

(Section B)

Q. No. 2 Part (I)

## Extraembryonic Membranes

These membranes arise from the trophoblast cells of the blastocyst and surround the embryo from outside which is why they are called extra embryonic membranes.

1-Chorion: It is the outermost layer that mingles with maternal endometrium via chorionic villi to form placenta which is the source of nutrients and gaseous exchange for the fetus.

2-Yolk sac: It is primarily the nutrient source in most organisms but it is empty in humans as the human embryo gets food from the mother. It is the first site of RBC formation and contributes to umbilical cord too.

3-Allantois: It forms circulatory system

4-Amnion: It is the protective layer that surrounds and cushions embryo from shock and damage.

Q. No. 2 Part (II) Telomeres and Aging

Telomeres are present at ends of chromosomes. They are considered the genetic factor that influences aging.

Influence: When a cell divides, the genetic information present in the telomeres gradually reduces (like the slow disintegration of pencil lead as we write). A time comes when these telomeres reach a **critical length**. At this point, the cell loses its ability to divide. So, when any cell is damaged, it is unable to recover. This loss of cell division and recovery is the onset of aging.

It is a natural process where physiological changes become negative. Actually, it was observed that outside the body, cells cannot divide more than 100 times due to the disintegration and reduction in length of telomeres.

04

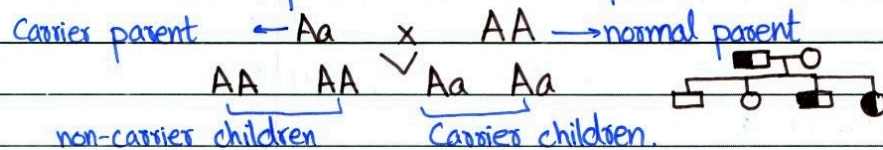
Answer the Question (Part) at the space specified.

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(Section B)

Q. No. 2 Part (III) Sickle Cell Anemia

A carrier parent is heterozygous and a normal parent is homozygous dominant. In this case, there is no chance of an affected child since it is an autosomal recessive disorder. But the chance of a carrier is as follows:



We see that the ratio of carrier to normal is 1:1. This means that there is a 50% chance of the child being a carrier. The probability will be:

$$\text{Probability} = \frac{n}{a} = \frac{2}{4} = \frac{1}{2}$$

Note: it is not possible to have an affected child in the case mentioned.

So, the probability will be  $\frac{1}{2}$ .

Q. No. 2 Part (iv)

## Sex Influenced Trait

These traits are more common in one gender as compared to another. This difference is mainly due to hormonal differences.

Inheritance Pattern: Let us consider the male balding pattern. The gene for this case acts dominant in males but recessive in females. This means that a male will be affected even in heterozygous condition but the female will only be affected if she is homozygous recessive. This is given in the table:

<u>Genotype</u>	<u>Male</u>	<u>Female</u>
BB ( $B_1B_1$ )	Normal	Normal
Bb ( $B_1B_2$ )	Bald	Normal
bb ( $B_2B_2$ )	Bald	Bald

(Section B)

Q. No. 2 Part (ix)

## Primary Function

The primary function of the human respiratory system is gaseous exchange with the environment or the inhalation of oxygen and exhalation of carbon dioxide.

### Benefits of Function:

- Respiration is an important energy producing process of the human body that breaks down glucose and gives  $CO_2$ . This  $CO_2$  would harm the body if it stays inside so the respiratory system expels it.
- Apart from energy production, oxygen is also needed for the proper functioning of all the body organs. Too much  $CO_2$  in blood can lead to serious respiratory and circulatory problems. So, gaseous exchange is essential for life and proper functioning of the body.

## Pedigree

Q. No. 2 Part (v)

Mother =  $X^R X^r$  (carrier)      Father =  $X^r Y$  (affected)

$X^r X^R$  ×  $X^r Y$

$X^r X^r$        $X^R Y$        $X^r Y$        $X^R X^r$

affected female      normal son      affected son      carrier daughter

We see that the child 3 (second daughter) is not affected. She is a carrier. The given pedigree is an X-linked recessive disorder pedigree where the father is affected and the mother is a carrier. In this case, there is 50% chance among all children to be affected. The probability of an affected daughter and a carrier daughter are also  $\frac{1}{2}$  or 50%. From the above given cross, we can see that the probability of child 3 to be affected is half but she is most likely carrier.

