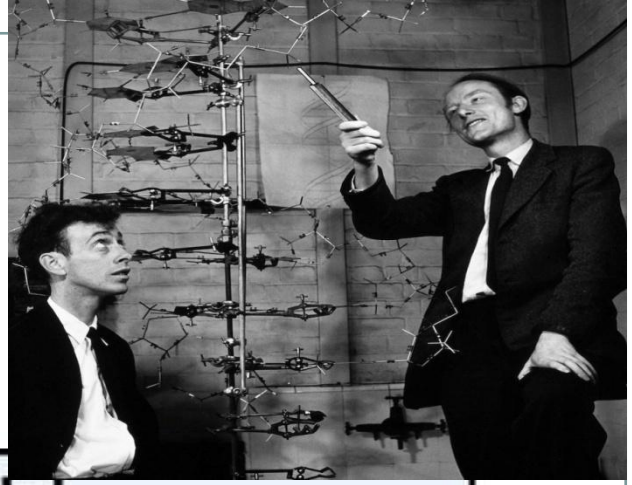
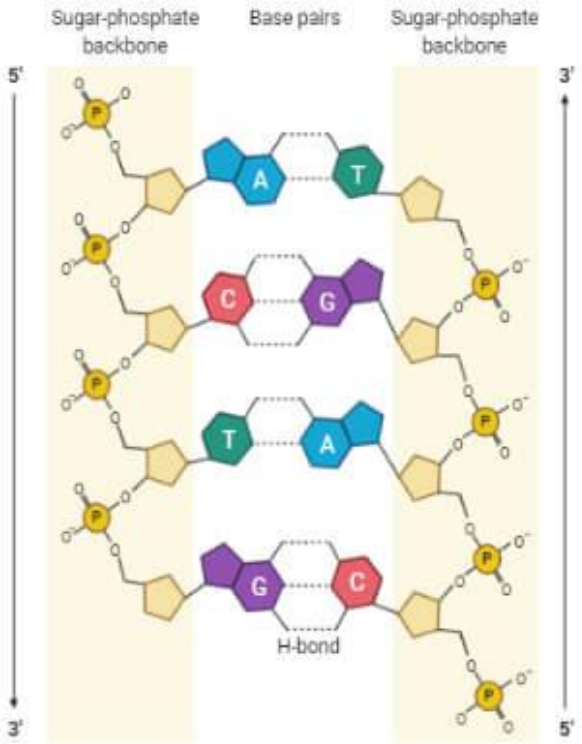
CLASS 12 BIOLOGY

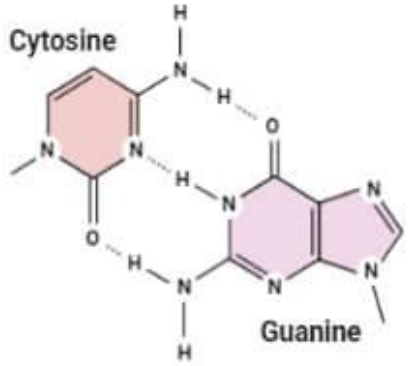
Molecular basis of inheritance

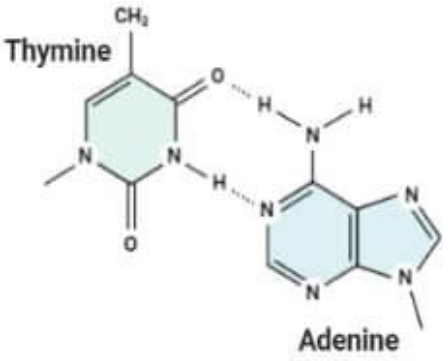
The Search Of Genetic Material.....



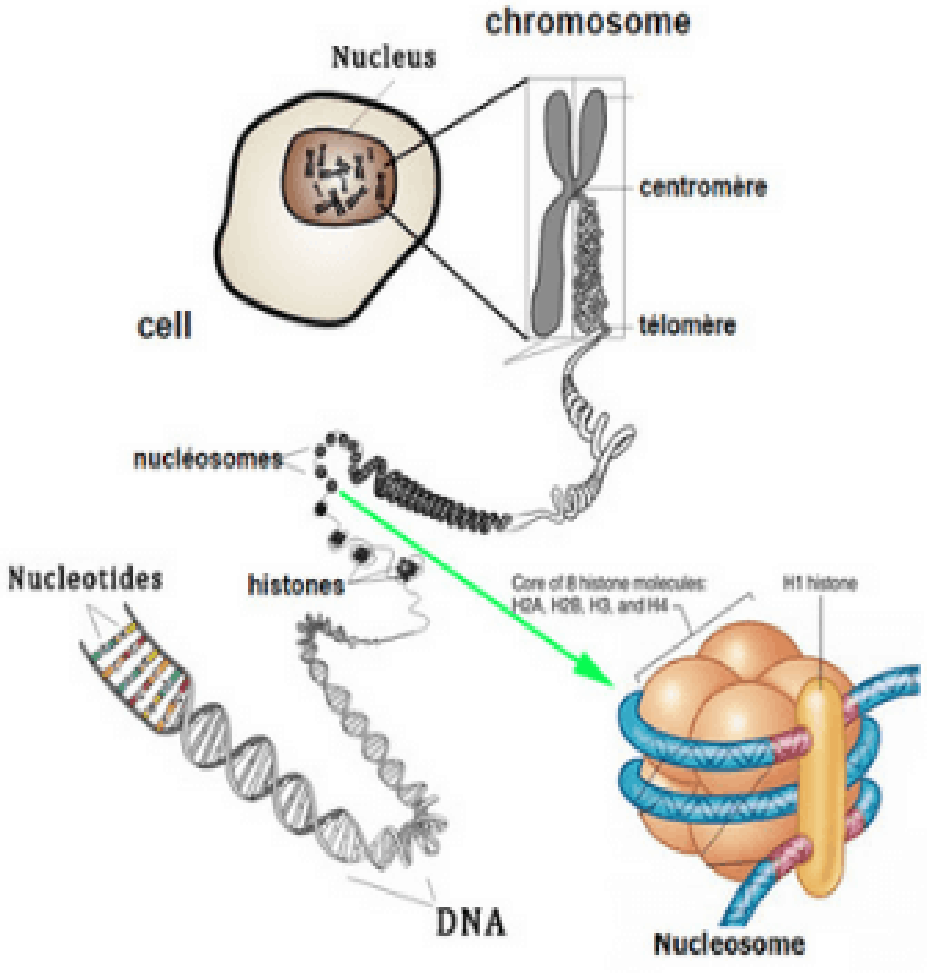
Watson and Crick DNA Model

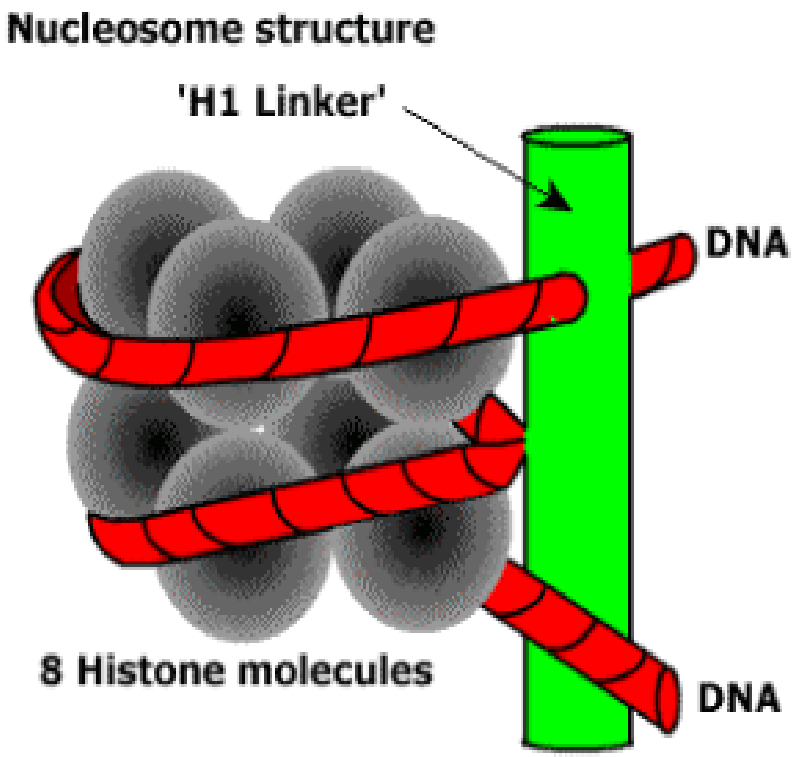






Nucleosome Model of Chromosome





A **shy** but **intelligent** Swiss boy with **hearing handicap**

↓

Isolated a unique chemical substance from nucleus of the WBCs called **nuclein, now known as DNA**

↓

Characterized nuclein to have phosphorus and nitrogen, but not sulfur

↓

Never believed his own discovered nuclein to be hereditary molecule

GRIFFITH'S "TRANSFORMING PRINCIPLE"

