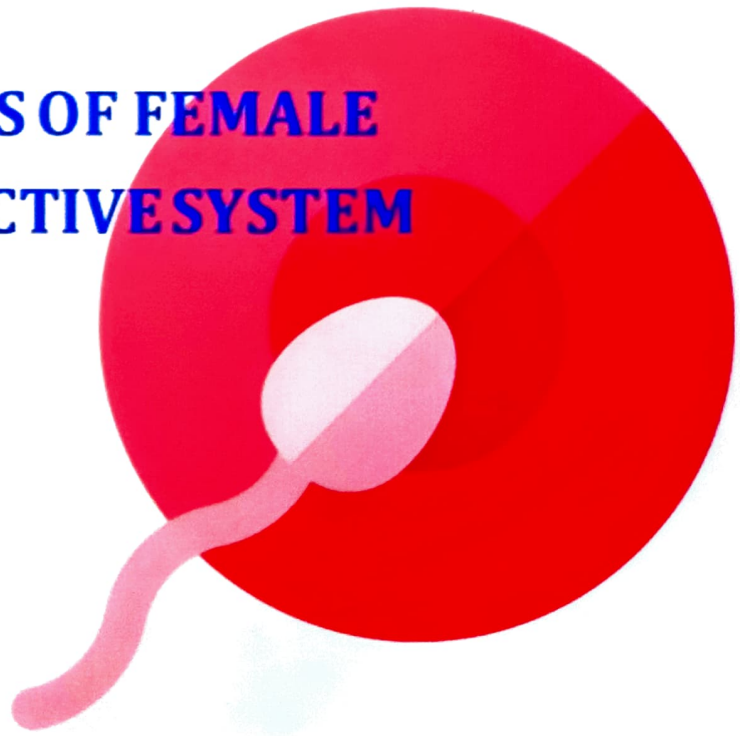
REPRODUCTIVE SYSTEM

PART - 1

Points to be covered in this topic

1. ANATOMY OF MALE REPRODUCTIVE SYSTEM
2. FUNCTIONS OF MALE REPRODUCTIVE SYSTEM
3. ANATOMY OF FEMALE REPRODUCTIVE SYSTEM
4. FUNCTIONS OF FEMALE REPRODUCTIVE SYSTEM



ANATOMY OF MALE REPRODUCTIVE SYSTEM

❑ MALE REPRODUCTIVE SYSTEM

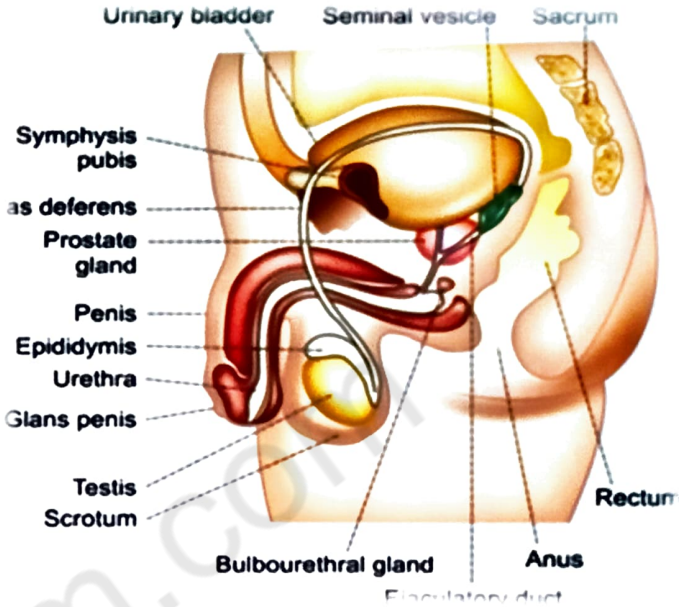
The organs of male reproductive system can be classified into:

❖ **External genital organs**

- ✓ Penis containing the urethra

❖ **Internal genital organs**

- ✓ Testes
- ✓ Vas deferens
- ✓ Seminal vesicles
- ✓ Prostate gland
- ✓ Scrotum
- ✓ Ejaculatory ducts

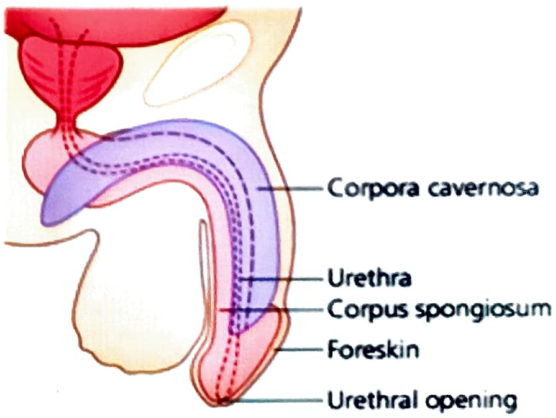


❖ External genital organs

✓ Penis:

- It is the copulatory organ and also contains the terminal part of urethra
- The parts of penis are:

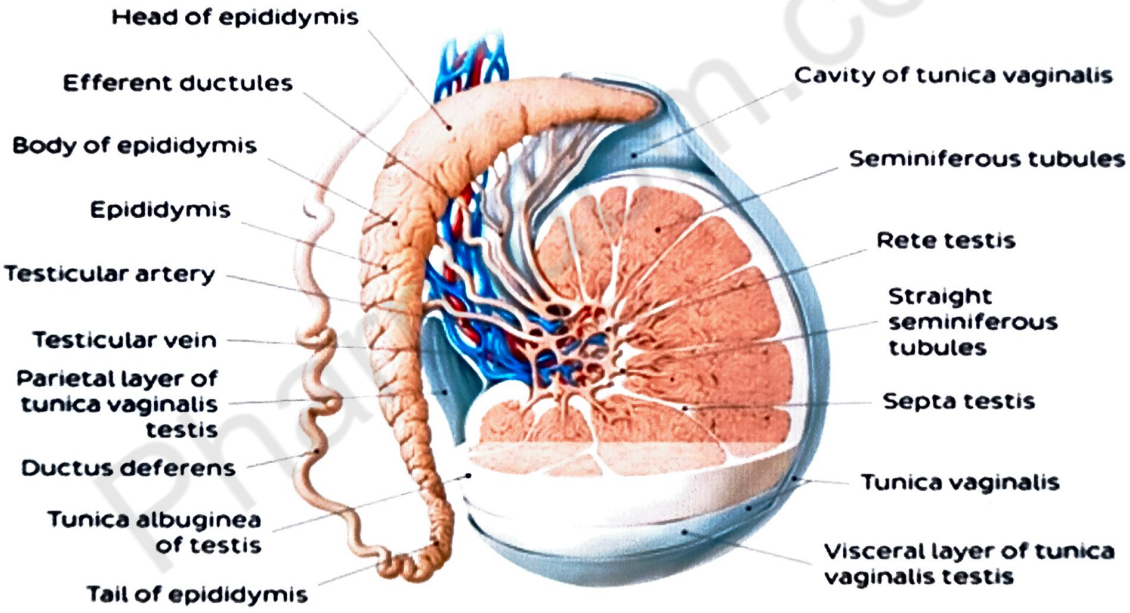
1. **Corpora cavernosa** : Which are two pillars of erectile tissue lying side by side under the skin of penis.
2. **Corpora spongiosum** : Contains the urethra and it lies below corpora cavernosa.
3. **Glans penis** : Which is the enlarged tip of penis.
4. **Propuce** : Which is a fold of skin covering the glans penis.



❖ Internal genital organs

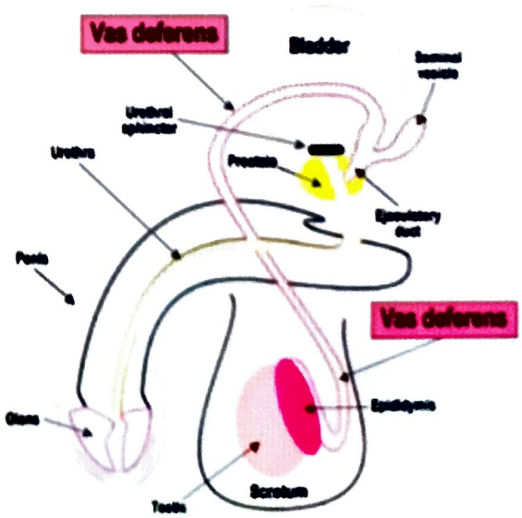
✓ Testes

- Testes are the **male reproductive** organs which produce **spermatozoa**.
- The testes are **two oval** shaped bodies lying one on each side in the **scrotum**.
- Each testis is **enclosed** in a sac called **tunica vaginalis** which is derived from peritoneum
- Each testis contains a number of **tubules** called **seminiferous tubules**
- **Spermatozoa** are formed in the walls of these tubules



✓ Vas deferens (seminal duct)

- They are **two** in number one for each testis And begins from the **epididymis** at the upper end of testis
- It travels **upwards** within the **spermatic cord** and enters the **abdominal cavity** through the inguinal canal situated at the back of the bladder.

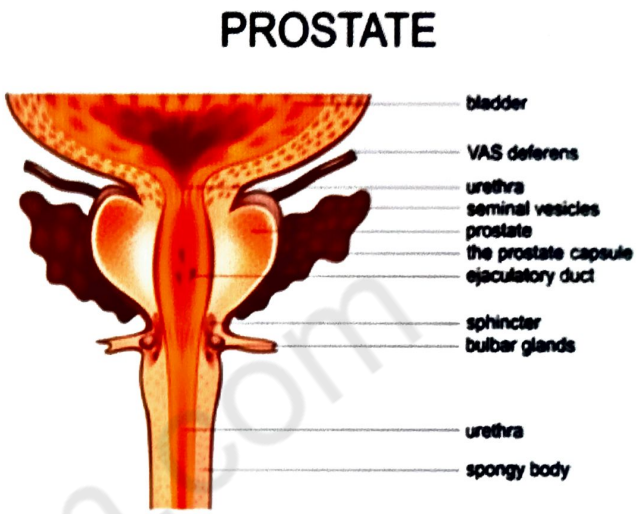


✓ Seminal vesicles

- They are **two** in number, each one lies at the **side** of the terminal part of **vas deferens** and forms the ejaculatory duct
- The function of seminal vesicles is to **produce** a thick secretion which is added to the **spermatozoa** during ejaculation

✓ Prostate gland

- It lies **below** the bladder and it **surrounds** the first part of urethra.
- It is **pyramidal** in shape. Its **base** is directed above and the **apex** is directed downward



✓ Scrotum

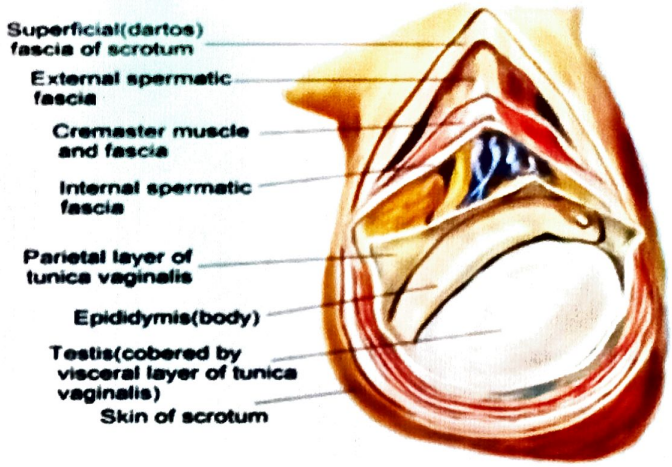
- The scrotum is an **outpouching** of the lower part of the anterior abdominal wall.
- It contains the **testes**, the **epididymides**, and the lower ends of the **spermatic cords**.
- It is divided on its surface into two compartments by a **raphe**, which is continued forward to the under surface of the penis, and backward, along the middle line of the perineum to the anus.
- Each compartment contains one of the **two testes**, and one of the **epididymides**.
- Normal sperm production requires a temperature of about **2-3°C** below body temperature.

