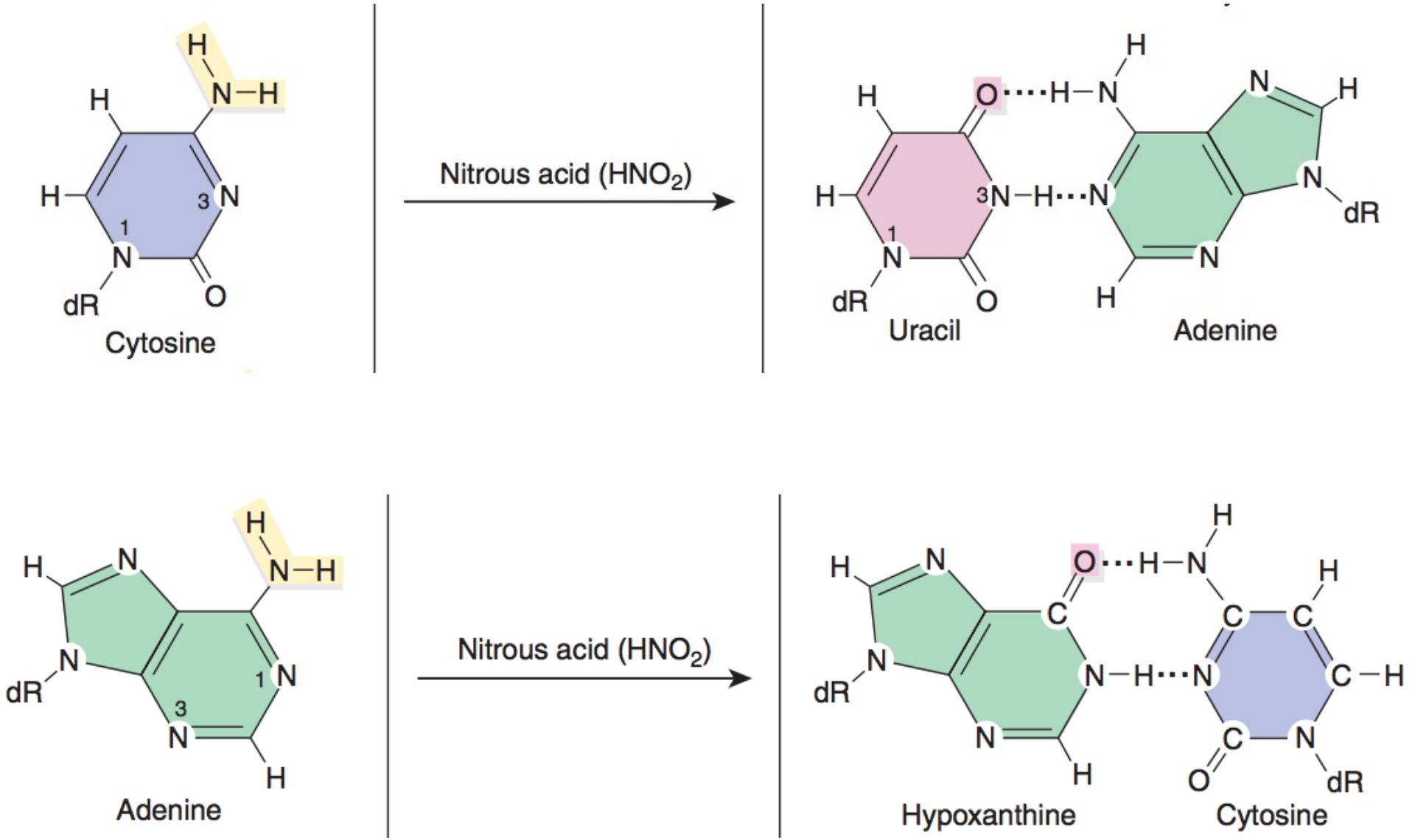


c) Mutagenic action of 5BU

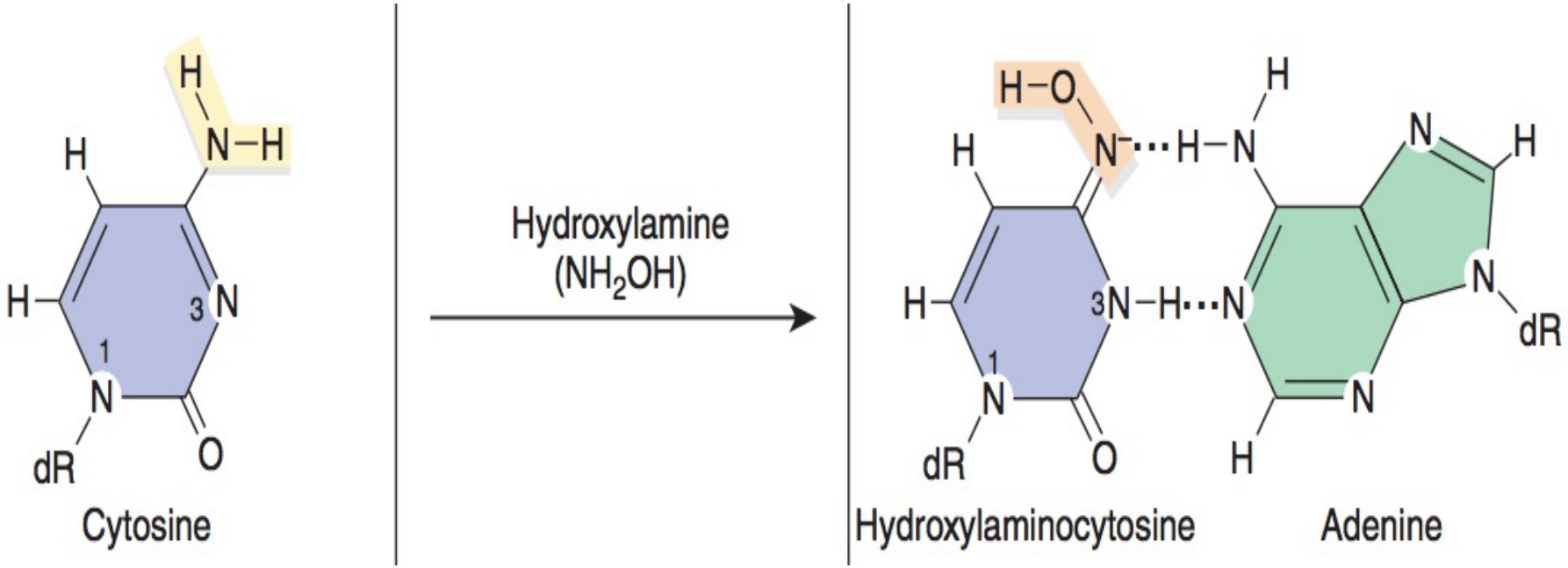


Deaminating Agent

