

AP INTER 2nd YEAR

ZOOLOGY

8

**MARKS
MOST IMP
QUESTIONS**

and Answers

IPE-2026

Krishna Sir





Chapters



4

Body fluids and Circulation ✓

Excretory Products and Elimination ✓

Human Reproductive System ✓

Genetics ✓

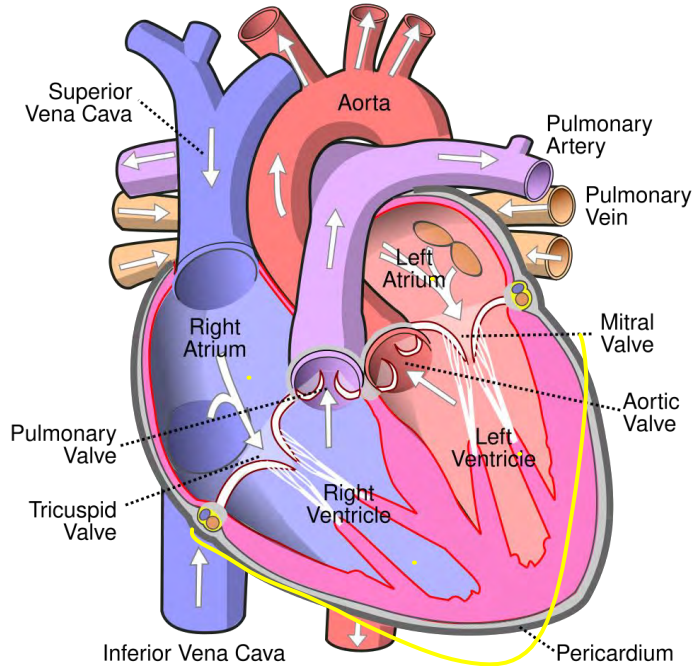


8 M LAQ List



Name of the Chapter	List of Questions
Body fluids and Circulation	Structure of Heart ✓ Functioning of Heart ✓
Excretory Products and Elimination ✓	Excretory system of a man ✓ Structure of Nephron ✓ Urine Formation ✓
Human Reproductive System ✓	Male Reproductive system ✓ Female Reproductive System ✓
Genetics ✓ <i>genetic disorders</i>	Multiple Alleles ✓ Chromosome theory of Sex determination ✓ Criss Cross inheritance ✓ DNA Fingerprinting ✓

Describe the structure of heart with neat labelled diagram



Thoracic cavity, between lungs ✓

Pericardium ✓ →

Epicardium: ✓ outer

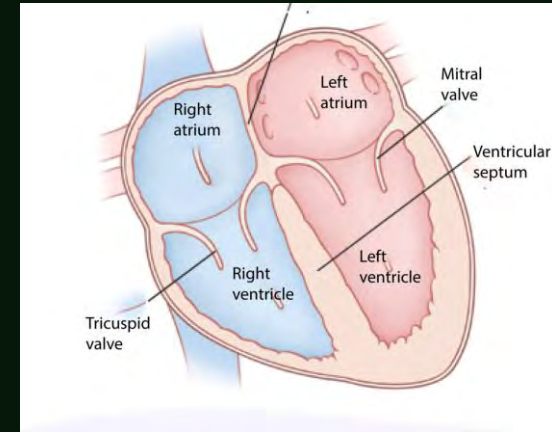
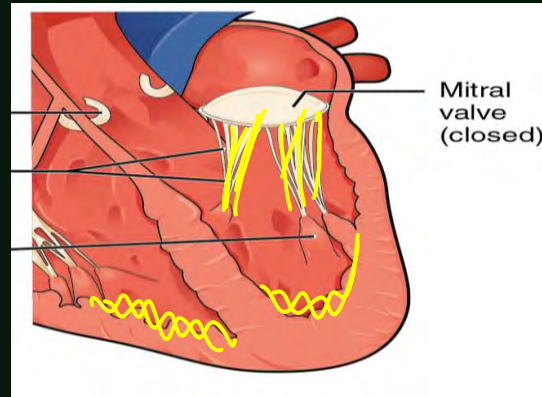
Myocardium: Middle

Endocardium: Inner

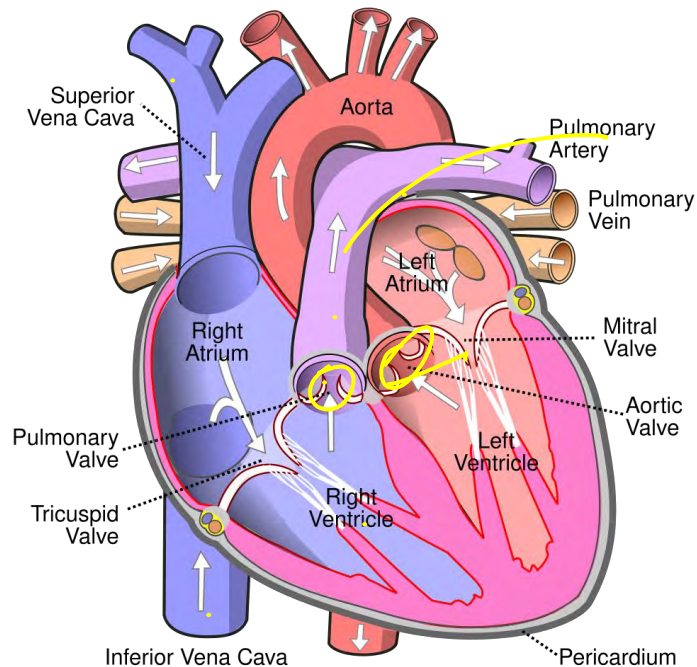
2 Atria:

2 Ventricles:

Structure	Description
Interatrial septum ✓	Separates atria;
Interventricular septum ✓	Separates ventricles
Chordae tendineae ✓	Collagen cords connecting valve cusps to papillary muscles
Columnae carneae ✓✓	Muscular ridges in ventricles



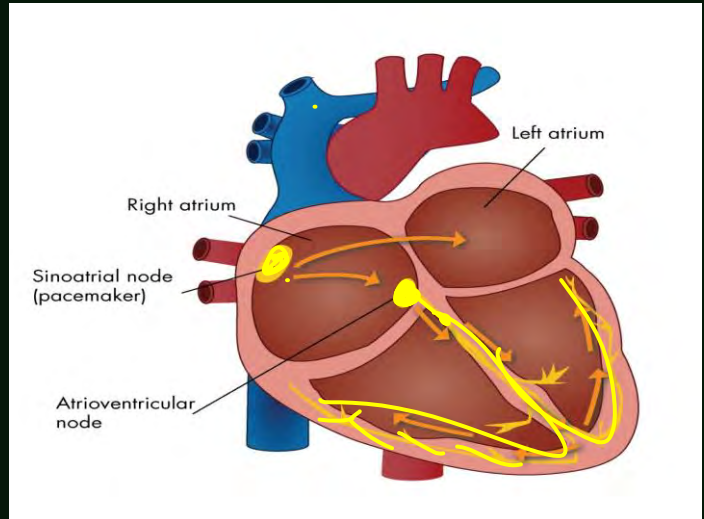
✓ L BW



Tricuspid ✓ RA/RV
 Mitral ✓ LA/LV
 Pulmonary ✓
 Aortic ✓

Vena cavae ✓ deoxygenated
 Pulmonary artery ✓ deoxygenated → lungs
 Pulmonary veins → oxygenated blood → L.A.
 Aorta — oxygenated blood — body part

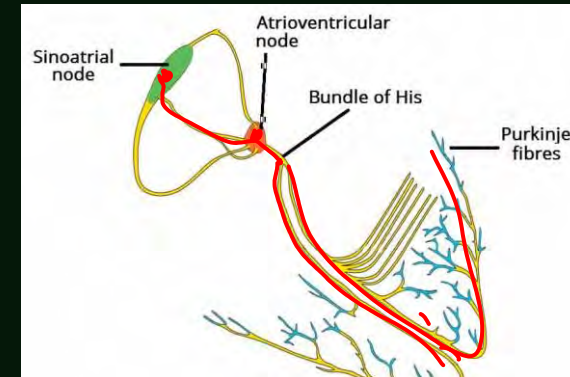
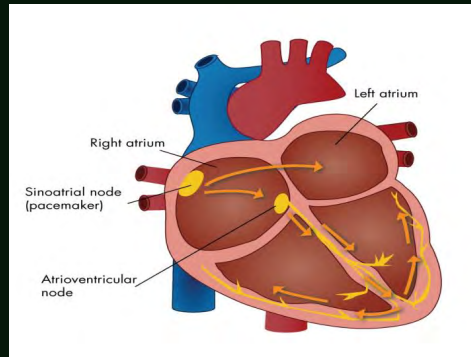
Part	Location
SA Node (Pacemaker)	Right atrium near superior vena cava
AV Node	Interatrial septum
AV (Bundle of His)	Interventricular septum
Purkinje Fibers	Walls of ventricles