Q. No. 2 Part (vi)

## DNA Sequencing

The three major steps involved in any DNA sequencing technique are:

**1- Fragments:** Creating various fragments of DNA that start at the same point but end at separate points.

**2- Gel Electrophoresis:** In this technique, the DNA fragments are separated on the basis of their different sizes.

**3- Analysis of Results:** The results are then analysed and probably incorporated via autoradiography or other X-ray techniques.

These three basic steps are used in every DNA sequencing technique including RFLP investigation, paternity establishment, etc. and the results are also analysed carefully.

## Purpose

Q. No. 2 Part (vii)

DNA Ligase: This "molecular glue" is used to create the recombinant DNA by joining the fragments of the gene of interest and the vector molecule. It ensures the binding of base pairs so that the recombinant DNA is formed and transferred to the next generation.

Vector: It is the "carrier". The vector is used to carry or transfer this recombinant DNA or gene of interest into the expression system or host. A vector is specific and should have an origin of replication site, restriction sites of various enzymes and antibiotic resistance.

Q. No. 2 Part (viii)

## Gene Therapy

Gene therapy helps to "replace" the faulty gene with normal one. The following two methods are employed:

In vivo method: The corrective genes are inserted directly into the body via carrier to the target organs.

Ex vivo method: Cells are first extracted from the affected area, the gene is inserted. The cells are cloned and then these modified cells are inserted back into the body to the affected organ.

Both techniques are equally valid and effective in their mode of action:

in in vivo method, cells are modified inside the body while they are modified outside the body in ex-vivo.

Cells take up the corrective genes for in vivo treatment.

Q. No. 2 Part (x)

## Molecular Biology

All organisms show similarities in their molecular makeup.

→ Cytochromes are found in all organisms. Organisms share introns too. Almost 90% of DNA is similar (99%).

We see that all organisms use the same primary molecules (proteins, carbohydrates, lipids, nucleic acids) as well as large similarities in their usage too. ATP is the main source of energy for all organisms.

In all these similarities, we see that there must have been a common ancestor from which all organisms had arisen. Thus, evidence from molecular biology support Darwin's concept of evolution and descent with modification.

(Section B)

Q. No. 2 Part (xi)

## Hardy Weinberg Theorem

### Conditions:

- 1- Population must be large.
- 2- There should be no selection.
- 3- There should be no mutation.
- 4- There should be no gene flow (migration).
- 5- Mating should be random.
- 6- Population must be isolated and sexually reproducing.

The Hardy Weinberg Theorem states that if all these conditions are met, the population will be in genetic equilibrium.

"Evolution is the deviation from Hardy-Weinberg Equilibrium".

Q. No. 2 Part (xii)

## Significance of Hypothalamus

It is the master control centre of the endocrine glands. It has neurosecretory cells that produce and release releasing and inhibiting factors. These factors influence the release of hormones from the pituitary gland. In addition, hypothalamus also produces two hormones: oxytocin and ADH. These two hormones are stored in the posterior pituitary and released when stimulated by the hypothalamus. So, the three roles of hypothalamus are:

- 1- Serves as link between nervous and endocrine system.
- 2- Controls secretions of anterior pituitary.
- 3- Production of oxytocin and ADH.

(Section B)

Q. No. 2 Part (xiii)

## Integrated Disease Management

It is the utilization of all possible techniques and process to control the spread of a dangerous disease or a group of local diseases and to prevent their further onset.

Actually, the main purpose is disease control and integrated disease management uses all possible methods to achieve this goal. For example, public awareness is important to prevent spread of a disease so integrated disease management can be used to organise walks, seminars and podcasts for this purpose. In addition, vaccination, preparation and administration of medicines, TV programmes for awareness are also included.

Q. No. 2 Part (xiv)

## Role of Diaphragm

It is the dome shaped skeletal muscle that separates the thoracic and abdominal cavity.

Inspiration: During inspiration, the diaphragm contracts and moves downward to become less dome shaped. This causes an increase in thoracic volume and decrease in air pressure due to which air enters lungs.