➢ The wall of the scrotum has the following layers

1. Skin

- The skin of the scrotum is thin, wrinkled, and pigmented and forms a single pouch.

2. Superficial fascia



- This is continuous with the **fatty** and **membranous layers** of the anterior abdominal wall.
- The fat is replaced by smooth muscle called the **dartos muscle.**

3. Spermatic fasciae

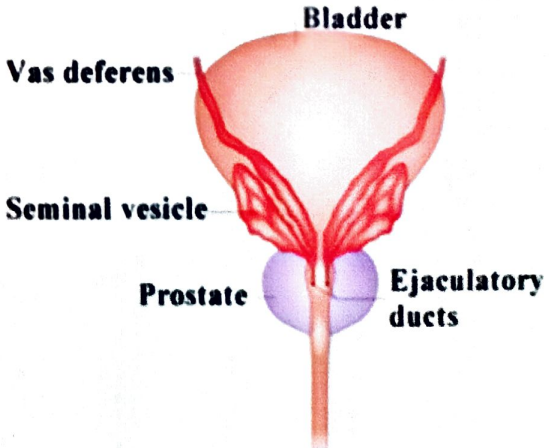
- It has **three layers** which lie beneath the superficial fascia and are derived from the three layers of the anterior abdominal wall on each side.
    a. The **external spermatic fascia** is derived from the aponeurosis of the external oblique muscle.
    b. The **cremasteric fascia** is derived from the internal oblique muscle.
    c. The internal spermatic fascia is derived from the **fascia transversalis.**

4. Tunica vaginalis

- This lies within the spermatic fasciae and covers the anterior, medial, and lateral surfaces of each testis.

✓ Ejaculatory ducts

- The two ejaculatory ducts are each less than 1 in. (2.5 cm) long and are formed by the **union of the vas deferens** and the **duct of the seminal vesicle.**
- The ejaculatory ducts pierce the posterior surface of the **prostate** and open into the prostatic part of the urethra, close to the margins of the prostatic utricle; their function is to **drain** the **seminal fluid** into the prostatic urethra.



FUNCTIONS OF MALE REPRODUCTIVE SYSTEM

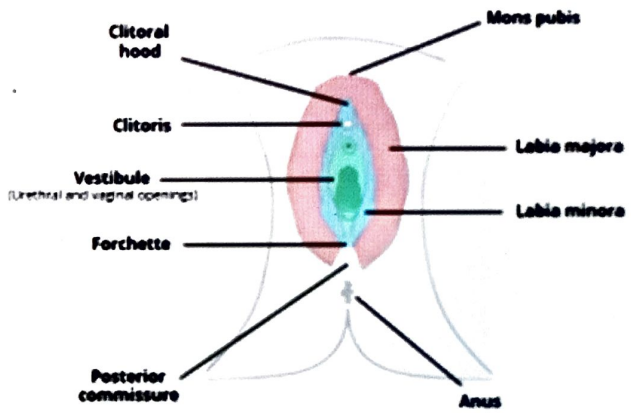
- ✓ **Testis** produces **sperm and testosterone**
- ✓ **Epididymis** matures and stores sperm
- ✓ **Sperm duct** carries sperm from the epididymis to the urethra
- ✓ **Seminal vesicles, cowper's gland and prostate gland** produces seminal fluid which feeds the sperm and allows them to swim.
- ✓ Sperm and seminal fluid are collectively called semen
- ✓ **Urethra** allows the passage of either urine or sperm
- ✓ **Penis** places sperm inside the body of a female
- ✓ **Scrotum** keeps testes at a lower temperature **35°C** this is the optimum temperature for meiosis to occur.

ANATOMY OF FEMALE REPRODUCTIVE SYSTEM

Female reproductive system comprises of:

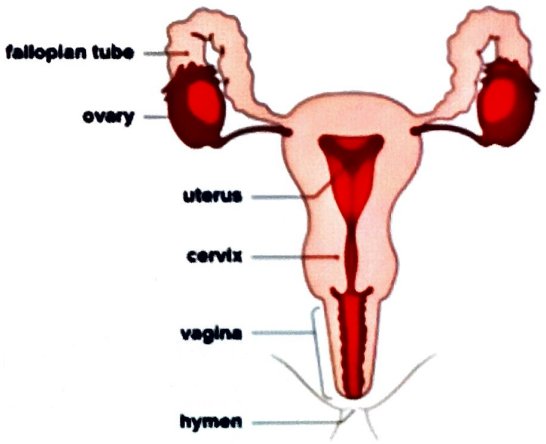
❑ EXTERNAL GENITAL (Vulva)

- ❖ Labia majora
- ❖ Labia minora
- ❖ Clitoris
- ❖ Perineum
- ❖ Vestibular glands



❑ INTERNAL GENITAL

- ❖ Fallopian tubes
- ❖ Uterus
- ❖ Cervix
- ❖ Vagina
- ❖ ovaries



❑ SECONDARY SEXUAL ORGANS

- ❖ Breast or mammary glands

❑ EXTERNAL GENITAL (Vulva)

- The external genital are collectively called the **vulva** that consist of the following structure

1. Labia majora

- These are the **two large folds** forming the boundary of the vulva.
- They are composed of **skin**, **fibrous tissue** and **fat** and contain large numbers of **sebaceous** and **eccrine sweat glands**.
- Labia majora or "greater lips" are the part around the **vagina** containing two glands **(Bartholin's glands)** which helps **lubrication** during intercourse.

2. Labia minora

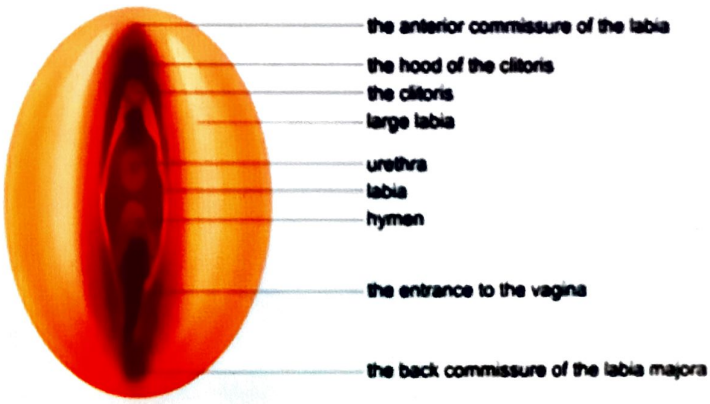
- These are **two smaller folds** of skin between the labia majora, containing numerous **sebaceous** and **eccrine sweat glands**.
- Posteriorly the labia minora are fused together to from the fourchette.
- The area between the labia minora is called the **vestibule**.
- Vestibule is **homologous** to membranous **urethra** of male.

3. Clitoris

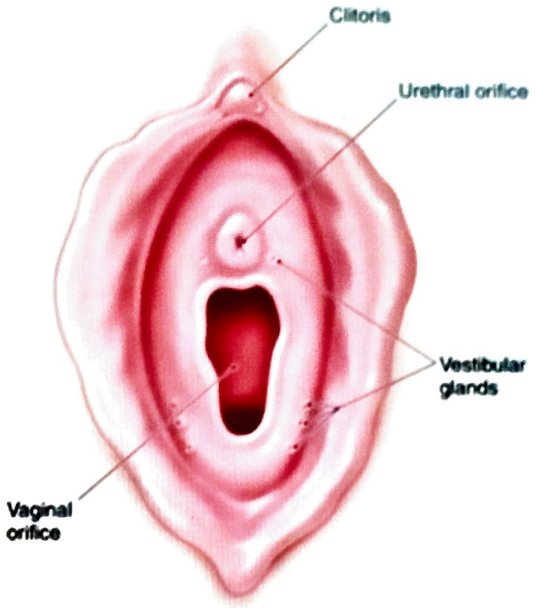
- The clitoris corresponds to the penis in the male and contains **sensory nerve endings** and **erectile tissue**.
- It is a small **pea-shaped** structure. It plays an important part in sexual excitement in females.

4. Perineum

- It is the area which extends from the **fourchette** to the **anus**.



5. Vestibular glands



✓ **These are of two types**

1. The **lesser** vestibular gland **numerous** minute glands that are present on either side of the **urethral orifice** (opening) these glands are homologous to the male prostate and secrete mucus.
2. The greater vestibular glands are paired glands, situated one on each side of the vaginal orifice (opening)

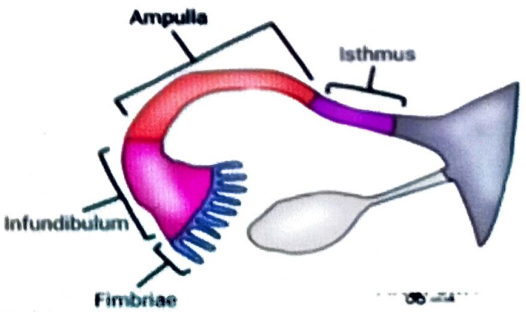
❑ INTERNAL GENITAL

❖ Two uterine tubes (fallopian tube)

- The uterine tubes (also called fallopian tubes or oviducts) serve as the conduit of the **oocyte** from the ovary to the uterus.
- Each of the two uterine tubes is close to, but not directly connected to, the ovary and **divided** into sections.

✓ Functions of fallopian tube

- Fertilization of the ovum generally takes place in the **upper** portion of the fallopian tube(ampulla).
- Provide a suitable environment for **fertilization**, and **transport** the egg from the ovary.
- The main function of the fallopian tube is to **collect the mature ovum** from ovaries and provide **passage to the fertilized** ovum to reach the uterus for **implantation**.

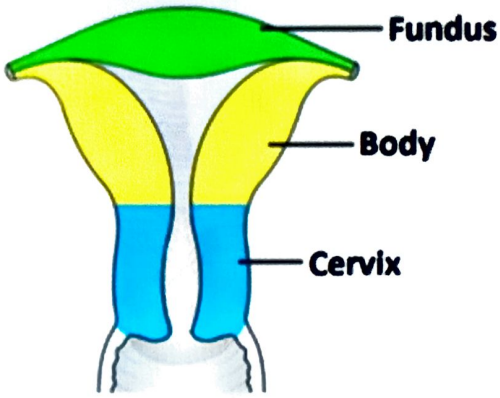


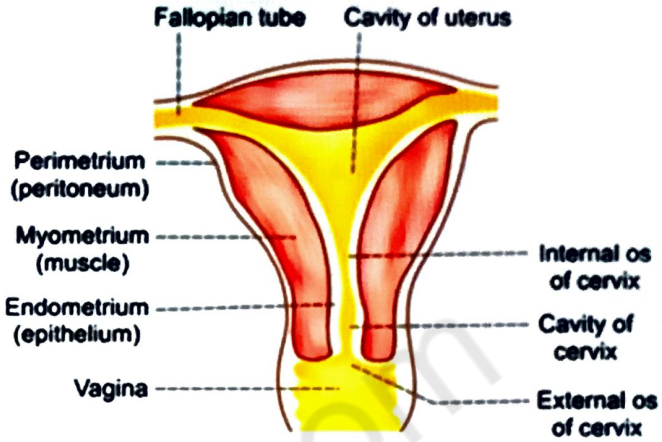
✓ **Three named parts of the Fallopian tube**

1. Isthmus
2. Ampulla
3. Infundibulum

❖ Uterus(womb)

- It lies in the **pelvic cavity**, in between the rectum and urinary bladder
- Uterus is a **hollow** muscular organ with a **thick** wall.
- It has a **central cavity**, which opens into vagina through cervix