Write the notes on Working heart of man

Conduction

Part	Location	Function
SA Node (Pacemaker) ✓	Right atrium near superior vena cava ✓	Initiates heart beat (72/min, myogenic) ✓
AV Node ✓	Interatrial septum ✓	Relays impulse to ventricles ✓
AV (Bundle of His) ✓	Interventricular septum ✓	Transmits signal from AVN to branches ✓
Purkinje Fibers	Walls of ventricles	Spread impulse causing ventricular contraction

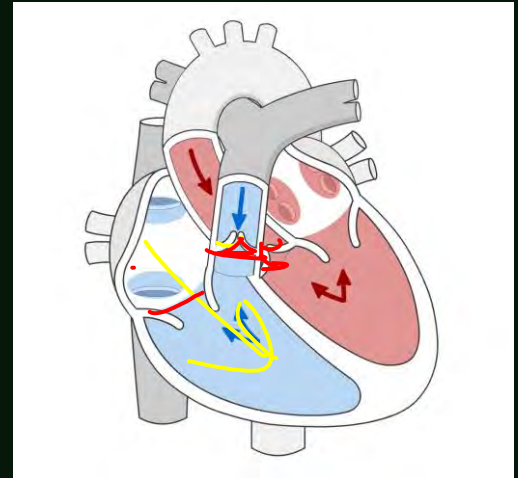


Cardiac Cycle

72/60 sec
 0.8 sec $\rightarrow$ 1 min AV $\leftarrow$ Tricuspid
 Bicuspid

Phase	Duration	Event Description
Atrial Systole	<u>0.1 sec</u> ✓	Atria contract $\rightarrow$ 30% more blood enters ventricles
Ventricular Systole	<u>0.3 sec</u>	Ventricles contract $\rightarrow$ <u>AV valves</u> close ("Lub") $\rightarrow$ blood ejected ✓
Joint Diastole	<u>0.4 sec</u>	All chambers relax $\rightarrow$ <u>semilunar valves</u> close ("Dup")

Systole - contraction
Diastole Relaxation

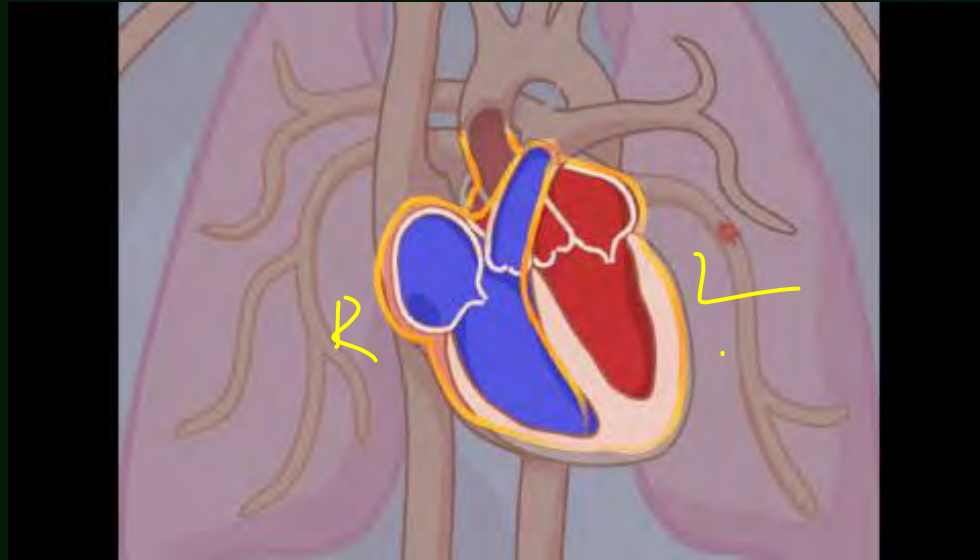


Cardiac Output

Term	Value / Formula
Stroke Volume	~70 mL/beat
Heart Rate	~72 beats/min
Cardiac Output	Stroke Volume $\times$ Heart Rate = ~5040 mL/min or ~5 L/min

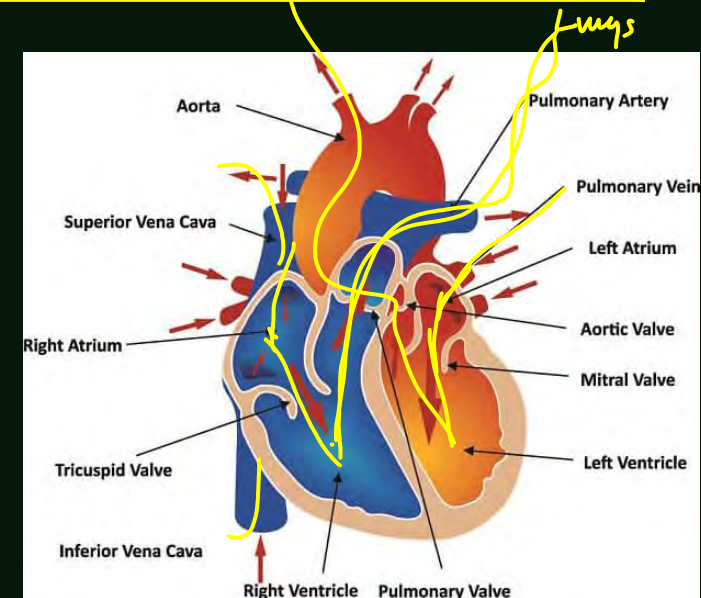
Stroke Volume mL / Beat
 70 mL / Beat
 72×70
 $= 5040 = \underline{5L}$

Pulmonary, Systemic circulation



Double Circulation

Vessel/Arch	Origin	Function
Pulmonary Circulation	Right ventricle	Sends deoxygenated blood to lungs ✓
Systemic Circulation ✓	Left ventricle	Sends oxygenated blood to body



