Shri Swami Vivekanand Shikshan Sanstha's

Vivekanand College, Kolhapur (Empowered Autonomous)

Department of Zoology

INDEX

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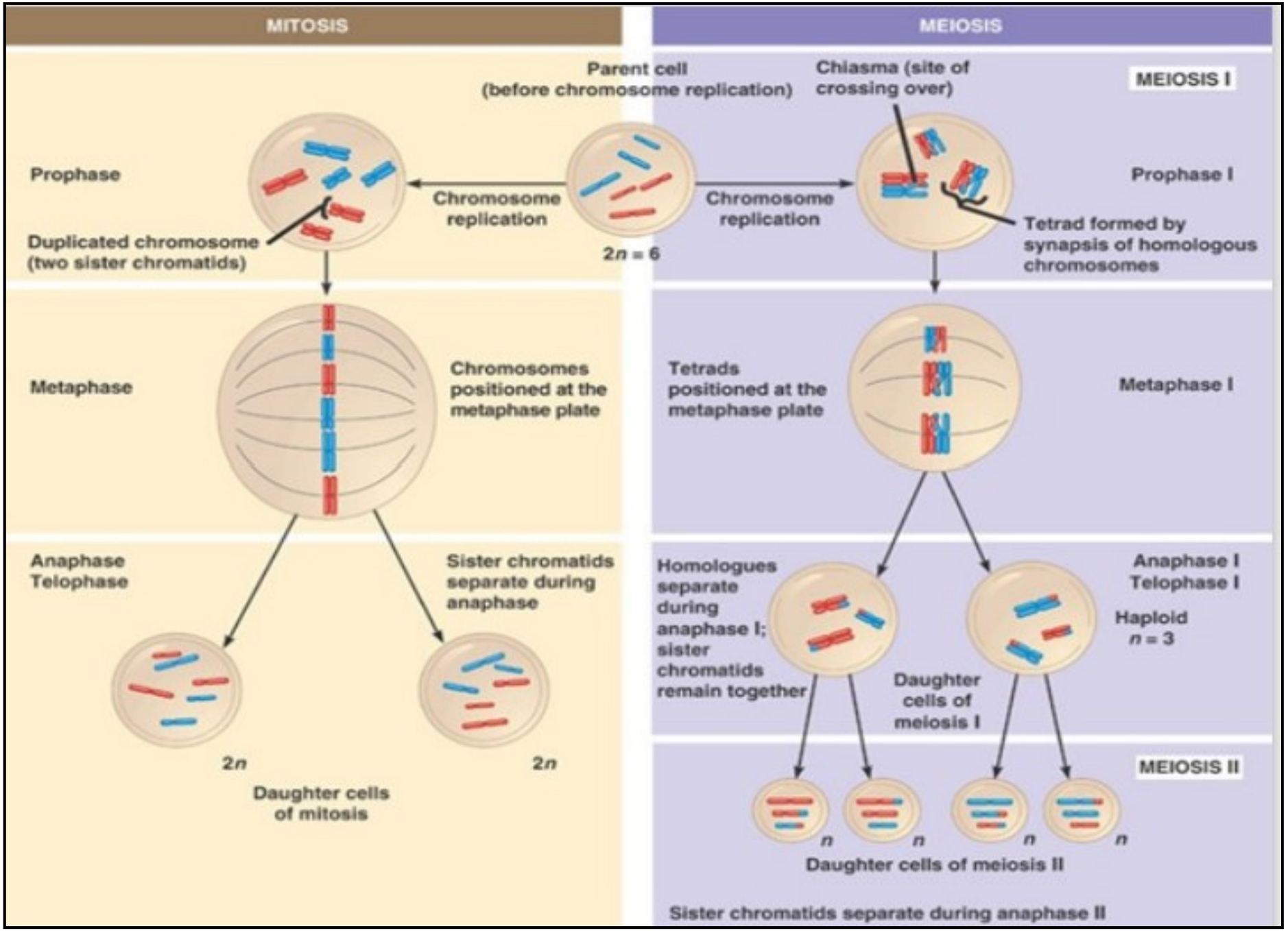
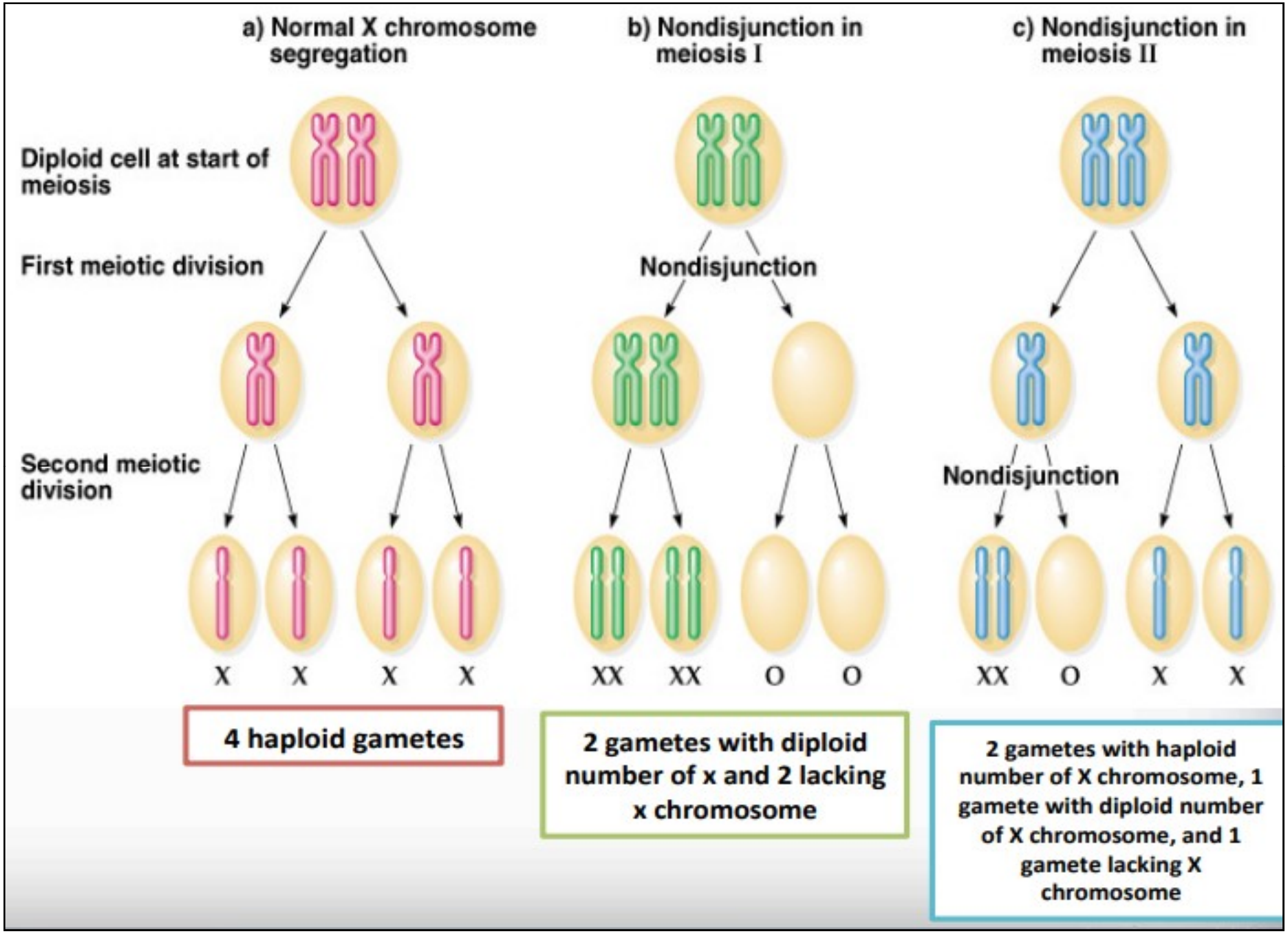
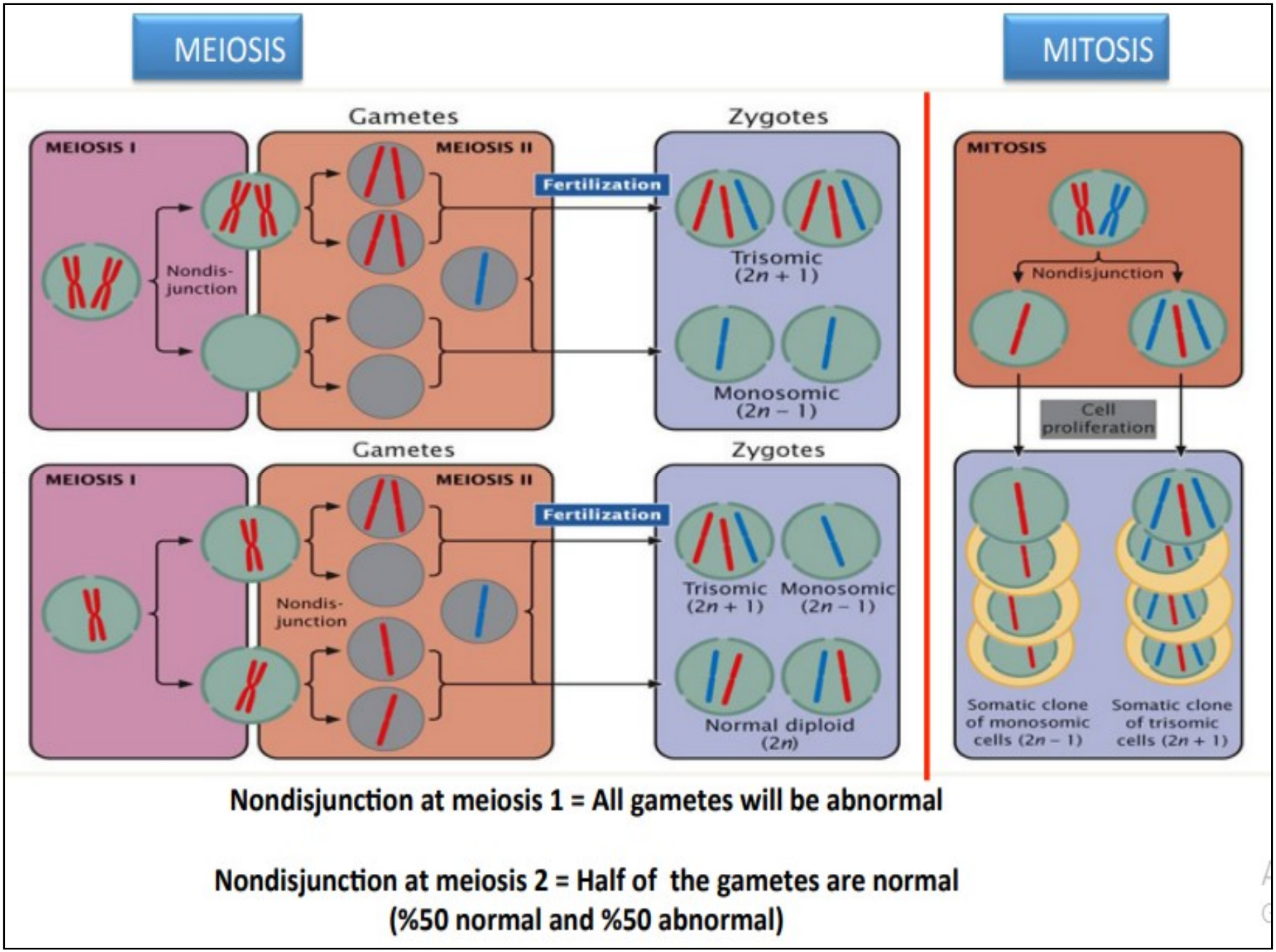
Klinefelter, Turner & Down Syndrome

Non-disjunction in Meiosis:

- Non-disjunction "not coming apart" is the failure of a chromosome pair to separate properly during meiosis 1, or of two chromatids of a chromosome to separate properly during meiosis 2 or mitosis.
- Can effect each pair.
- Not a rare event.
- As a result, one daughter cell has two chromosomes or two chromatids and the other has none
- The result of this error is ANEUPLOIDY.


MITOSIS
MEIOSIS
Parent cell
(before chromosome replication)
Chiasma (site of crossing over)
MEIOSIS I
Prophase
Chromosome replication
Chromosome replication
Prophase I
Duplicated chromosome
(two sister chromatids)
2n = 6
Tetrad formed by synapsis of homologous chromosomes
Metaphase
Chromosomes positioned at the metaphase plate
Tetrads positioned at the metaphase plate
Metaphase I
Anaphase
Telophase
Sister chromatids separate during anaphase
Homologues separate during anaphase I; sister chromatids remain together
Anaphase I
Telophase I
Haploid
n = 3
Daughter cells of meiosis I
2n
2n
Daughter cells of mitosis
MEIOSIS II
n
n
n
n
Daughter cells of meiosis II
Sister chromatids separate during anaphase II


a) Normal X chromosome segregation
b) Nondisjunction in meiosis I
c) Nondisjunction in meiosis II
Diploid cell at start of meiosis
First meiotic division
Second meiotic division
Nondisjunction
Nondisjunction
X X X X
XX XX O O
XX O X X
4 haploid gametes
2 gametes with diploid number of x and 2 lacking x chromosome
2 gametes with haploid number of X chromosome, 1 gamete with diploid number of X chromosome, and 1 gamete lacking X chromosome

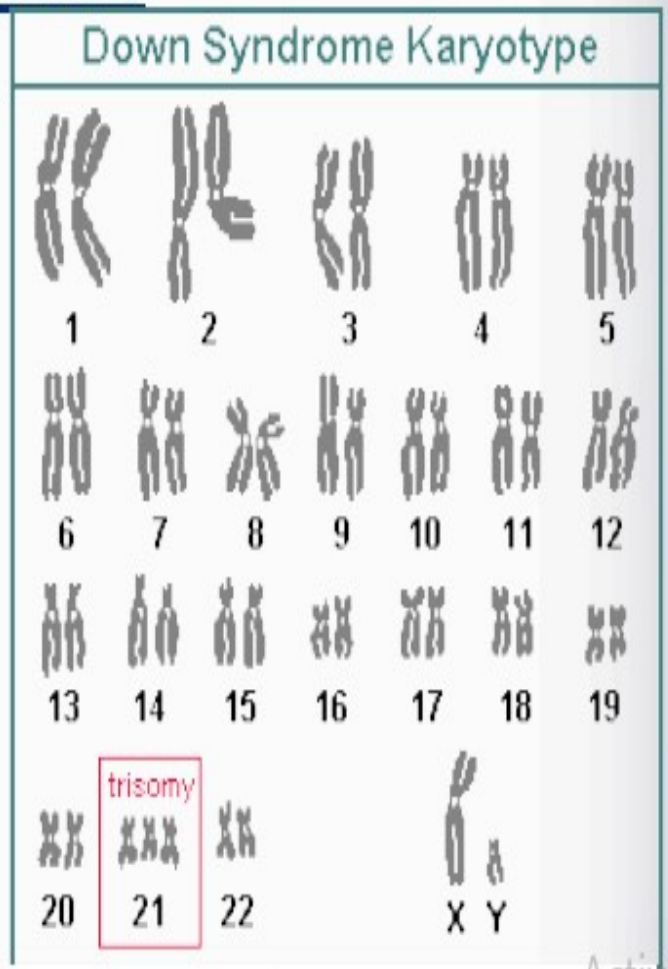


Nondisjunction at meiosis 1 = All gametes will be abnormal

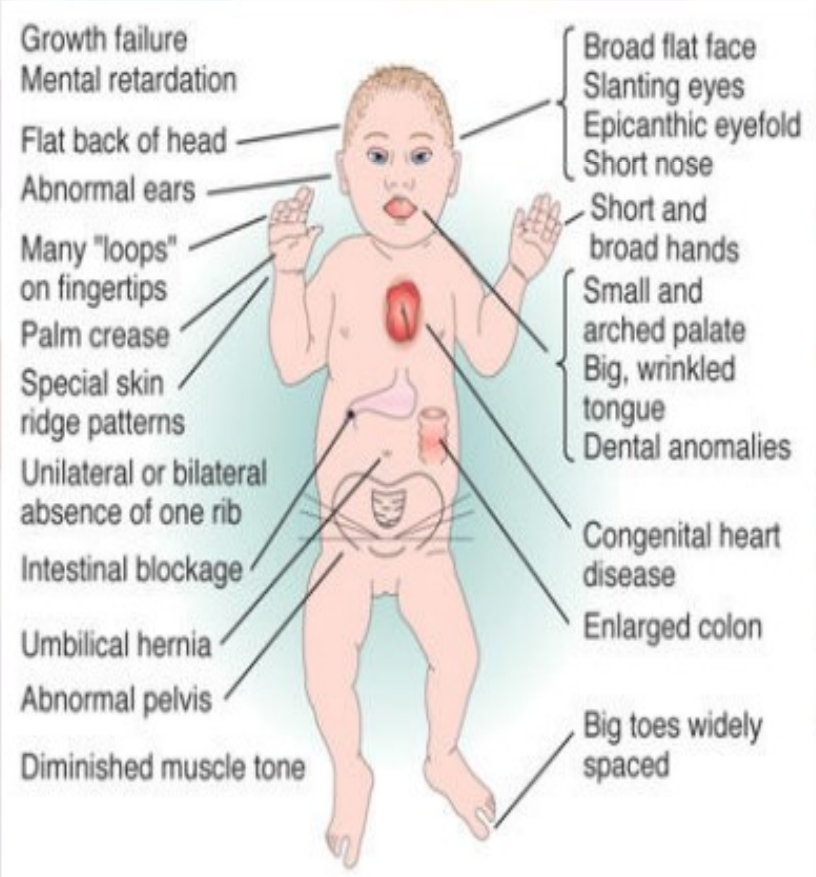
Nondisjunction at meiosis 2 = Half of the gametes are normal (%50 normal and %50 abnormal)

Down's Syndrome

- Karyotype: 47, XY, +21
  Three copies of chromosome 21 (21 trisomy)
- The incidence of trisomy 21 rises sharply with increasing maternal age (above 37), but Down syndrome can also be the result of nondisjunction of the father's chromosome 21 (%15 of cases)
- A small proportion of cases is mosaic* and probably arise from a non-disjunction event in early zygotic division.