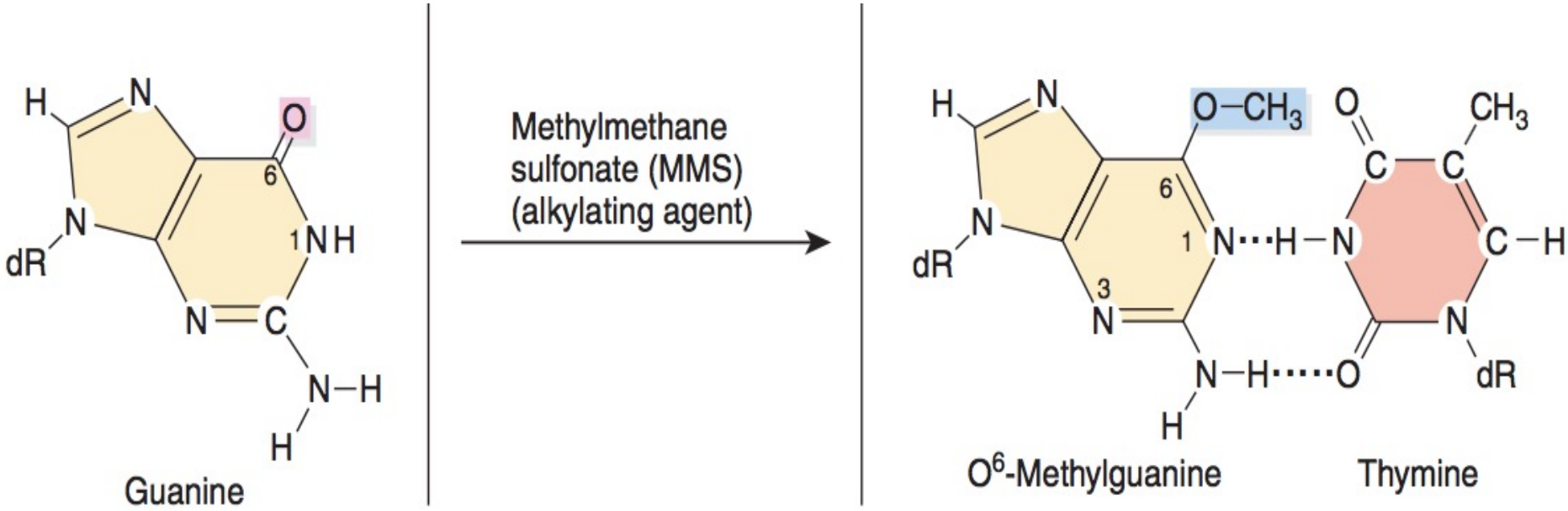



Nitrous acid (HNO_2)
Cytosine
Uracil
Adenine
Nitrous acid (HNO_2)
Adenine
Hypoxanthine
Cytosine