Pulmonary Circulation

Blood pumped to

Pulmonary Artery ✓

Nature of blood

Deoxygenated ✓

Destination

Lungs ✓

Gas exchange

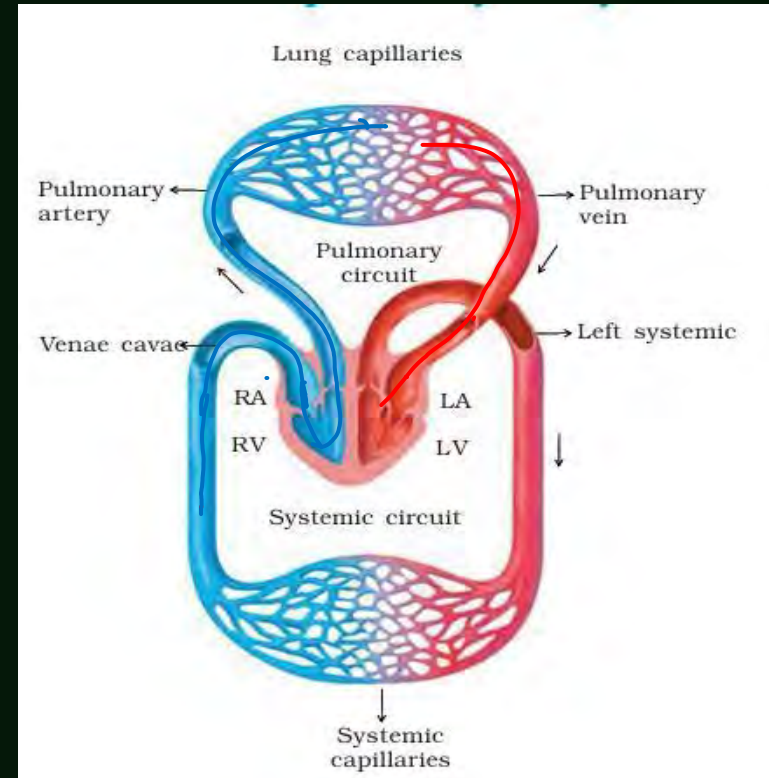
CO₂ is released; O₂ is absorbed ✓

Returns to

Left Atrium via Pulmonary Veins

Nature of returning blood

Oxygenated



Systemic Circulation

Blood pumped to

Nature of blood

Carried to

Function

Returns to

Nature of returning blood

Aorta ✓

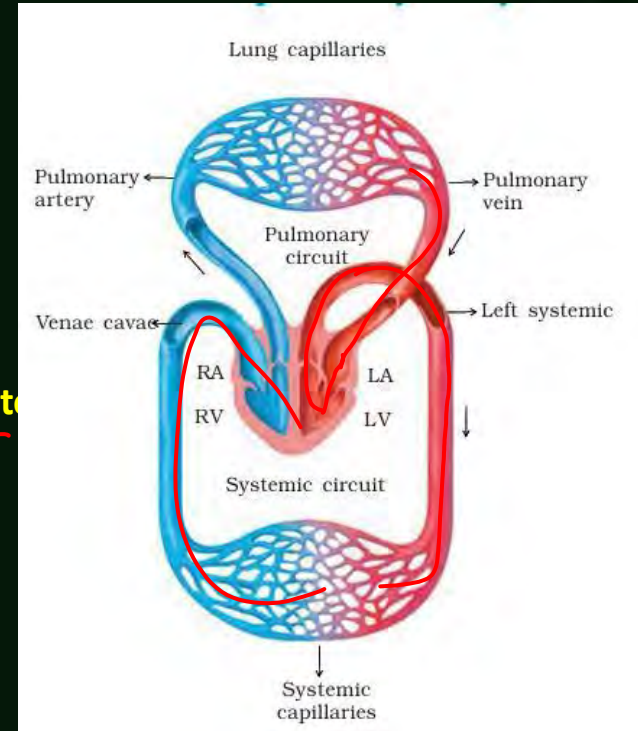
Oxygenated ✓

All body tissues ✓

Supplies O_2 , nutrients; removes CO_2 , waste ✓

✓ **Right Atrium via Venae Cavae** ✓

✓ **Deoxygenated**



Describe the excretory system of man, giving the structure of a nephron

Human Excretory System

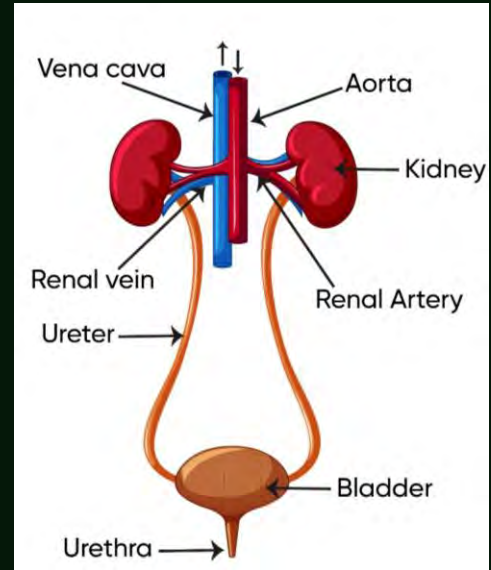
It consists of four parts:

A pair of kidneys ✓

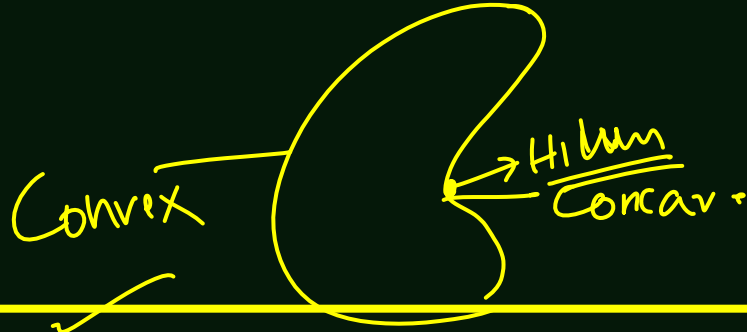
A pair of ureters ✓

Urinary bladder ✓

Urethra ✓

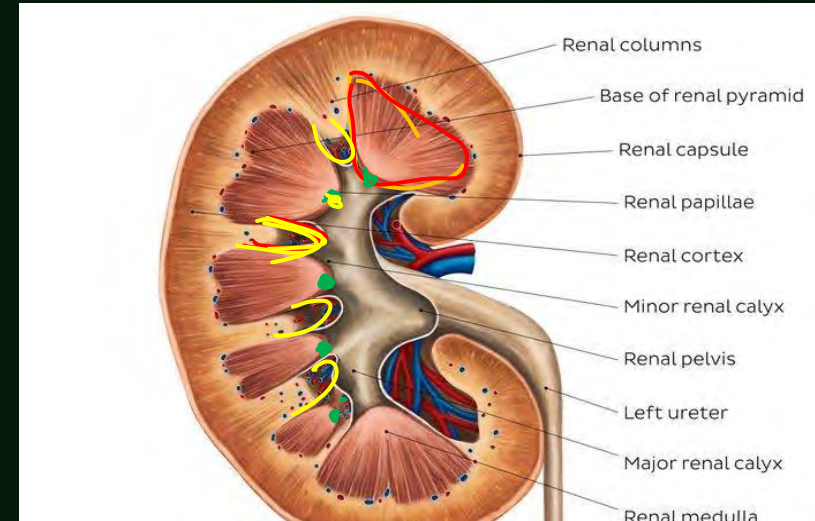


Kidney



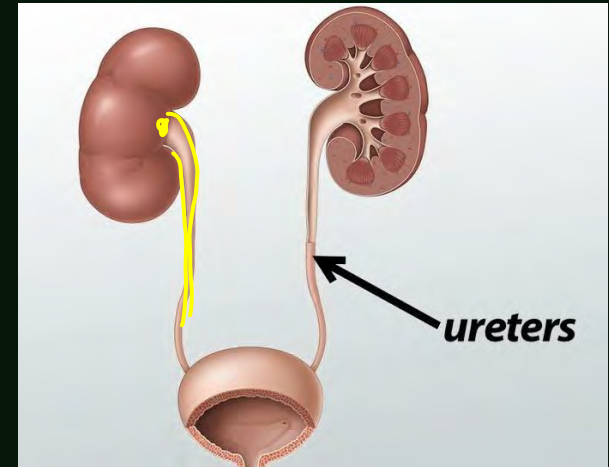
Shape & position	Reddish-brown, retroperitoneal, bean-shaped organs
Location	Present on either side of the vertebral column at the level of anterior lumbar vertebrae
Surface features	Outer surface convex; inner surface concave with a notch called hilum

Region	Details
Cortex ✓	Outer region
Medulla	Inner region; contains renal pyramids
Renal Pyramids	Cone-shaped structures in medulla
Renal Columns (Columns of Bertin)	Cortical projections between pyramids
Renal Papilla	Tip of each pyramid; opens into minor calyx



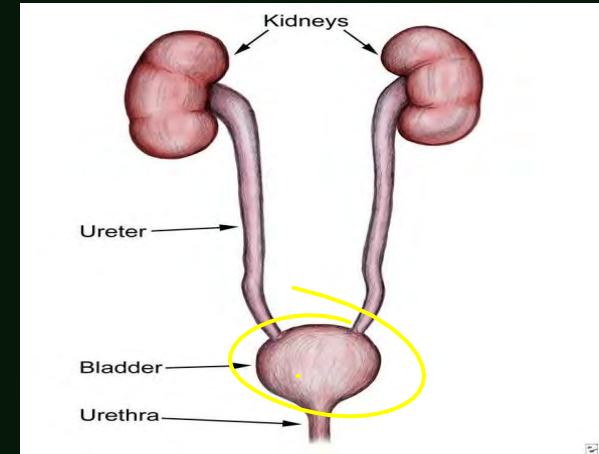
Ureters

Feature	Origin	Details
Structure		Begin at renal pelvis
Lining		<u>Slender, whitish, muscular tubes</u>
Path		<u>Transitional epithelium</u> Run down from <u>kidneys</u> to <u>urinary bladder</u>



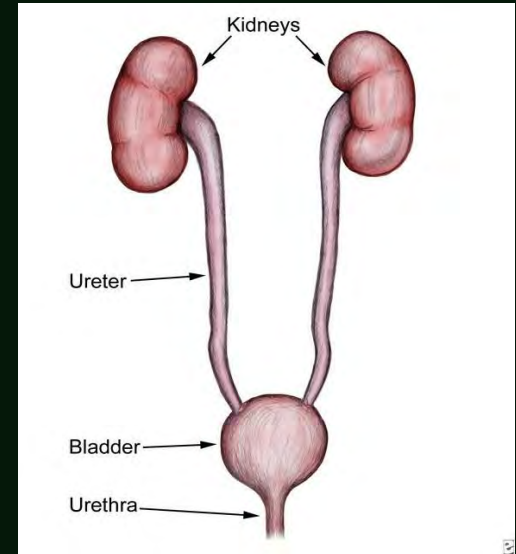
Urinary Bladder

Location	Lower abdominal cavity (pelvic region)
Function	Temporary storage of urine
Wall	Thick, muscular, distensible
Lining	Transitional epithelium
Sphincters	- <u>Internal sphincter</u> : Smooth muscle - <u>External sphincter</u> : Striated muscle



Urethra

Function	Conducts urine from bladder to outside
Female Urethra	Opens near vaginal orifice
Male Urethra	Passes through penis



Nephron

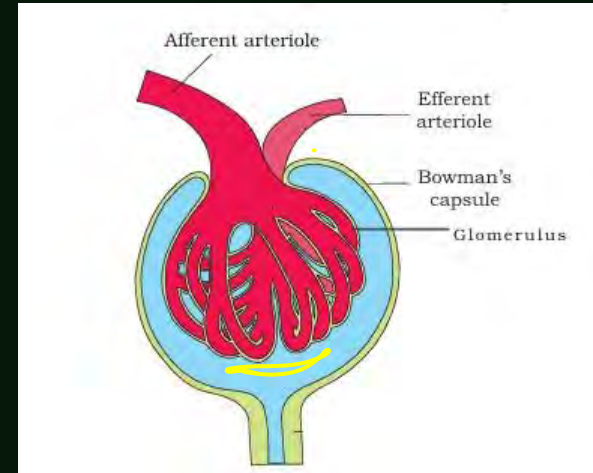
Feature	Details
Total per kidney	~1million nephrons
Basic unit of	Structure & function of kidney
Two main parts	1 Renal corpuscle (Malpighian body) 2 Renal tubule