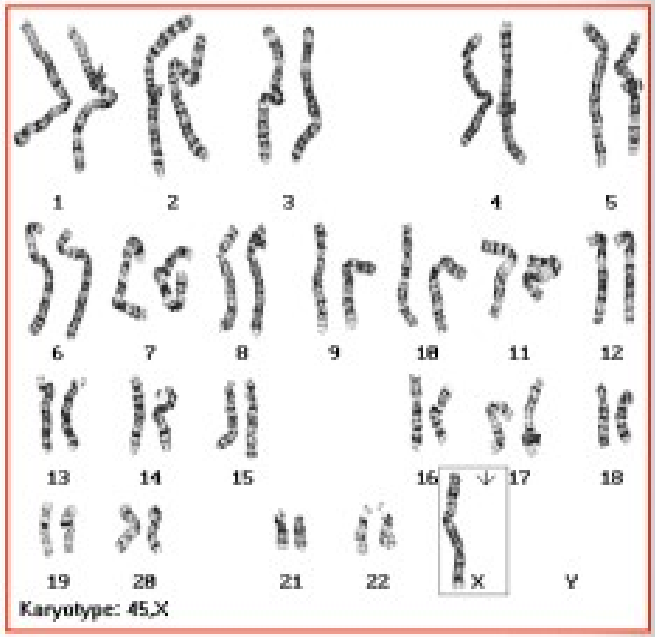
Features of Down's Syndrome



- Low muscle tone
- Rough skin
- Head and facial malformations: Small round face, protruding tongue
- Abnormalities of the extremities: Short broad hands, stubby fingers
- Developmental delays (Mental retardation)
- Heart malformations
- Increased risk of infectious diseases
- Short life span (Early death)

Turner's Syndrome

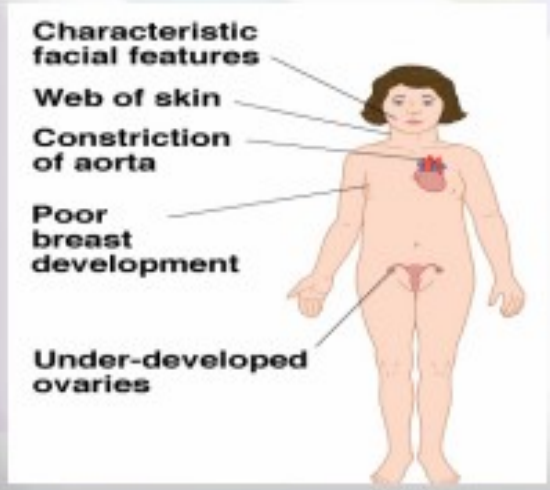
- Karyotype: Monosomy X: 45, XO
- Monosomy of sex chromosome (only one X chromosome present)
- The only viable monosomy in humans
- Occurring in 1 in 2500 phenotypic females
- NO developmental delays
- Turner syndrome is commonly treated with growth hormones, and estrogen replacement therapy.

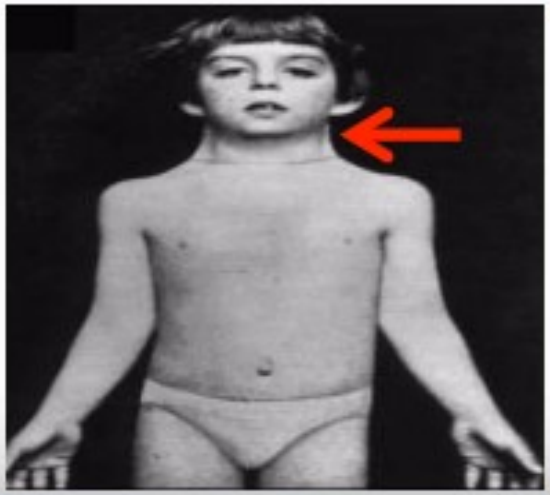


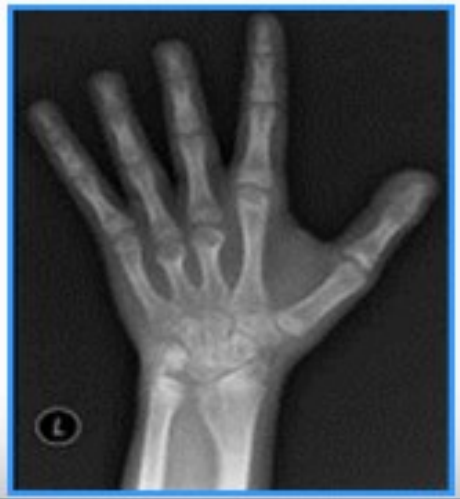
Features of Turner's Syndrome

Cardiovascular	Skeletal	Reproductive
▪ Cardiovascular constriction ▪ Bicuspid aortic valve ▪ Coarctation of the aorta ▪ Thoracic aortic aneurysm	▪ Short stature ▪ Neck Abnormalities (webbing of the neck) ▪ Osteoporosis (due to lack of estrogen) ▪ Scoliosis ▪ Short 4th metacarpal/metatarsal bone. (+ \- short 3rd & 5th)	▪ Lack of ovarian development ▪ Women with Turner syndrome are almost universally infertile. ▪ Reproductive technology can help women with Turner syndrome become pregnant

Others: Diabetes, kidneys and thyroid problems

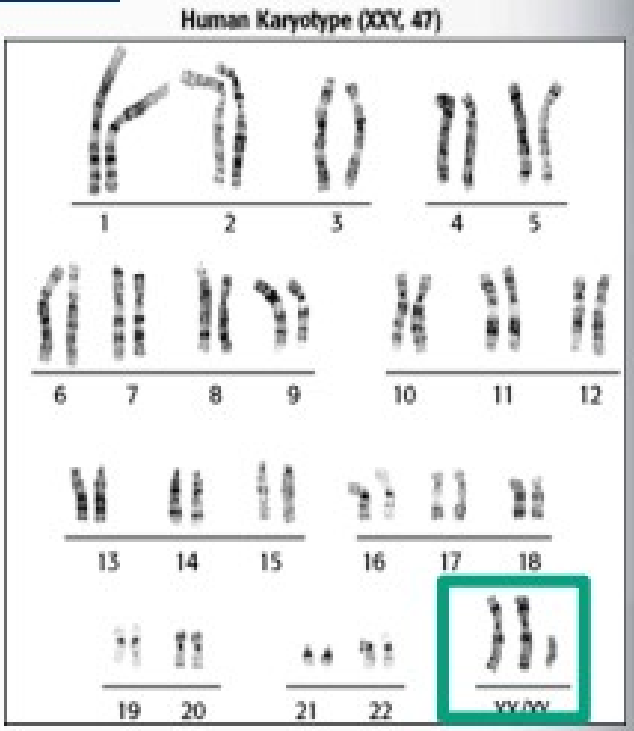






Klinefelter's Syndrome

- Karyotype: (XXY, 47) males
  Nondisjunction (23 trisomy)
- 1 in 1,100 births
- Klinefelter syndrome is a genetic condition that results when a boy is born with an extra copy of the X chromosome.
- Very rarely more extreme forms of Klinefelter syndrome occur where the patient has 48, XXXY or even 49, XXXXY karyotype.
  These individuals are generally severely retarded.
- Klinefelter syndrome often isn't diagnosed until adulthood. Most men with Klinefelter syndrome produce little or no sperm. But assisted reproductive procedures may make it possible for some men with Klinefelter syndrome to father children.



Features of Klinefelter's Syndrome

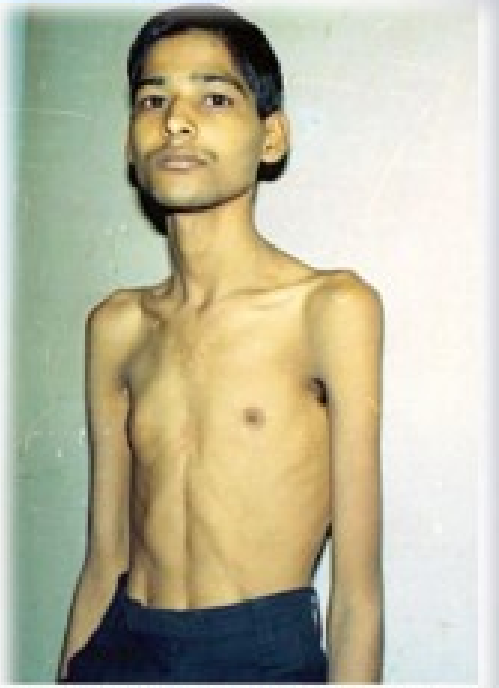
- Unusually small testes → low production of testosterone → Gynaecomastia, reduced body hair and other feminine body characteristics.

- No spermatogenesis → sterile (in some cases, testicular function is preserved)
- Low mental abilities (a slight reduction in IQ but generally they have normal intelligence)
- Delays in speech and motor skills as well as deficits in attention, auditory processing and social skills.

- Patients are taller and thinner (Reduced muscle mass) than usual with long fingers and arms
- Normal lifespan
- Brown spots (nevi)

- Increased risk of autoimmune disorders, breast cancer, osteoporosis, leg ulcers, depression, and dental problems

❖Treatment for these problems includes: testosterone therapy and assisted learning.



Photograph showing development of gynecomastia in
ld male after 2 months of isoniazid containing Catego
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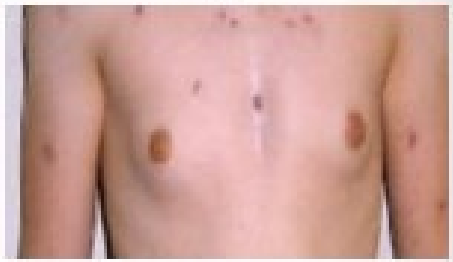


Sex chromosome unbalance is much less deleterious

- **47, XYY:**
  - ✓ May be without any symptoms.
  - ✓ Males are tall but normally proportioned.
  - ✓ 10 - 15 points reduction in IQ compared to siblings.

- **XXX females:**
  - ✓ It seems to do little harm,
  - ✓ individuals are fertile and do not transmit the extra chromosome.
  - ✓ They do have a reduction in IQ comparable (similar) to that of Kleinfelter's males.

When to do a chromosomal test

- **Prenatal:**
  A. maternal age>37yrs; Ultrasound scan changes; Family history
  B. Triple test (measuring the alpha fetoprotein (AFP), human chorionic gonadotropin (hCG), and estriol): if positive it indicates an increased risk of having diseases due to chromosomal anomalies

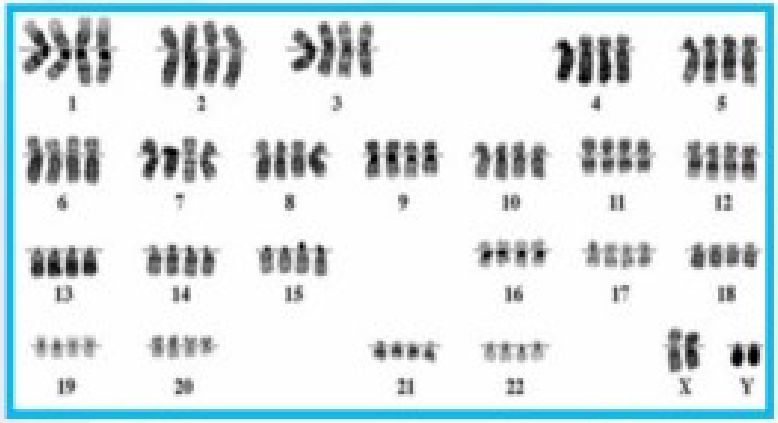
- **Postnatal:** Learning & developmental disability; growth retardation

- **Infertility:** Recurrent miscarriage, primary infertility

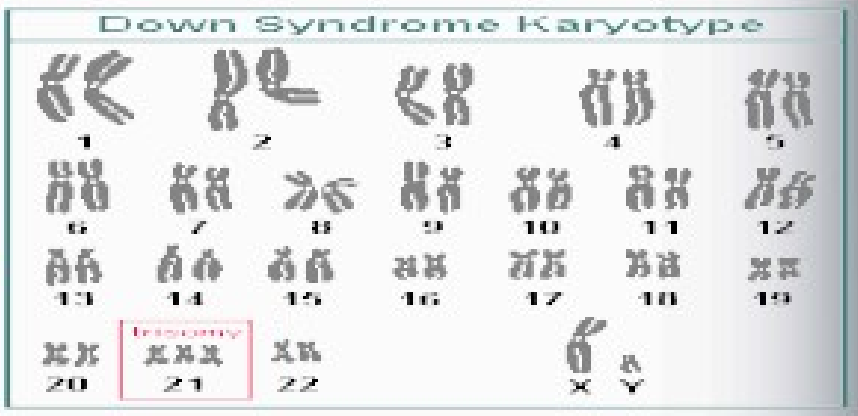
Aneuploidy

- Aneuploidy refers to a numerical change in PART OF the chromosome set.
- Aneuploidy could be:
  1. Autosomal: Trisomy 21 (Down syndrome)
  2. Sex chromosome:
     - 47XXY (Klinefelter syndrome)
     - 45X (Turner syndrome)

- Polyploidy refers to a numerical change in the WHOLE SET of chromosomes



VS



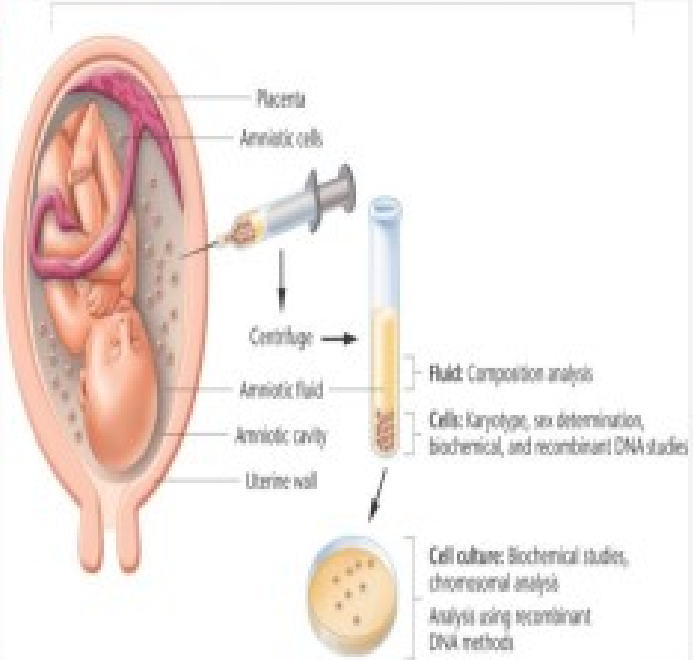
Polyploidy

Aneuploidy

Aneuploidy Screening

1. Rapid Aneuploidy screening by FISH

- Available on amniocentesis sample
- Uncultured amniocytes
- FISH probes for X,Y, 21
- Result in 24-48 hours
- Proceed onto full karyotype (11-14 days)