LIVE R-STRAIN

MOUSE LIVED

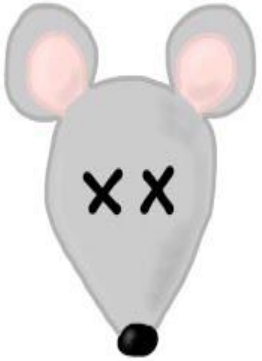
LIVE S-STRAIN

MOUSE DIED

HEAT KILLED S-STRAIN

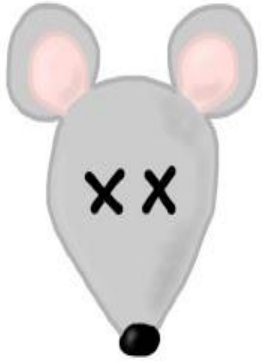
MOUSE LIVED

HEAT KILLED S-STRAIN LIVE R-STRAIN



MOUSE DIED

LIVE S-STRAIN FROM GROUP 4

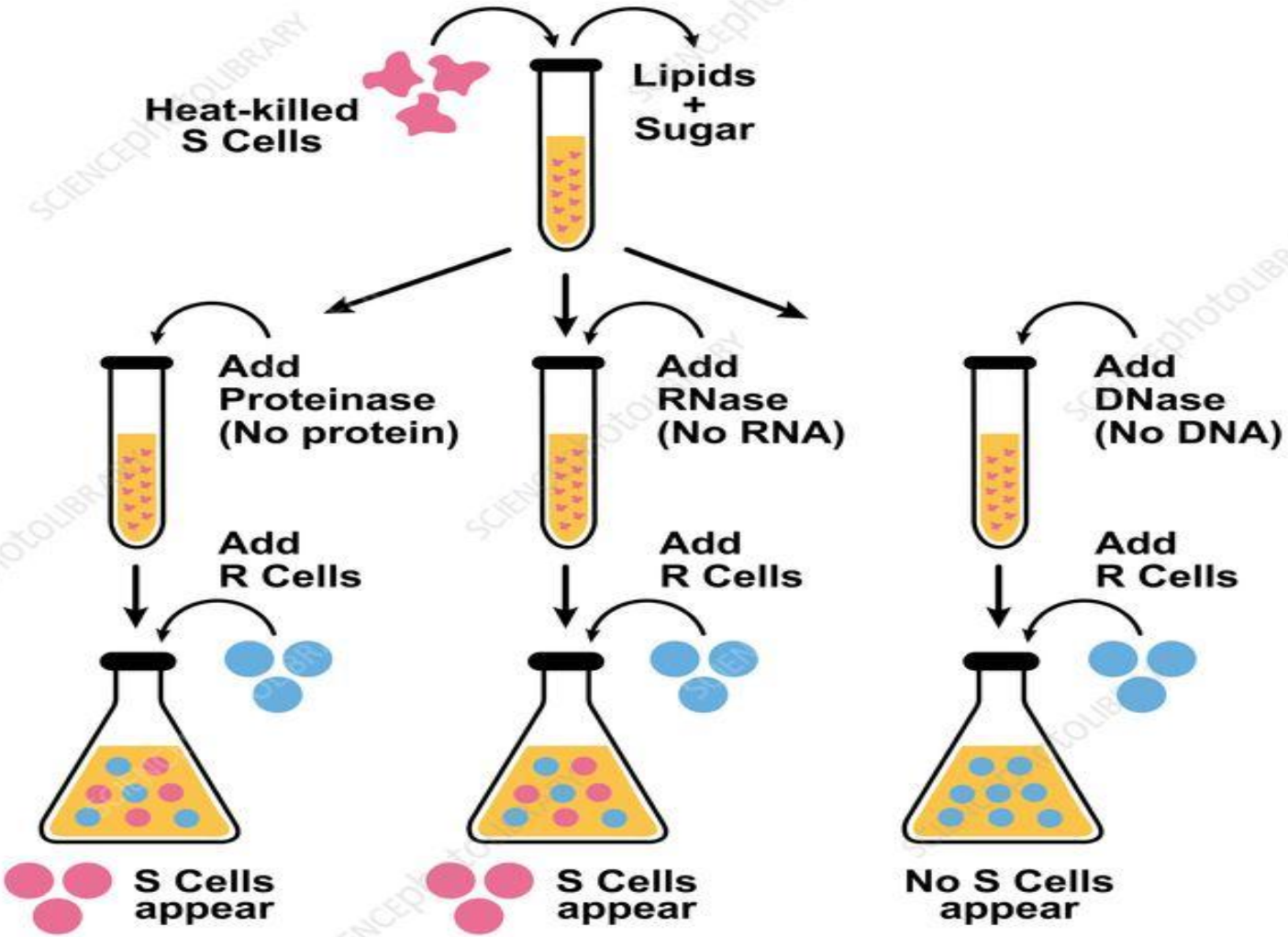


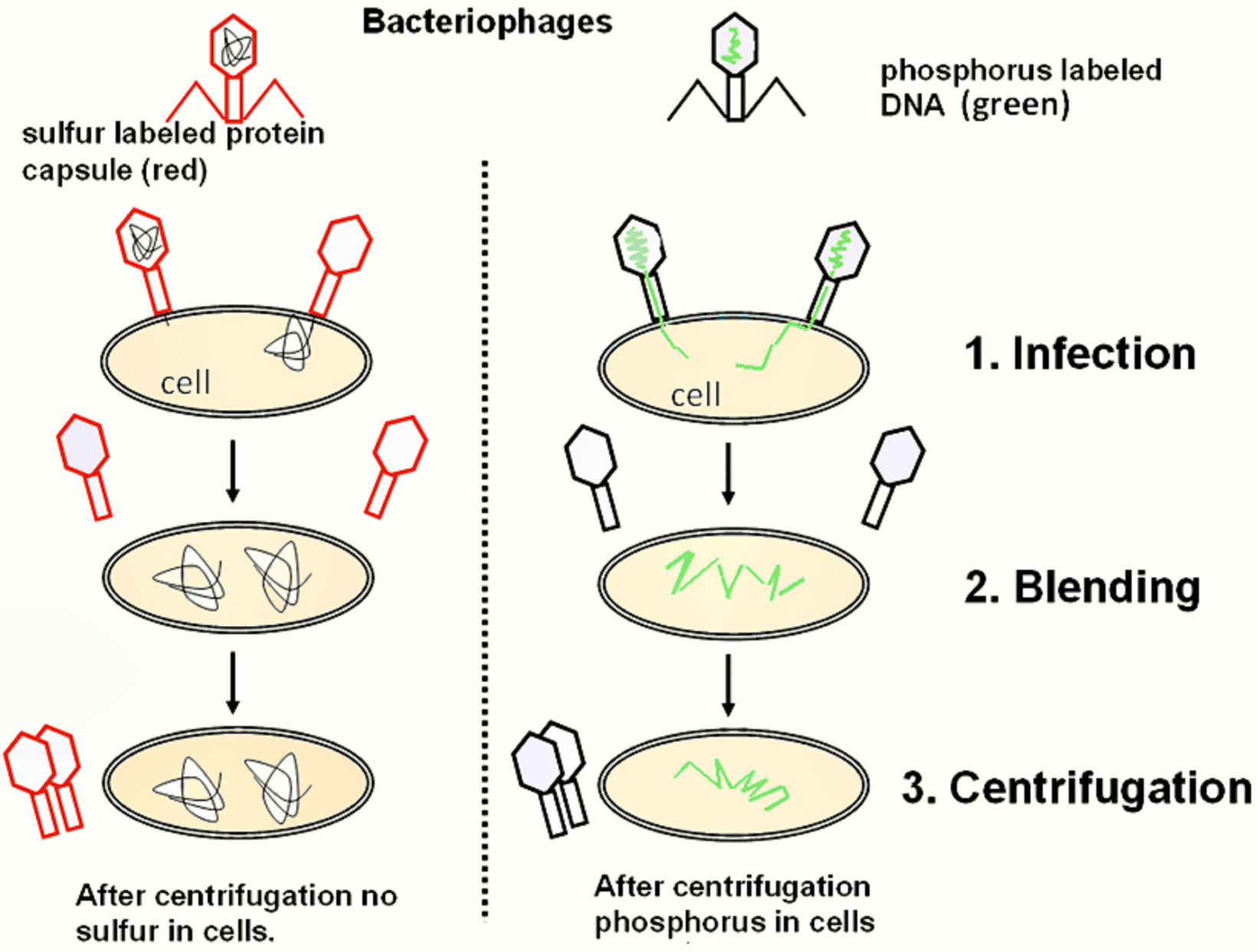
MOUSE DIED

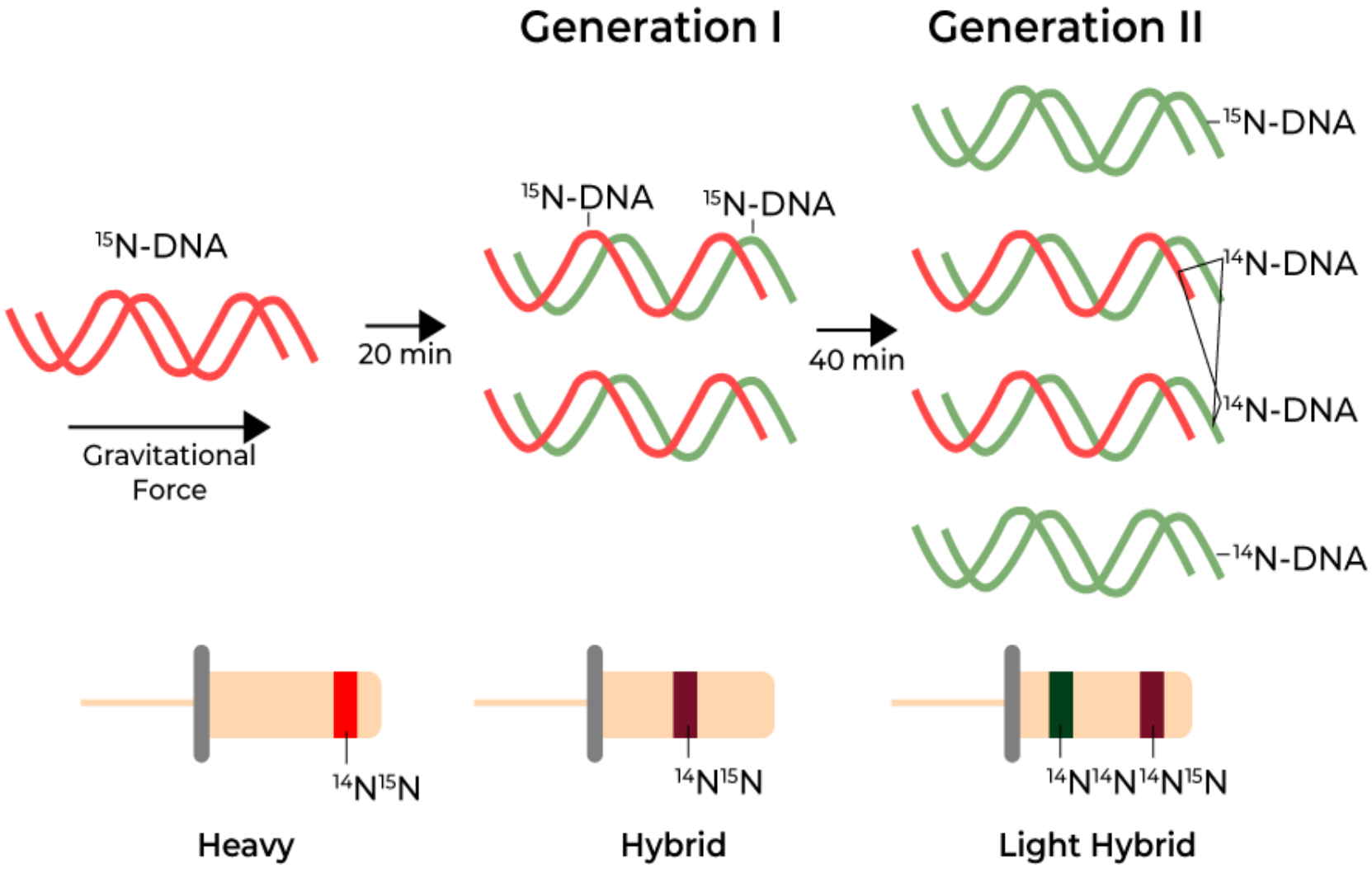