N < RC
 RT

Renal Corpuscle (Malpighian Body)

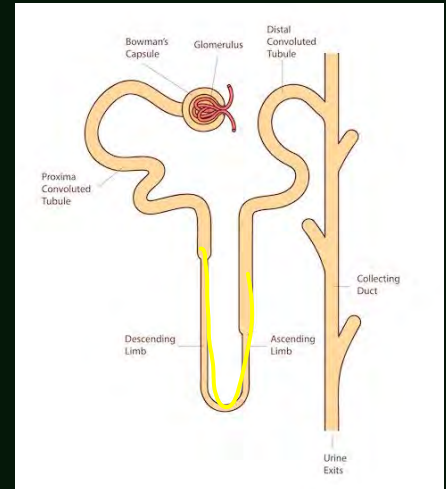
Component	Details
Glomerulus	Capillary tuft; formed by afferent arteriole , drained by efferent arteriole
Bowman's Capsule	Double-walled cup surrounding glomerulus
Podocytes	Specialized cells on inner wall of Bowman's capsule
Filtration slits	Gaps between podocyte foot processes – allow filtration

R < Glomerulus
 Bowman's capsule
 SSSSS



Renal Tubule Structure

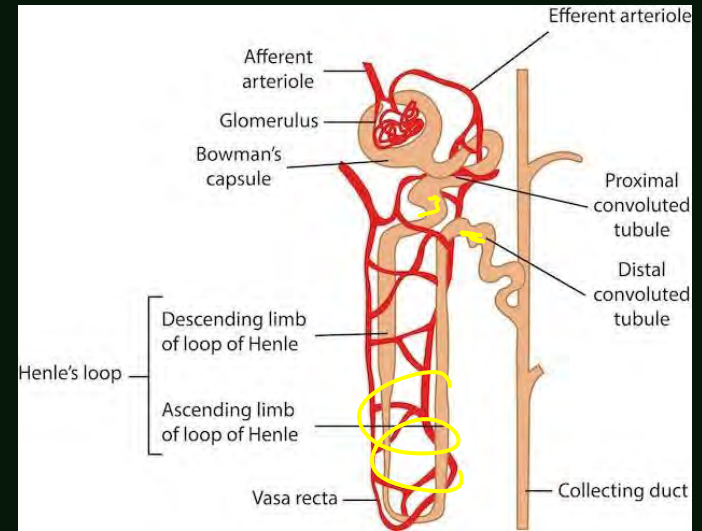
Segment	Description
PCT (Proximal Convoluted Tubule) ✓	Highly coiled, in cortex; major site of reabsorption
Loop of Henle ✓	Hairpin-shaped; has descending limb and ascending limb
DCT (Distal Convoluted Tubule) ✓	Located in cortex; continues into collecting duct
Collecting Duct ✓	Begins in cortex, passes into medulla.



Blood Vessels around Nephron

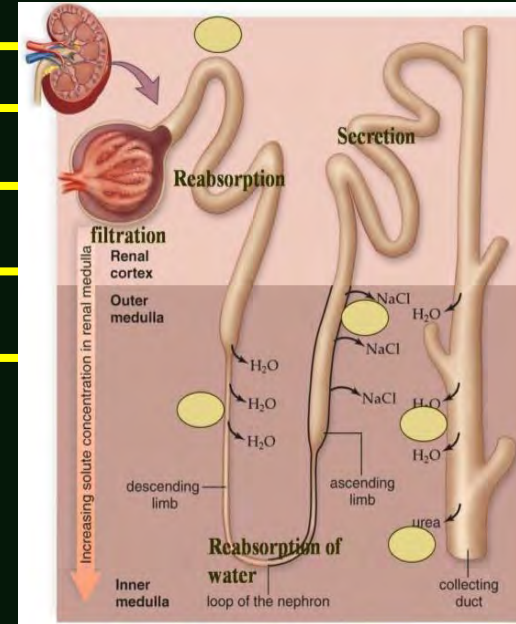
Vessel	Function
Afferent Arteriole	Brings blood to glomerulus
Efferent Arteriole	Drains glomerulus
Peritubular Capillaries	Surround PCT & DCT (mainly in cortical nephrons)
Vasa Recta	Straight capillaries around loop of Henle (in juxtamedullary nephrons)

Afferent E



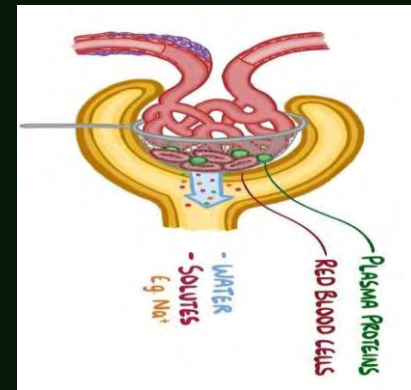
Discuss the various steps involved in urine formation

Name	Description
Glomerular Filtration	Blood filtered from glomerulus into Bowman's capsule
Selective Reabsorption	Nutrients and water reabsorbed into blood
Tubular Secretion	Ions/toxins actively secreted into tubule

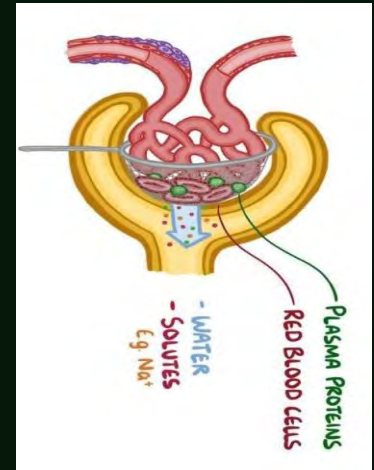


Glomerular Filtration

Site	Bowman's capsule (Glomerulus → Bowman's space)
Afferent Arteriole ✓	Wider → Brings blood into glomerulus
Efferent Arteriole ✓	Narrow → Takes blood away from glomerulus
Pressure Condition	High hydrostatic pressure inside glomerulus
Process Type	Ultrafiltration (passive, pressure driven)
Filtered Components	Water, Glucose, Amino acids, Na^+ , Cl^- , Urea & other small solutes



Not Filtered	Blood cells & Plasma proteins (large size)
Filtrate Produced per Minute (GFR)	125 mL/min
Filtrate Produced per Day	~180 Liters
Resulting Fluid Name	Glomerular Filtrate



Tubular Reabsorption and Secretion

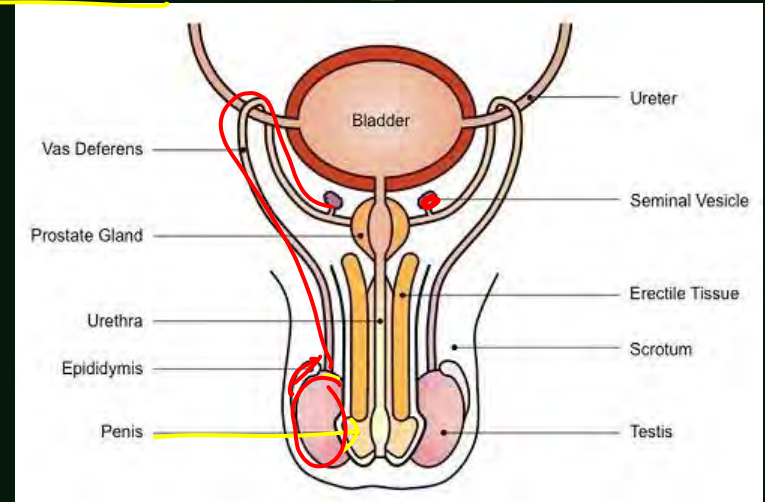
Segment	Reabsorbs	Secretes
PCT ✓	Glucose, amino acids, Na^+ , Cl^- , water ✓	H^+ , NH_3 ✓
LoH	Desc: Water Asc: NaCl ✓	—
DCT	Na^+ , HCO_3^- , Water (facultative) ✓	H^+ , K^+ , NH_3 ✓
CD	Water, Urea (recycling) ✓	H^+ , K^+ ✓

Discuss the Male Reproductive system

The male reproductive system consists of organs that produce male gametes (sperms) and male sex hormones

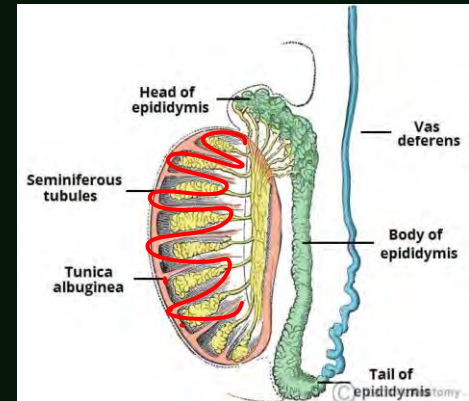
(androgens)

- ✓ Testes ✓
- ✓ Epididymis ✓
- ✓ Vas deferens ✓
- ✓ Seminal vesicles ✓
- ✓ Prostate gland ✓
- ✓ Bulbo-urethral (Cowper's) glands ✓
- ✓ Urethra ✓
- ✓ Penis ✓



Testes

- Primary sex organs; paired, ovoid, pinkish, ~4–5 cm long, 2–3 cm wide.
- Location: Outside abdominal cavity in the scrotum → maintains temp. ~2–2.5°C lower than body (essential for spermatogenesis).
- Coverings:
 - Tunica vaginalis – serous covering
 - Tunica albuginea – dense fibrous capsule, forms septa → divides testis into ~250 lobules.