➢ **Divisions of uterus** : Three portions

1. Fundus (above the entrance points of fallopian tubes)
2. Body (between fundus and isthmus)
3. Cervix (below isthmus).

➢ **Structure of uterus** : Three layers

1. Serous or outer layer
2. Myometrium or middle muscular layer
3. Endometrium or inner mucus layer

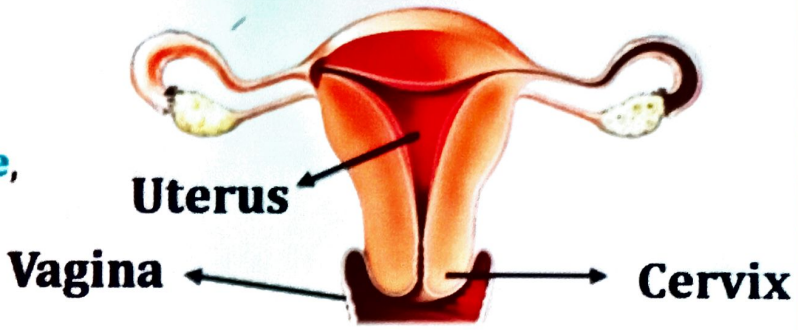
❖ Caervix

- It is the **lower constricted** part of uterus
- It is divided into **two** portions:

Upper Supra vaginal portion	Lower Vaginal portion
Communicates with body of uterus through internal os (orifice) of cervix.	Communicates with vagina through external os (orifice) of cervix
Mucus membrane of this portion has glandular follicles, which secrete mucus	Mucus membrane of this portion is formed by stratified epithelial cells

❖ Vagina

- It is a **short tubular** organ
- It is lined by **mucus membrane**, which is formed by stratified epithelial cells

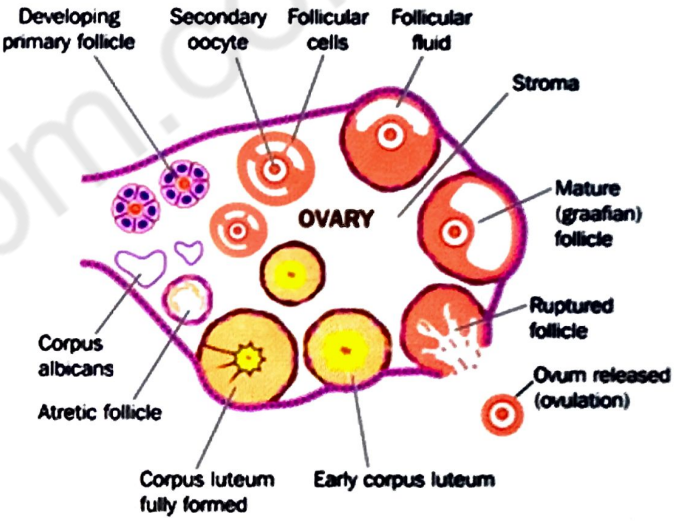


❖ Ovaries

- They are **Two** in number and lie **both** of the uterus
- A fold of **peritoneum** attaches the **ovaries** to **posterior** aspect of broad ligament
- The ovaries lie **below** the **fallopian tube** of each side

➢ **The ovary contains:**

1. A central soft issue called **stroma**
2. An outer surface called **germinal epithelium**



- The germinal epithelium contains the **graffian follicles**. The graffian follicles contain the **ova**.
- Beneath the epithelium is the tunica albuginea-a layer of connective tissue
- The ovarian stroma consists of a dense outer layer called the cortex and a less dense inner portion called the medulla.

❑ SECONDARY SEXUAL ORGANS

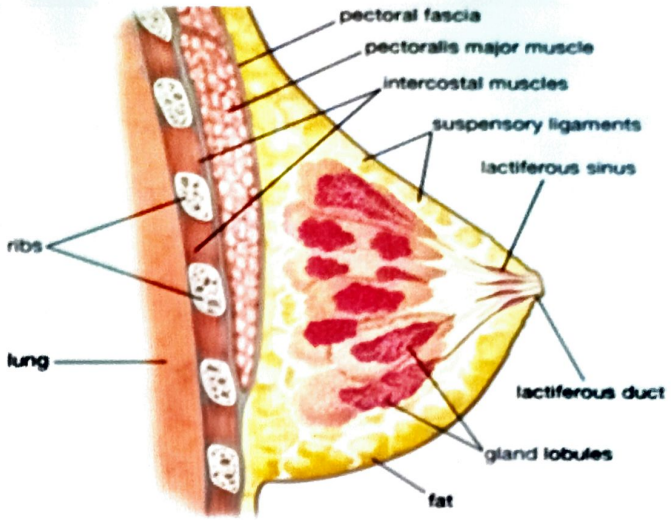
❖ Breast or mammary glands

- The mammary gland is a gland located in the breasts of females that is responsible for **lactation**, or the **production** of milk.
- Both males and females have glandular tissue within the breasts; however, in females the glandular tissue begins to develop after puberty in response to estrogen release.

Functions of mammary glands

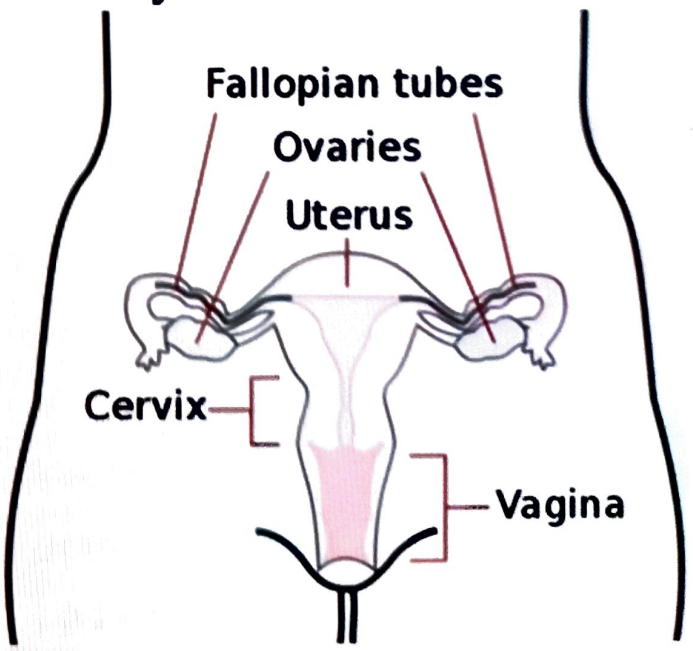
i. Synthesis, Secretion and ejection of milk is called as lactation.

ii. Milk production is stimulated by prolactin hormone secreted by anterior pituitary.



FUNCTIONS OF FEMALE REPRODUCTIVE SYSTEM

- ✓ **Germinal epithelial cells** of the **ovary produce** ova (oogenesis).
- ✓ Fertilization takes place in the **Fallopian tube** (oviduct).
- ✓ After puberty the **uterus** goes through the **menstrual cycle**
- ✓ **Implantation and prenatal growth** take place in the **uterus**
- ✓ The **vagina** receives the **seminal fluid** during copulation.
- ✓ **Parturition** process of birth of child is also **important function** of the female reproductive system
- ✓ **Mammary glands** of the female **secrete milk** after parturition.

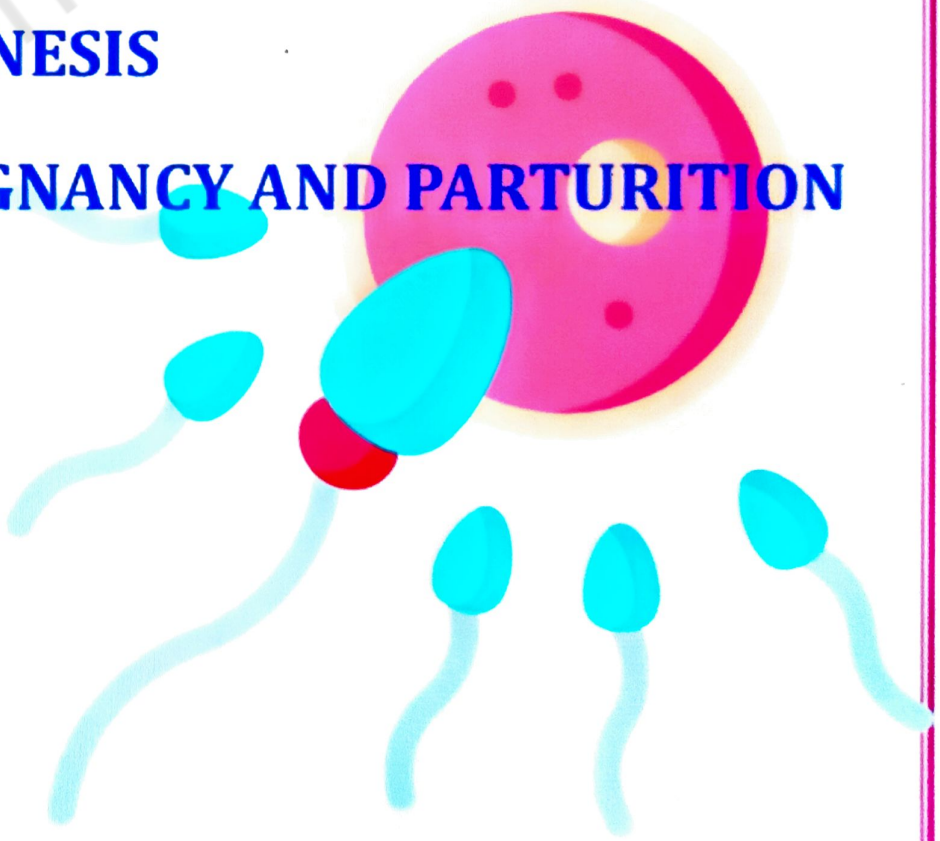


REPRODUCTIVE SYSTEM

PART - 2

Points to be covered in this topic

5. SEX HORMONES
6. PHYSIOLOGY OF MENSTRUATION
7. FERTILIZATION
8. SPERMATOGENESIS
9. OOGENESIS
10. PREGNANCY AND PARTURITION



SEX HORMONES

❑ Male sex hormones

- **Interstitial cells** of the Testes secrete male sex hormones, which are collectively called the androgens.
- **Androgens** secreted by testes are:
  - ❖ **Testosterone**
  - ❖ **Dihydrotestosterone**
  - ❖ **Androstenedione**

Among these three androgens, testosterone is secreted in large quantities. However, dihydrotestosterone is more active

✓ Functions of male sex hormones

1. Stimulation of **spermatogenesis**.
2. **Growth** of penis, scrotum and prostate.
3. **Development** of secondary sex characters.

❑ Female sex hormones

❖ INTRODUCTION

- Female sex hormones, or sex steroids, play vital roles in **sexual development, reproduction,** and **general health.**
- Sex hormone levels change over time, but some of the most significant changes happen during **puberty, pregnancy,** and **menopause.**
- Ovary secretes the female sex hormones **Estrogen** and **Progesterone**

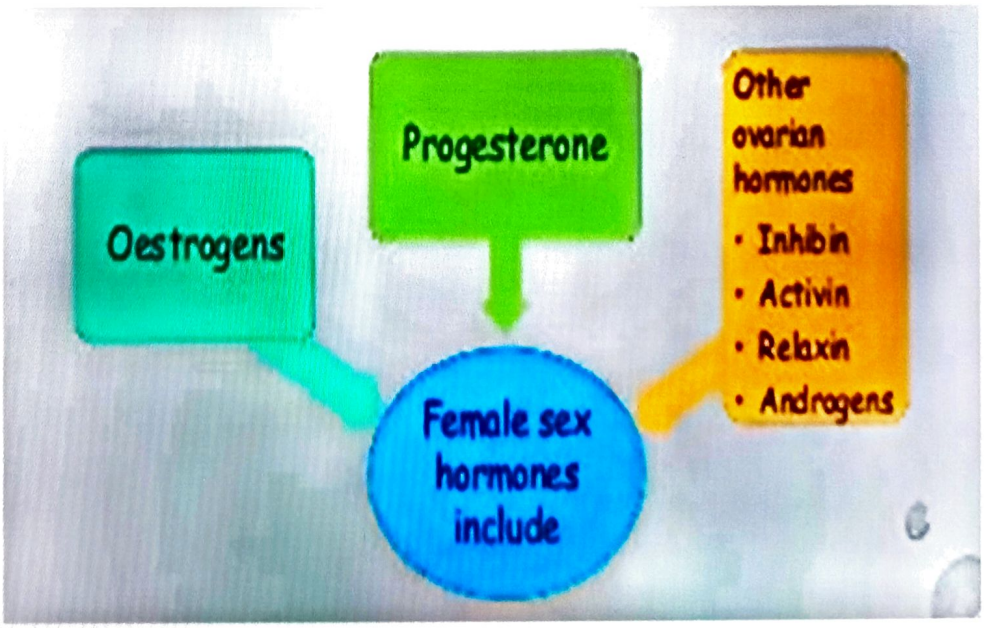
❖ Types of female sex hormone

1. Estrogen

- Estrogen is the most well-known **sex hormone.**
- The majority of estrogen production occurs in the **ovaries**, the **adrenal glands** and fat cells produce small amounts of estrogen.
- Estrogen plays a crucial role in **reproductive** and **sexual development**, which begins when a person reaches puberty.