2. New Techniques

A. Quantitative Fluorescence PCR: to measure number of copies of a chromosome

B. Cell-free Fetal DNA:

at 6-8 weeks of gestation. It is a non-invasive prenatal diagnostic tool for chromosomal aneuploidy. It can be used to determine the fetus sex– (look for presence of Y chromosome)


THANK
YOU

PHYLUM PORIFERA

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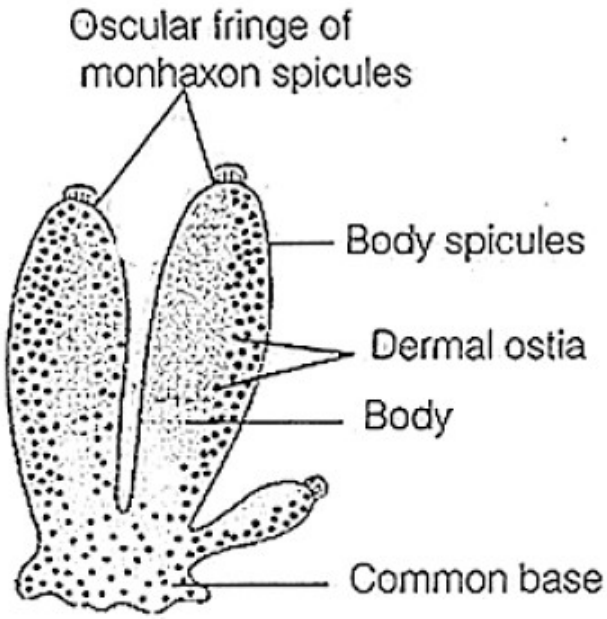
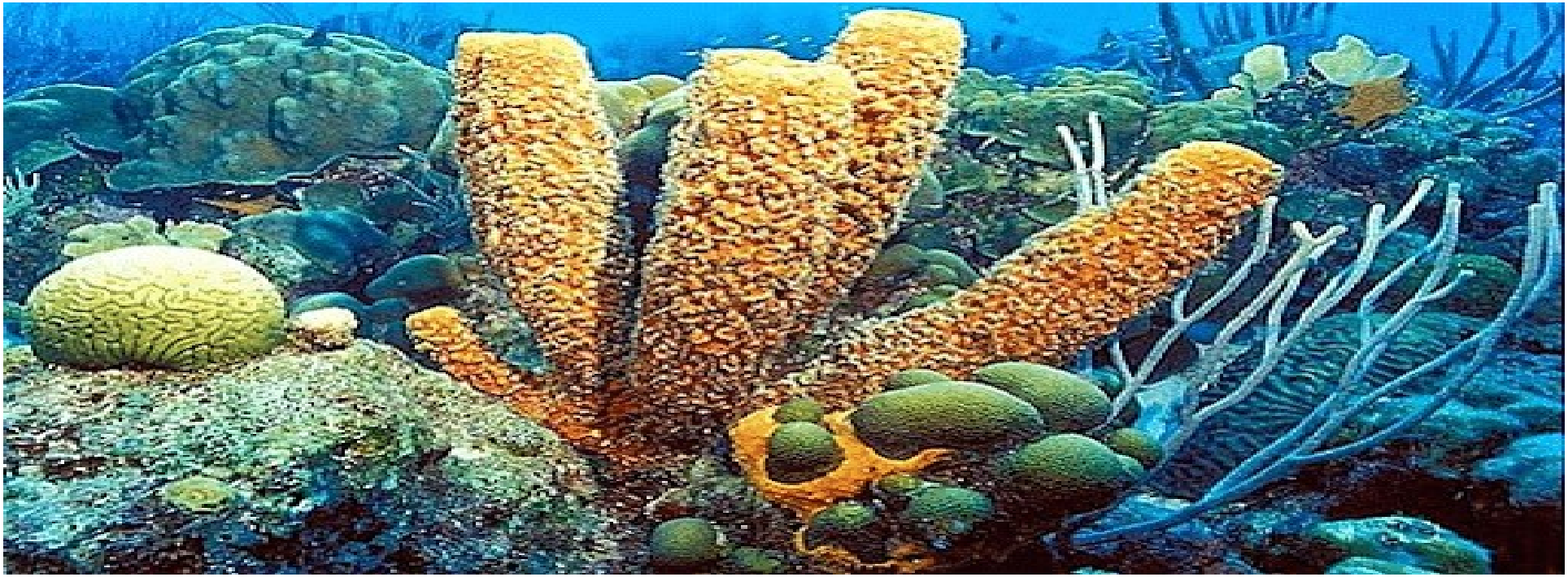
Sycon



Phylum: Porifera
Class: Calcarea
Order: Heterocoela
Family: Sycettidae
Genus: Sycon



- Sponges are referred to as the members of the phylum Porifera.

- Sycon is a type of sponge which is generally marine in nature and is mostly asymmetrical in nature.

- Sycon possesses a water transport canal system wherein the water enters via the minute pores [ostia] in the body wall into the central cavity [spongocoel] from where it goes out through the osculum.

- The Canal system present in Sycon is helpful in gathering food, respiration and removal of waste in sycon via water transport.

The following are the characteristics which are commonly observed in the case of Sycons:

- The length of the body of the organism varies from 2.5 centimetres to 7.6 centimetres.

- Their bodies are either radially symmetrical or asymmetrical.

- They have the capability to regenerate their lost body parts.

- Needle-like spines called spicules cover their body which gives it a bristle-like appearance.

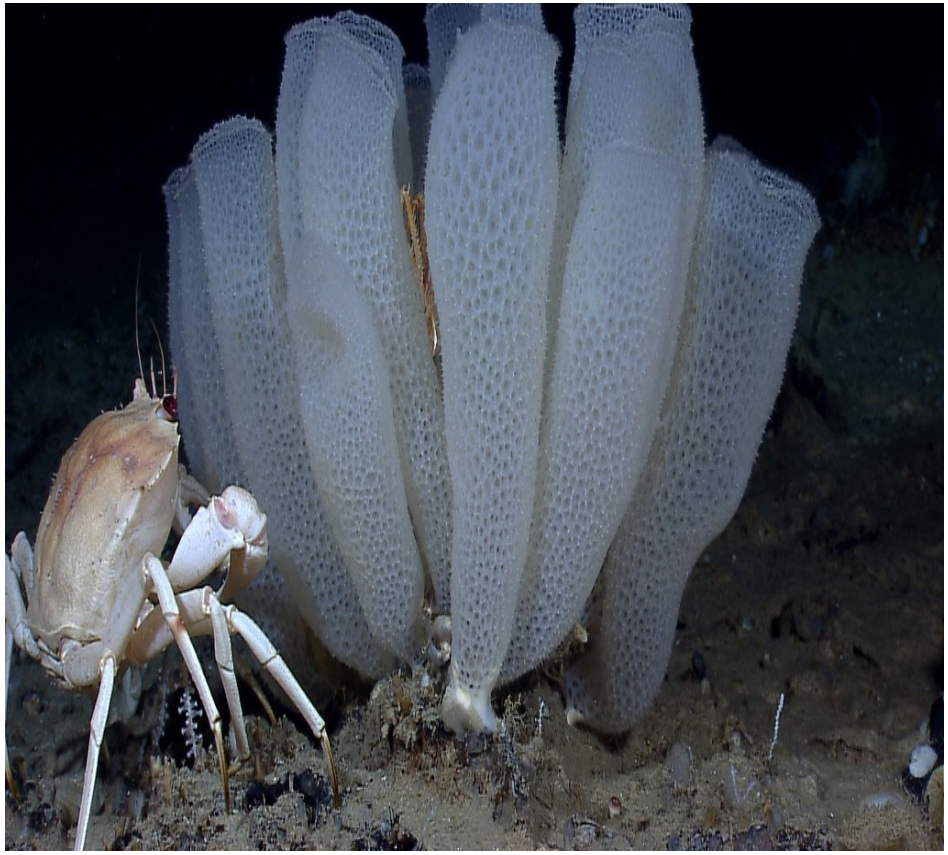
Euplectella

•Euplectella is categorized under the Porifera phylum. They belong to the Hexactinellida class, distinguished by the skeleton of triaxon spicules with six rays.

An example of a very common species of Euplectella is *Euplectella aspergillum*.

Classification of euplectella:-

Kingdom	Animalia
Phylum	Porifera
Class	Hexactinellida
Order	Lyssacinosida
Family	Euplectellidae
Genus	Euplectella



Structure and Characteristics of Euplectella

- The body is cylindrical and basket-like, connected to the sea bottom by the tuft of fibres.

- Triaxon spicules make up the skeleton. The silica spicules give them a glassy appearance.

- The true Ostia is not present. Numerous perforations are parietal gaps on the outer surface.

- For water circulation, a well-connected canal system is present. The canal system is found to be syconoid.

- Within the body and outside, the incurrent channels are attached to radial channels and open into spongocoel.

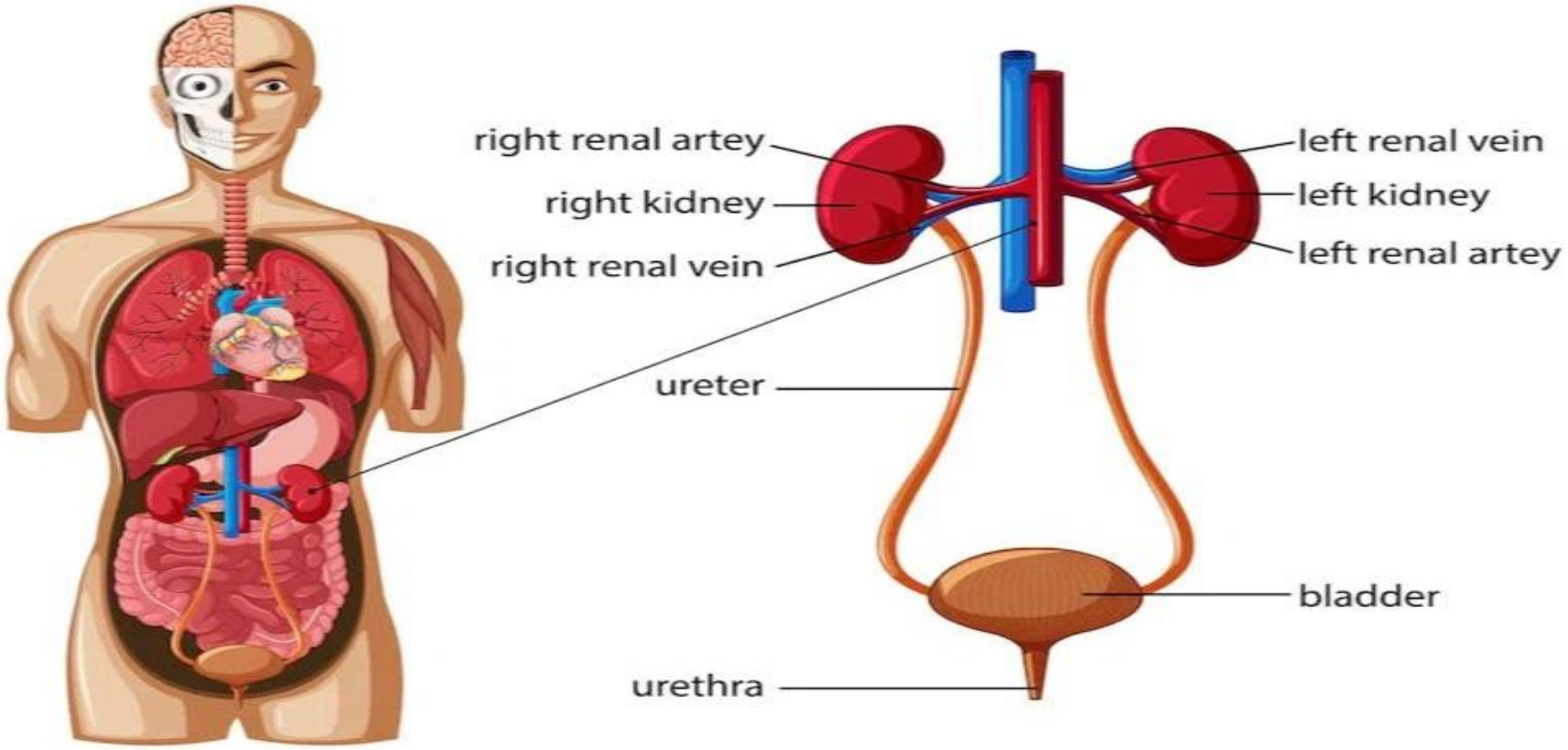
- Throughout the lives of males and females, a pair of sponge cola shrimps are found inside the sponge's body. Their tiny offspring come out to find their basket. Shrimp receives the food from the basket and cleans the basket from the inside, in exchange.

- They reproduce both asexually and sexually.

- Euplectella's glassy fibrous binding to the ocean bed is being examined to make more durable optical fibres for fibre optics. This can also be used to make solar cells that are low-cost and more efficient.

THANK YOU

Excretory System



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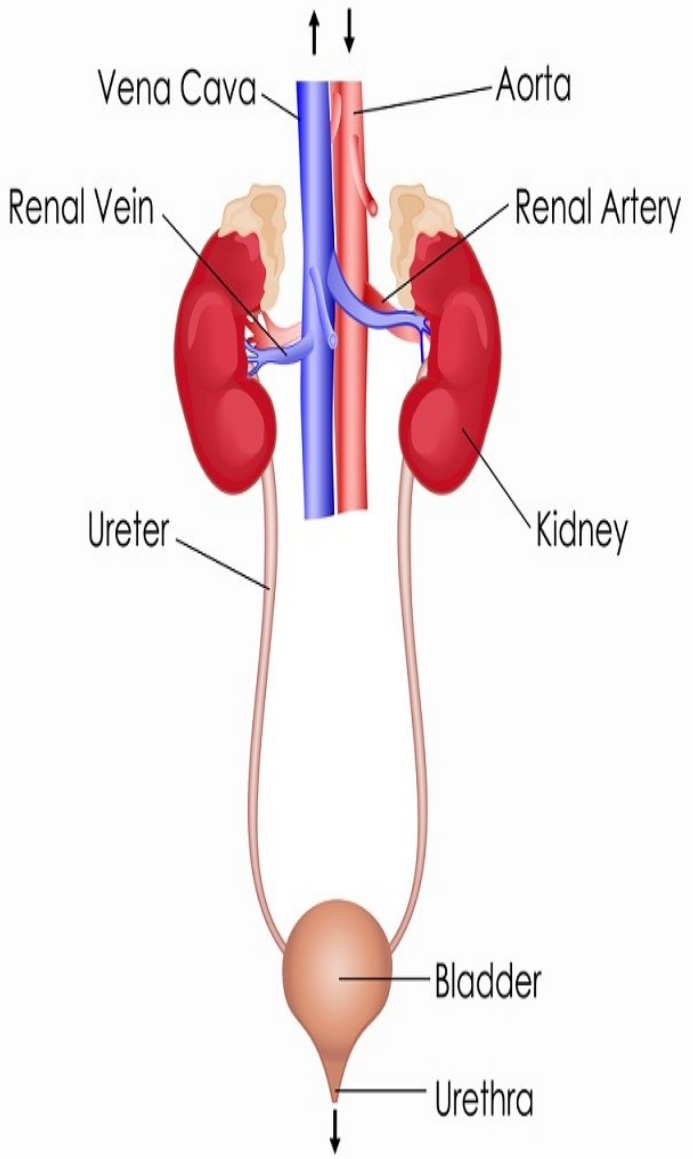
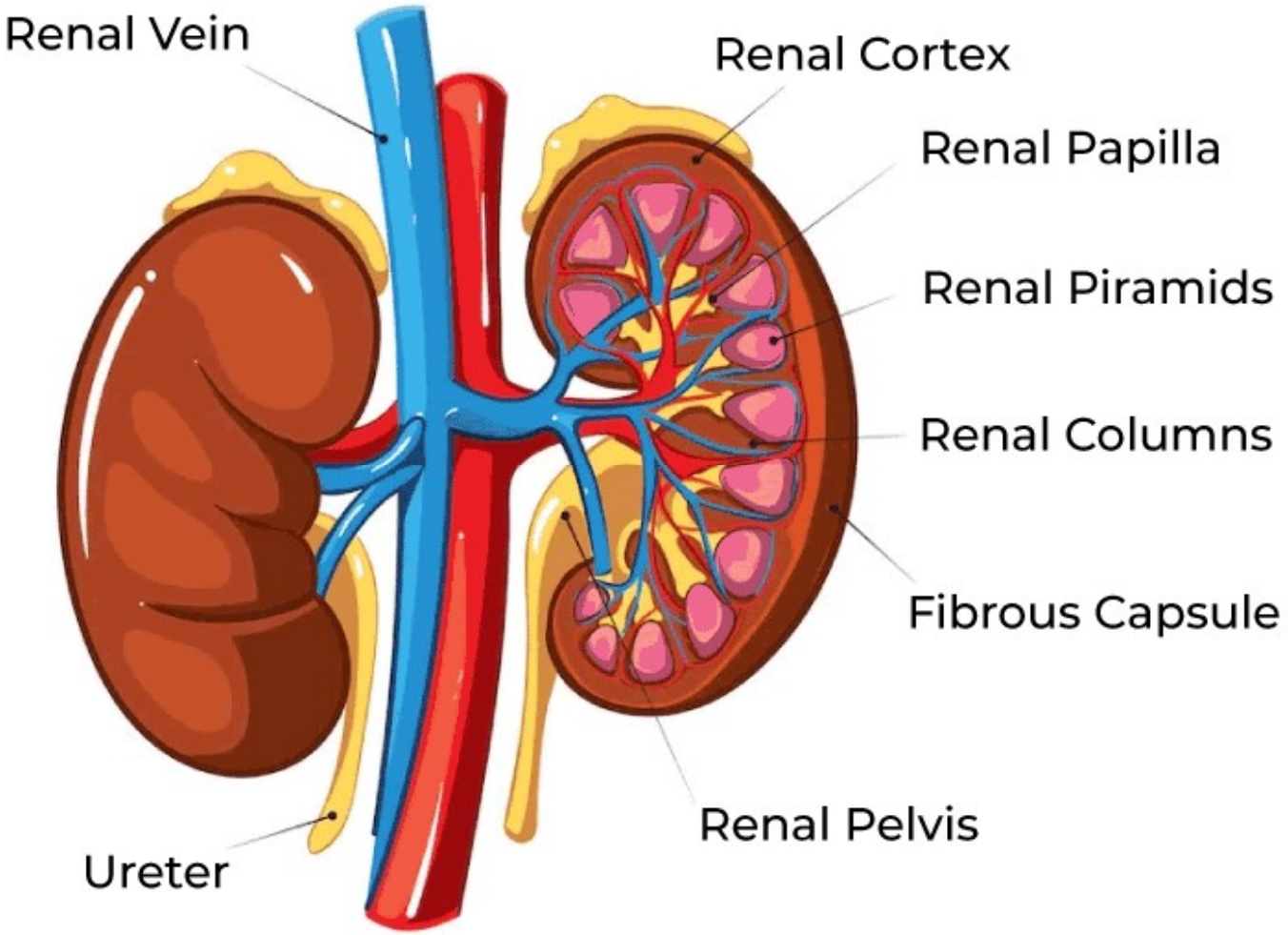
Excretory System Organs