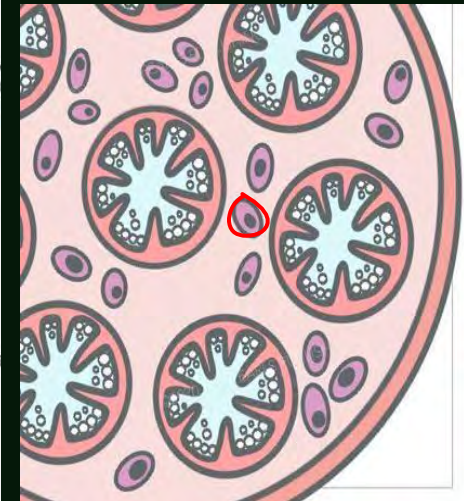
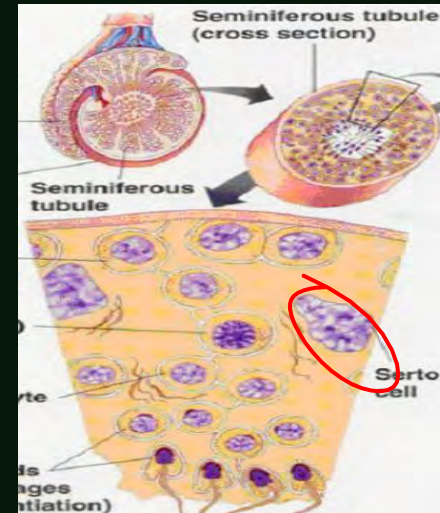
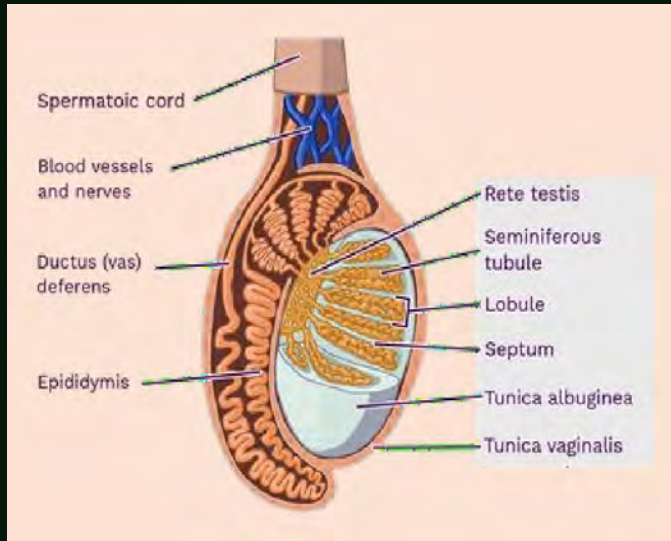
Each lobule contains 1–3 seminiferous tubules (site of sperm formation).

Seminiferous tubules:

Cells present:

Sertoli cells : Provide nutrition to developing sperms.

Interstitial spaces: Contain Leydig (interstitial) cells Produce testosterone ✓



Epididymis

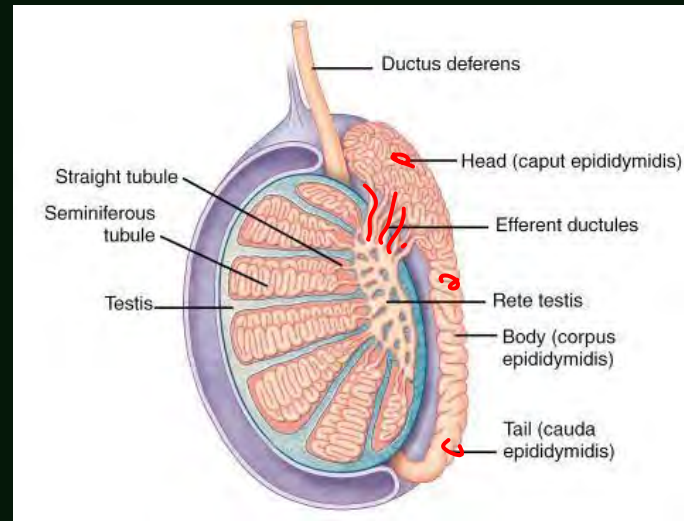
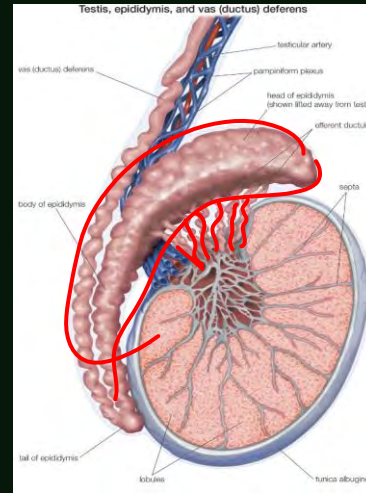
Highly coiled duct (~6 m if uncoiled), posterior surface of testis.

Parts:

Caput (head) – receives sperm from vasa efferentia.

Corpus (body) – site of sperm maturation (acquisition of motility, fertilizing capacity).

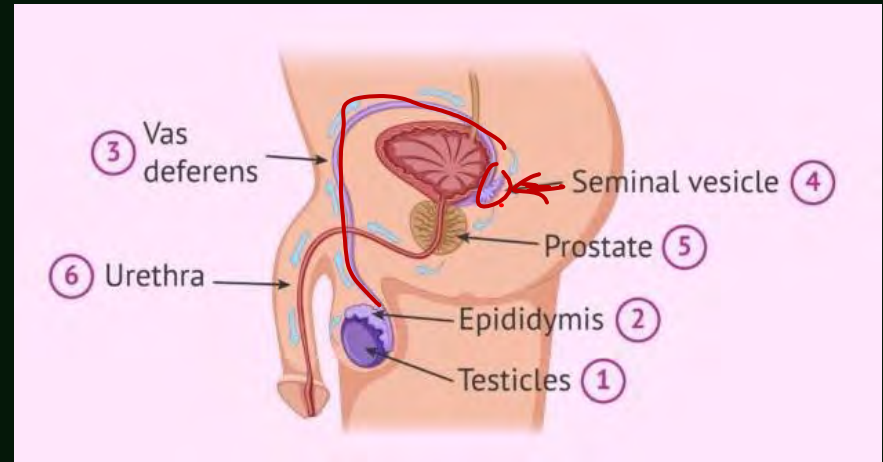
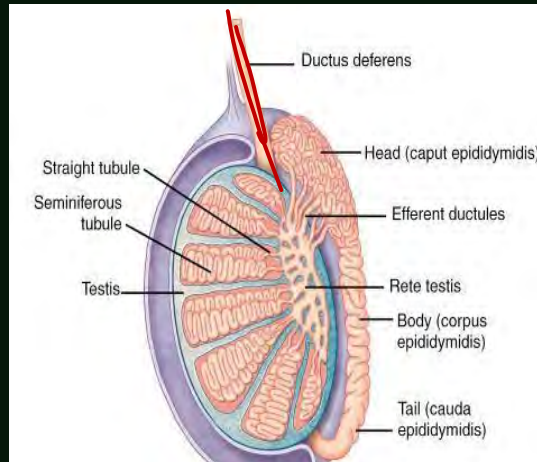
Cauda (tail) – major sperm storage site.



Vas deferens

Vas deferens (Ductus deferens) Long muscular tube .

Joins duct of seminal vesicle → forms ejaculatory duct. ✓

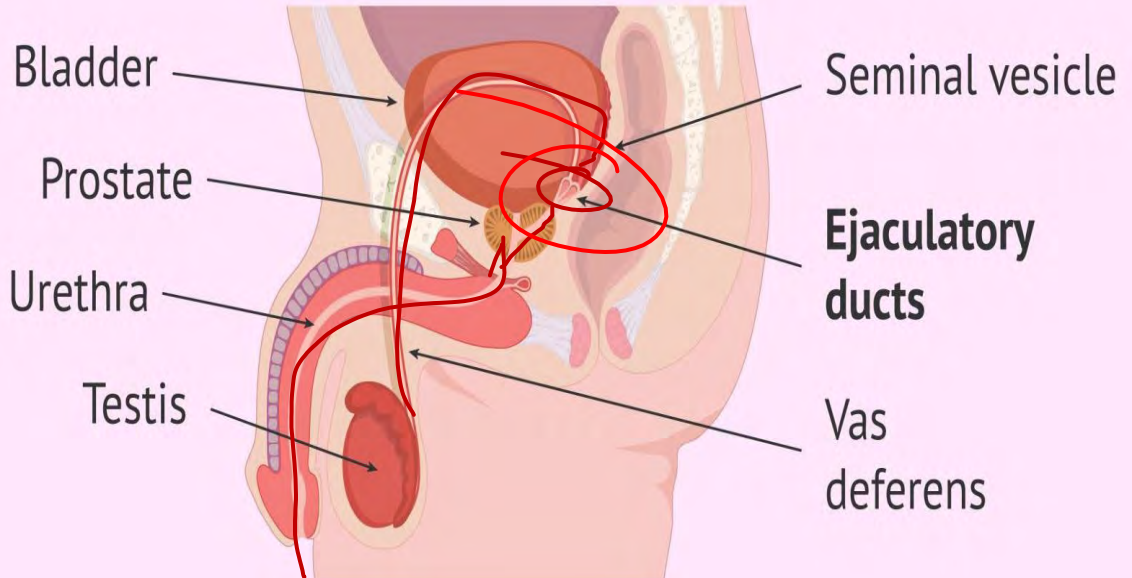


Ejaculatory ducts

Short ducts (2) formed by union of vas deferens + seminal vesicle duct.