A-VIRULENT R-STRAIN

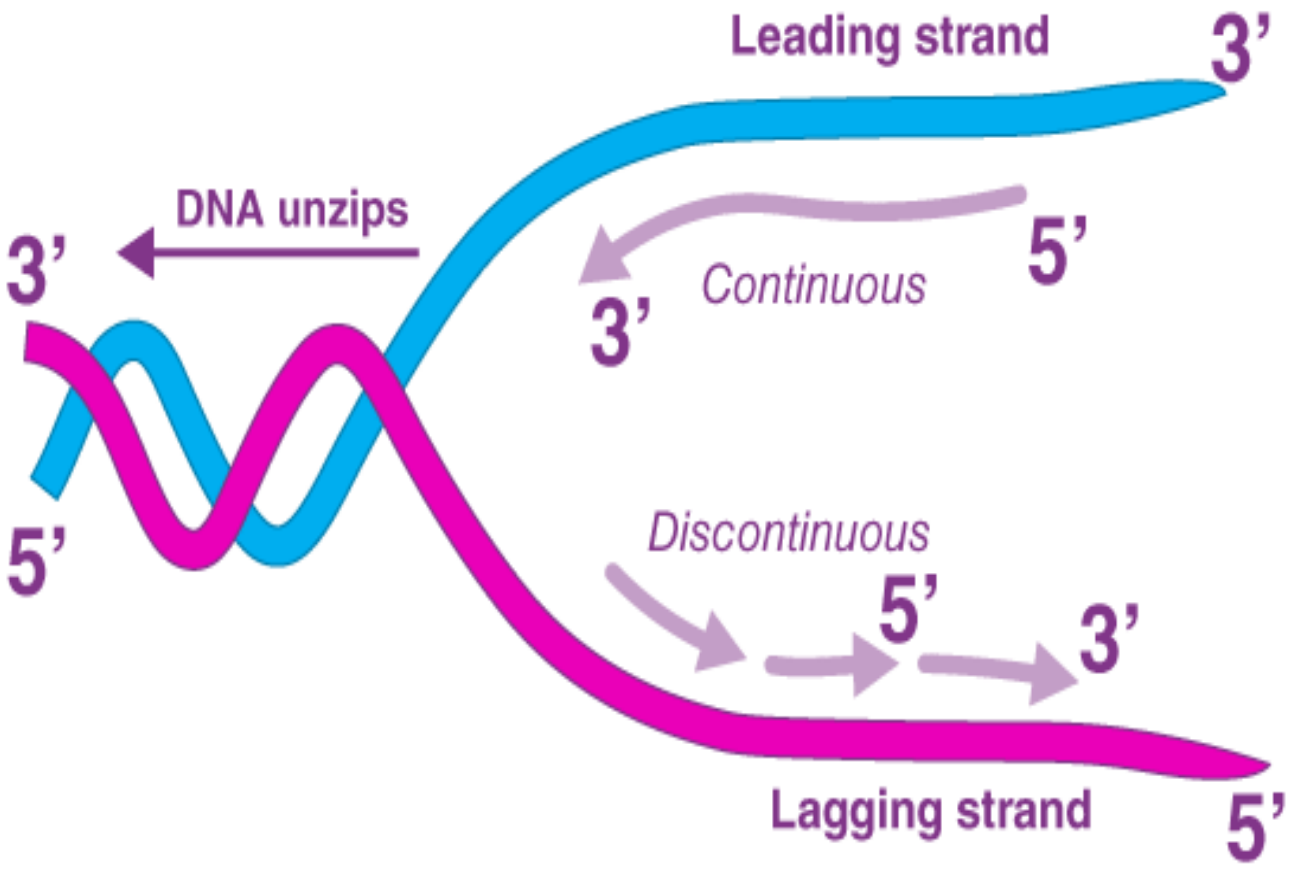
VIRULENT S-STRAIN

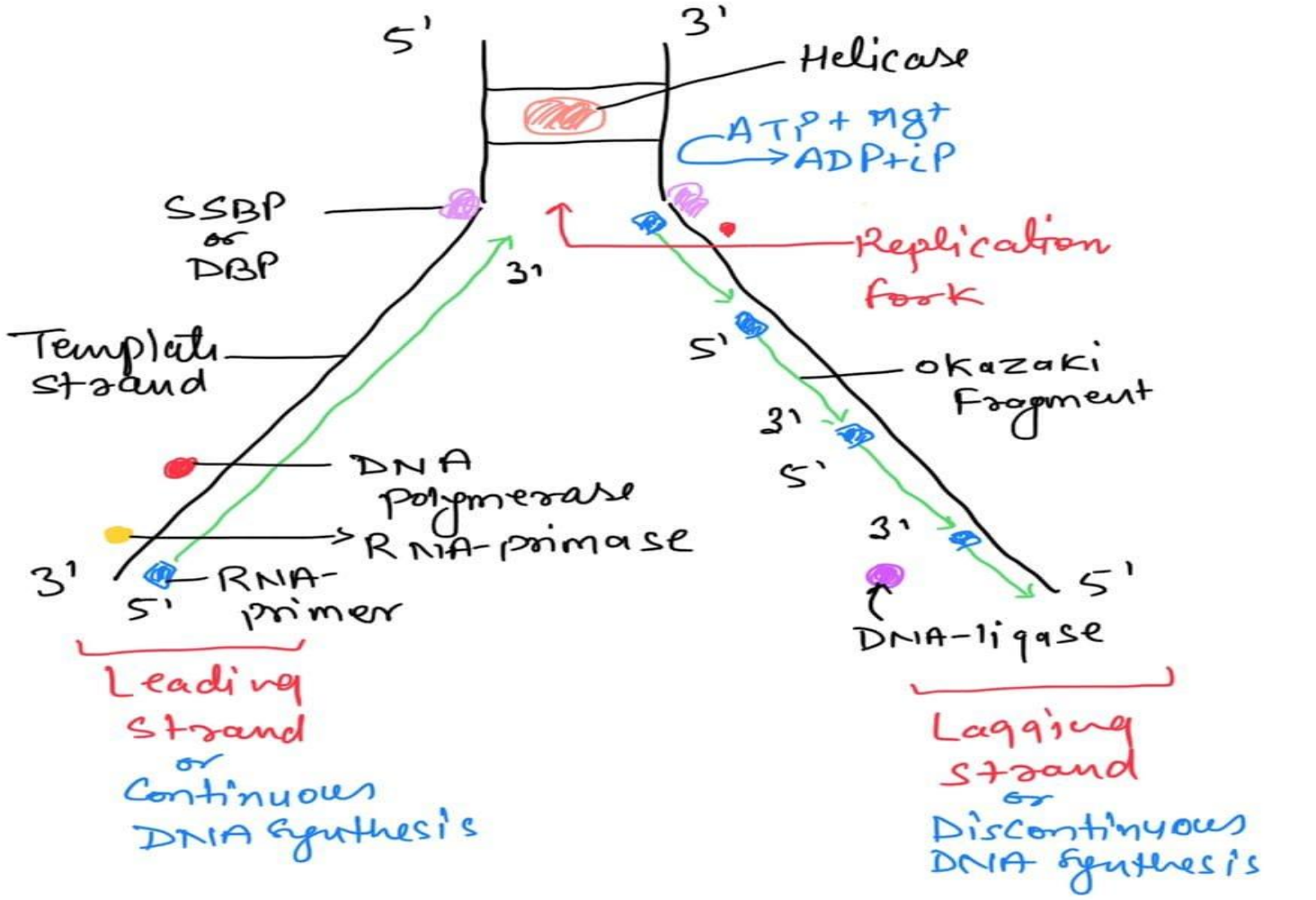
Avery MacLeod McCarty Experiment



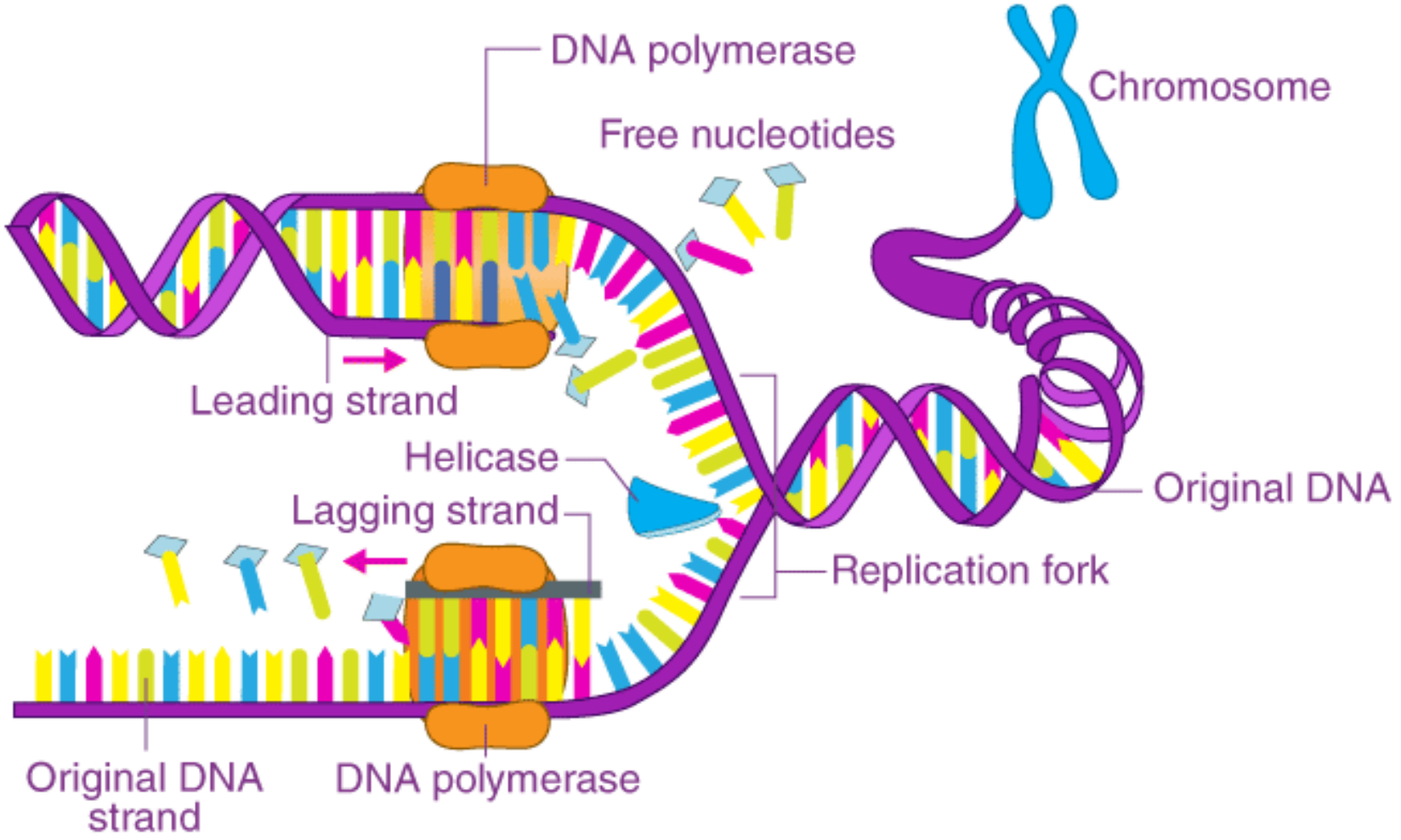

Bacteriophages
sulfur labeled protein capsule (red)
phosphorus labeled DNA (green)
cell
cell
1. Infection
2. Blending
3. Centrifugation