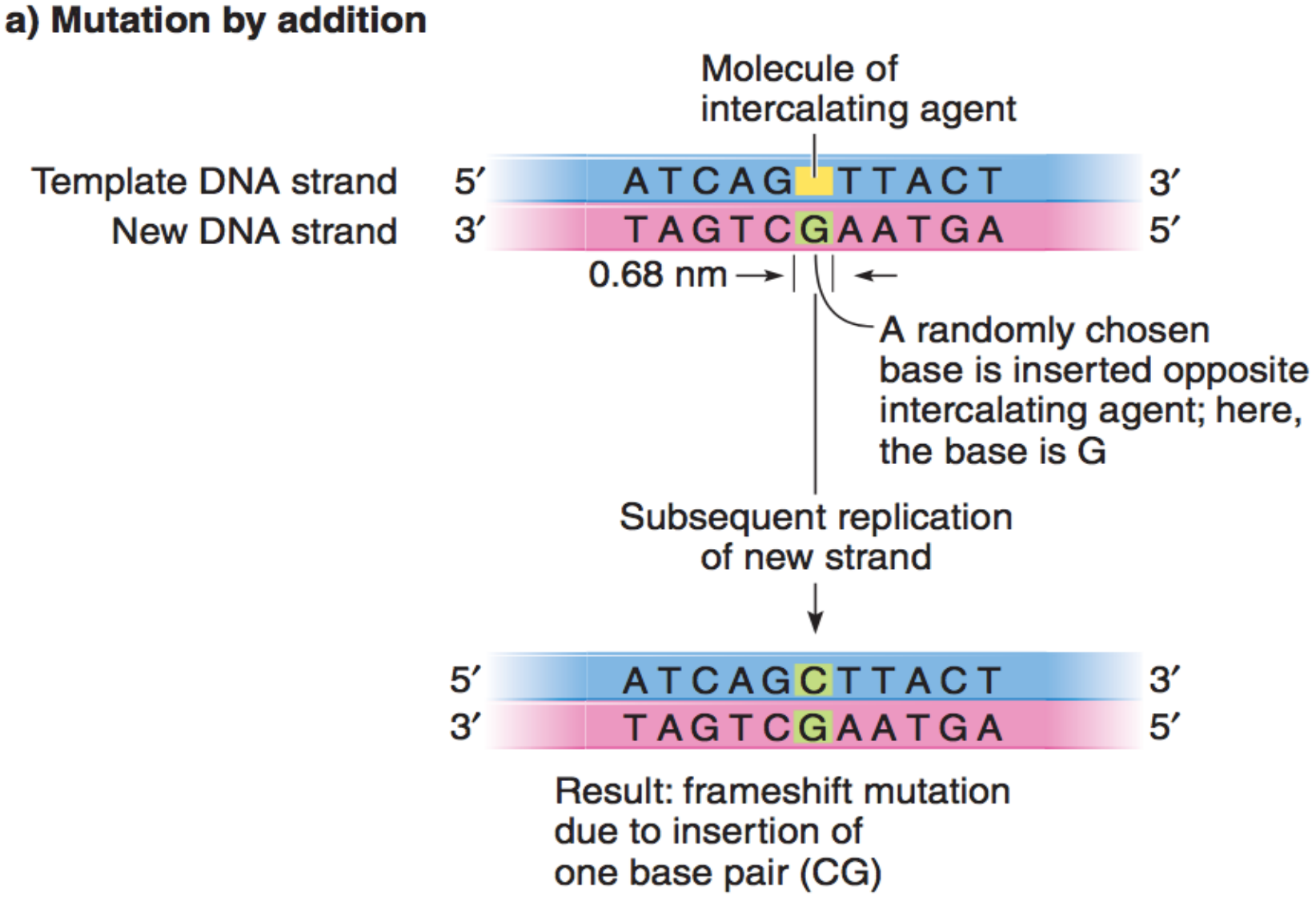
Hydroxylating Agent



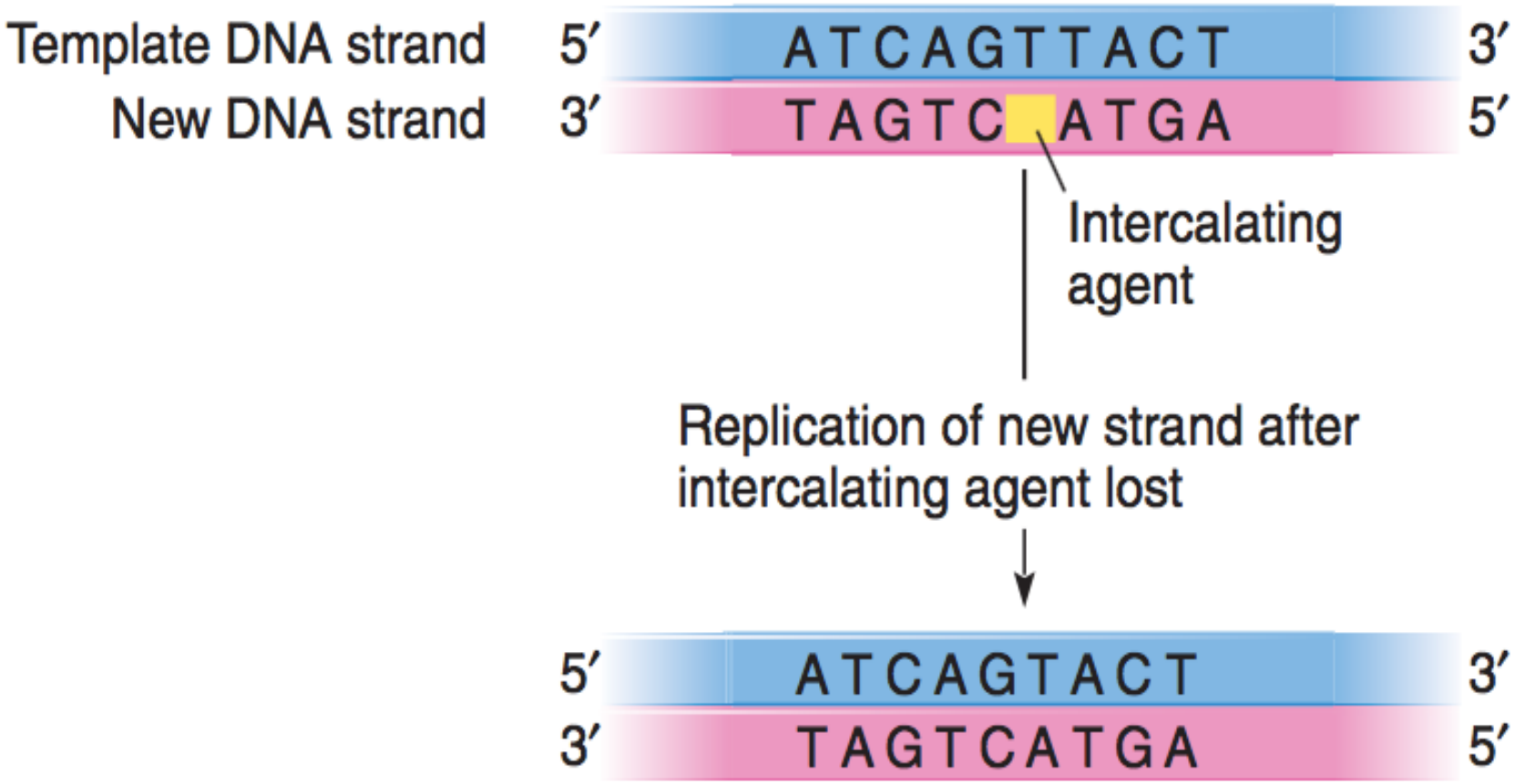
Alkylating Agent



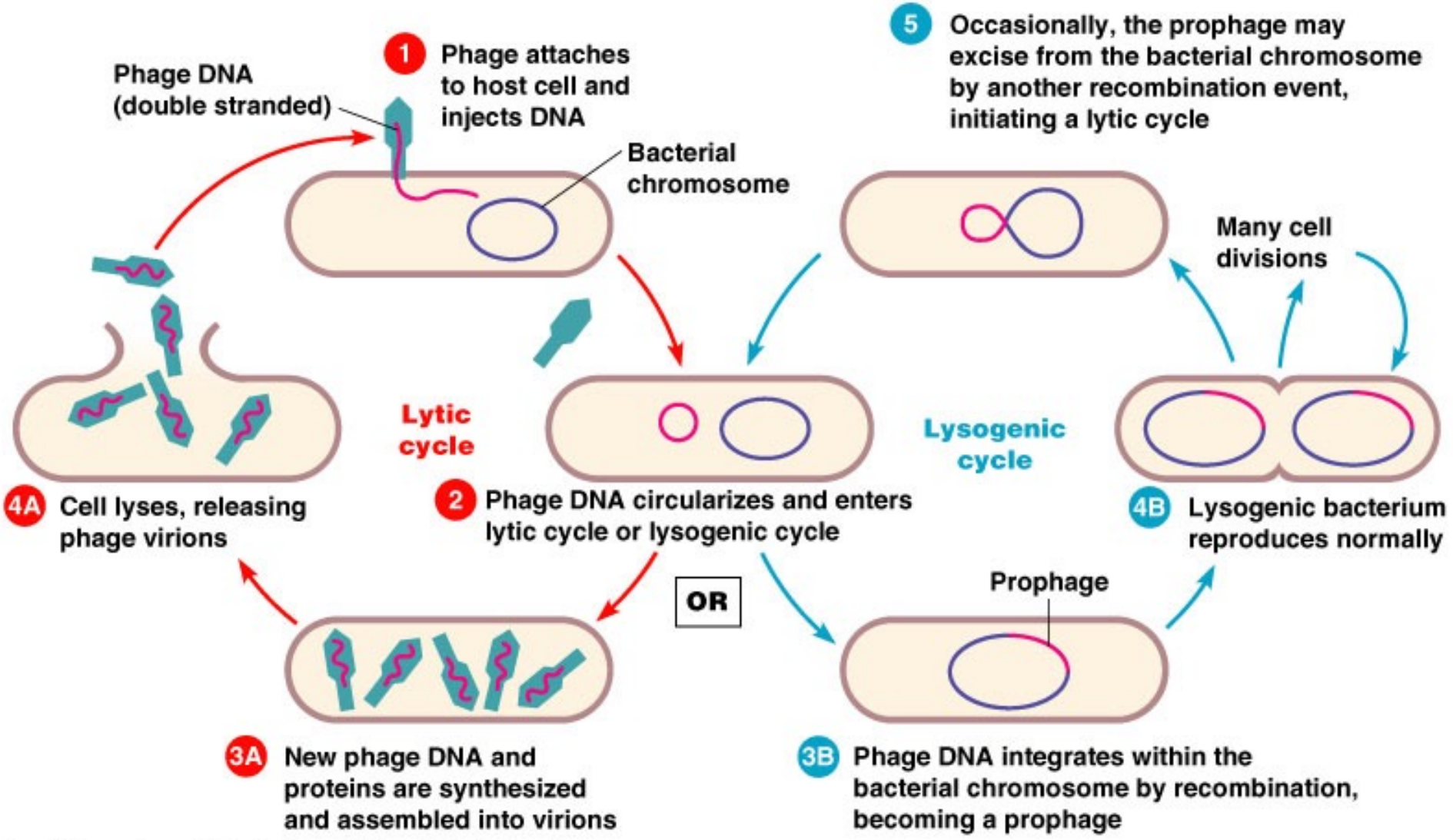