2. Progesterone

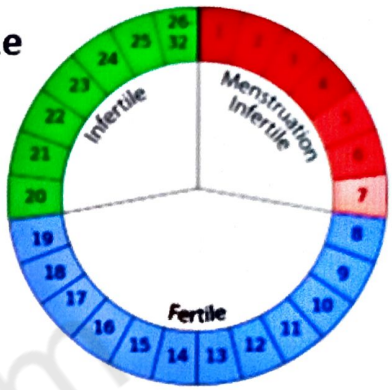
- The ovaries, adrenal glands, and placenta produce the hormone **progesterone**.
- Progesterone levels increase during ovulation and spike during pregnancy.
- Progesterone helps stabilize **menstrual cycles** and prepares the body for **pregnancy**.



PHYSIOLOGY OF MENSTRUATION

❑ INTRODUCTION

- Menstrual cycle is defined as **cyclic events** that take place in a rhythmic fashion during the reproductive period of a woman's life.
- It starts at the age of **12** to **15** years, which marks the **onset** of **puberty**.
- The commencement of menstrual cycle is called **menarche**.
- Menstrual cycle ceases at the age of **45** to **50** years.
- Permanent cessation of menstrual cycle in old age is called **Menopause**



❖ The menstrual cycle consists of following four phases

1. **Menstrual Phase (Bleeding phase or menses)**
2. **Follicular phase (Proliferative phase)**
3. **Ovulatory phase**
4. **Luteal phase (Secretary phase)**

1. Menstrual phase (bleeding phase)

- In a 28 day menstrual cycle, the menses takes place on cycle **days 3-5.**
- The production of LH from the anterior lobe of the pituitary gland is considerably reduced.
- The withdrawal of this hormone causes **degeneration of the corpus luteum** and, therefore, progesterone production reduced.
- Production of **estrogen** is also reduced in this phase.
- The endometrium of the uterus **breaks** down and **menstruation begins.**
- The cells of **endometrium secretions, blood** and the **unfertilized ovum** constitute the menstrual flow.

2. Follicular phase (Proliferative phase)

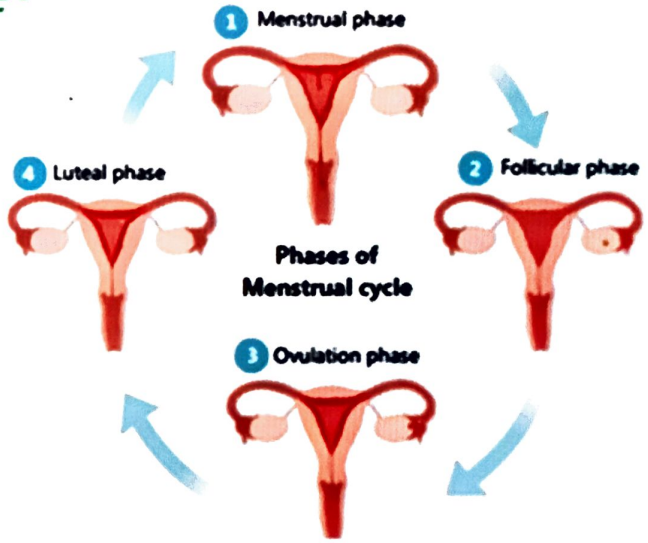
- This phase usually includes cycle days **6-13 or 14 in a 28 day cycle.**
- The follicle stimulating hormone (FSH) secreted by the anterior lobe of the pituitary gland stimulate the **ovarian follicle** to secrete estrogen.
- Estrogen stimulate the **proliferation** of the **endometrium** of the uterine wall.
- The endometrium becomes thicker by rapid cell multiplication and this is accompanied by an **increase** of **uterine glands** and **blood vessels.**

3. Ovulatory phase

- Both LH and FSH attain a peak level in the middle of cycle about **14th day).**
- Rapid secretion of LH induces **rupturing** of Graafian follicle and thereby the release of ovum in human beings **secondary oocyte** is released.
- This is called ovulation in fact LH causes ovulation.

4. Luteal phase (Secretory phase)

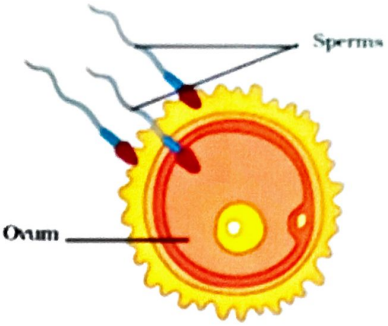
- It is the phase lasting for the next **14 days**
- During this phase, the **endometrium** becomes thick and vascular to receive the **fertilized ovum**



- Now, the corpus luteum secretes **progesterone**
- If conception occurs by **implantation** of the fertilized ovum, the secretion of progesterone **continues** throughout pregnancy. Otherwise the corpus luteum begins to degenerate and does not produce progesterone.
- Later, the capillaries of **endometrium burst** and **menstruation** occurs

FERTILIZATION

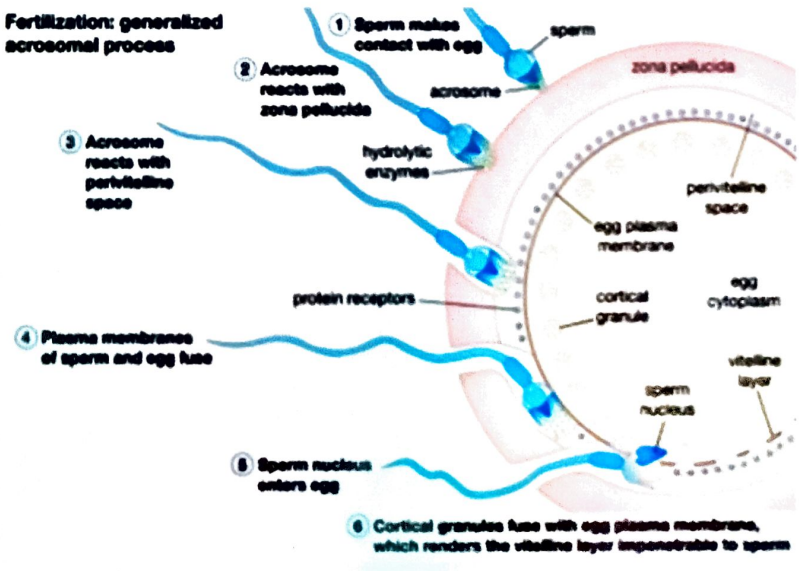
❑ INTRODUCTION



Fertilization refers to **fusion** (union) of male and female gametes (sperm and ovum) to form a **new offspring.**

❖ Steps in fertilization

1. After sexual intercourse, **semen** is ejaculated in the **vagina**, the sperms travel through the **vagina** and **uterus** to reach the **fallopian tube.**
2. Movement of the sperm through uterus is facilitated by the **antiperistalsis contractions** of **uterine muscles.**
3. **Uterine contractions** are induced by **oxytocin**, which is secreted from posterior pituitary by neuroendocrine reflex during sexual intercourse
4. Among 200 to 300 millions of sperms entering female genital tract, only a few thousand sperms reach the spot near the ovum. Among these few thousand sperms, **only one** succeeds in fertilizing the ovum.
5. During fertilization, the sperm enters the ovum by **penetrating** the multiple layers of granulosa cells known as **corona radiata** present around the **ovum**.
6. It is facilitated by **hyaluronidase** and **proteolytic enzymes** present in acrosome of sperm.
7. Proteolytic enzymes from acrosome of the successful sperm diffuse through the structures of **zona pellucida** and **inactivate** the other sperms entering the ovum.



8. Immediately after fertilization, ovum, which is in **secondary oocyte stage,** divides into a **matured ovum** and a **second polar body**. Second polar body is expelled.
9. Nucleus of matured ovum becomes **female pronucleus** with 23 chromosomes, which include 22 autosomes and one sex chromosome called X chromosome.
10. Simultaneously, head of sperm swells and becomes **male pronucleus.**
11. Then 23 chromosomes of the sperm and 23 chromosomes of ovum arrange thmselves to reform the **23 pairs** of chromosomes in the **fertilized ovum.**

SPERMATOGENESIS

- It is the process by which the male gametes called **spermatozoa (sperms)** are formed from the primitive **spermatogenic cells** in the testis .
- It takes 74 days for the formation of sperm from a primitive germ cell

❖ **Stages of Spermatogenesis** four stages

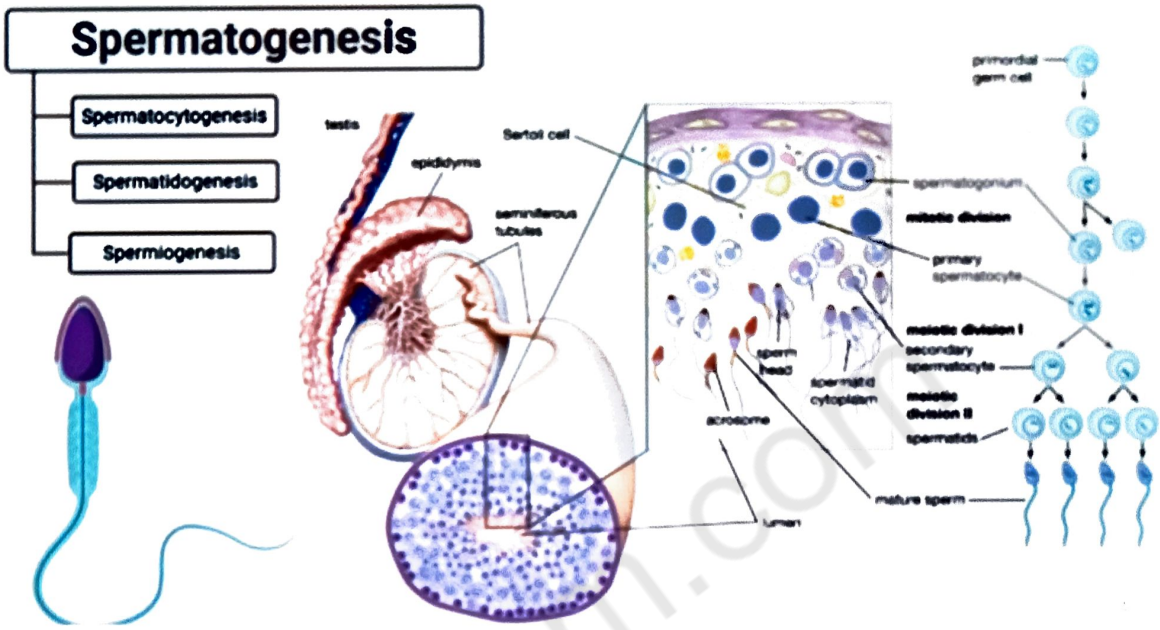
1. **Stage of proliferation**
2. **Stage of growth**
3. **Stage of maturation**
4. **Stage of transformation**

1. Stage of Proliferation

- In this phase, **spermatogonia** divide by **mitosis**, without any change in chromosomal number.
- In man, there are usually **seven generations** of spermatogonia The **last generation** enters the stage of growth as **primary spermatocyte.**
- During this stage, the **spermatogonia** migrate along with **Sertoli cells** towards the lumen of **seminiferous tubule.**

2. Stage of Growth

- In this stage, the **primary spermatocyte** grows into a **large cell**
- There is **no** other change in **spermatocyte** during this stage.