After centrifugation no sulfur in cells.
After centrifugation phosphorus in cells


Generation I
Generation II
15N-DNA
Gravitational Force
20 min
15N-DNA
15N-DNA
40 min
15N-DNA
14N-DNA
14N-DNA
14N-DNA
14N15N
14N15N
14N14N14N15N
Heavy
Hybrid
Light Hybrid

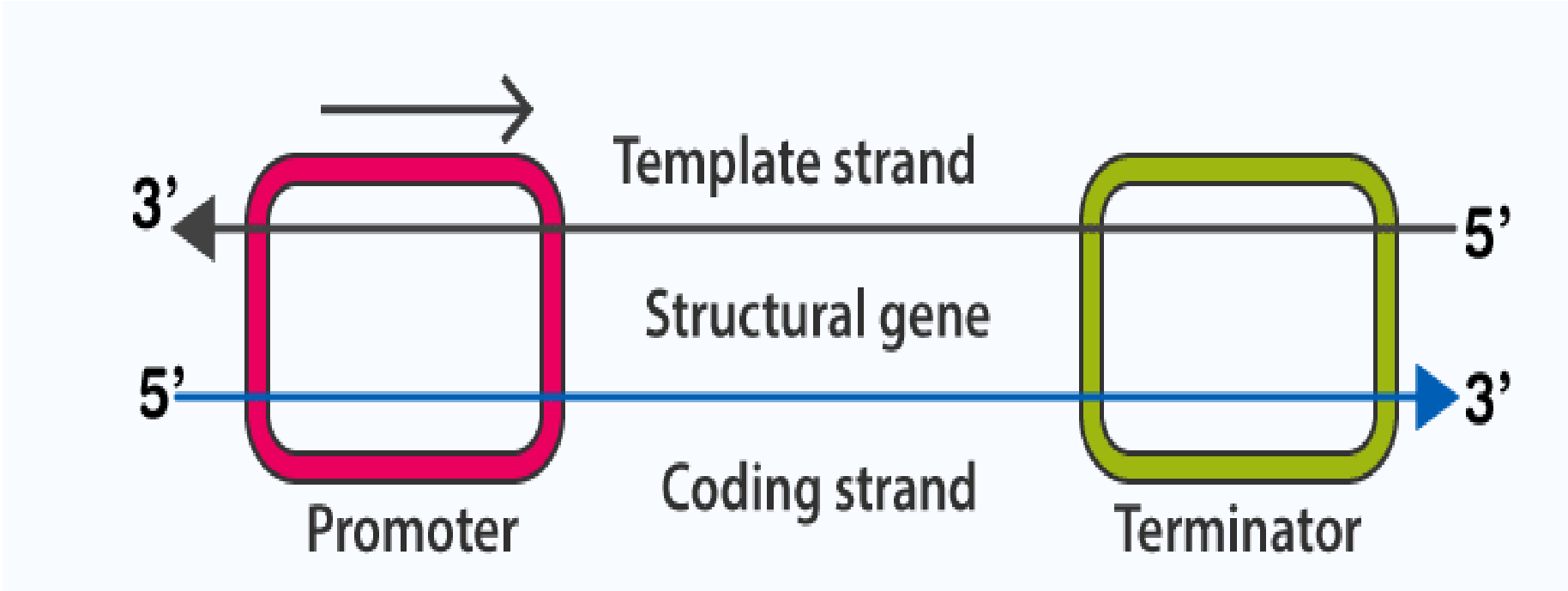

Leading strand
3’
DNA unzips
3’
5’
3’
Continuous
5’
Discontinuous
5’
3’
Lagging strand
5’