Expiration: Diaphragm relaxes and moves upwards back to its dome shape. So, the thoracic volume decreases and air pressure increases which expels air out of lungs.

Thus, diaphragm helps in ventilation by changing pressure and volume in chest cavity along with the external and internal intercostal muscles.

## XX-XY Mechanism

Found in: This mechanism of sex determination is found in:

- Mammals (including man).
- *Drosophila* (with few differences).

### What is XX-XY?

It basically shows what type of gametes will be seen in the two different sexes of a particular specie.

### XX-XY in Man

In Man:

XX: It determines the female offspring. When the sperm containing the X chromosome fertilizes an egg containing X chromosome too, the offspring is a female.

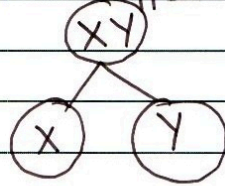
XY: It determines the male offspring. When the sperm containing the Y chromosome fertilizes an egg containing X chromosome, the offspring is a male.

Probability: The ratio of a son to a daughter is 1:1 which means that the probability for either gender in the next generation is  $\frac{1}{2}$  or 50%.

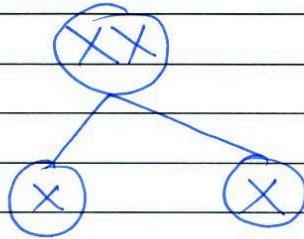
Deciding Factor: The gender of the offspring depends entirely on the type of sperm (X or Y) that fertilizes the egg.

Q. No. 3 (Page 2) Why males are heterogametic?

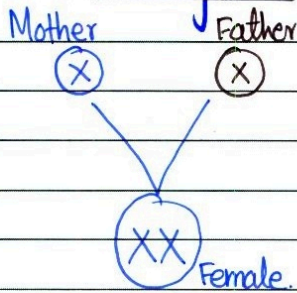
Males are considered to be heterogametic since their sperms contain two different types of chromosomes.



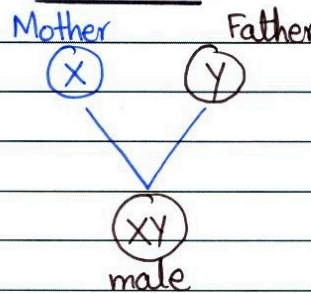
Since it produces two different kinds of gametes, therefore we consider a human male to be heterogametic and a human female to be homogametic, since she produces two same gametes.



Daughter



Son





# PCR

Q. No. 4 (Page 1)

Polymerase Chain Reaction is the technique used to produce thousands of copies of a gene via in vitro method.

## PCR Mixture and Thermocycler:

The PCR mixture consists of many dNTPs (free nucleotides), primers and Taq polymerase in a buffer solution.

Taq polymerase is the enzyme derived from *Thermophilus aquaticus* bacteria that lives in hot springs. It is highly temperature resistant.

Thermocycler is the machine that varies temperature of the mixture as per requirements of the process.

## Procedure

### 1) Denaturation:

Heat is used as the denaturation agent in PCR. Temperature is raised to about  $94^{\circ}\text{C}$  for a minute or two. This causes the two strands of DNA to completely separate from each other and become single stranded.

### 2) Primer Annealing:

Forward and backward primers are annealed or hybridized to the separated DNA strands. The temperature during this process is reduced to  $54^{\circ}\text{C}$ . The primers provide the points of attachment to which the Taq polymerase will attach the free nucleotides during extension phase.

## (Section C)

**Q. No. 4 (Page 2) 3) Extension/Amplification:**

The thermocycler increases the temperature to about  $72^{\circ}\text{C}$ . This increase favours the activity of the Taq polymerase and we get two copies of our original DNA.

The temperature may again be increased to about  $94^{\circ}\text{C}$  to denature the strands, make their copies and so on. We see that the process occurs in a cyclic manner and results in exponential ( $2 \rightarrow 4 \rightarrow 8 \rightarrow 16$ ) increase in number of DNA molecules.