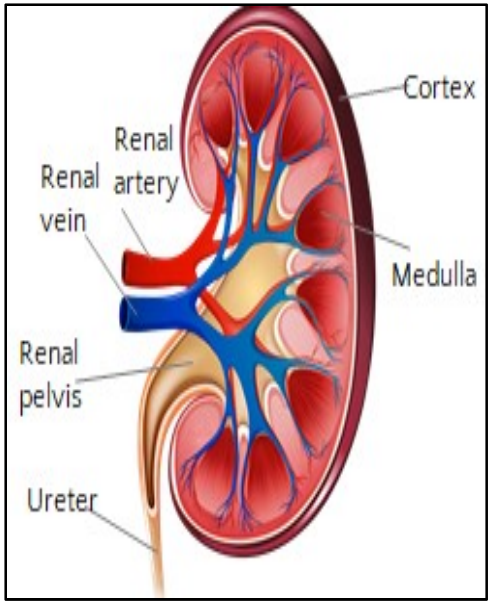
The human excretory system organs include:

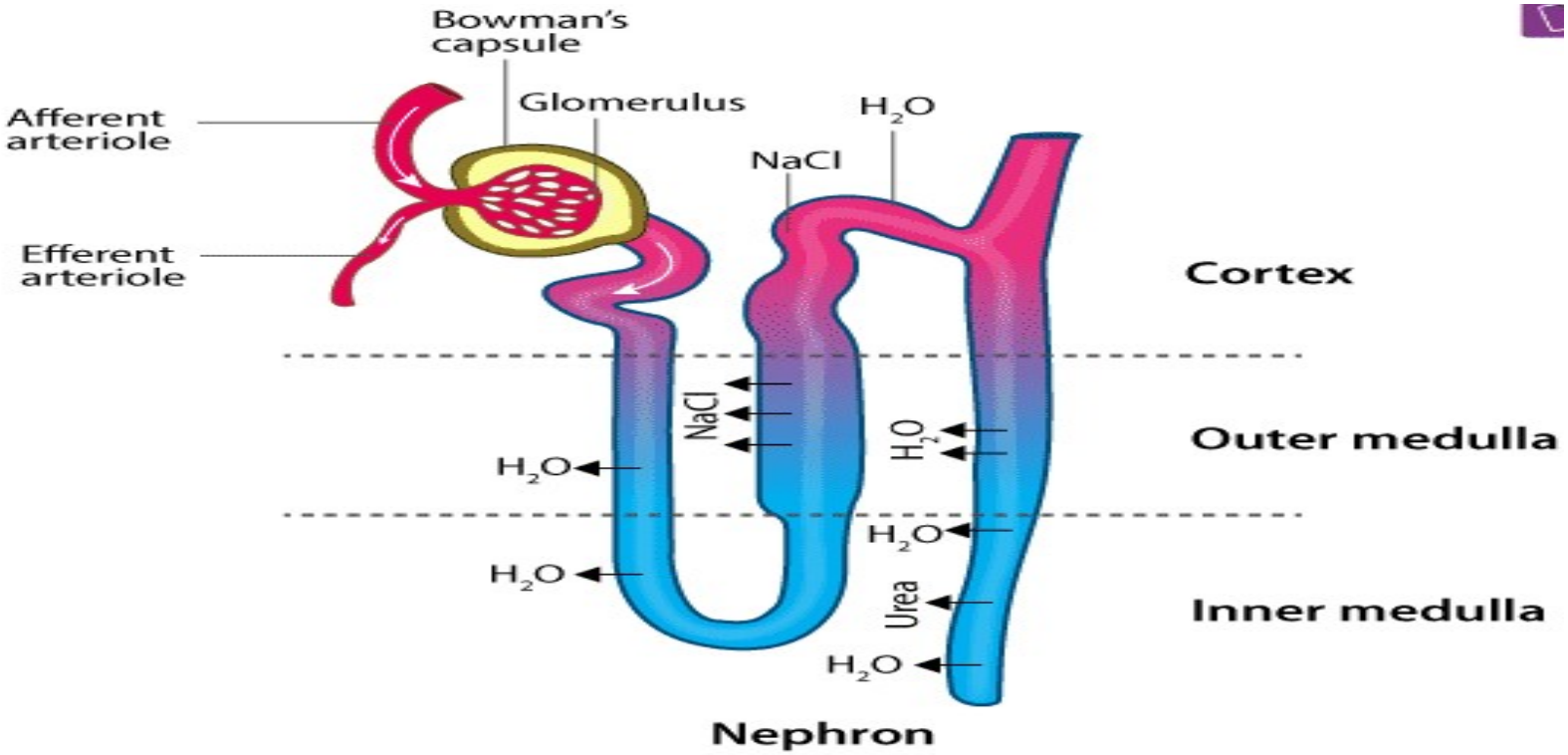
- A pair of kidneys
- A pair of ureters
- A urinary bladder
- A urethra

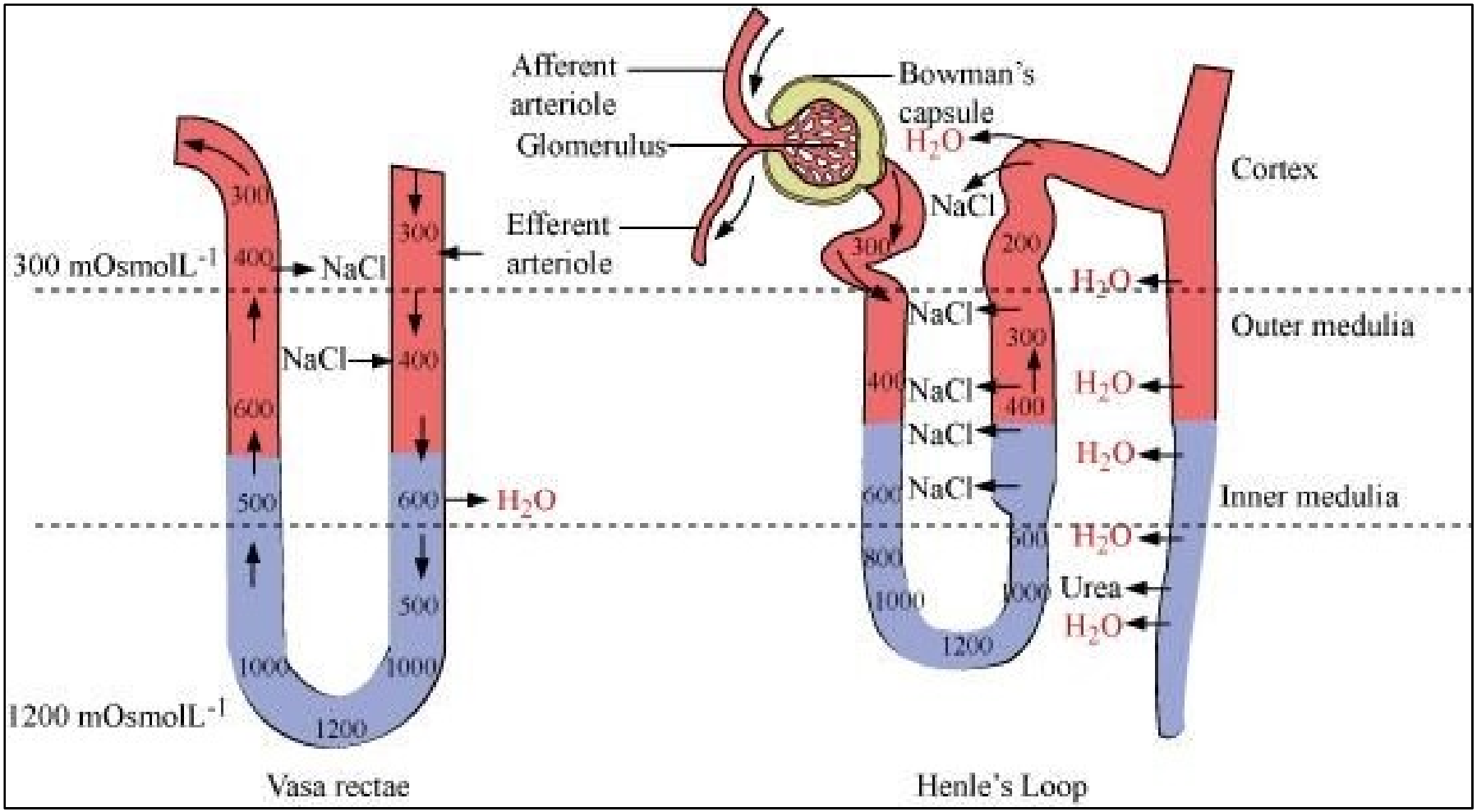


Anatomy of Kidney




Cortex
Renal artery
Renal vein
Medulla
Renal pelvis
Ureter


Bowman's capsule
Glomerulus
Afferent arteriole
Efferent arteriole
H_2O
NaCl
Cortex
Outer medulla
Inner medulla
Urea
Nephron



Counter-Current Mechanism of Urine Formation

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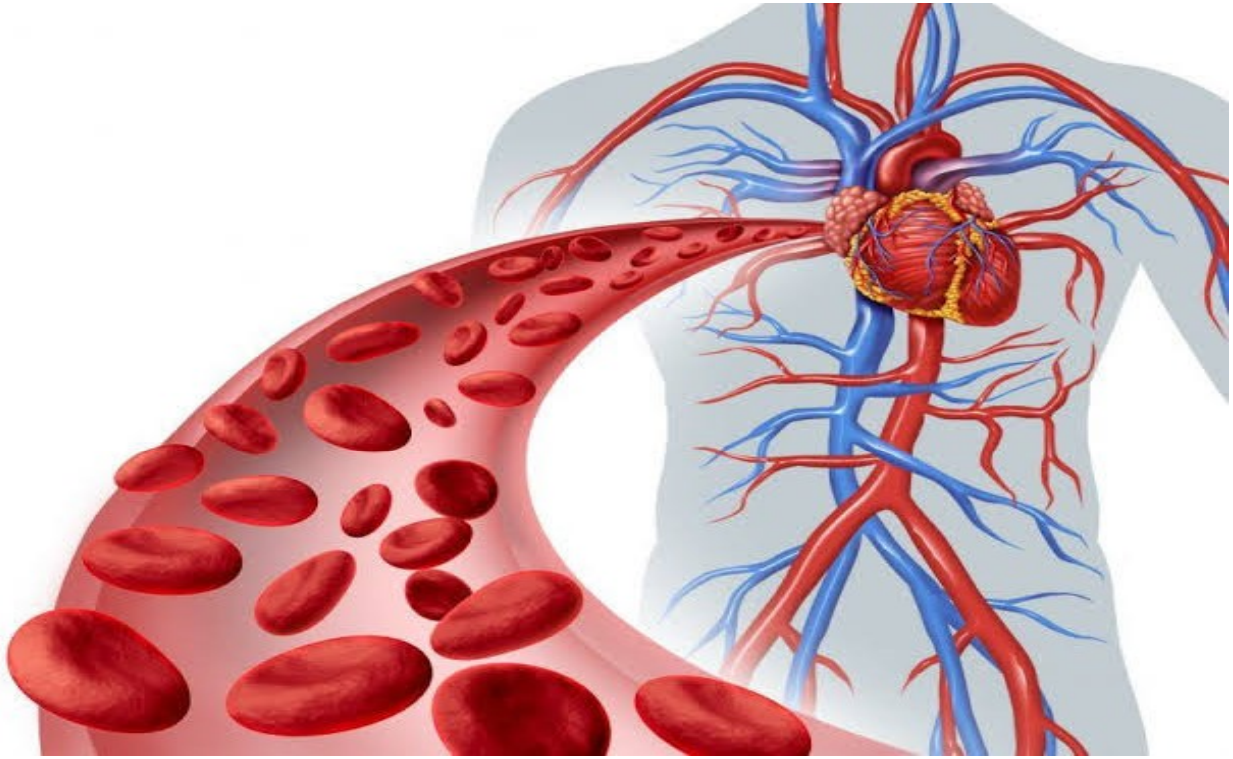
Circulatory System