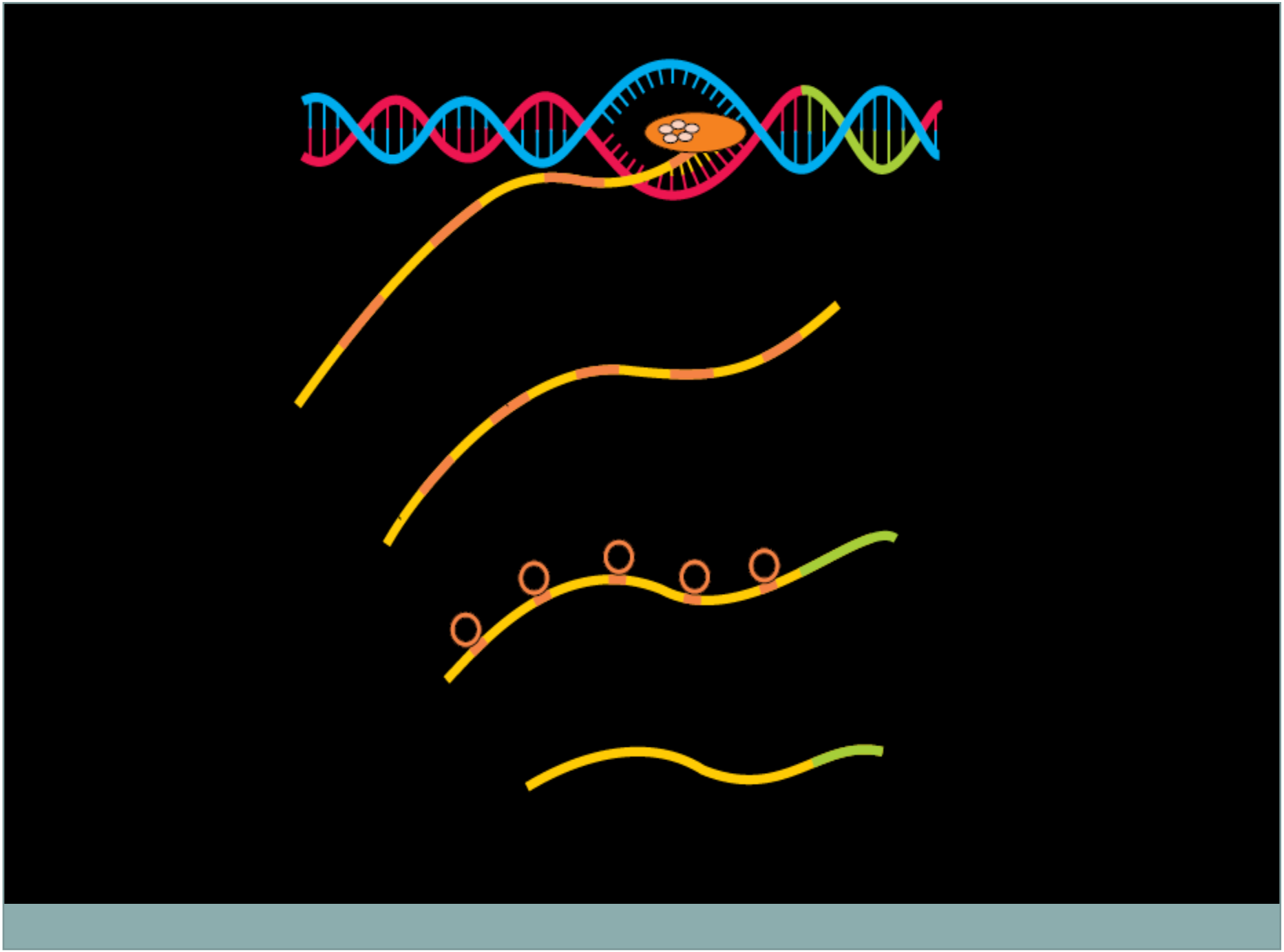


DNA Replication Simplified Diagram


DNA polymerase
Chromosome
Free nucleotides
Leading strand
Helicase
Lagging strand
Original DNA
Replication fork
Original DNA strand
DNA polymerase
Adenine
Thymine
Cytosine
Guanine

Enzyme	Function
DNA polymerase III	The 'replicase' enzyme
DNA polymerase I	A repair enzyme involved in removing errors and filling in gaps in the sequence
DNA ligase	Seals 'nicks' in the sugar-phosphate backbone
RNA polymerase or RNA primase	Make RNA primers to get the DNA polymerase III started
RNase H	Removes the RNA primers once they have completed their function
DNA helicase and DNA gyrase	Unwind the DNA duplex
Helix-destabilizing protein	Prevents the unwound DNA from immediately rewinding