3. Stage of Maturation

- After reaching the full size, each primary **spermatocyte** quickly undergoes **meiotic** or maturation division, which occurs in **two** phases:

➢ **First phase** : each **primary** spermatocyte divides into two **secondary** spermatocytes

➢ **Second phase** : each **secondary** spermatocyte undergoes **second meiotic** division, resulting in two smaller cells called **spermatids.**

4. Stage of Transformation

- There is no further division
- Spermatids are **transformed** into **matured spermatozoa** (sperms), by means of **spermiogenesis** and released by **spermination**

OOGENESIS

❑ INTRODUCTION

- Oogenesis is the process of **formation** of **female gametes**. This process begins inside the fetus before birth.
- The steps in oogenesis up to the production of **primary oocytes** occur before birth.
- Primary oocytes do not divide further. They either become **secondary oocytes** or degenerate.
- Oogenesis occurs in the **outermost** layers of the ovaries.
- Oogenesis starts with a **germ cell** called **oogonium** and undergoes mitosis to increase in number.

❖Key Terms

✓ **Spermatocyte**: a **male gametocyte**, from which a spermatozoon develops

✓ **Oocyte**: a cell that develops into an egg or ovum; **a female gametocyte**

✓ **Polar body**: one of the small cells that are **by-products** of the meiosis that forms an egg

✓ **Mitosis**: the **division** of a **cell nucleus** in which the genome is copied and separated into two identical halves. It is normally followed by cell division

✓ **Meiosis**: cell division of a **diploid cell** into four haploid cells, which develop to produce gametes

❖ The **process of oogenesis** takes place in the following three stages:

1. **Pre-natal**
2. **Antral**
3. **Pre-ovulatory**

1. Pre-natal Stage

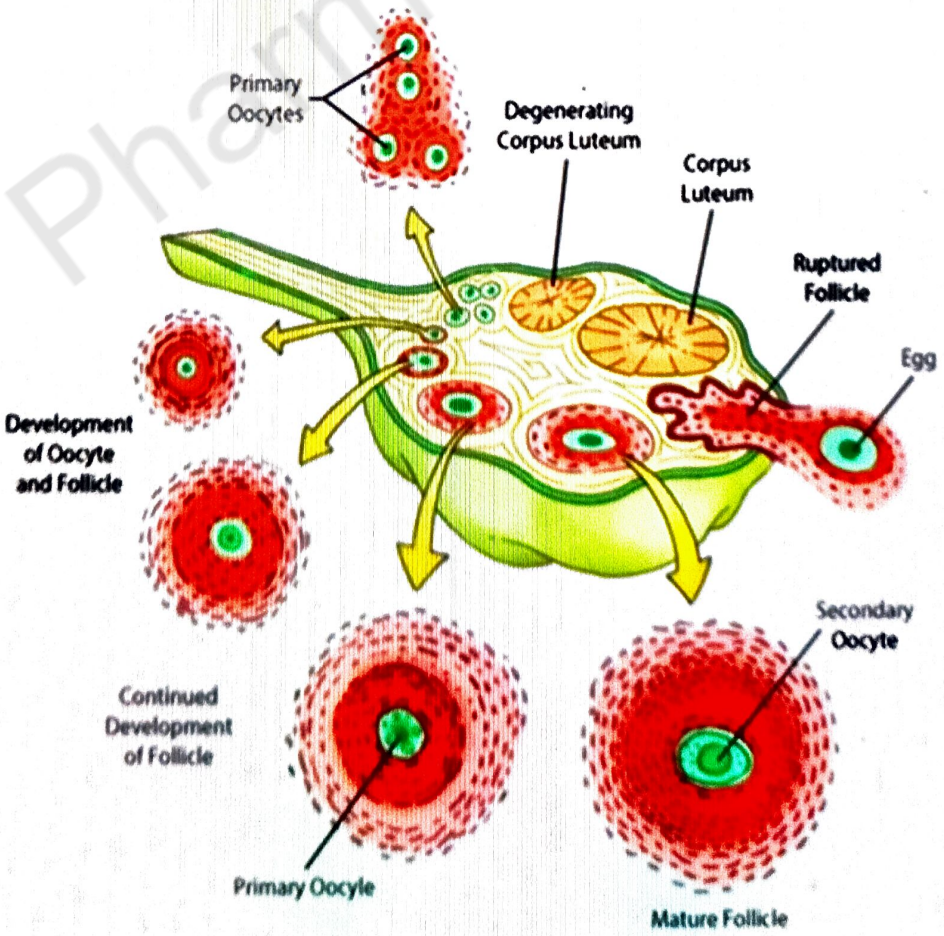
- The primary oocyte grows while being arrested in **meiosis-I.**
- The follicular cells proliferate and form a stratified cuboidal epithelium. Such cells are known as **granulosa cells.**
- These cells secrete glycoproteins to form **zona pellucida** around the primary oocyte.

2. Antral Stage

- The fluid-filled area, present between granulosa cells, combines to form a central fluid-filled space called the antrum. These are known as secondary follicles.
- In every month cycle, these secondary follicles develop under the influence of follicle-stimulating hormone and luteinizing hormone.

3. Pre-Ovulatory Stage

- This stage is induced by LH surge, and meiosis-I completes here.
- Two haploid cells of unequal sizes are formed within the follicle. One of the daughter cells that receive less cytoplasm forms a polar body.
- This cell does not participate in ovum formation. The other daughter cell is known as the secondary oocyte.
- The two daughter cells undergo meiosis-II.
- The polar body replicates to form two polar bodies, while the secondary oocyte arrests in the metaphase stage of meiosis-II.



PREGNANCY AND PARTURITION

- **Pregnancy** occurs when the **sperm** fertilizes the **egg**, and the **zygote** is formed.
- The **zygote** gets **implanted** in the wall of the **uterus** which needs nourishment and care
- The **zygote** develops into an **embryo** which further grows into a **baby**.
- After **implantation** takes place, the embryo starts showing the **triploblastic nature** of human beings and develops the **new** organs
- All these steps lead to the **development** of the baby and it takes nine months. The development is very **gradual** and **steady.**
- By the **end** of 9 months, the **fetus** is completely developed

❑ STEPS IN PREGNANCY

❖ Fertilization of the ovum

- It refers to **fusion** of male and female **gamates** (sperm and ovum) to form a **new offspring**
- Fertilization takes place in the **fallopian tubes,** Fertilization happens when a sperm cell successfully **meets** an **egg cell** in the fallopian tube. Once fertilization takes place, this newly fertilized cell is called a **Zygote**

❖ Implantation

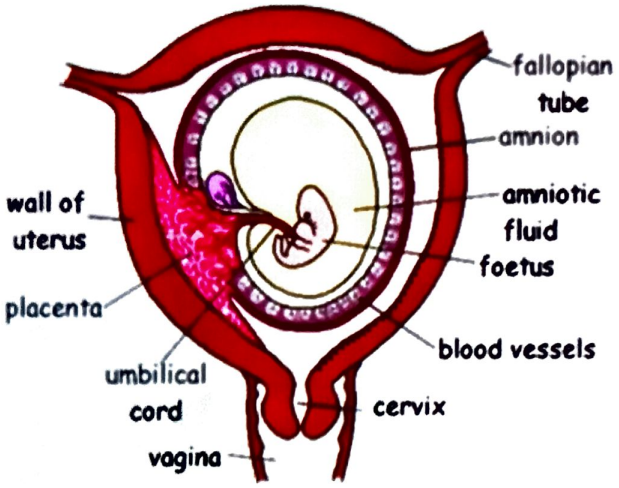
- Implantation is the process by which the fertilized ovum called **zygote implants** (gets attached) in the **endometrial lining** of uterus.
- Zygote takes 3 to 5 days to reach the **uterine cavity** from fallopian tube.

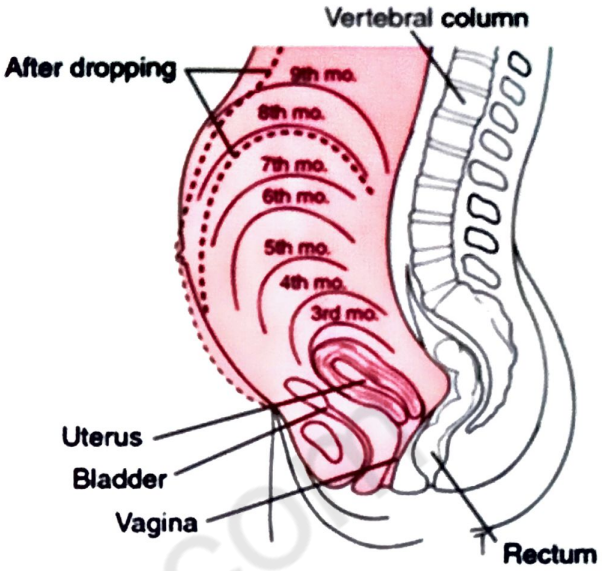
❖ Development of Placenta and Embryo

- After implantation, **placenta** develops between **morula** and **endometrium**
- When implantation occurs, there is further **increase** in the **thickness** of endometrium because of continuous **secretion** of progesterone from corpus luteum.
- An **embryo** is the early stage of development of a multicellular

❖ Gestation period

- It refers to the **pregnancy period** and measured in weeks
- A normal pregnancy can range from **38 to 42 weeks**.
- Infants born before 37 weeks are considered **premature**





❑ PARTURITION

- Parturition is the **expulsion** or **delivery** of the fetus from the mother's body
- It occurs at the **end** of pregnancy
- The process by which the delivery of fetus occurs is called **labor**. It involves **various activities** such as contraction of uterus, dilatation of cervix and opening of vaginal canal.

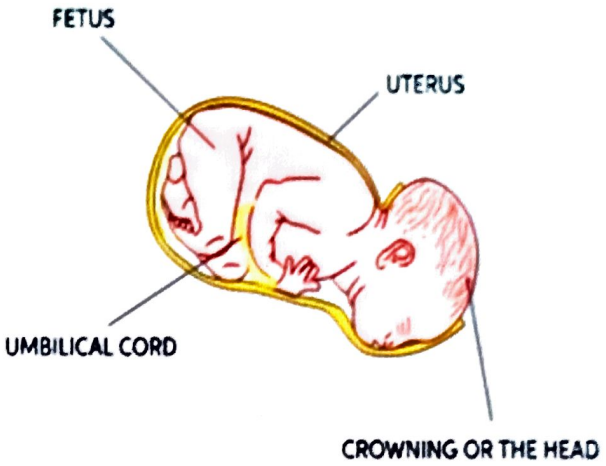
❖ STAGES OF PARTURITION : Three stages

✓ First Stage

- The strong uterine contractions called **labor contractions** begins.
- Labor contractions arise from **fundus** of uterus and move downwards which results in dilatation of **cervix** and opening of **vaginal canal.**
- This stage extends for a **variable** period of time.