Intercalating Agent



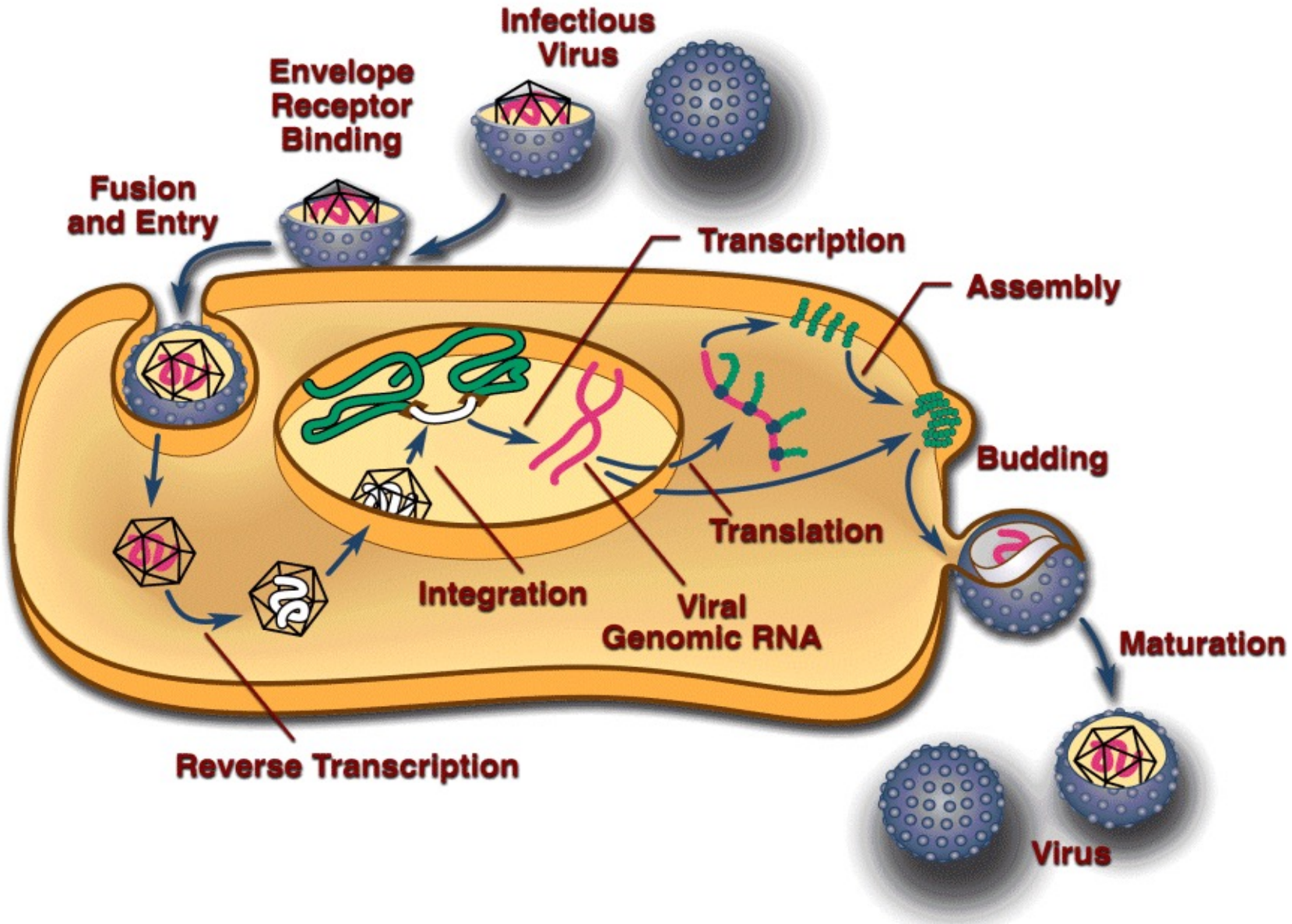
b) Mutation by deletion



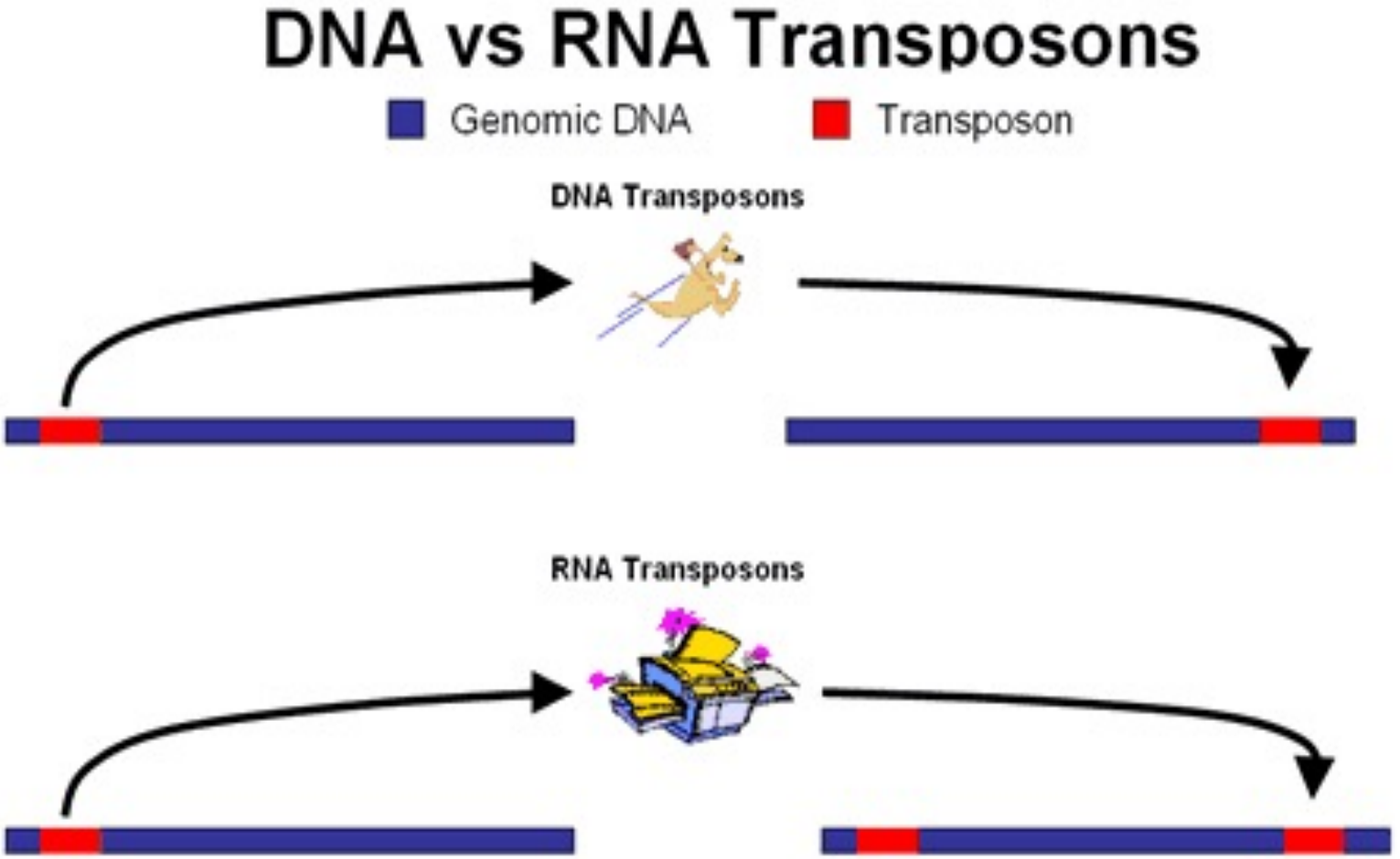