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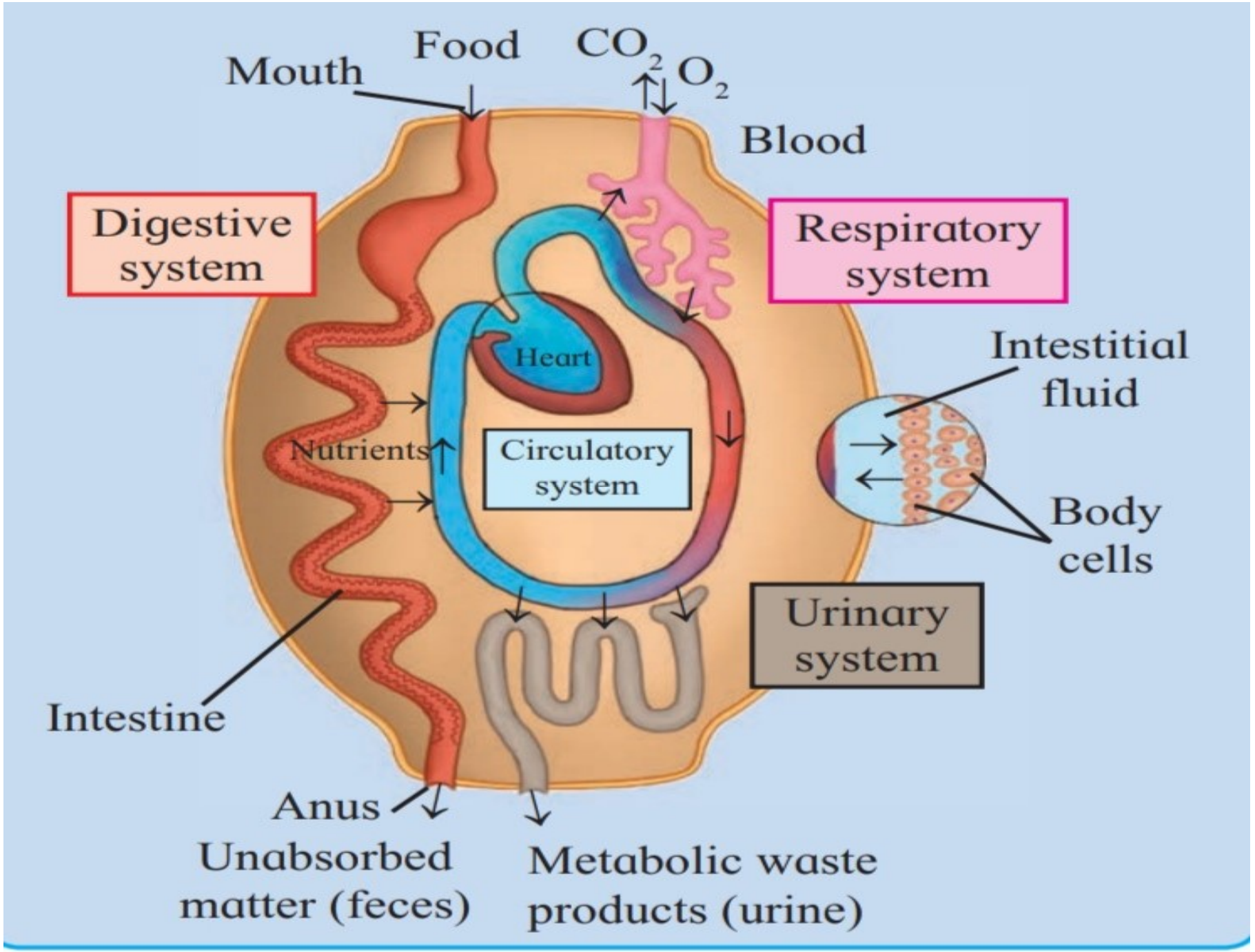
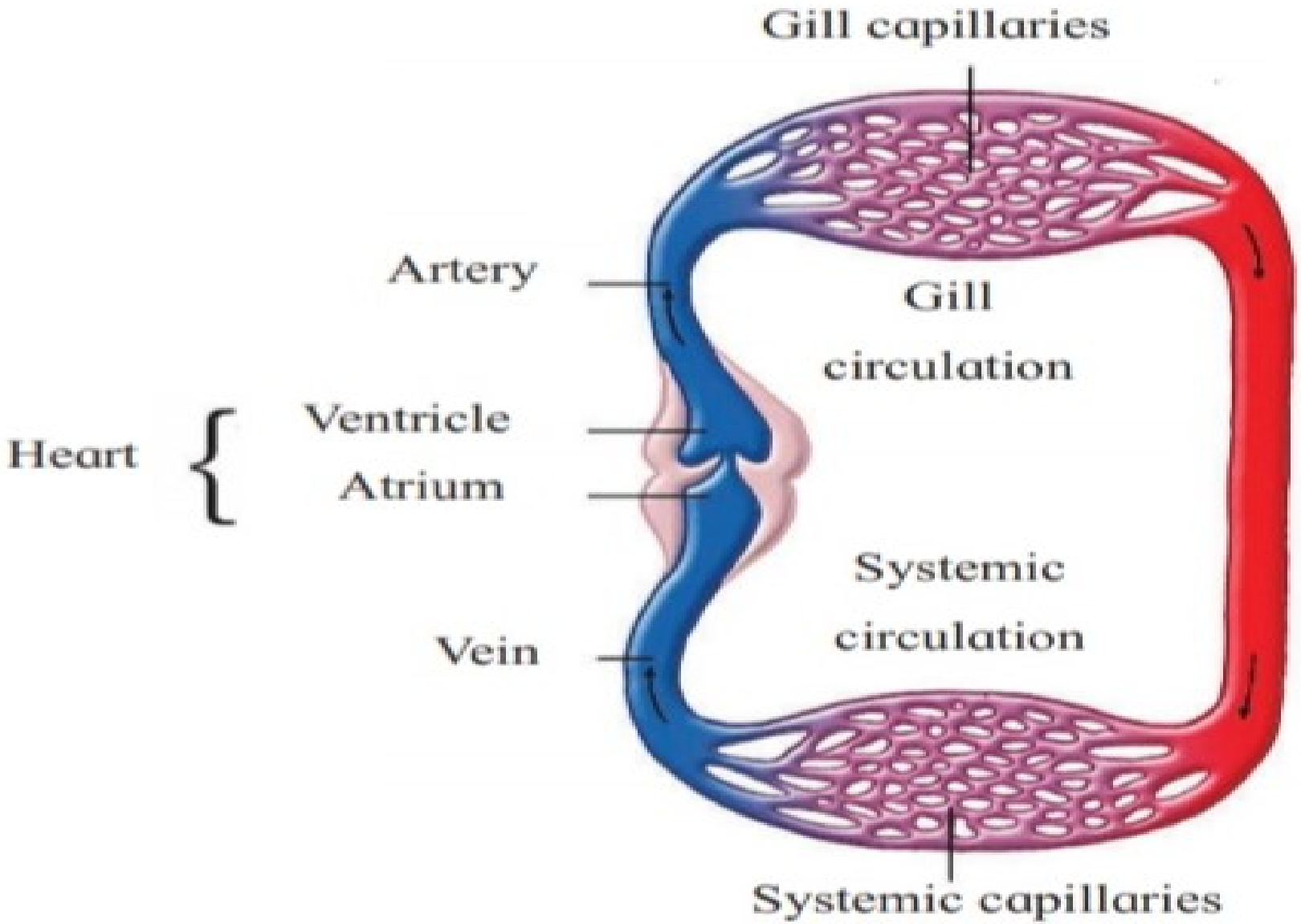

Mouth
Food
CO_2
O_2
Blood
Digestive system
Respiratory system
Heart
Intestitial fluid
Nutrients
Circulatory system
Body cells
Urinary system
Intestine
Anus
Unabsorbed matter (feces)
Metabolic waste products (urine)



Single Circulation

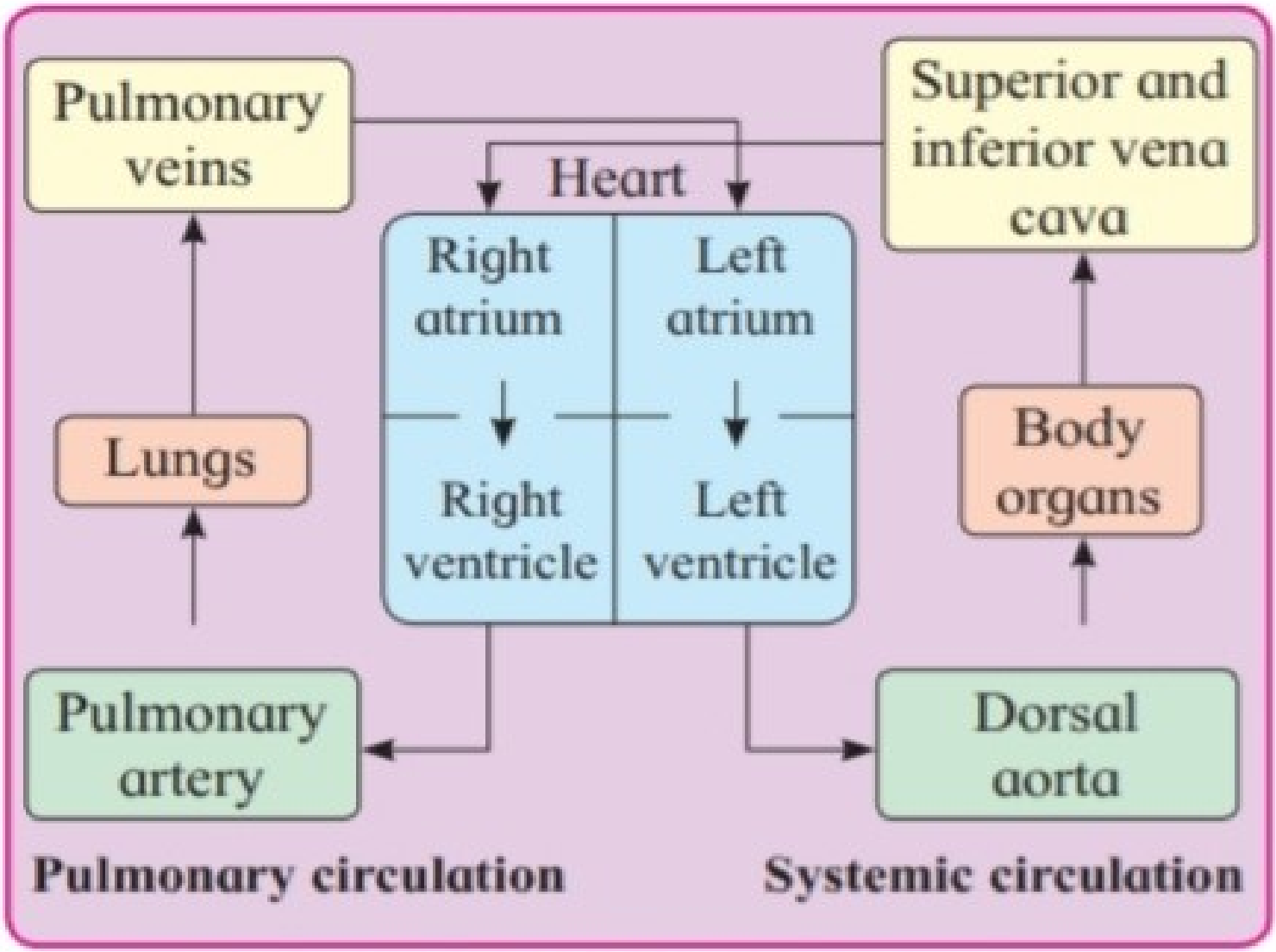
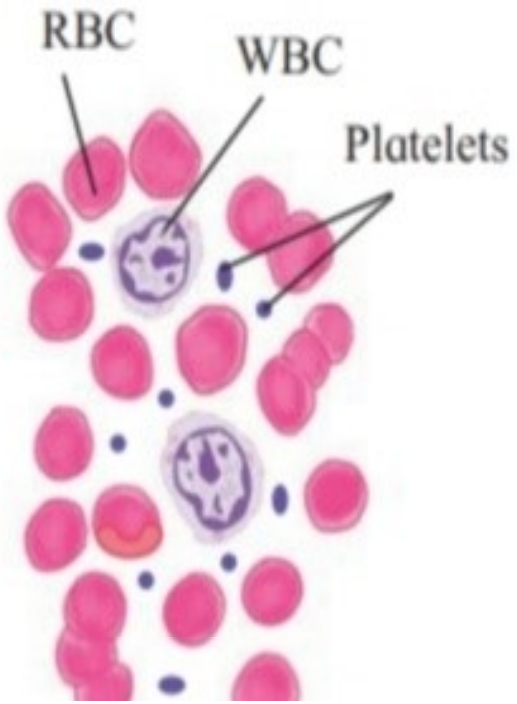
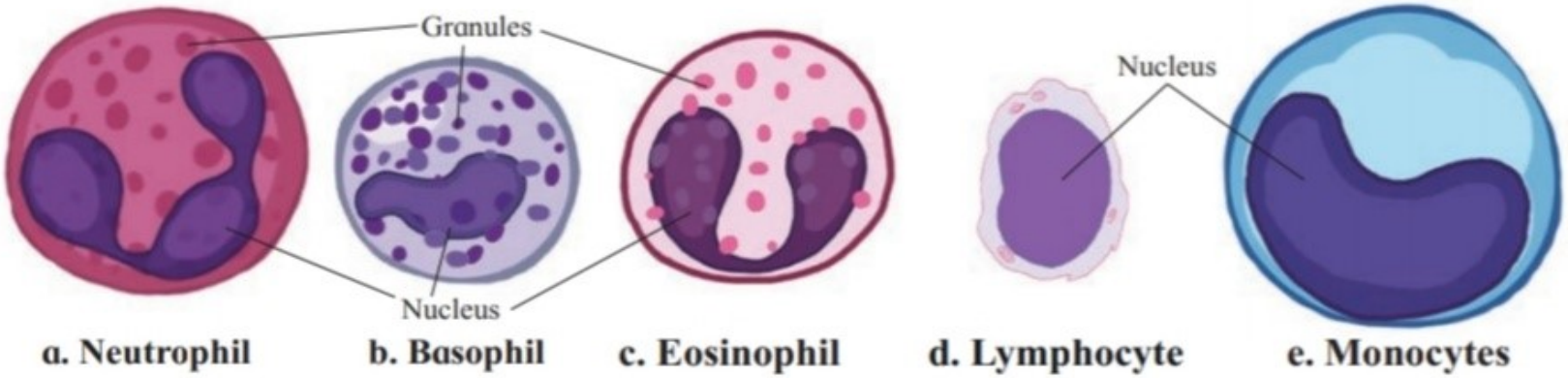


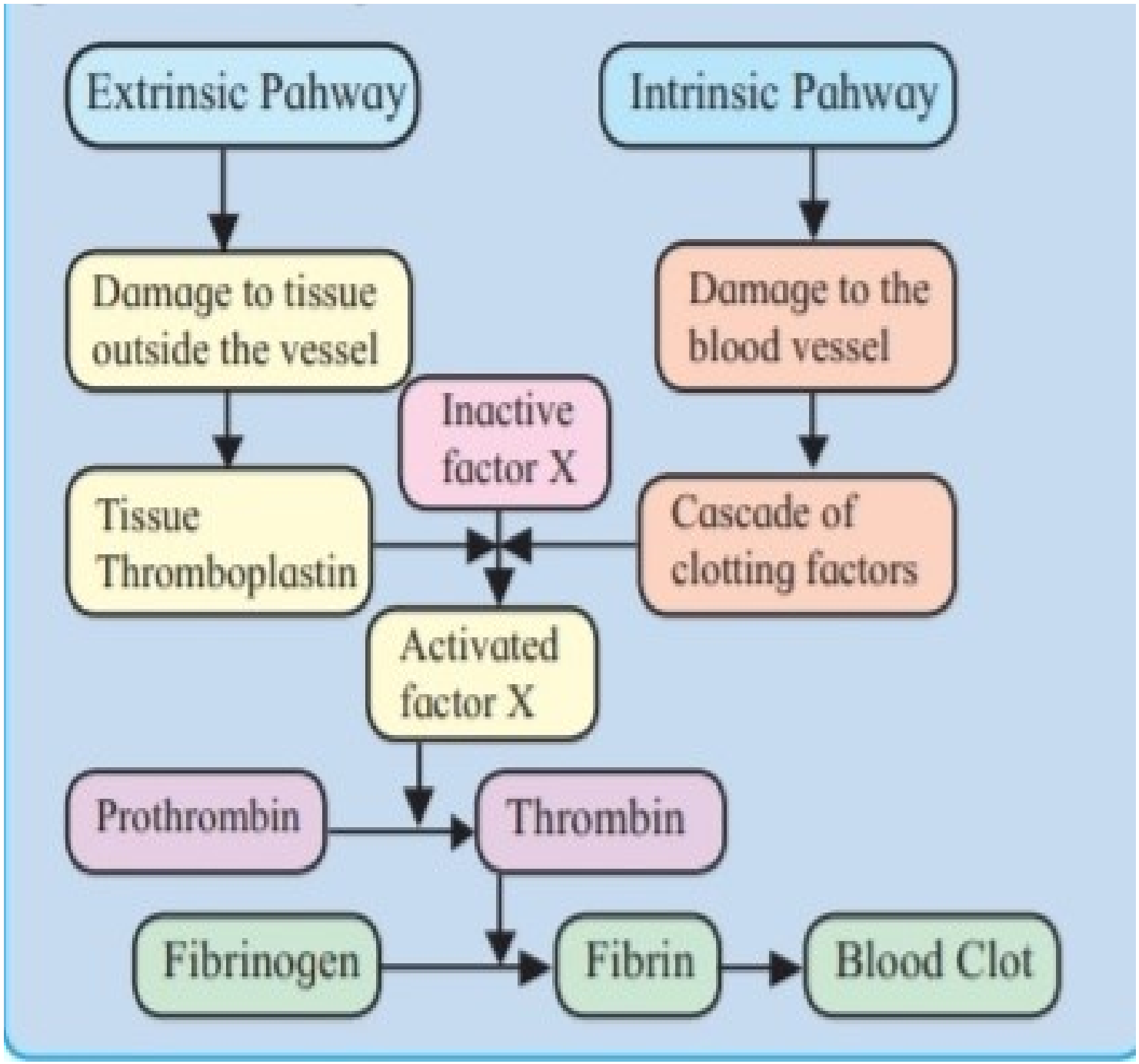
Fig. 8.13 : Double circulation (Diagrammatic)