✓ Second stage

- In this stage, the fetus is **delivered** out from uterus through cervix and vaginal canal.
- This stage lasts for about **1 hour**



✓ Third stage

- During this stage, the **placenta** is **detached** from the decidua and is **expelled** out from uterus
- It occurs within **10 to 15 minutes** after the delivery of the child

INTRODUCTION TO GENETICS

Points to be covered in this topic

1. CHROMOSOMES
2. GENES AND DNA
3. PROTEIN SYNTHESIS
4. GENETIC PATTERN OF INHERITANCE

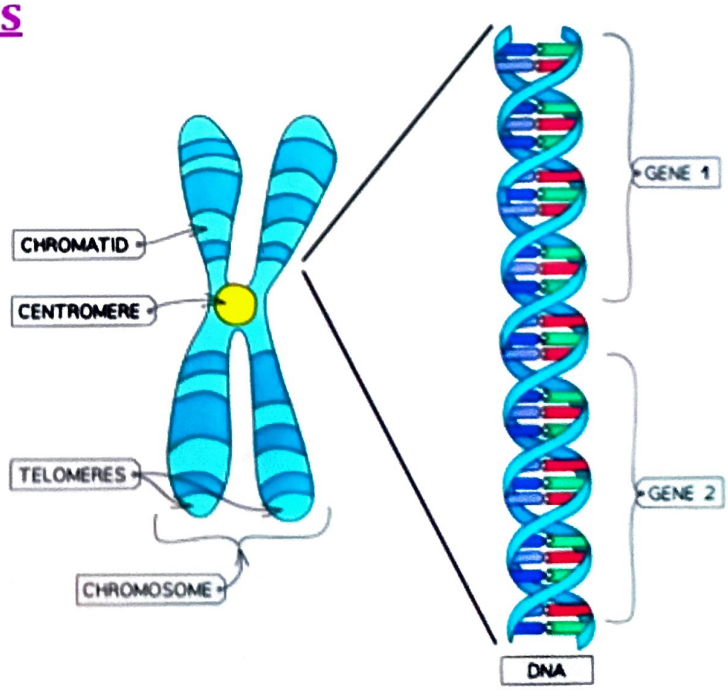
CHROMOSOMES

❑ INTRODUCTION

- The term chromosome comes from the Greek words for **color** (chroma) and **body** (soma) because of the ability to **stained** by some **colorful dyes** used in research
- Chromosomes are **thread-like structures** located inside the **nucleus** of animal and plant cells.

❖ Structure of Chromosomes

- Chromosome is the **rod-shaped nuclear** structure that carries a complete blueprint of all the **hereditary characteristics** of that species.
- A chromosome is formed from a **single** DNA molecule **coiled** around **histone** molecules.

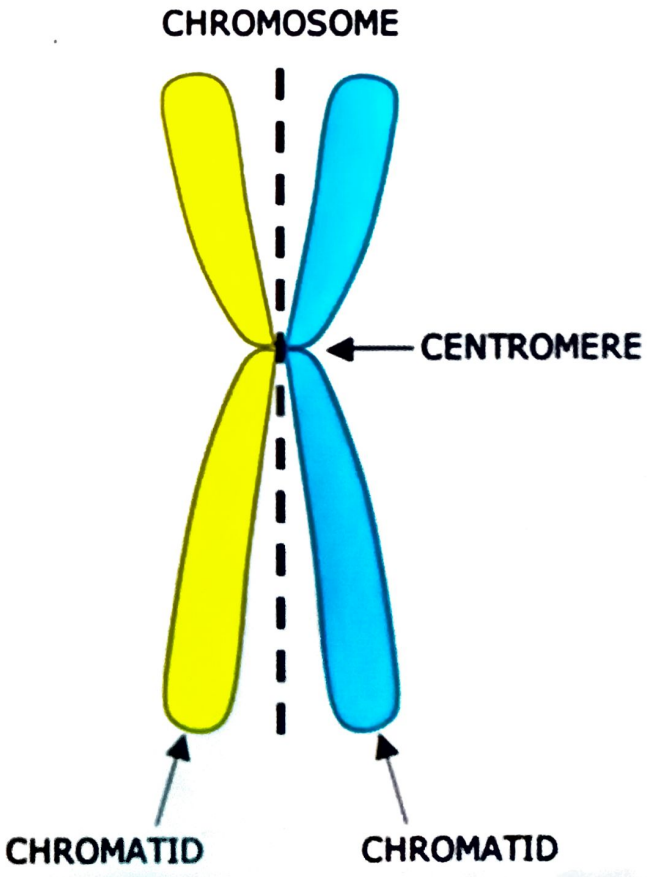


- Each DNA contains many **genes**.
- All the **dividing** cells of the body except reproductive cells contain **23 pairs** of chromosomes.
- Each pair consists of one **chromosome** inherited from **mother** and one from **father**.
- The cells with **23 pairs** of chromosomes are called **diploid cells.**
- The reproductive cells called **gametes** or **sex cells** contain only 23 single chromosomes.
- These cells are called **haploid cells**

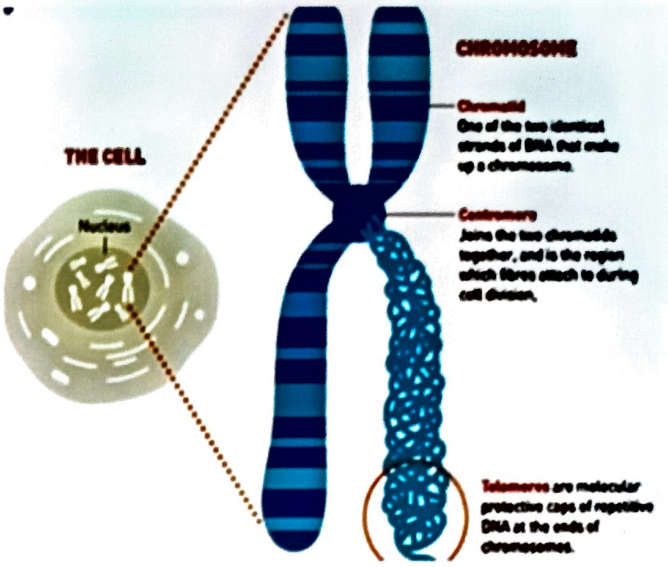
The chromosomes are not visible in the nucleus under microscope. Only during cell division, the chromosomes are visible under microscope. This is because DNA becomes more tightly packed just before cell division, which makes the chromosome visible during cell division

✓ Centromeres

- The **constricted region** of linear chromosomes is known as the centromere.
- It usually is not located exactly in the center of the chromosome and, in some cases, is located almost at the **chromosome's end.**
- The regions on either **side** of the centromere are referred to as the **chromosome's arms.**
- Centromeres help to keep chromosomes properly **aligned** during the complex process of **cell division.**

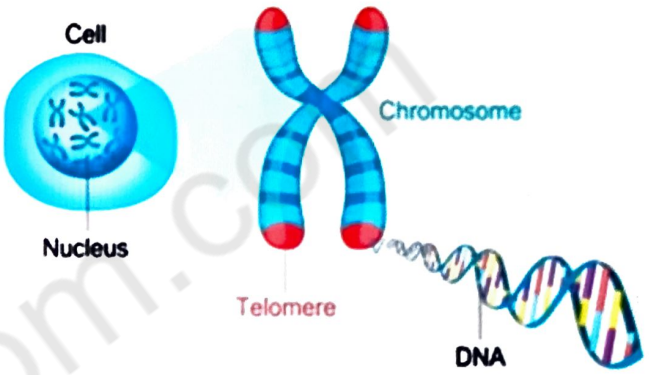


- Centromere serves as an attachment site for the two halves of each replicated chromosome, known as **sister chromatids.**



✓ Telomeres

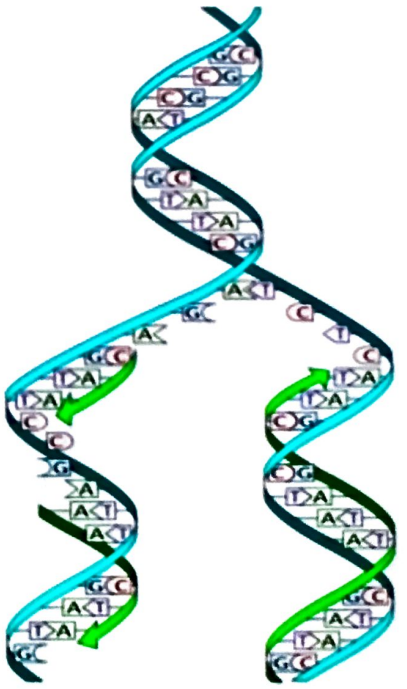
- Telomeres are **repetitive stretches** of DNA located at the ends of **linear chromosomes.**
- They **protect** the **ends** of chromosomes
- In many types of cells, telomeres **lose** a bit of their **DNA** every time a cell **divides**. Eventually, when all of the telomere DNA is gone, the cell cannot **replicate** and **dies**.



GENES AND DNA

❑ DNA- Deoxyribonucleic acid

- DNA is a **nucleic acid** that carries the genetic information to the **offspring** of an organism.
- DNA forms the **chemical** basis of **hereditary** characters.
- It contains the **instruction** for the **synthesis** of proteins in the ribosomes.
- Gene is a part of a DNA molecule.
- DNA is present in the **nucleus**



(chromosome) and **mitochondria** of the cell.

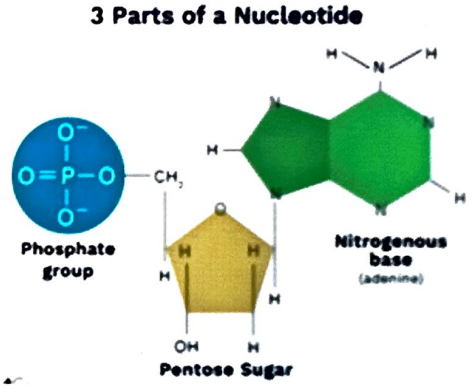
- The DNA present in the nucleus is responsible for the formation of **RNA**.
- RNA regulates the synthesis of **proteins** by **ribosomes**.
- DNA in mitochondria is called **non-chromosomal DNA.**

❖ Structure of DNA

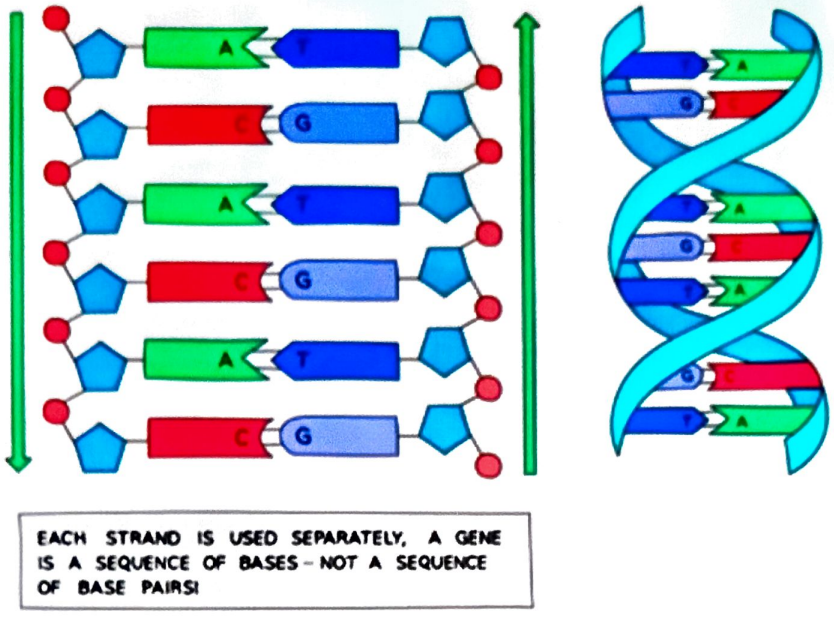
- DNA is a **double stranded** complex nucleic acid. formed by **deoxyribose, phosphoric acid** and **four** types of **bases.**
- Each DNA molecule consists of two **polynucleotide chains,** which are twisted around one another in the form of a double helix.
- The two chains are formed by the **sugar deoxyribose** and **phosphate** which form the **backbone** of DNA molecule.
- Both **chains** of DNA are connected with each other by some **organic bases** and consists of **nucleotide**

✓ Each **nucleotide** is formed by:
1. Deoxyribose – sugar
2. Phosphate
3. One of the nitrogenous bases:

Purines – Adenine (A) – Guanine (G)
Pyrimidines – Thymine (T) – Cytosine (C)



- The **strands** of DNA are arranged in such a way that both are **bound** by specific pairs of **bases**.
- The **adenine** of **one** strand binds specifically with **thymine** of **opposite strand.**
- Similarly, the **cytosine** of one strand binds with **guanine** of the other strand. DNA forms the component of chromosomes, which carries the **hereditary information.**
- The hereditary **information** that is **encoded** in DNA is called **genome.**
- Each DNA molecule is divided into discrete **units** called **genes.**



EACH STRAND IS USED SEPARATELY, A GENE IS A SEQUENCE OF BASES - NOT A SEQUENCE OF BASE PAIRS!