Template strand
3'
5'
Structural gene
5'
3'
Coding strand
Promoter
Terminator



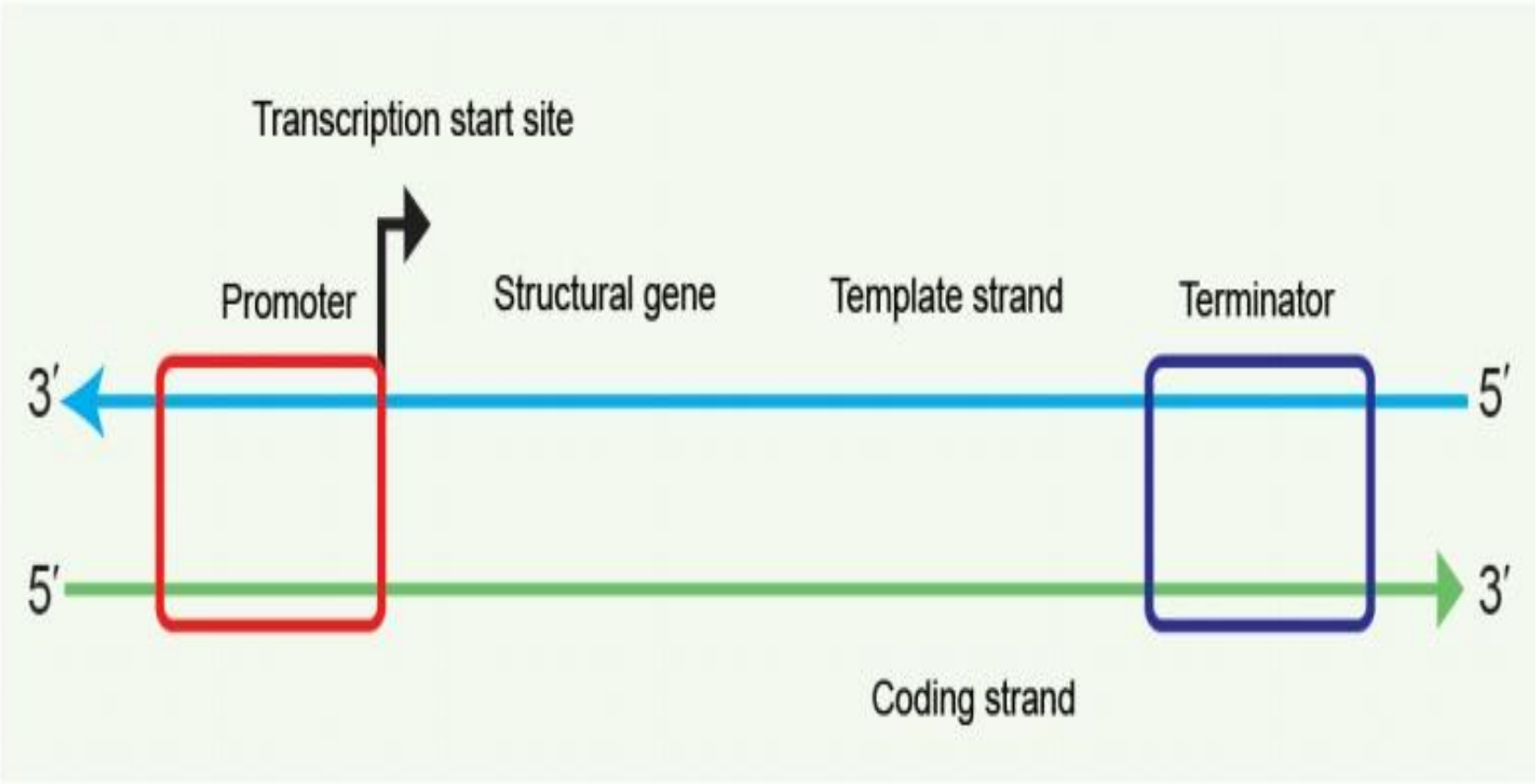
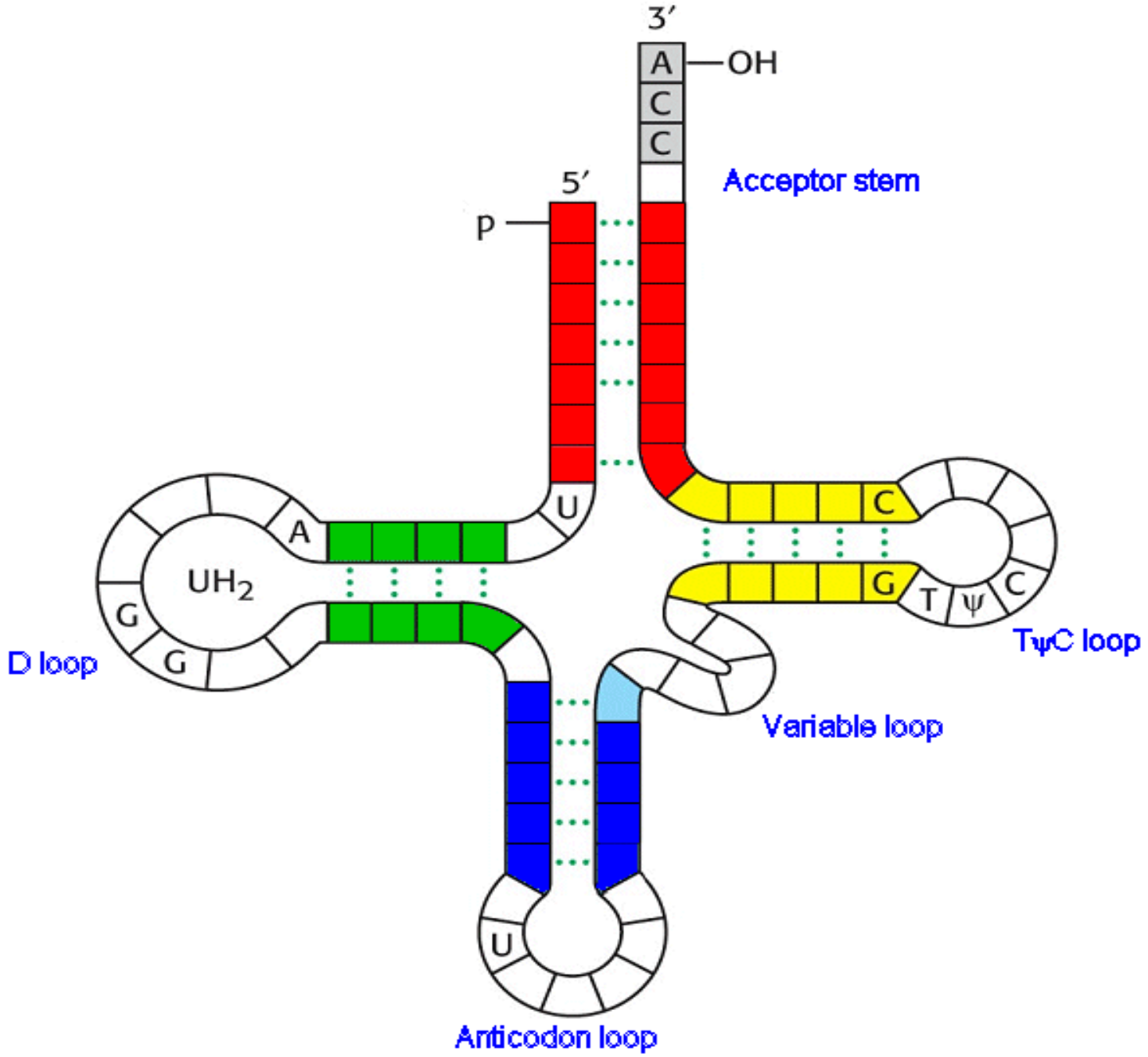
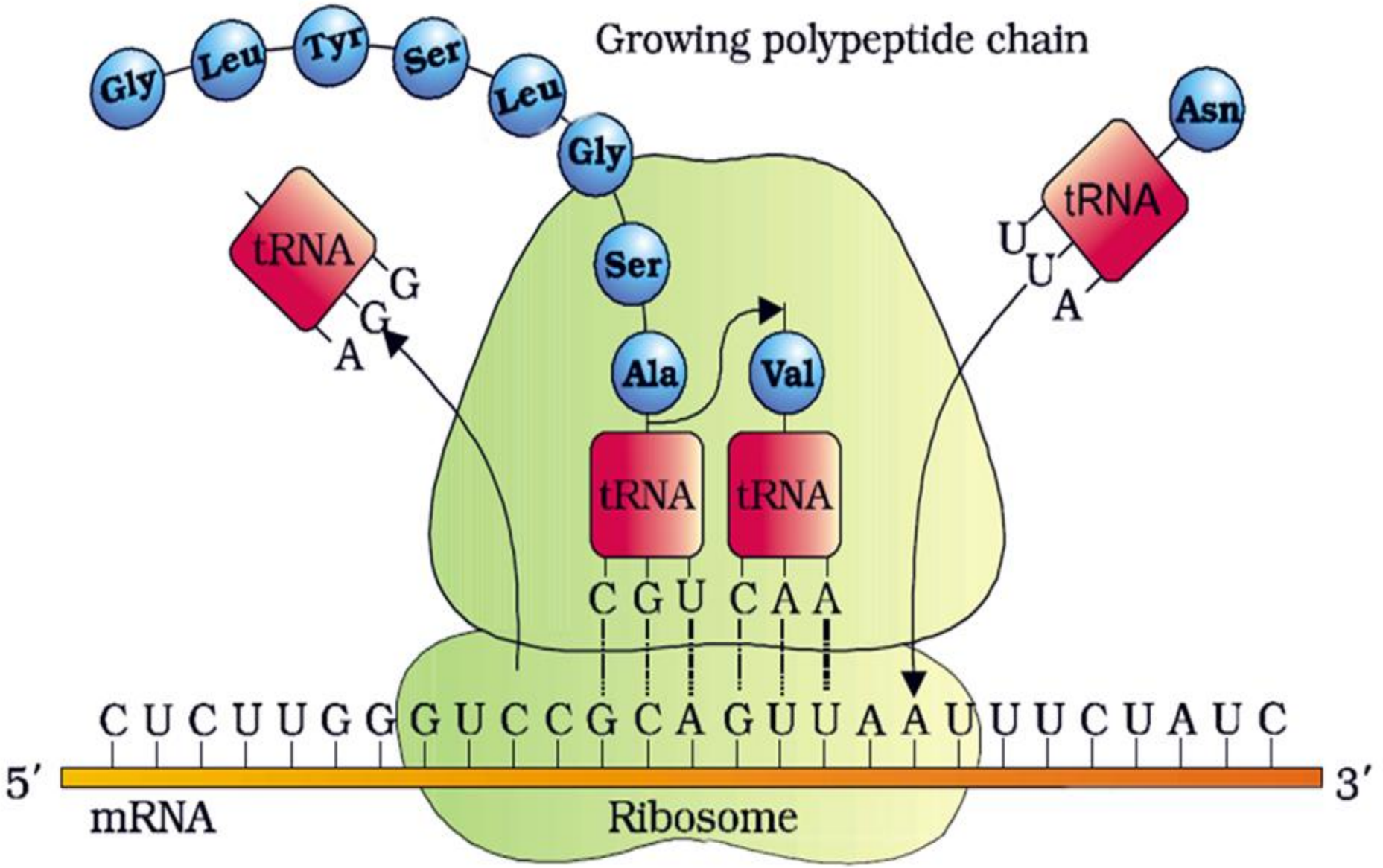
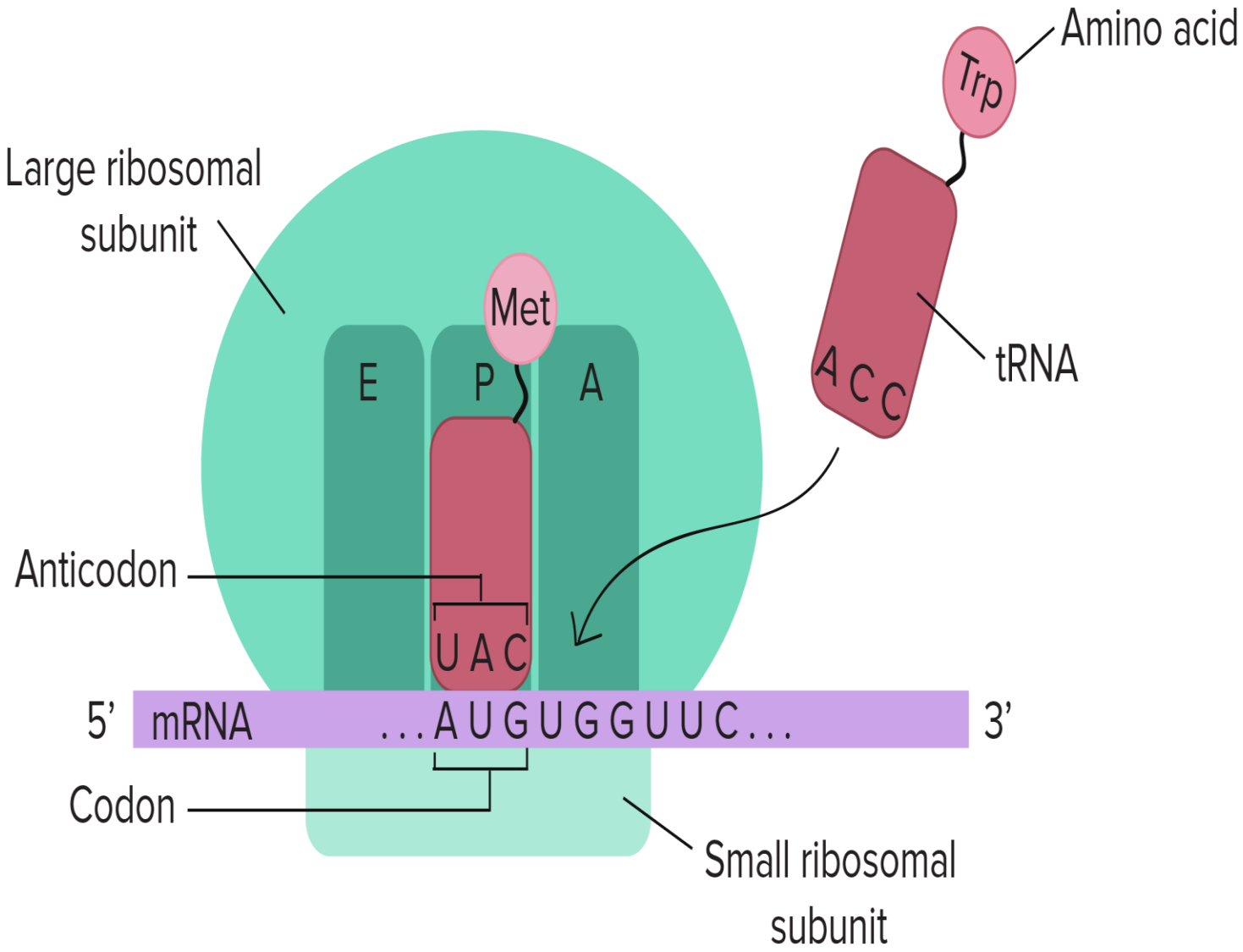