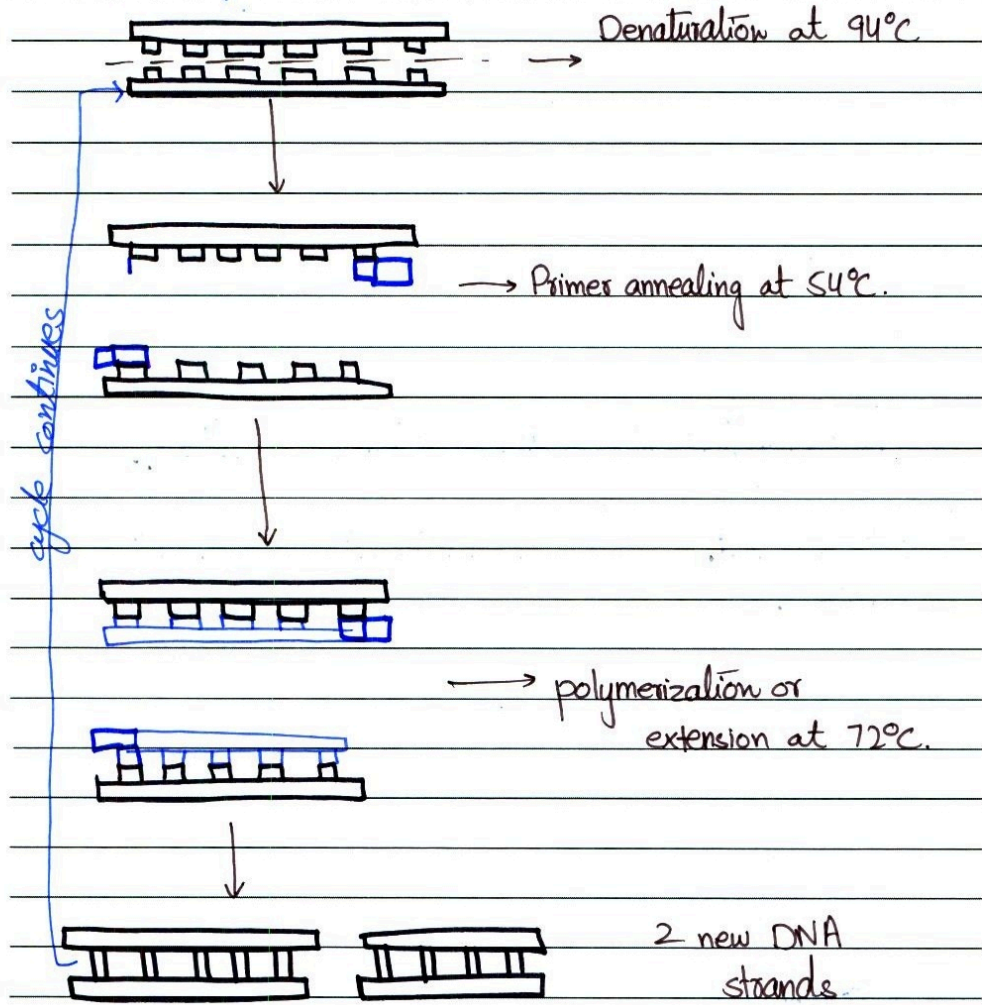
Thus, we say that PCR has amplified the gene.

## Applications

The applications of PCR are vast and enormous. Some are listed below:

- 1- Create millions of copies of a gene of interest in small amount of time.
- 2- Used to identify defects in gene. (study genes)
- 3- Used to highlight the disease causing bacteria and other harmful agents.
- 4- Used in forensic sciences to:
  - i) identify victims from tiny samples
  - ii) establish paternity cases.
  - iii) identify culprits.
- 5- PCR can also be used in genetic screening.
- 6- PCR can be used to make medicines and other useful compounds too, same as recombinant DNA Technology.
- 7- Synthesis of vaccines on a large scale.

Q. No. 4 (Page 3)



# Polymerase Chain Reaction

## Action Potential

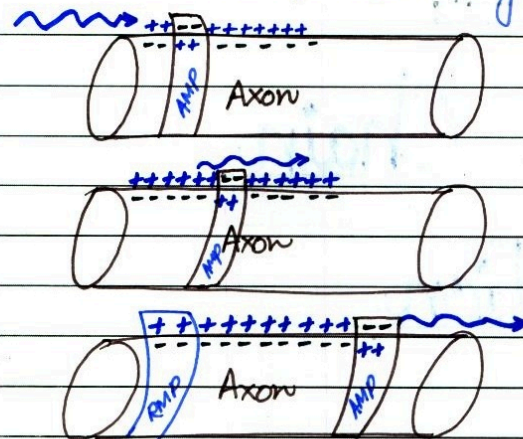
When the neuron membrane is depolarized, a nerve impulse passes through the neuron. This is called action potential or active membrane potential.