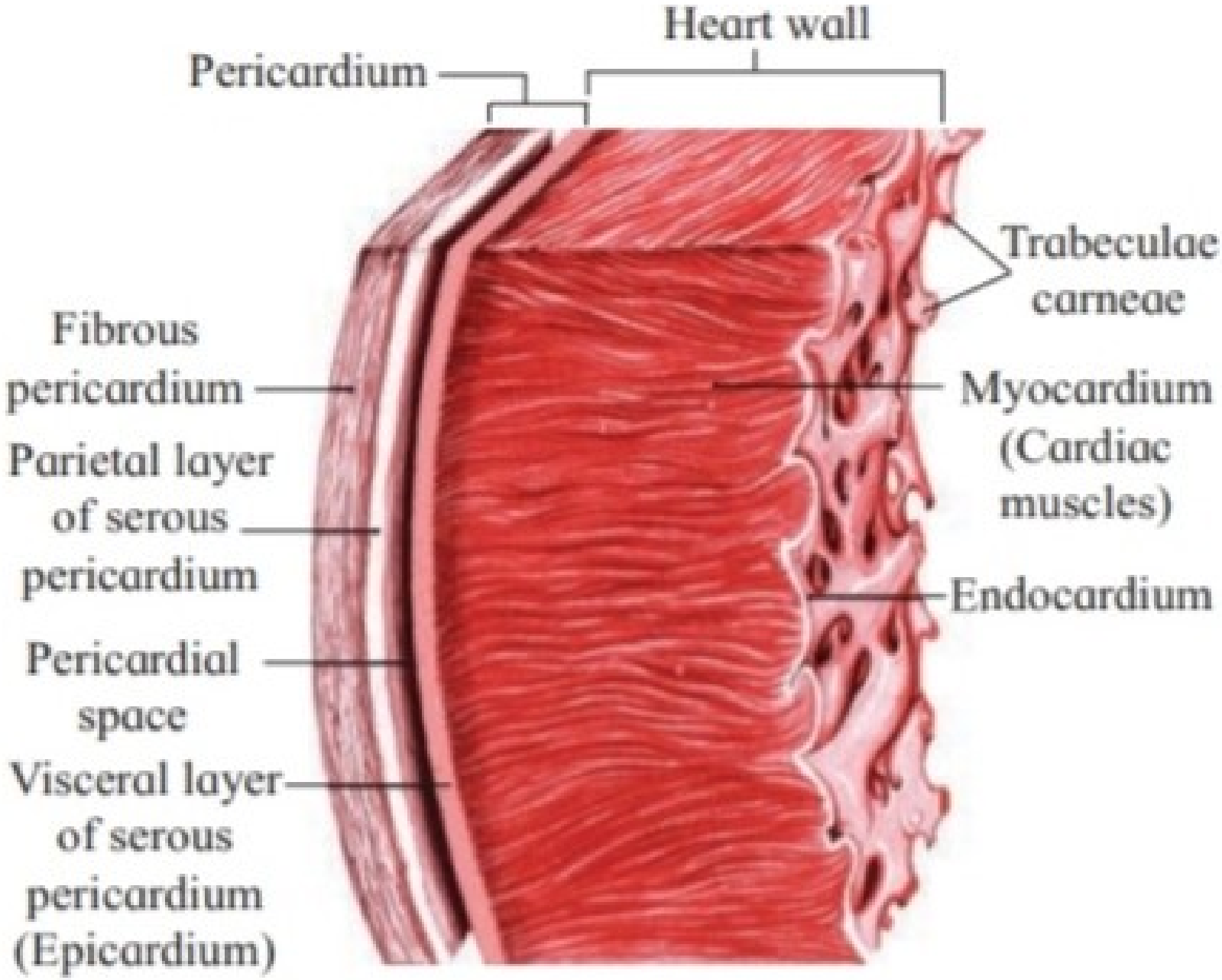
Contents	Percentage
1. Water	90 %
2. Proteins (albumen, globulin, properdin, prothrombin, fibrinogen)	7 to 8 %
3. Inorganic salts (Na, K, Mg, Ca, Fe, Mn and Cl^-, HCO^-_3 and PO^{3-}_4)	1 %
4. Others :	1 to 2 %
a. Food (glucose, amino acids, fatty acids, triglycerides)	
b. Wastes (urea, uric acid and creatinine)	
c. Regulators (hormones, enzymes, vitamins)	
d. Anticoagulants (heparin)	
e. Cholesterol and antibodies	
f. Dissolved gases (O_2, CO_2, N_2)	

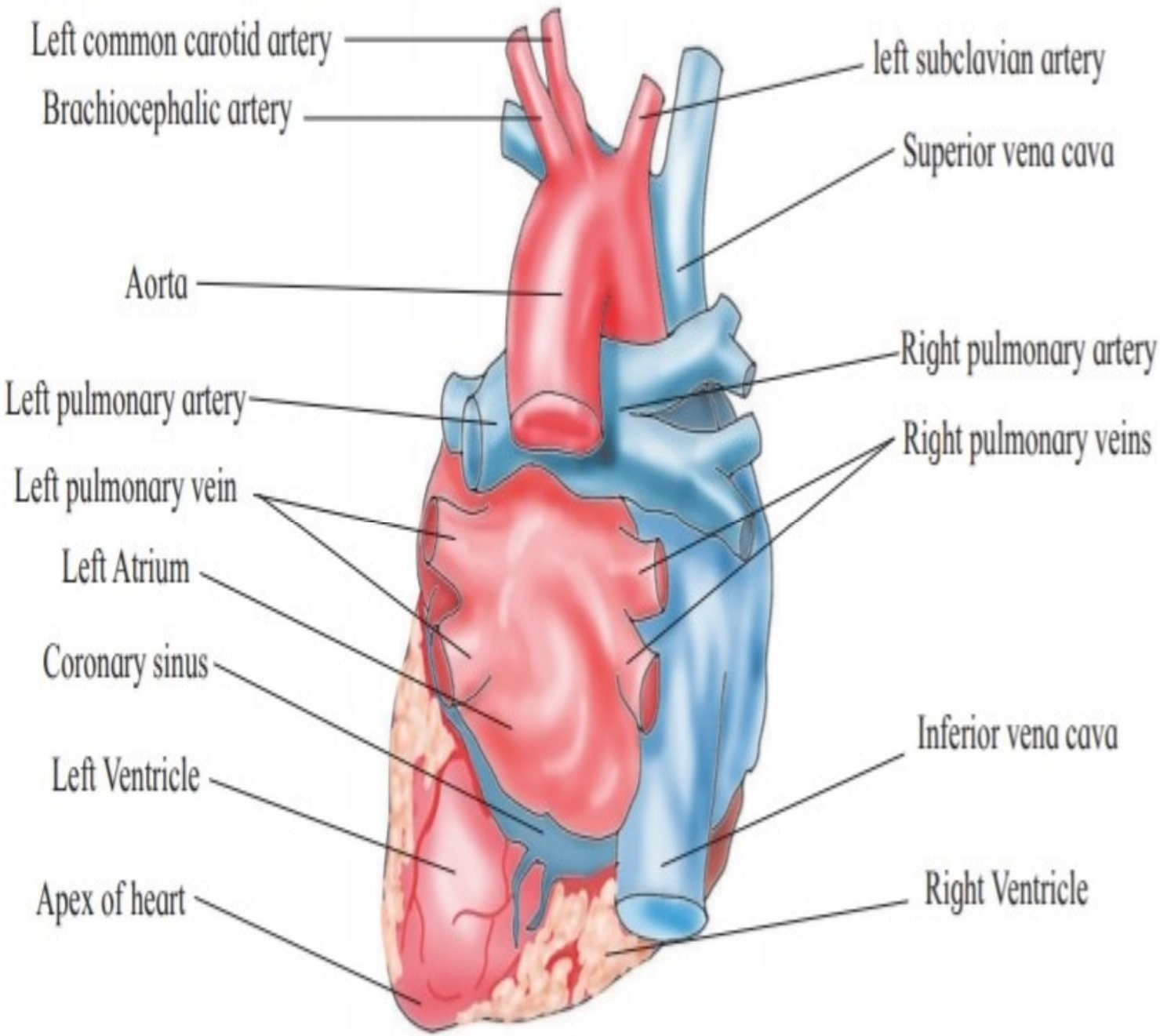


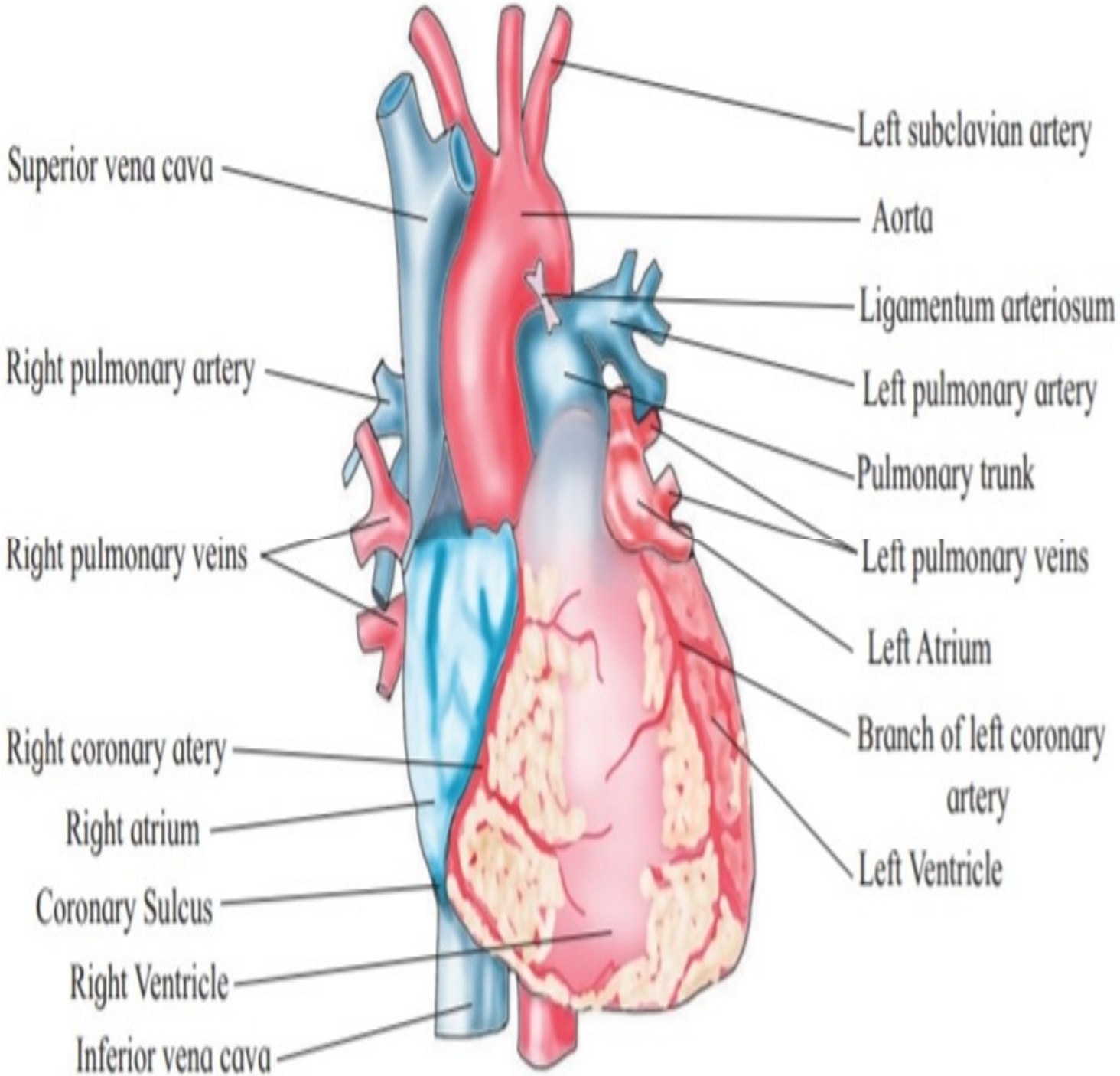


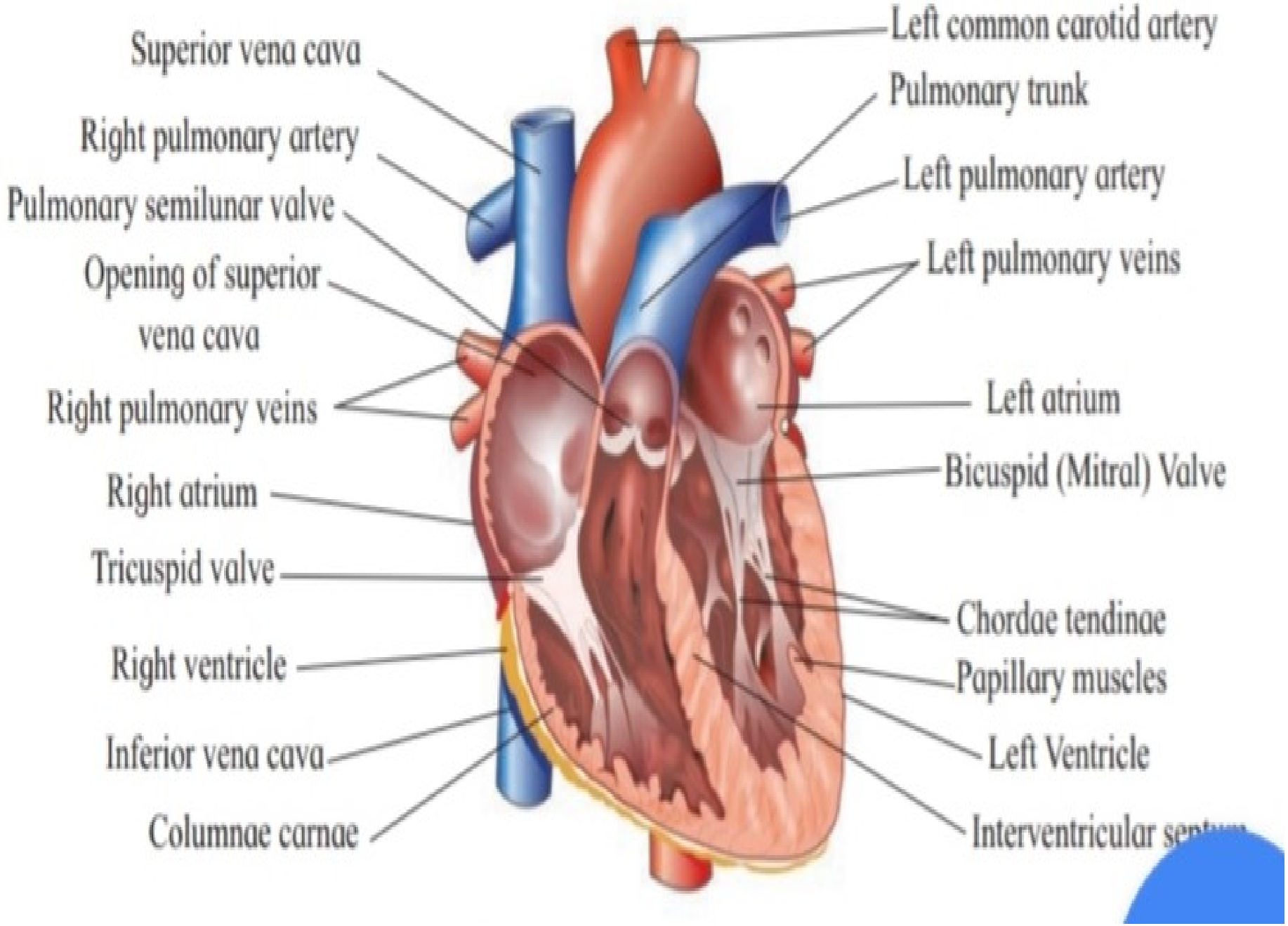
a. Neutrophil b. Basophil c. Eosinophil d. Lymphocyte e. Monocytes