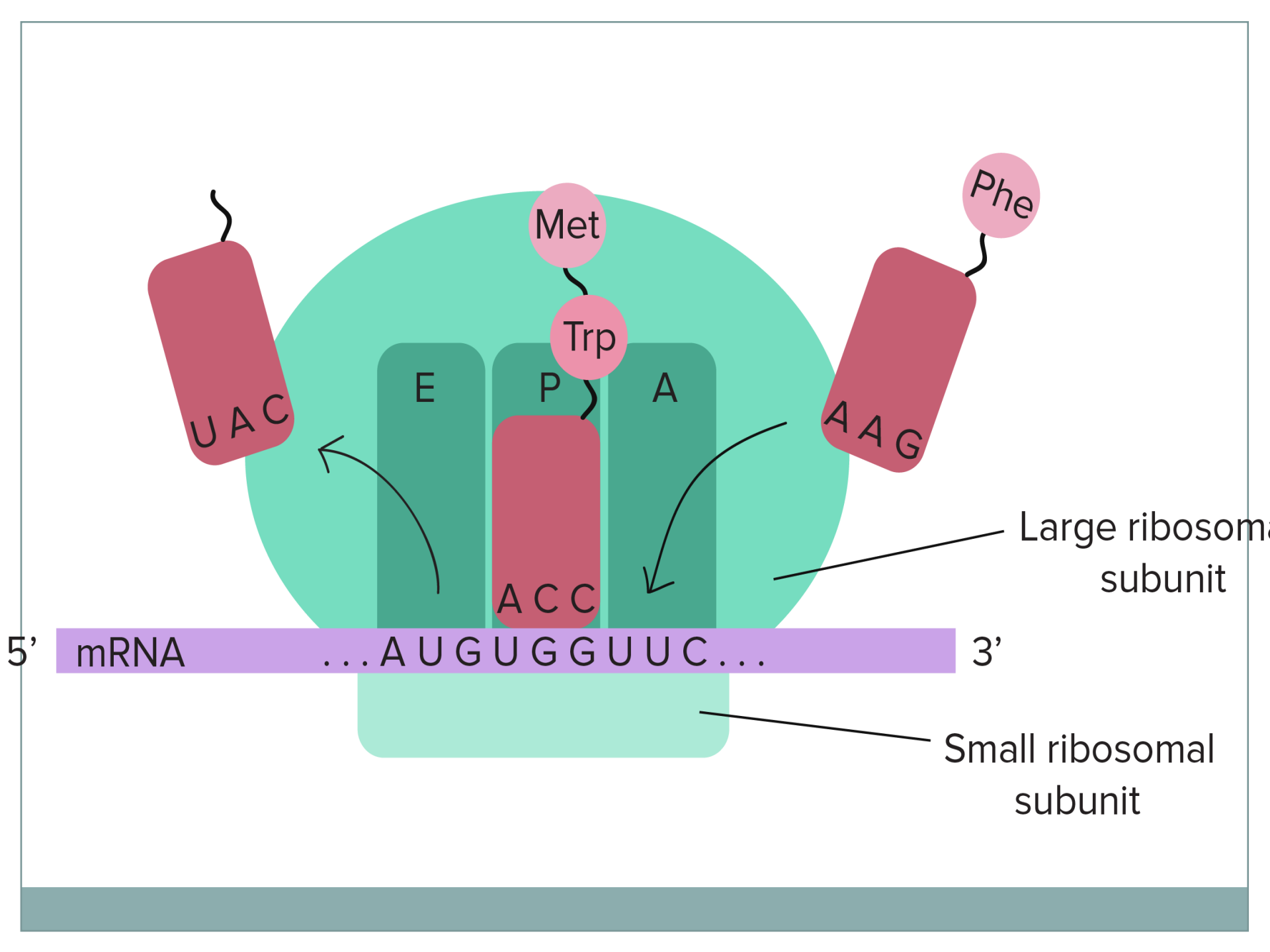
Fig. 5. 7 Schematic structure of a transcription unit

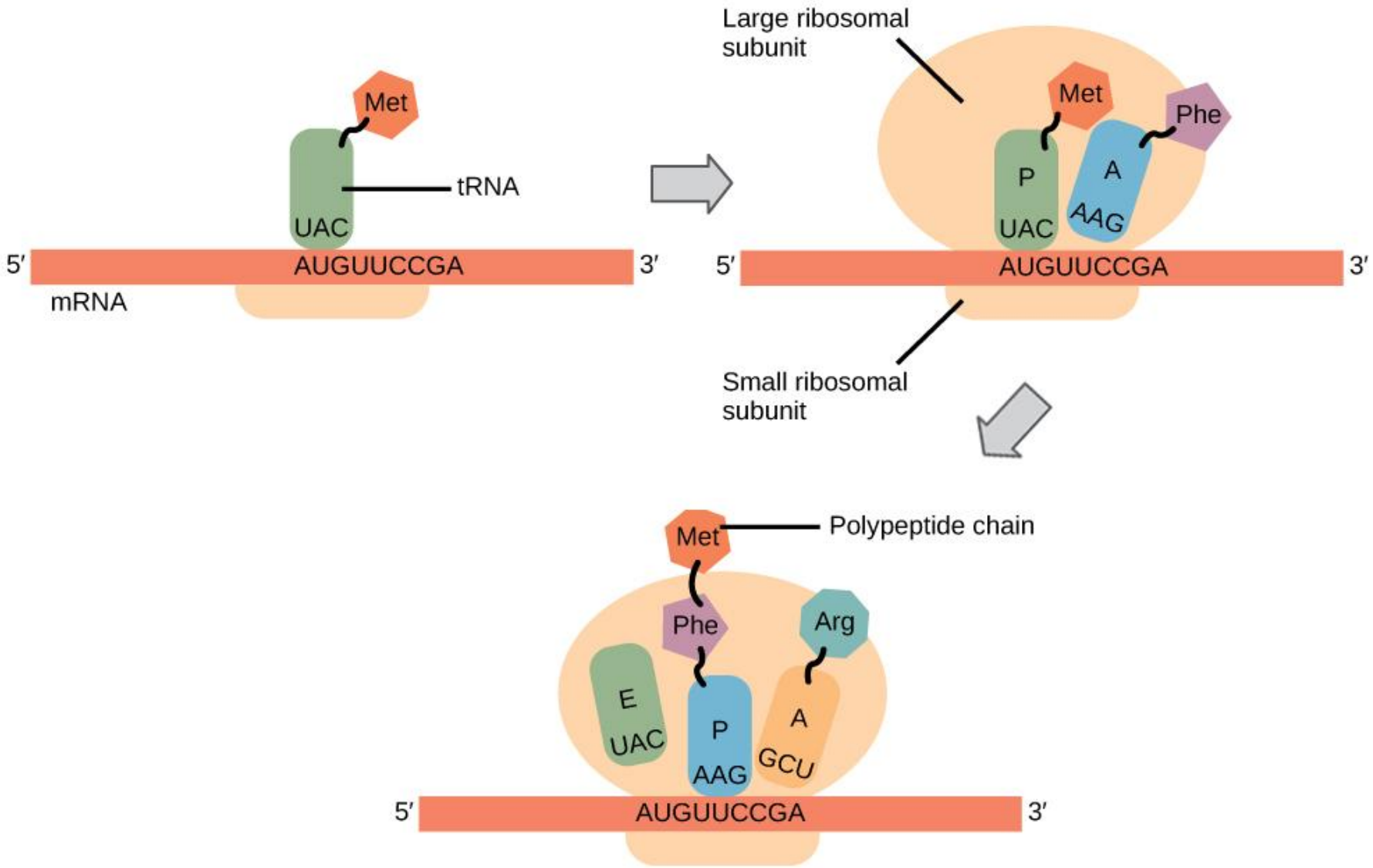
	U	C	A	G	
U	UUU, UUC } Phe UUA, UUG } Leu	UCU, UCC, UCA, UCG } Ser	UAU, UAC } Tyr **UAA Stop** **UAG Stop**	UGU, UGC } Cys **UGA Stop** UGG Trp	U C A G
C	CUU, CUC, CUA, CUG } Leu	CCU, CCC, CCA, CCG } Pro	CAU, CAC } His CAA, CAG } Gln	CGU, CGC, CGA, CGG } Arg	U C A G
A	AUU, AUC, AUA } Ile **AUG Met**	ACU,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

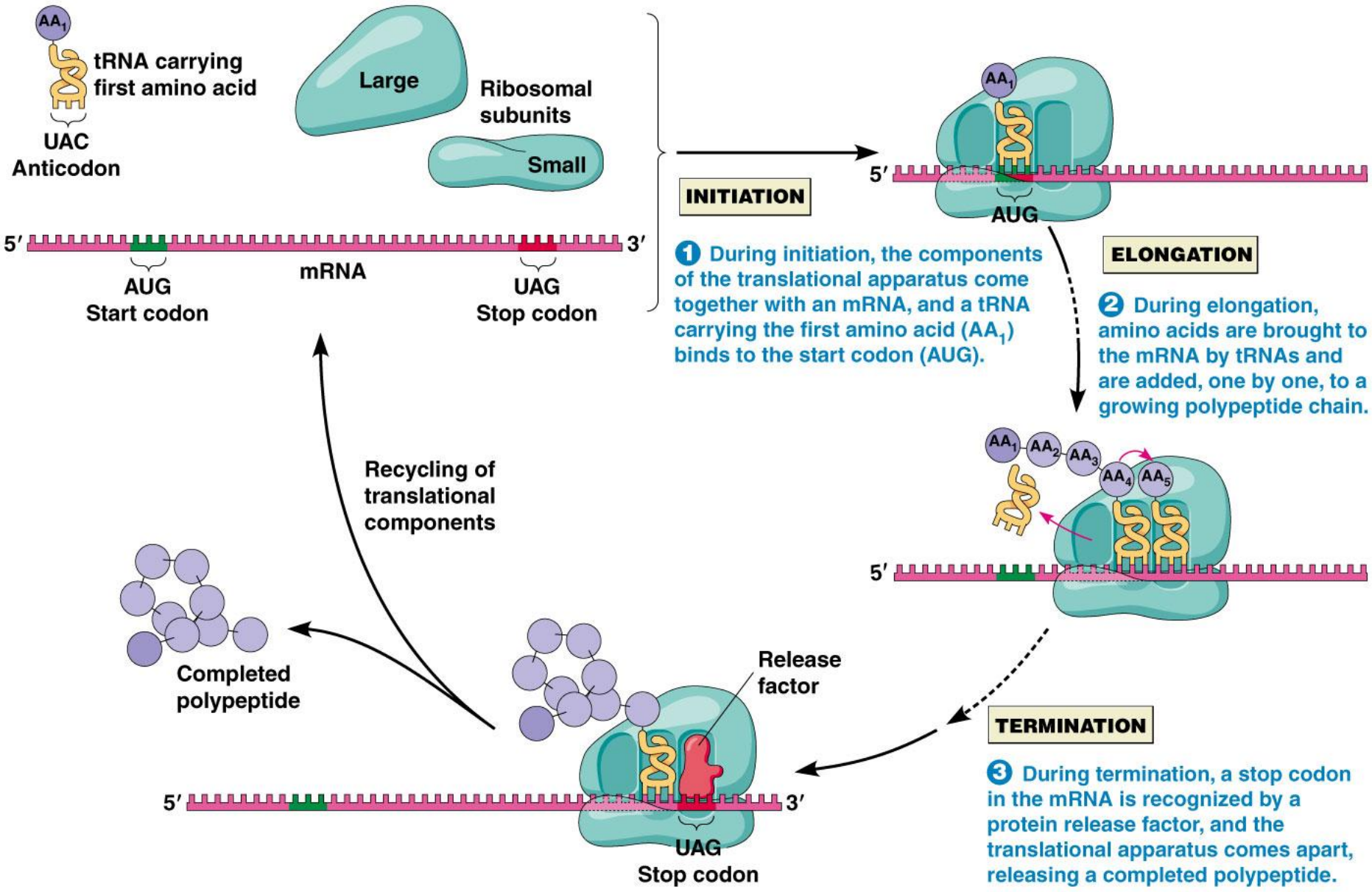

3′
A
C
C
OH
Acceptor stem
5′
P
U
A
UH2
G
G
D loop
C
G
T
ψ
C
TψC loop
Variable loop
U
Anticodon loop