Open into prostatic urethra.

Carry sperm + seminal vesicle secretion.



Urethra (Male)

Shared passage for **urine + semen**. ~20 cm long.

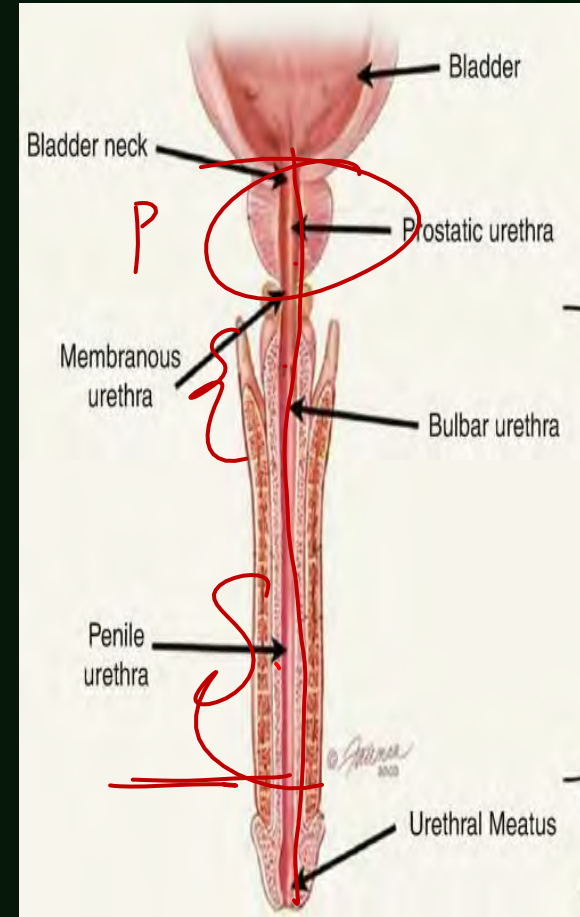
Divisions:

Prostatic urethra (inside prostate).

Membranous urethra (shortest, narrowest).

Spongy (penile) urethra (longest, runs inside penis).

Ends at **external urethral meatus** (tip of glans penis).



Penis

External genital organ; deposits sperm in female vagina.

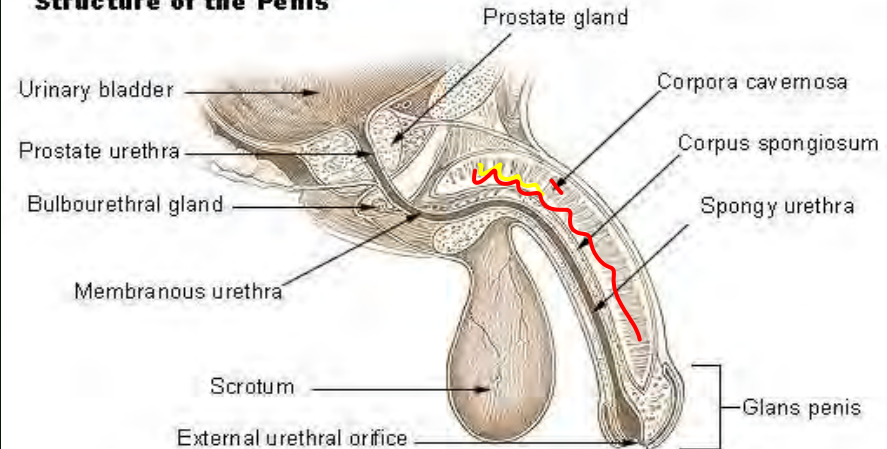
Structure:

Corpora cavernosa (2, dorsal) – erectile tissue.

Corpus spongiosum (1, ventral) – surrounds urethra, forms glans penis.

Glans penis: Enlarged tip, covered by prepuce (foreskin).

Structure of the Penis



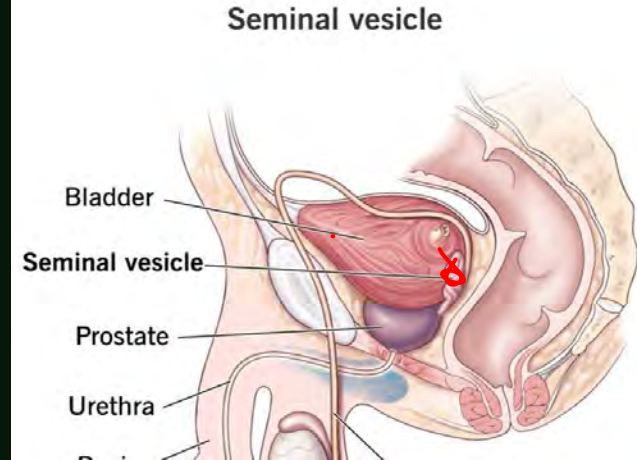
Seminal Vesicles

Paired, behind bladder.

Open into vas deferens.

Contribute ~60% of semen volume.

Secretion: alkaline, viscous, rich in fructose, prostaglandins, proteins, citric acid, potassium, phosphorus.



Bulbourethral (Cowper's) Glands

Bulbourethral (Cowper's) Glands

Small, pea-shaped, at base of penis.

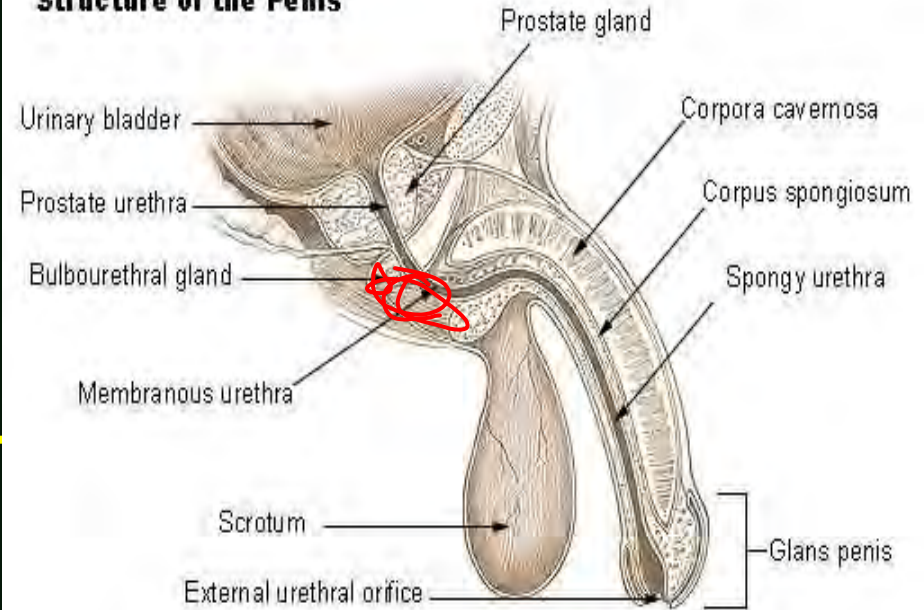
Secrete alkaline mucus before ejaculation.

Functions:

Lubricates urethra.