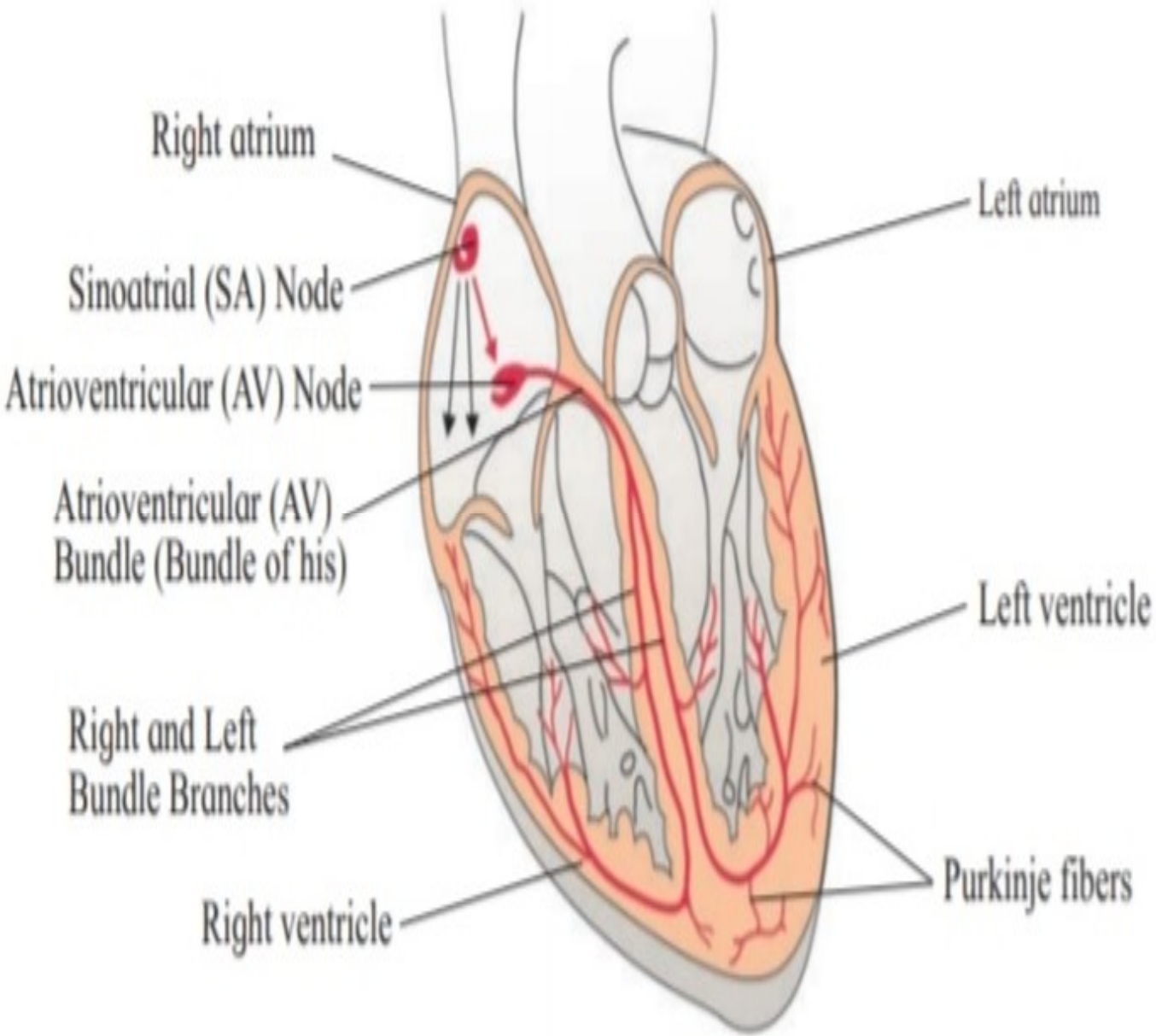

Extrinsic Pahway
Intrinsic Pahway
Damage to tissue outside the vessel
Damage to the blood vessel
Inactive factor X
Tissue Thromboplastin
Cascade of clotting factors
Activated factor X
Prothrombin
Thrombin
Fibrinogen
Fibrin
Blood Clot


Heart wall
Pericardium
Trabeculae carneae
Fibrous pericardium
Myocardium (Cardiac muscles)
Parietal layer of serous pericardium
Endocardium
Pericardial space
Visceral layer of serous pericardium (Epicardium)


Left common carotid artery
Brachiocephalic artery
left subclavian artery
Superior vena cava
Aorta
Right pulmonary artery
Left pulmonary artery
Right pulmonary veins
Left pulmonary vein
Left Atrium
Coronary sinus
Inferior vena cava
Left Ventricle
Apex of heart
Right Ventricle


Left subclavian artery
Superior vena cava
Aorta
Ligamentum arteriosum
Right pulmonary artery
Left pulmonary artery
Pulmonary trunk
Right pulmonary veins
Left pulmonary veins
Left Atrium
Right coronary atery
Branch of left coronary artery
Right atrium
Left Ventricle
Coronary Sulcus
Right Ventricle
Inferior vena cava


Superior vena cava
Right pulmonary artery
Pulmonary semilunar valve
Opening of superior vena cava
Right pulmonary veins
Right atrium
Tricuspid valve
Right ventricle
Inferior vena cava
Columnae carnae
Left common carotid artery
Pulmonary trunk
Left pulmonary artery
Left pulmonary veins
Left atrium
Bicuspid (Mitral) Valve
Chordae tendinae
Papillary muscles
Left Ventricle
Interventricular sept


Right atrium
Left atrium
Sinoatrial (SA) Node
Atrioventricular (AV) Node
Atrioventricular (AV) Bundle (Bundle of his)
Left ventricle
Right and Left Bundle Branches
Purkinje fibers
Right ventricle

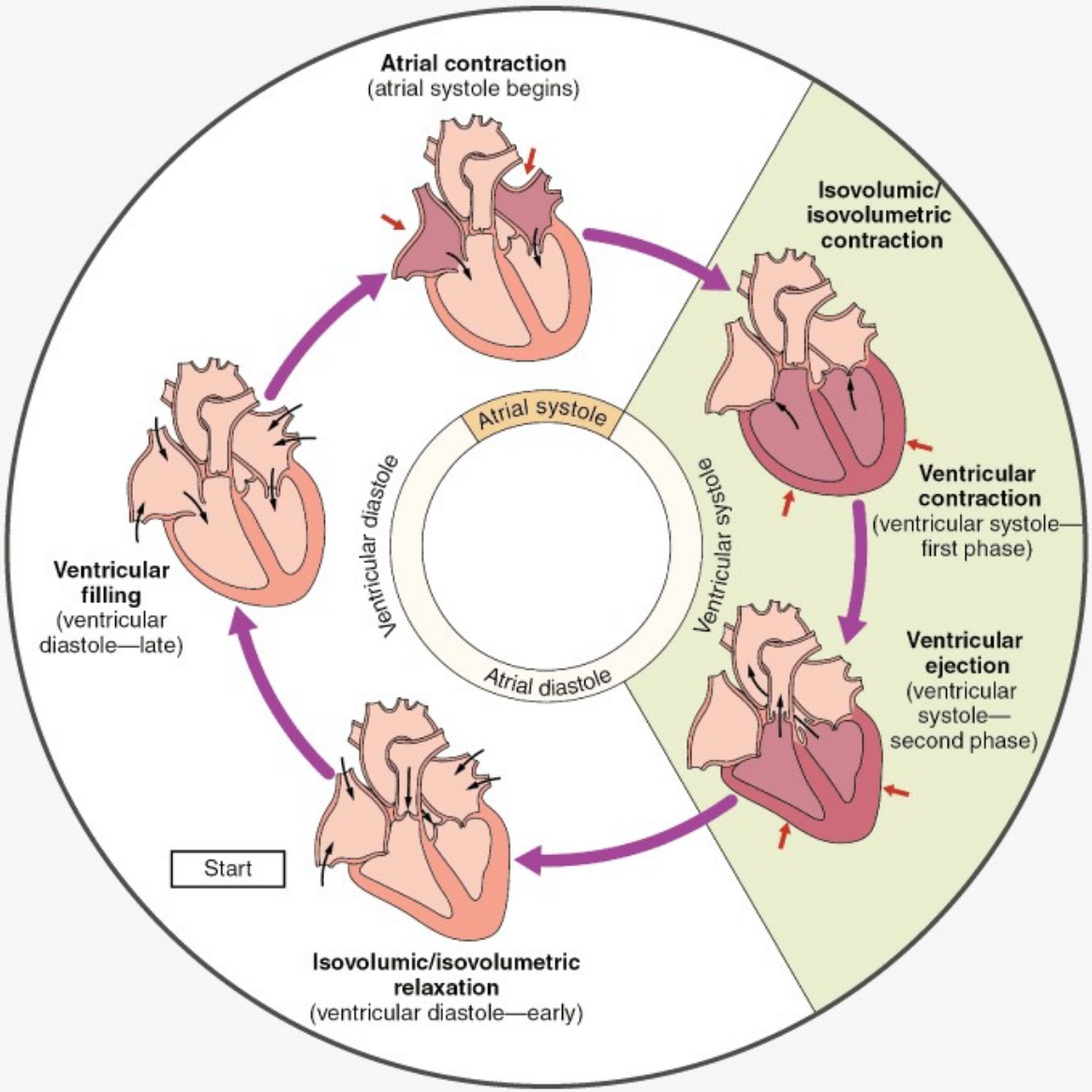
Cardiac Cycle

- The cardiac cycle attributes to a comprehensive heartbeat from its production to the commencement of the next beat.
- It comprises diastole, the systole, and the intervening pause.
- The occurrence of a cardiac cycle is illustrated by a heart rate, which is naturally indicated as beats per minute.

- A healthy human heart beats 72 times per minute which states that there are 72 cardiac cycles per minute.
- The cardiac cycle involves a complete contraction and relaxation of both the atria and ventricles and the cycle last approximately 0.8 seconds.

- **Cardiac Cycle Diagram**
- The diagram below represents the different phases of the cardiac cycle.
- The atrial systole, ventricular diastole, ventricular systole and ventricular diastole are clearly mentioned in the cardiac cycle diagram given below.

CARDIAC CYCLE



• Cardiac Cycle Phases

Following are the different phases that occur in a cardiac cycle:

- **Atrial Diastole**: In this stage, chambers of the heart are calmed. That is when the aortic valve and pulmonary artery closes and atrioventricular valves open, thus causing chambers of the heart to relax.

- **Atrial Systole**: At this phase, **blood cells** flow from atrium to ventricle and at this period, atrium contracts.

- **Isovolumic Contraction**: At this stage, ventricles begin to contract. The atrioventricular valves, valve, and pulmonary artery valves close, but there won't be any transformation in volume.

- **Ventricular Ejection**: Here ventricles contract and emptying. Pulmonary artery and aortic valve close.

- **Isovolumic Relaxation**: In this phase, no blood enters the ventricles and consequently, pressure decreases, ventricles stop contracting and begin to relax. Now due to the pressure in the aorta – pulmonary artery and aortic valve close.

- **Ventricular Filling Stage:** In this stage, blood flows from atria into the ventricles. It is altogether known as one stage (first and second stage). After that, they are three phases that involve the flow of blood to the pulmonary artery from ventricles.

- Duration of Cardiac Cycle
- In a normal person, a heartbeat is 72 beats/minute. So, the duration of one cardiac cycle can be calculated as:

- 1/72 beats/minute=.0139 minutes/beat

- At a heartbeat 72 beats/minute, duration of each cardiac cycle will be 0.8 seconds.

- Duration of different stages of the cardiac cycle is given below:
  - **Atrial systole:** continues for about 0.1 seconds
  - **Ventricular systole:** continues for about 0.3 seconds
  - **Atrial diastole:** continues for about 0.7 seconds
  - **Ventricular diastole:** continues for about 0.5 seconds

THANK YOU

RESTRICTION ENZYMES

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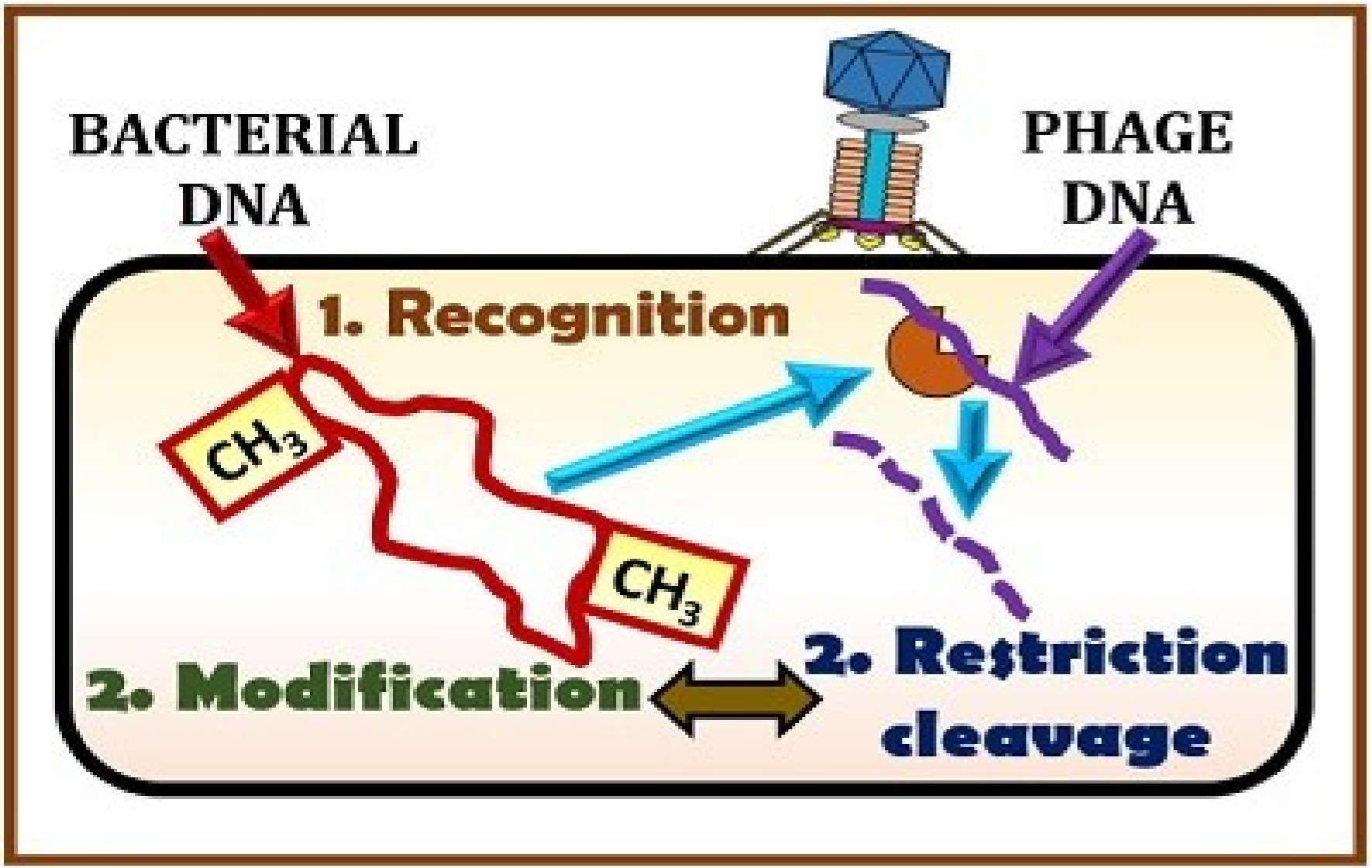
Restriction enzyme

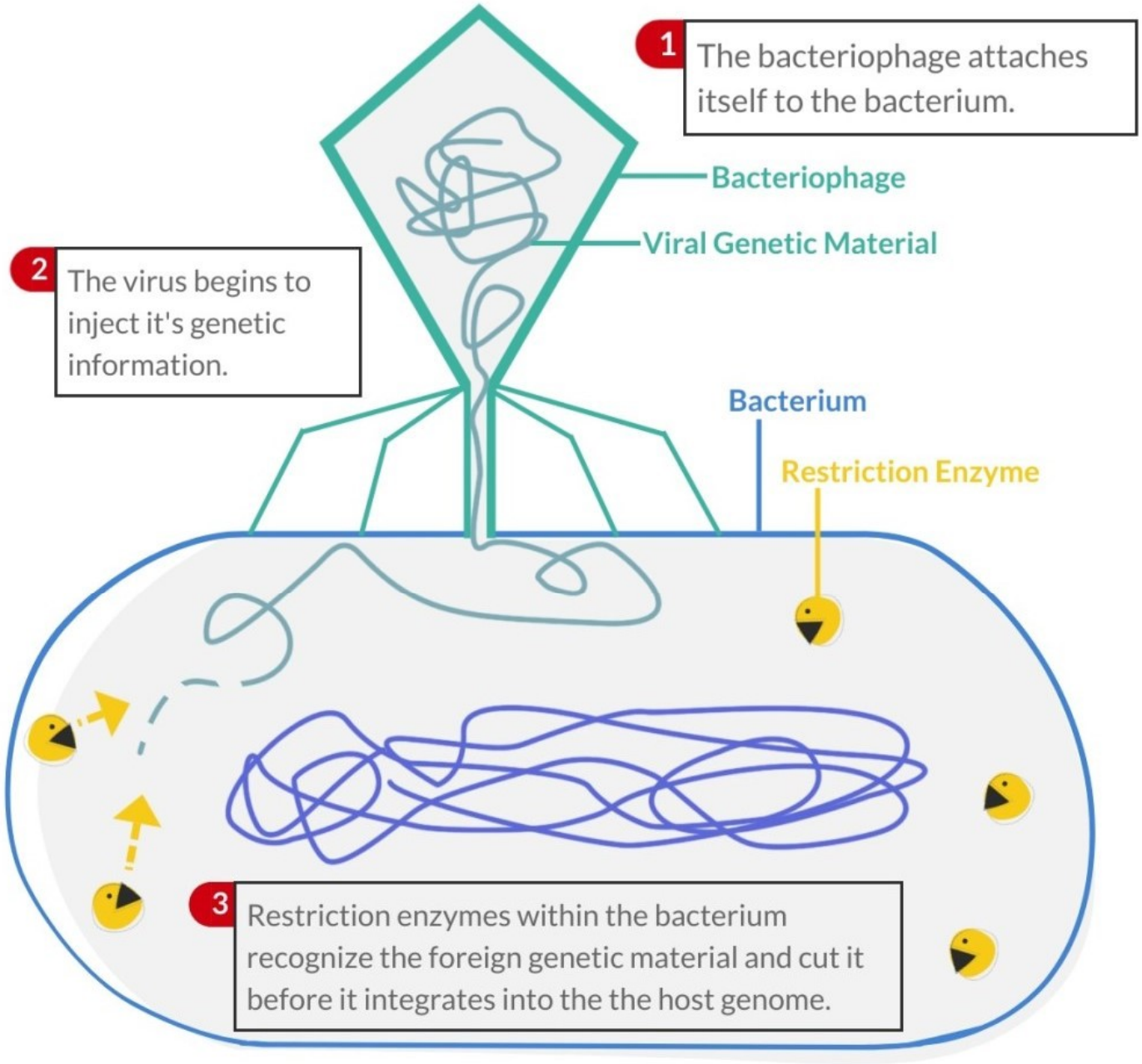
- **Restriction enzyme**, also called **restriction endonuclease**, a protein produced by bacteria that cleaves DNA at specific sites along the molecule.