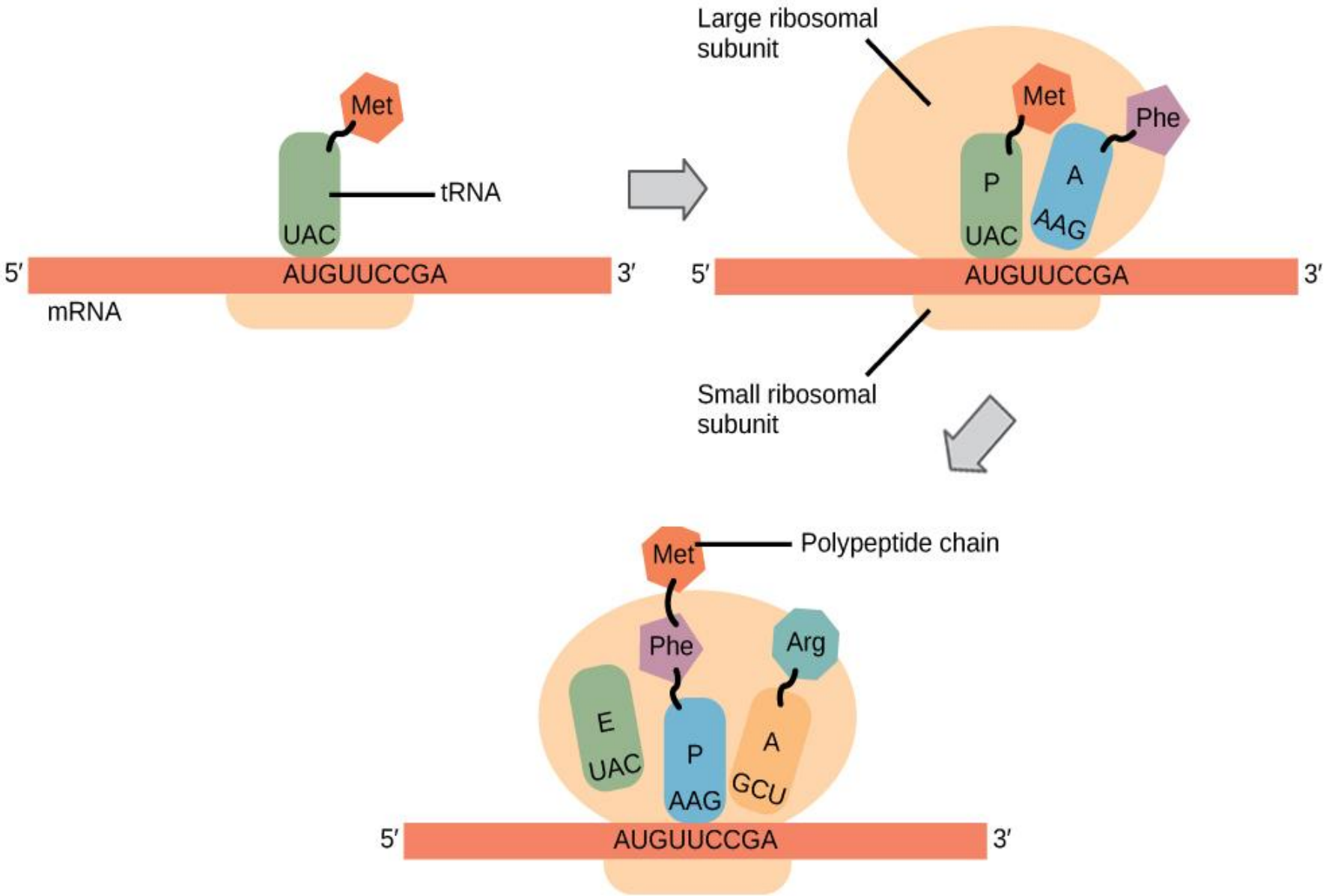

Growing polypeptide chain
Gly
Leu
Tyr
Ser
Leu
Gly
Ser
Ala
Val
tRNA
tRNA
tRNA
tRNA
G
G
A
U
U
A
Asn
C G U C A A
C U C U U G G G U C C G C A G U U A A U U U C U A U C
5′
3′
mRNA
Ribosome


Amino acid
Trp
Large ribosomal subunit
Met
E
P
A
A C C
tRNA
Anticodon
U A C
5’
mRNA
. . . A U G U G G U U C . . .
3’
Codon
Small ribosomal subunit


Met
Trp
Phe
E
P
A
U A C
A A G
A C C
5’
mRNA
. . . A U G U G G U U C . . .
3’
Large ribosomal subunit
Small ribosomal subunit


Met
tRNA
UAC
5′
AUGUUCCGA
3′
mRNA
Large ribosomal subunit
Met
Phe
P
A
UAC
AAG
5′
AUGUUCCGA
3′
Small ribosomal subunit
Met
Polypeptide chain
Phe
Arg
E
UAC
P
AAG
A
GCU
5′
AUGUUCCGA
3′




Met
tRNA
UAC
5′
AUGUUCCGA
3′
mRNA
Large ribosomal subunit
Met
Phe
P
A
UAC
AAG
5′
AUGUUCCGA
3′
Small ribosomal subunit
Met
Polypeptide chain
Phe
Arg
E
UAC
P
AAG
A
GCU
5′
AUGUUCCGA
3′

HUMAN GENOME PROJECT

•**Goal:**

To map and sequence the entire human genome, which consists of approximately 3 billion DNA base pairs and an estimated 20,000-25,000 genes.

•**International Collaboration:**

The HGP involved scientists from around the world, including the U.S. Department of Energy and the National Institutes of Health, along with other international partners.

•**Timeline:**

The project formally began in 1990 and was declared complete in 2003.

•**Key Outcomes:**

Some of the important goals of HGP were as follows:

(i) Identify all the approximately 20,000-25,000 genes in human DNA;

(ii) Determine the sequences of the 3 billion chemical base pairs that make up human DNA;

(iiii) Store this information in databases;

(iv) Improve tools for data analysis;

(v) Transfer related technologies to other sectors, such as industries;

(vi) Address the ethical, legal, and social issues (ELSI) that may arise from the project.

1. Sequence annotation is the process of identifying and describing features within a DNA, RNA, or protein sequence. It involves marking specific regions or sites of interest and attaching descriptive information about their structure, function, or other characteristics.

2. Expressed Sequence Tags (ESTs) are short sequences of DNA derived from expressed genes. They represent a portion of a gene transcript and are used to identify genes that are actively being transcribed into RNA and translated into proteins. ESTs are valuable tools for gene discovery, expression profiling, and mapping genes to specific chromosomal locations.

5.9.1 Salient Features of Human Genome

Some of the salient observations drawn from human genome project are as follows:

(i) The human genome contains 3164.7 million bp.

(ii) The average gene consists of 3000 bases, but sizes vary greatly, with the largest known human gene being dystrophin at 2.4 million bases.

(iii) The total number of genes is estimated at 30,000–much lower than previous estimates of 80,000 to 1,40,000 genes. Almost all (99.9 per cent) nucleotide bases are exactly the same in all humans.

(iv) The functions are unknown for over 50 per cent of the discovered genes.

(v) Less than 2 per cent of the genome codes for proteins.

(vi) Repeated sequences make up very large portion of the human genome.

(vii) Repetitive sequences are stretches of DNA sequences that are repeated many times, sometimes hundred to thousand times. They are thought to have no direct coding functions, but they shed light on chromosome structure, dynamics and evolution.

(viii) Chromosome 1 has most genes (2968), and the Y has the fewest (231).

(ix) Scientists have identified about 1.4 million locations where singlebase DNA differences (**SNPs – single nucleotide polymorphism,** pronounced as 'snips') occur in humans. This information promises to revolutionise the processes of finding chromosomal locations for disease-associated sequences and tracing human history.

DNA FINGERPRINTING

The technique of DNA Fingerprinting was initially developed by Alec Jeffreys. He used a satellite DNA as probe that shows very high degree of polymorphism. It was called as Variable Number of Tandem Repeats (VNTR). The technique, as used earlier, involved Southern blot hybridisation using radiolabelled VNTR as a probe. It included
(i) isolation of DNA,
(ii) digestion of DNA by restriction endonucleases,
(iii) separation of DNA fragments by electrophoresis,
(iv) transferring (blotting) of separated DNA fragments to synthetic membranes, such as nitrocellulose or nylon,
(v) hybridisation using labelled VNTR probe, and
(vi) detection of hybridised DNA fragments by autoradiography.

DNA Fingerprinting