It is the state in which the neuron conducts an impulse. We can call action potential the impulse or electrochemical change that passes along neuron.

### Identification and Explanation

1) Process X: It is depolarization of neuron membrane.

When the threshold stimulus reaches the neuron, it causes the  $\text{Na}^+$  gates open. This is called influx of  $\text{Na}^+$  ions. Due to this influx, the positive charge inside the membrane becomes more than the negative charge so we say that the neuron membrane is depolarized. The potential of neuron membrane increases from  $-70 \text{ mV}$  to  $0 \text{ mV}$  and then to  $+50 \text{ mV}$ , as shown by the peak in the graph.



Conduction  
of  
Action  
Potential  
in a  
Neuron

Q. No. 5 (Page 2) 2) Process 1: It is repolarization of neuron membrane. The  $K^+$  gates open just after the  $Na^+$  gates and  $K^+$  ions start to move out of the neuron membrane. This works to bring down the membrane potential from +50mV value and to return the neuron to its polarized state of more positive outside and more negative inside.

3) Process 2: It shows the hyperpolarization of neuron membrane followed by refractory period. The  $K^+$  ions keep leaving the neuron. Due to a delay in the closing of  $K^+$  ion gate, there is an excess loss of +ve ions and the potential reduces to almost -90mV.

At this point, the sodium-potassium pump starts to work to bring the concentration of  $K^+$  ions to normal.

In the refractory period, we see a reversal of ions position. This is because here, a neuron has more  $Na^+$  inside and more  $K^+$  outside while a normal neuron in RMP has more  $Na^+$  outside and more  $K^+$  inside. So, the sodium potassium pump uses ATP to restore the original ionic balance by pumping  $Na^+$  out and  $K^+$  in.

At this point, it does not conduct a nerve impulse. This period lasts for about 2-4ms.

Thus, we see that a nerve impulse is conducted along the neuron membrane as action potential and various changes occur and are reversed to maintain the normal resting state of a neuron.





Q. No. 6 (Page 1)

# Types of Muscles

## Identification

Type A: Cardiac Muscle

Branching and Striation shows clear distinction.

Type B: Skeletal Muscle

Cylindrical and striated.

Type C: Smooth Muscle

Spindle shaped and uninucleated.

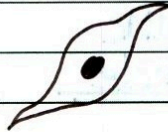
## Comparison of Properties

<u>Property</u>	<u>Smooth Muscle</u>	<u>Cardiac Muscle</u>	<u>Skeletal muscle</u>
Shape	Fusiform or Spindle shaped	Branched Cylindrical	Cylindrical or rod shape.
Number of nuclei	Single nucleus	Single Nucleus	Multi-nucleated.
Connections	May have gap junctions	Intercalated disks	No gap junctions
Control	Involuntary	Involuntary	Voluntary

(Section C)

Q. No. 6 (Page 2)

<u>Property</u>	<u>Smooth Muscle</u>	<u>Cardiac Muscle</u>	<u>Skeletal Muscle</u>
Nervous Control	Yes (Autonomic NS)	No (Spontaneous Contractions)	Yes (Somatic NS)
Striations	No	Yes (Irregular)	Yes (regular)
Found in	Stomach, intestine and various visceral organs.	Heart only	Connected to bones (mostly in limbs).



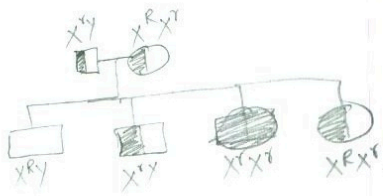
(Section C)

Q. No. 6 (Page 3)

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Space for rough work



$Aa \times Aa$   
 $Aa, Aa, Aa, aa$   
 $\frac{25\%}{}$

Human  $\rightarrow$  insulin  $\rightarrow$  engineering (not cloning)  
 $\hookrightarrow$  Recombinant DNA

Recombinant DNA Technology

2) Negative Feedback