A = Adenine
C = Cytosine
G = Guanine
P = Phosphate
S = Sugar
T = Thymine

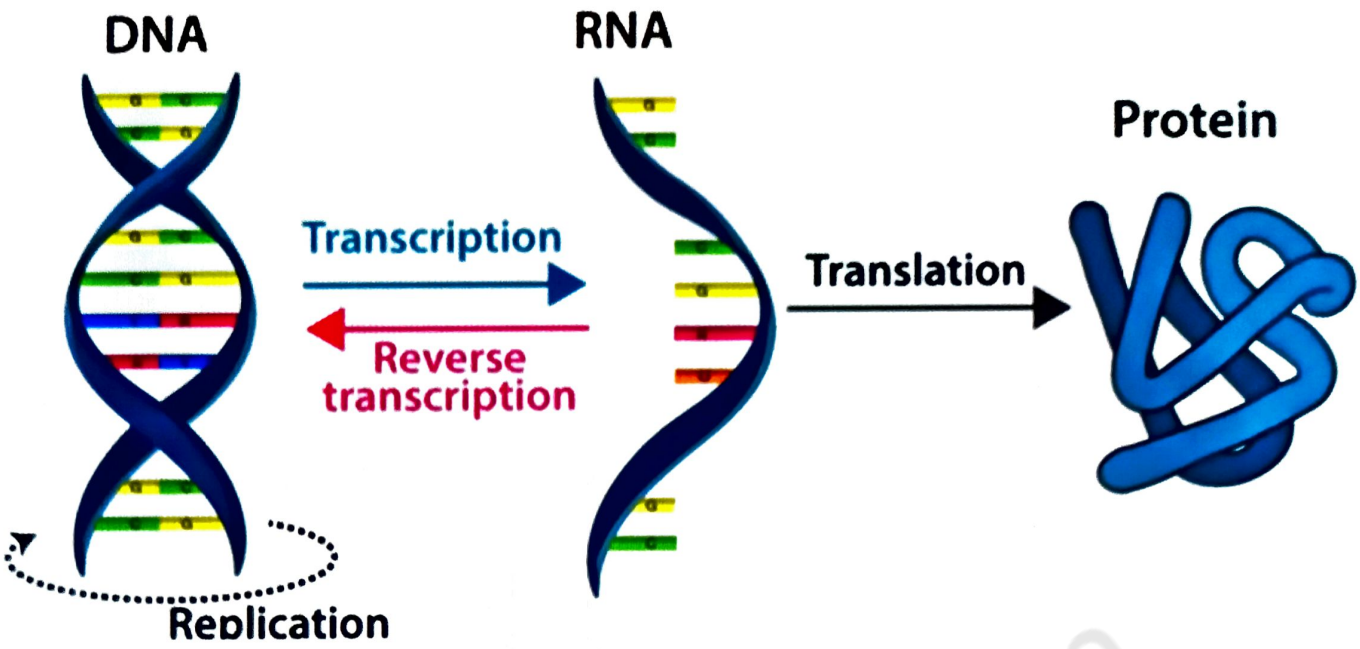
PROTEIN SYNTHESIS

- Protein synthesis is **the process of creating protein molecules.**
- In biological systems, it involves **amino acid synthesis, transcription, translation, and post-translational events.**
- The **proteins** determine the **physical** and **chemical** characteristics of cells
- Some **proteins** help assemble **cellular structures** such as the plasma membrane, the cytoskeleton, and other organelles
- Others serve as **hormones**, antibodies, and contractile elements in **muscular tissue.**
- Still others act as **enzymes**, regulating the rates of the numerous chemical reactions that occur in cells, or transporters, carrying various materials in the blood.

❖ STEPS OF PROTEIN SYNTHESIS

1. Transcription

- Transcription or **RNA synthesis**, is the process of **creating an equivalent RNA copy of a sequence of DNA.**
- Both **RNA and DNA are nucleic acids**, which **use base pairs of nucleotides complementary** language that as can a be **converted back and forth from** DNA to RNA in the presence of the correct enzymes.



- During transcription, a **DNA sequence is read** by **RNA polymerase,** which **produces a complementary, antiparallel RNA strand.**
- As opposed to DNA replication, transcription results in an RNA compliment that includes uracil (U) in all instances where thymine (T) would have occurred in a DNA compliment.

❖ **Transcription is divided into 3 stages:**

1. Initiation

2. Elongation

3. Termination

1. Initiation

- Initiation in bacteria, transcription begins with the binding of RNA polymerase to the promoter in DNA.
- RNA polymerase is a core enzyme consisting of five subunits: 2 α subunits, 1 β subunit, 1β subunit, and 1 ω subunit.
- At the start of initiation, the core enzyme is associated with a sigma factor that aids in finding the appropriate -35 and-10 base pairs downstream of promoter sequences.

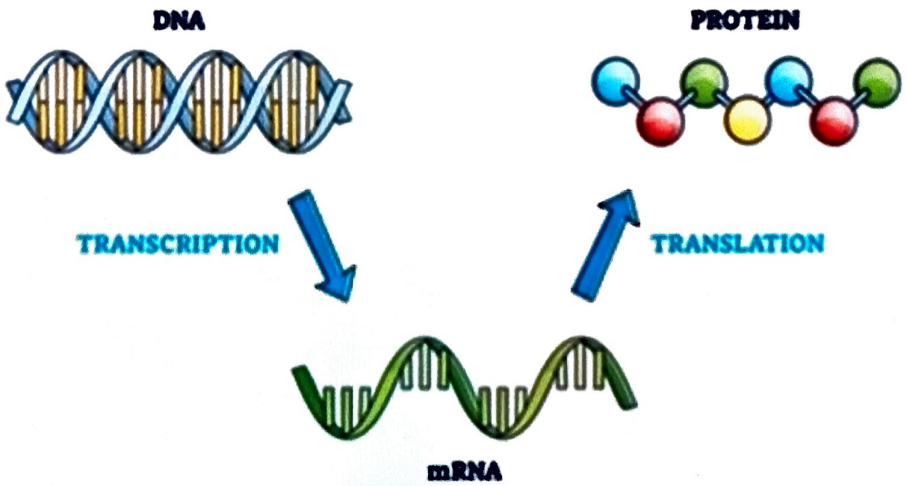
2. Elongation

- One strand of DNA, the template strand (or noncoding strand), is used as a template for RNA synthesis.
- As transcription proceeds, RNA polymerase traverses the template strand and uses base pairing complementarity with the DNA template to create an RNA copy.
- Although RNA polymerase traverses the template strand from 3' ⟶ 5', the coding (non-template) strand and newly-formed RNA can also be used as reference points, so transcription can be described as occurring 5'⟶ 3'.

❖ GENE EXPRESSION

- In this process, a gene's DNA is used as a template for synthesis of a specific protein.
- First, In Transcription, the information encoded in a specific region of DNA is transcribed (copied) to produce a specific molecule of RNA (ribonucleic acid).
- In a second process, referred to as Translation, the RNA attaches to a ribosome, where the information contained in RNA is translated into a corresponding sequence of amino acids to form a new protein molecule

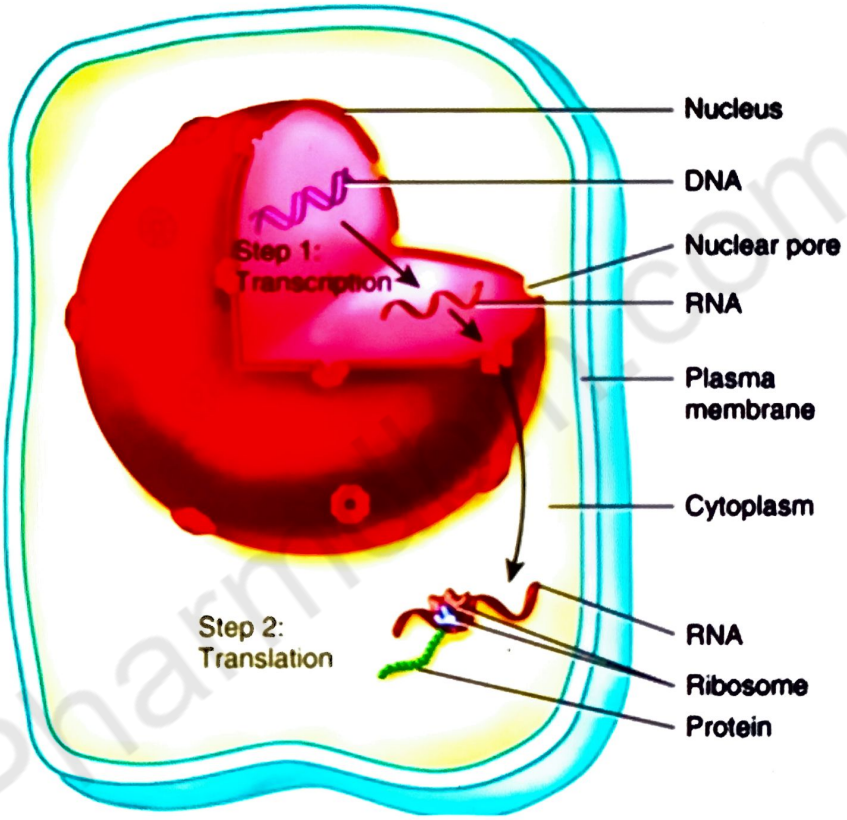
GENE EXPRESSION



✓ Overview of gene expression

Synthesis of a specific **protein** requires **transcription** of a gene's DNA into RNA and translation of RNA into a corresponding sequence of amino acids

- Transcription occurs in the **Nucleus**
- Translation occurs in the **Cytoplasm**

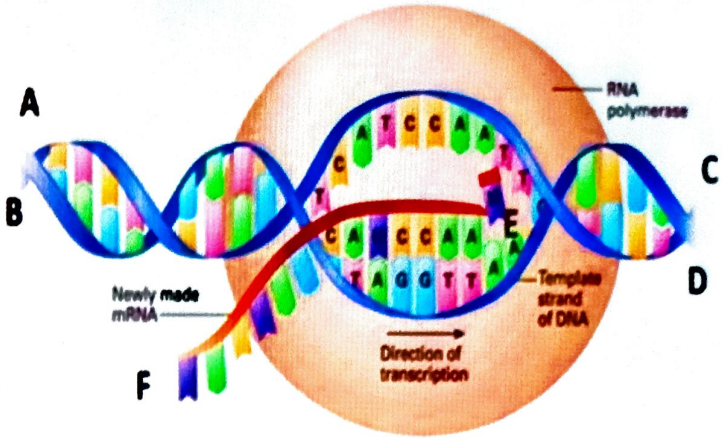


✓ Points to Remember

- **DNA** and **RNA** store genetic information as sets of **Three nucleotides.**
- A **sequence** of three such **nucleotides** in DNA is called a **Base Triplet.**
- Each **DNA base triplet** is transcribed as a **complementary** sequence of **Three** nucleotides, called a **Codon.**
- A given **codon** specifies a particular **amino acid.**
- The **genetic code** is the set of **rules** that relate the base triplet sequence of DNA to the corresponding **codons** of **RNA** and the **amino acids** they specify.