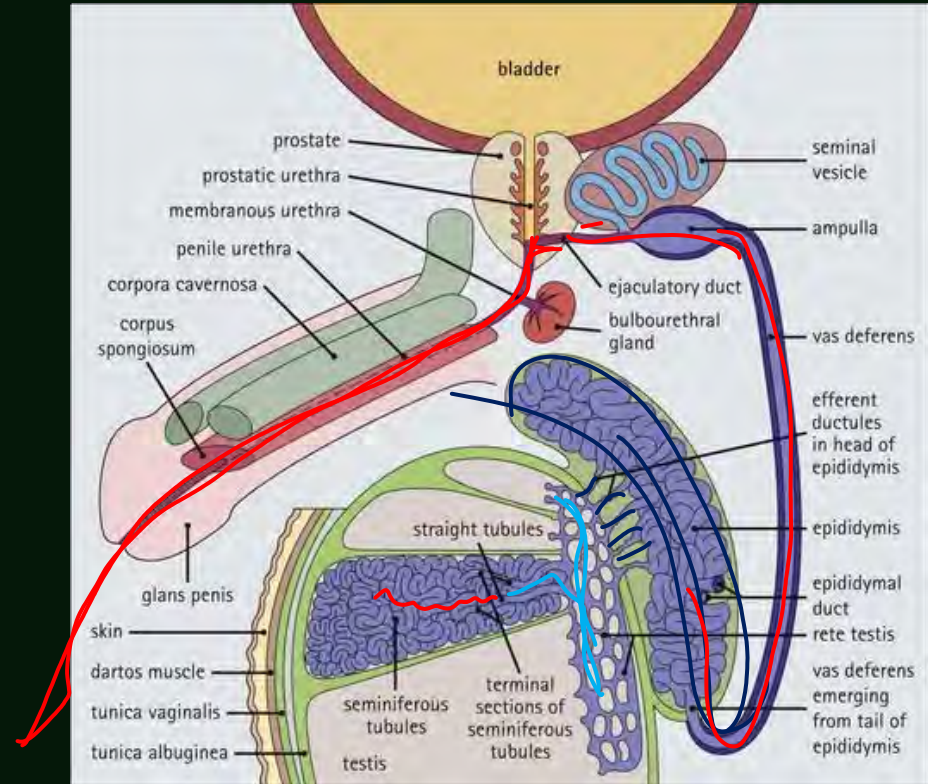
Neutralizes acidic urine traces.

Structure of the Penis



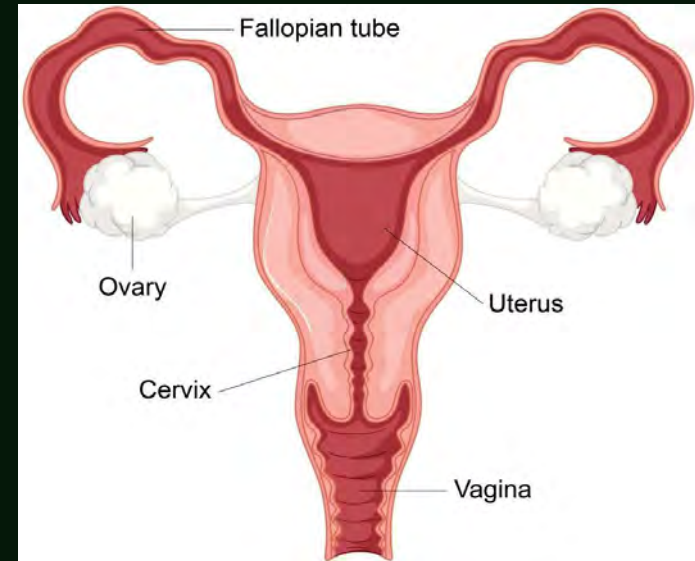
Passage of Sperm

Seminiferous tubules → Rete testis → Vasa efferentia → Epididymis → Vas deferens → Ejaculatory duct → Urethra → Penis



Discuss the female Reproductive system

Consists of: A pair of ovaries , A pair of oviducts (fallopian tubes), Uterus, Vagina, External genitalia (vulva) and accessory Glands



Ovaries



Ovaries

Primary female sex organs.

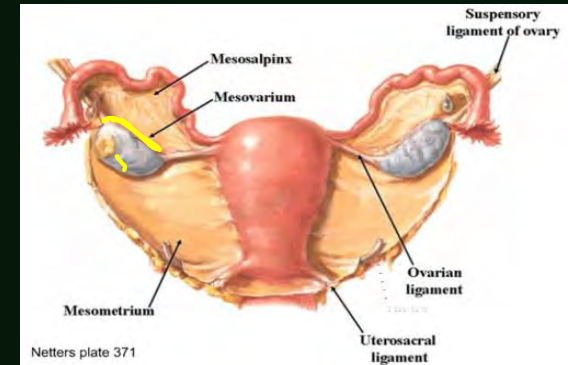
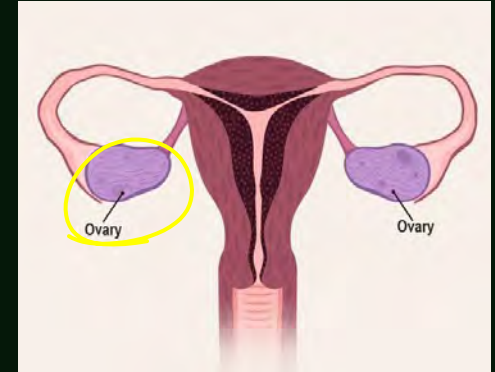
Functions:

Produce female gametes (ova).

Secrete steroid hormones (estrogen, progesterone).

Location: One on each side of lower abdomen.

Attachment: Connected to abdominal wall by mesovarium



Coverings & Structure:

Germinal (ovarian) epithelium – simple cuboidal epithelium (visceral peritoneum).

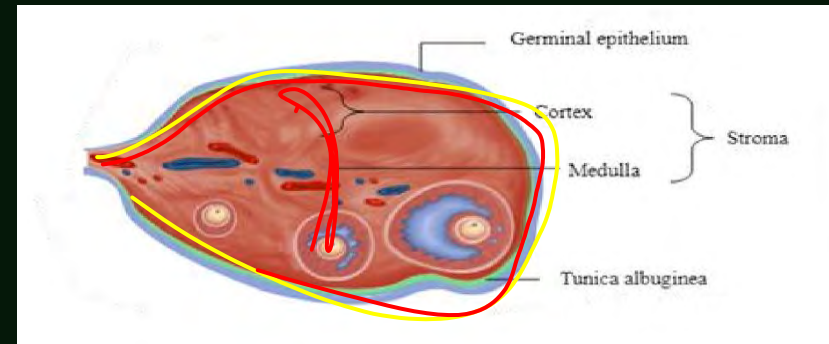
Tunica albuginea – dense connective tissue capsule.

Stroma – two zones:

Cortex (outer) – dense, contains ovarian follicles in various developmental stages.

Medulla (inner) – loose connective tissue with blood vessels, lymphatics, nerves.

Follicles – infoldings of germinal epithelium.



Fallopian Tubes (Oviducts)

Function: Conduct ovum/zygote to uterus

peristalsis.

Length: ~10–12 cm.

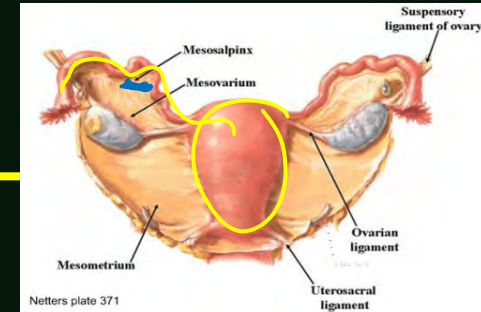
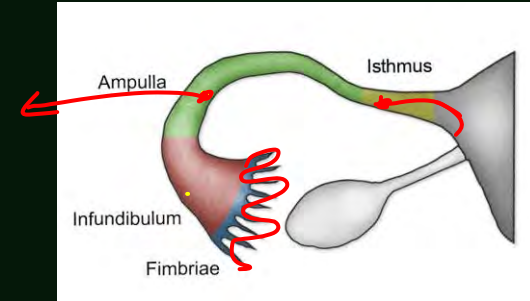
Parts:

Infundibulum – funnel-shaped, near ovary; has fimbriae (finger-like projections to collect ovum).

Ampulla – wide central part; site of fertilization (ampulla–isthmus junction).

Isthmus – narrow lumen, connects to uterus.

Attachment: Connected to abdominal wall by mesosalpinx (peritoneal fold).

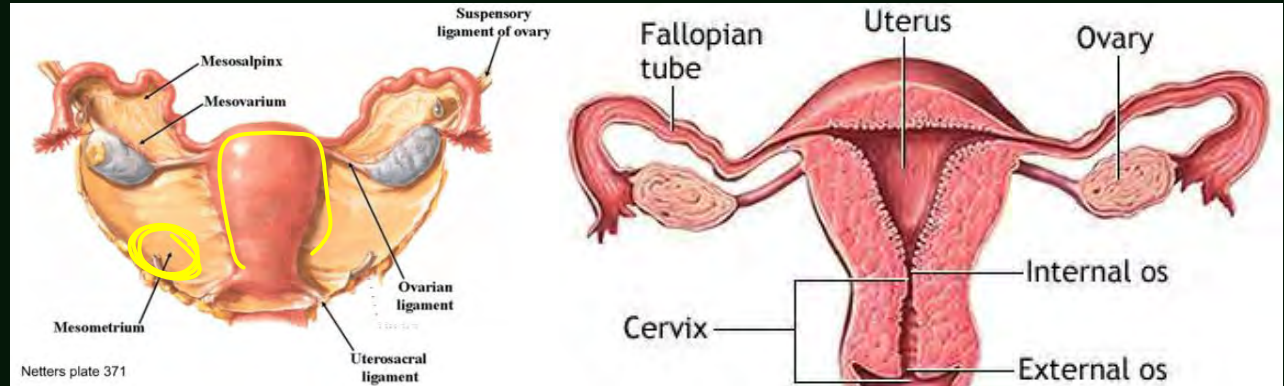


Uterus

Structure: Single, large, muscular, vascular, inverted pear-shaped organ. ✓

Location: In pelvis, between urinary bladder and rectum.