Biological Agents

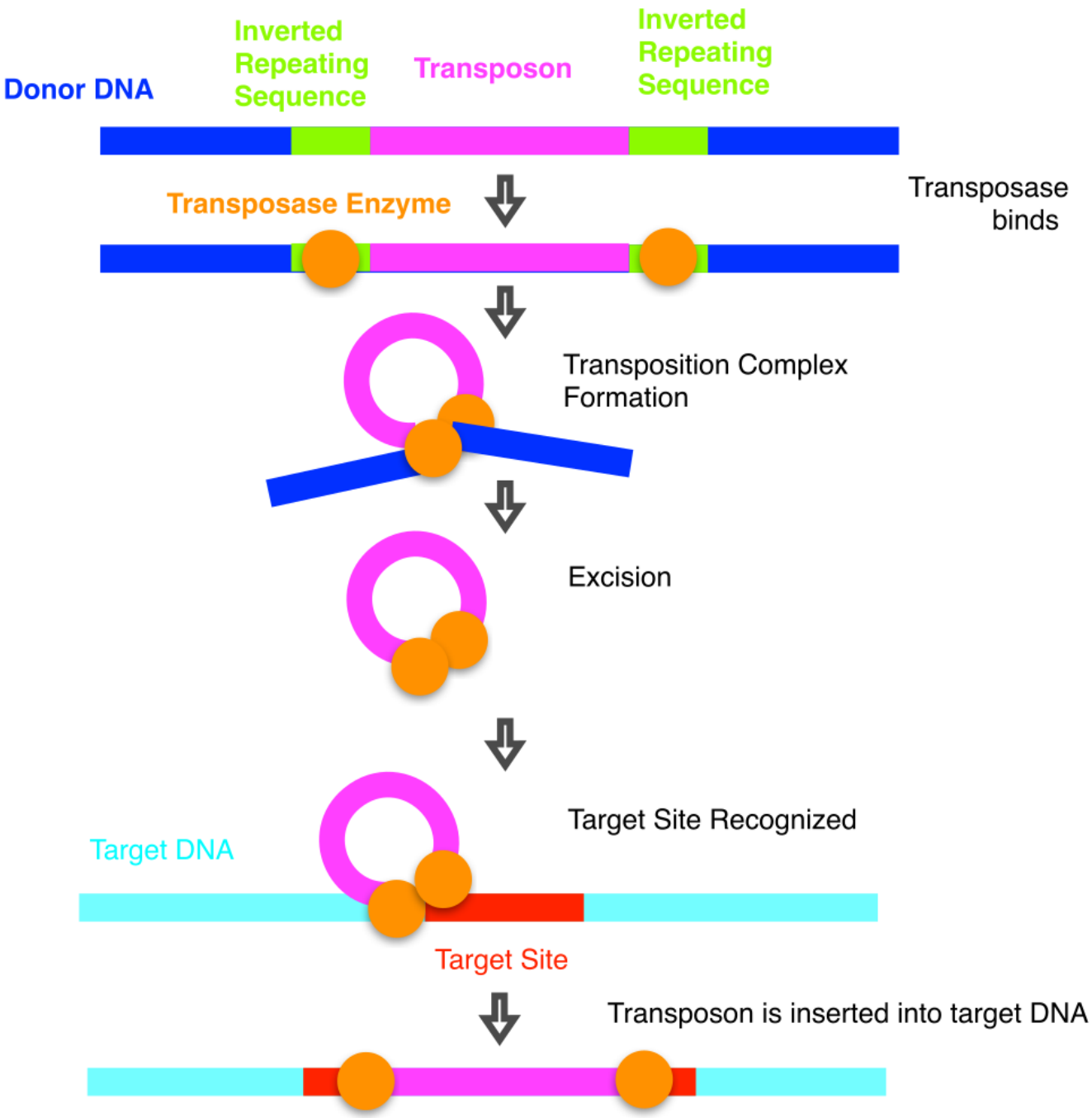


Retrovirus


Infectious Virus
Envelope Receptor Binding
Fusion and Entry
Transcription
Assembly
Budding
Translation
Integration
Viral Genomic RNA
Maturation
Reverse Transcription
Virus

Transposon


DNA vs RNA Transposons
Genomic DNA
Transposon
DNA Transposons
RNA Transposons


Donor DNA
Inverted Repeating Sequence
Transposon
Inverted Repeating Sequence
Transposase Enzyme
Transposase binds
Transposition Complex Formation
Excision
Target Site Recognized
Target DNA
Target Site
Transposon is inserted into target DNA