1. TRANSCRIPTION OF GENETIC CODE

- The word transcription means **copying**. which indicates the copying of **genetic code** from DNA to RNA.
- The **proteins** are synthesized in the **ribosomes** which are present in the **cytoplasm**.



- The **synthesis** of different **proteins** depends upon the information **(sequence of codon)** encoded in the **genes** of the DNA
- DNA is a **macromolecule**, it cannot pass through the **pores** of the nuclear membrane and **enter** the cytoplasm.
- But, the information from DNA must be sent to **ribosome**.
- So, the gene has to be **transcribed** (copied) into **mRNA** which is developed from **DNA**.

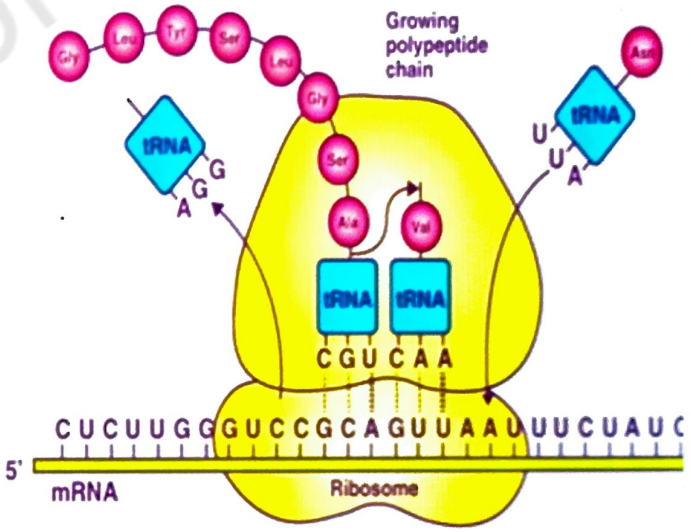
Three types of RNA are made from the DNA template:

1. **Messenger RNA** (mRNA) directs the synthesis of a protein.
2. **Ribosomal RNA** (rRNA) joins with ribosomal proteins to make ribosomes.
3. **Transfer RNA** (tRNA) binds to an amino acid and holds it in place on a ribosome until it is incorporated into a protein during translation

- In **transcription** of genetic code, It involves the **formation** of **mRNA** and copying or transfer of information from DNA to mRNA.
- The mRNA enters the cytoplasm from the nucleus and activates the **ribosome** resulting in protein synthesis.
- The formation of mRNA from DNA is facilitated by the **Enzyme RNA polymerase**

2. TRANSLATION OF GENETIC CODE

- Translation is the **process** by which **protein synthesis** occurs in the **ribosome** of the cell under the **direction** of genetic instruction carried by **mRNA** from **DNA**.
- It is the process by which the mRNA is read by ribosome to produce a **protein**.
- This involves the role of other two types of **RNA**, namely **tRNA** and **rRNA**.
- The mRNA moves out of **nucleus** into the **cytoplasm**.
- Now, a group of ribosomes called **polysome** gets attached to mRNA.
- The sequence of **codons** in mRNA are **exposed** and **recognized** by the complementary sequence of base in tRNA.
- The **complementary** sequence of base is called **anticodon**.
- According to the sequence of bases in anticodon, different amino acids are transported from the **cytoplasm** into the **ribosome** by tRNA that acts as a **carrier**.
- **rRNA** helps the protein molecules in assembling from **amino acids.**
- The protein synthesis occurs in the ribosomes which are attached to **Rough endoplasmic reticulum.**

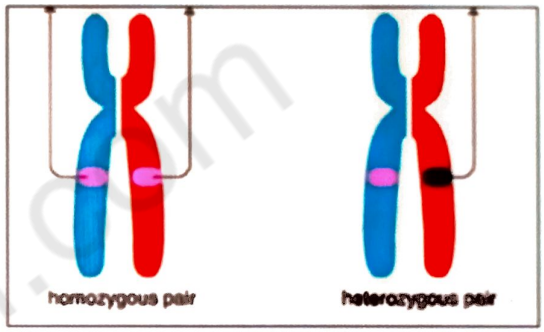


GENETIC PATTERN OF INHERITANCE

❑ INTRODUCTION

- Inheritance is the passage of **hereditary traits** from **one** generation to the **next**.
- It is the process by which we acquired our **characteristics** from **parents**
- The branch of biology that deals with **inheritance** is called **genetics.**
- The area of **health care** that offers advice on **genetic problems** (or potential problem) is called **genetic counseling**

❖ Some Important Definitions

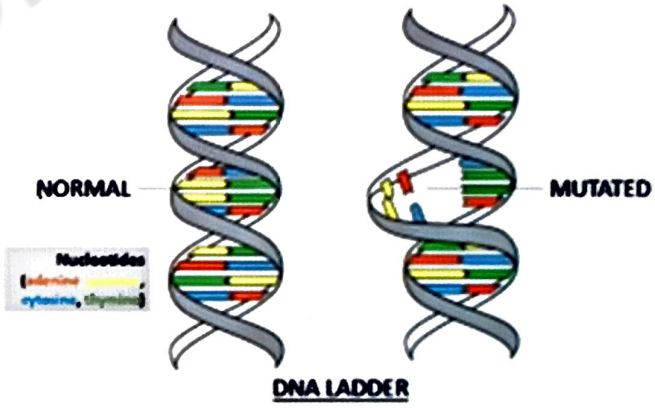


✓ ALLELES

- Alternative forms of a gene that **code** for the **same trait** and are at the **same location** on **homologous** chromosomes are called alleles .

✓ MUTATION

- A mutation is a **permanent** heritable change in an allele that produces a different **variant** of the **same trait.**



✓ GENOTYPE

- It refer a particular gene or **set** of genes which are carried by an individual.
- Genotype of an individual is its complete **heritable genetic identity,** which is **unique** to an organism or individual.
- It also refers to the alleles or **variants** of a gene, which are carried by an organism.

✓ PHENOTYPE

- Phenotype refers to how the **genetic makeup** is expressed in the body
- It is the **physical** or **outward** expression of a gene.

✓ DOMINANT ALLELE

- An allele that **dominates** or masks the presence of **another** allele and is fully **expressed** is said to be a dominant allele,

✓ RECESSIVE TRAIT

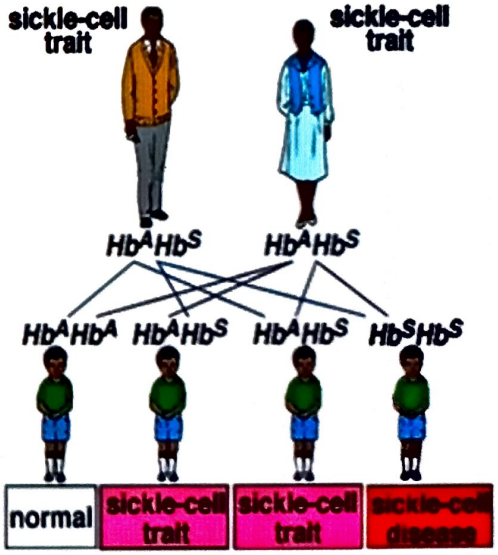
- The trait **expressed** is called a dominant trait.
- The allele whose **presence** is **completely** masked is said to be a recessive allele, and the trait it controls is called a recessive trait.

❖ Variations on dominant–recessive inheritance

- Most inherited traits are influenced by more than one gene, and, to complicate matters, most genes can influence more than one trait.
- **Variations** on dominant–recessive inheritance include **incomplete dominance, multiple-allele inheritance,** and **complex inheritance.**

✓ Incomplete Dominance

- In incomplete dominance, neither member of a pair of **alleles** is **dominant** over the other, and the **heterozygote** has a phenotype intermediate between the **homozygous** dominant and the **homozygous** recessive phenotypes.
- An example of incomplete dominance in humans is the inheritance of **sickle-cell disease.**
- People with the homozygous **dominant** genotype HbAHbA form normal hemoglobin
- Those with the homozygous **recessive** genotype HbS HbS have sickle-cell disease and severe **anemia.**



- Although they are usually healthy, those with the heterozygous genotype HbAHbS have minor problems with anemia because half their **hemoglobin** is normal and half is not.
- **Heterozygotes** are carriers, and they are said to have **sickle-cell trait**

✓ Multiple-allele Inheritance

- A single **individual** inherits only **two alleles** for each gene, some genes may have more than two **alternative** forms this is the basis for multiple-allele inheritance
- One example of multiple-allele inheritance is the inheritance of the **ABO blood group.**
- The four blood types (phenotypes) of the ABO group— **A, B, AB, and O**—result from the inheritance of six combinations of three different alleles of a single gene called the I gene:

  (1) allele I A produces the A antigen,

  (2) allele I B produces the B antigen, and

  (3) allele i produces neither A nor B antigen.

- Each person inherits two I-gene alleles, one from each parent, that give rise to the various phenotypes.

Genotype	Blood type
$I_A I_A$	A
$I_A I_B$	AB
$I_A i$	A
$I_B I_B$	B
$I_B i$	B
ii	O

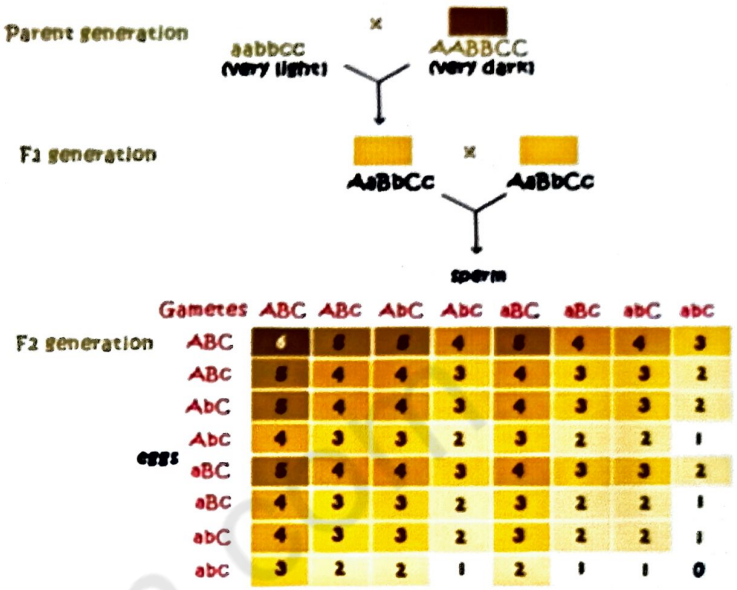
✓ Complex Inheritance

- Most inherited traits are **not** controlled by one **gene**, but instead by the **combined** effects of two or more genes, a situation referred to as polygenic inheritance (poly- many).
- The **combined effects** of many genes and **environmental factors**, a situation referred to as complex inheritance.
- Examples of **complex traits** include **skin color, hair color, eye color, height, metabolism rate, and body build.**

- In complex inheritance, one **genotype** can have many possible **phenotypes**, depending on the **environment**, or one phenotype can include many possible genotypes

✓ Sex-linked Inheritance

- In addition to determining the sex of the **offspring**, the **sex chromosomes** are responsible for the **transmission** of several nonsexual traits.
- Many of the **genes** for these traits are present on **X chromosomes** but are **absent** from Y chromosomes.



- This feature produces a pattern of **heredity** called sex-linked inheritance.

Genotype	Phenotype
X^CX^C	Normal female
X^CX^c	Normal female (but a carrier of the recessive gene)
X^cX^c	Red–green color-blind female
X^CY	Normal male
X^cY	Red–green color-blind male

- One example of sex-linked inheritance is **red–green color blindness**
- It is characterized by a deficiency in either red- or green **sensitive cones**, so red and green are seen as the same color (either red or green, depending on which cone is present).
- The gene for red–green color blindness is a **recessive** one designated c. Normal color vision, designated **C**, dominates.
- The **C/c genes** are located only on the X chromosome, so the ability to see **colors** depends entirely on the X chromosomes.