- In the bacterial cell, restriction enzymes cleave foreign DNA, thus eliminating infecting organisms.

- Restriction enzymes can be isolated from bacterial cells and used in the laboratory to manipulate fragments of DNA, such as those that contain genes; for this reason they are indispensible tools of recombinant_DNA_technology (genetic engineering).



ROLE OF RESTRICTION ENZYME


1
The bacteriophage attaches itself to the bacterium.
Bacteriophage
Viral Genetic Material
2
The virus begins to inject it's genetic information.
Bacterium
Restriction Enzyme
3
Restriction enzymes within the bacterium recognize the foreign genetic material and cut it before it integrates into the the host genome.

- A bacterium uses a restriction enzyme to defend against bacterial viruses called bacteriophage or phages.

- When a phage infects a bacterium, it inserts its DNA into the bacterial cell so that it might be replicated. The restriction enzyme prevents replication of the phage DNA by cutting it into many pieces.

- Restriction enzymes were named for their ability to restrict, or limit, the number of strains of bacteriophage that can infect a bacterium.

- Each restriction enzyme recognizes a short, specific sequence of nucleotide bases (the four basic chemical subunits of the linear double-stranded DNA molecule—adenine, cytosine, thymine, and guanine).

- These regions are called recognition sequences, or recognition sites, and are randomly distributed throughout the DNA.

- Different bacterial species make restriction enzymes that recognize different nucleotide sequences.

- When a restriction endonuclease recognizes a sequence, it snips through the DNA molecule by catalyzing the hydrolysis (splitting of a chemical bond by addition of a water molecule) of the bond between adjacent nucleotides.

- Bacteria prevent their own DNA from being degraded in this manner by disguising their recognition sequences. Enzymes called methylases add methyl groups (—CH_3) to adenine or cytosine bases within the recognition sequence, which is thus modified and protected from the endonuclease. The restriction enzyme and its corresponding methylase constitute the restriction-modification system of a bacterial species.

SUMMARY:

- A restriction enzyme is a **nuclease enzyme** that cleaves DNA sequence at a random or specific recognition sites known as **restriction sites.**

- In bacteria, restriction enzymes form a combined system (**restriction + modification system**) with modification enzymes that methylate the bacterial DNA.

- Methylation of bacterial DNA at the recognition sequence typically protects the own DNA of the bacteria from being cleaved by restriction enzyme.

- There are two different kinds of restriction enzymes:

- (1) Exonucleases catalyses hydrolysis of **terminal** nucleotides from the end of DNA or RNA molecule either **5'to 3' direction or 3' to 5' direction.**
- Example:
- exonuclease I, exonuclease II etc.

- (2) **Endonucleases** can recognize specific base sequence (restriction site) within DNA or RNA molecule and cleave **internal phosphodiester bonds** within a DNA molecule. Example: EcoRI, Hind III, BamHI etc.

Sticky & Blunt Ends

EcoRI produces "sticky" ends,

SmaI restriction enzyme produces "blunt" ends

Restriction Endonuclease Nomenclature

- Restriction endonucleases are named according to the organism in which they **were discovered**, using a system of letters and numbers.

- For example, HindIII (pronounced "hindee-three")

  was discovered in Haemophilus influenza (strain d). Roman numerals are used to identify specific enzymes from bacteria that contain multiple restriction enzymes indicating the order in which restriction enzymes were discovered in a particular strain.

Named for bacterial genus, species, strain, and type

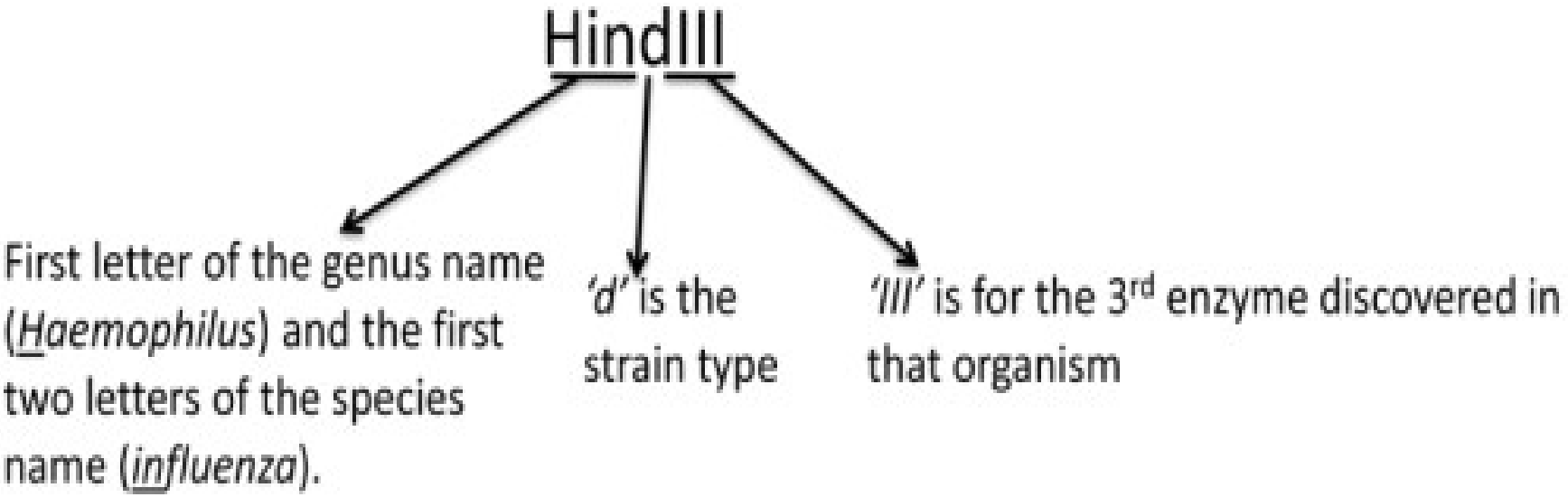
Example: EcoR1

Genus: Escherichia

Species: coli

Strain: R

Order discovered: 1



Classification of Restriction Endonucleases

- There are three major classes of restriction endonucleases based on the types of sequences recognized, the nature of the cut made in the DNA, and the enzyme structure:

  - Type I restriction enzymes
  - Type II restriction enzymes
  - Type III restriction enzymes

Type I restriction enzymes:

• These enzymes have both restriction and modification activities. Restriction depends upon the methylation status of the target DNA.

• Cleavage occurs approximately **1000 bp** away from the recognition site.

• The recognition site is asymmetrical and is composed of two specific portions in which one portion contain 3–4 nucleotides while another portion contain 4–5 nucleotides and both the parts are separated by a non-specific spacer of about 6–8 nucleotides.

• They require S-adenosylmethionine (SAM), ATP, and magnesium ions (Mg^{2+}) for activity.

• These enzymes are composed of mainly three subunits, a specificity subunit that determines the DNA recognition site, a restriction subunit, and a modification subunit

Type II restriction enzymes:

- Restriction and modification are mediated by separate enzymes so it is possible to cleave DNA in the absence of modification.

- Although the two enzymes recognize the same target sequence, they can be purified separately from each other.

- Cleavage of nucleotide sequence occurs at the restriction site.

- These enzymes are used to recognize rotationally symmetrical sequence which is often referred as palindromic sequence.

- These palindromic binding site may either be interrupted (e.g. BstEII recognizes the sequence 5´-GGTNACC-3´, where N can be any nucleotide) or continuous (e.g. KpnI recognizes the sequence 5´-GGTACC-3´).

- They require only Mg2+ as a cofactor and ATP is not needed for their activity.

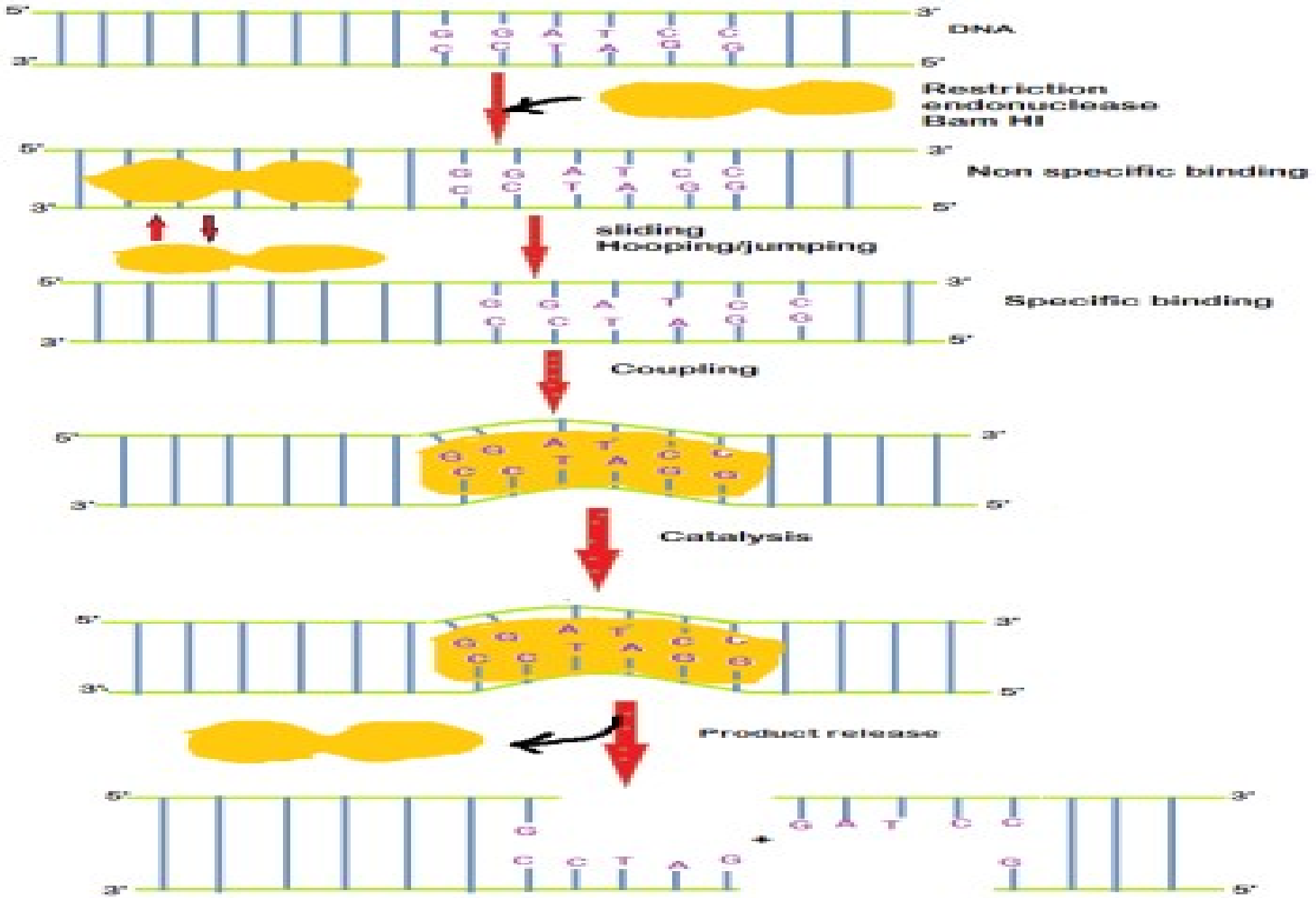


Fig 2-1.4.2: Structures of free, nonspecific, and specific DNA-bound forms of BamHI.

3. Type III restriction enzymes:

- These enzymes recognize and methylate the same DNA sequence but cleave 24–26 bp away.

- They have two different subunits, in which one subunit (M) is responsible for recognition and modification of DNA sequence and other subunit (R) has nuclease action.

- • Mg+2 ions, ATP are needed for DNA cleavage

- Cleave only one strand. Two recognition sites in opposite orientation are necessary to break the DNA duplex.

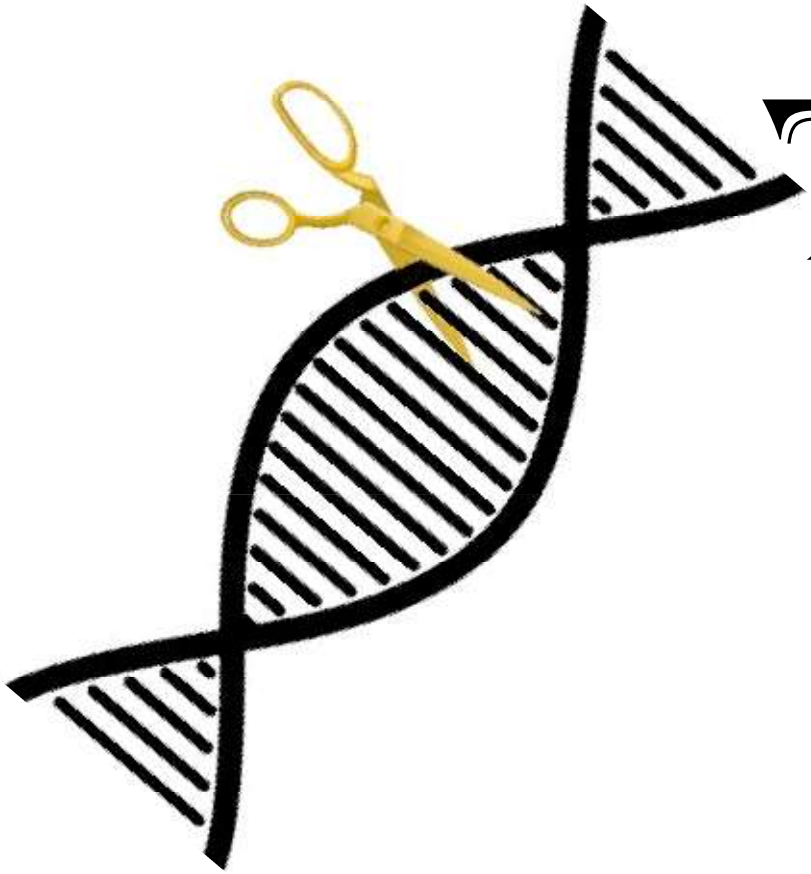
Applications:

- In various applications related to genetic engineering DNA is cleaved by using these restriction enzymes.

- They are used in the process of insertion of genes into plasmid vectors during gene cloning and protein expression experiments.

- Restriction enzymes can also be used to distinguish gene alleles by specifically recognizing single base changes in DNA known as single nucleotide polymorphisms (SNPs).
- This is only possible if a mutation alters the restriction site present in the allele.

- Restriction enzymes are used for Restriction Fragment Length Polymorphism (RFLP) analysis for identifying individuals or strains of a particular species.



THANK YOU