Attachment: To abdominal wall by mesometrium (peritoneal fold).

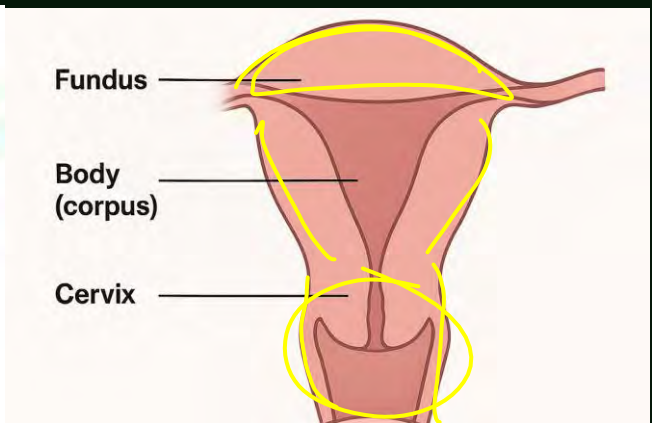
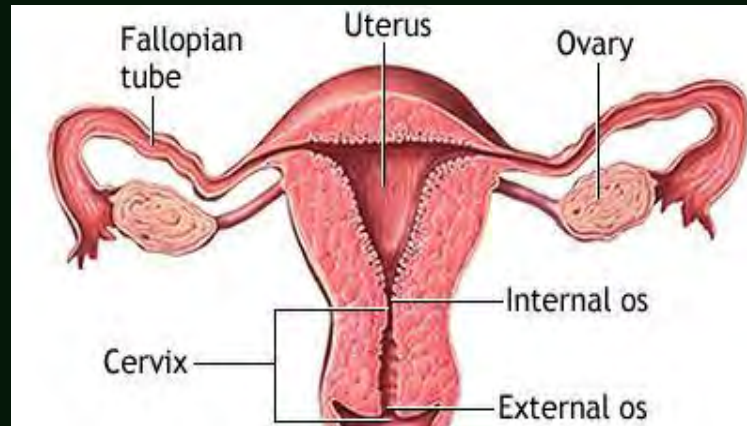


Regions:

Fundus - upper dome-shaped.

Body (Corpus) - central major part.

Cervix - lower narrow part opening into vagina.

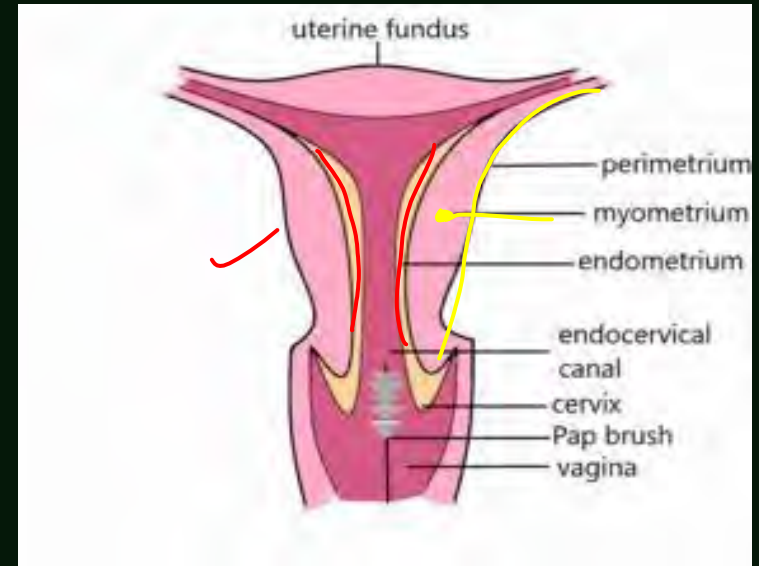


Uterine wall layers:

Perimetrium - outer thin membrane.

Myometrium - middle thick smooth muscle, shows strong **parturition contractions**.

Endometrium - inner glandular lining, undergoes **cyclic changes (menstrual cycle)**, site of implantation.



Vagina

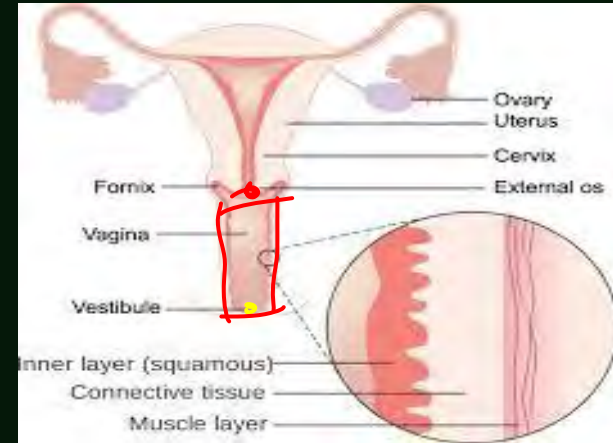
Fibro-muscular tube extending from cervix →
vestibule.

Opens into vestibule via vaginal
orifice.

Functions:

Serves as birth canal (with cervix).

Acts as copulatory organ.



Vulva (External Genitalia)

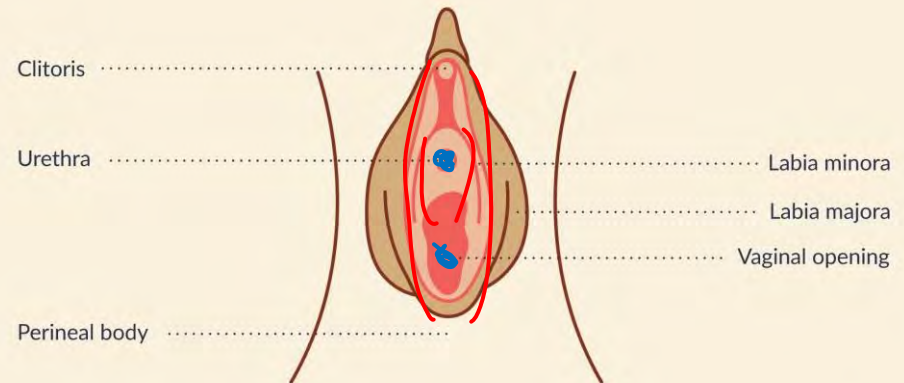
Components:

Vestibule – space between two labia minora
with two openings: Urethral orifice (upper).

Vaginal orifice (lower).

Vaginal orifice may be partially covered by hymen (mucous membrane).

outer lips
Labia Major
Inner lips
labia minor

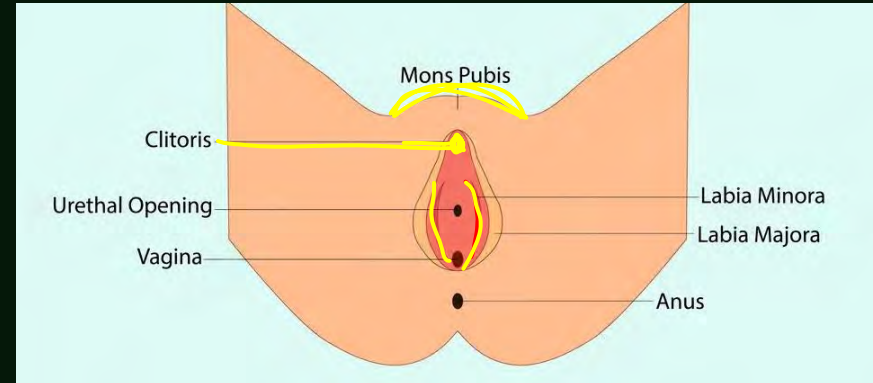


Labia minora – inner folds.

Labia majora – outer, larger folds of skin with pubic hair.

Clitoris – erectile, sensitive structure at upper junction of labia minora; homologous to penis.

Mons pubis – fatty tissue cushion above labia majora, covered with hair.

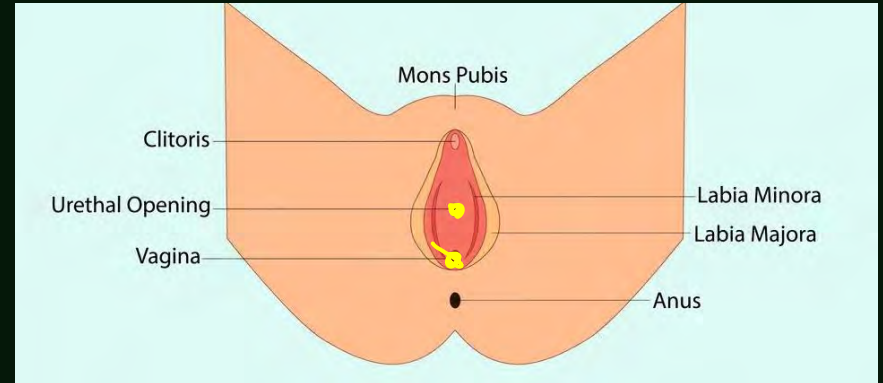


Accessory Reproductive Glands

Bartholin's (Greater vestibular glands)

Two, located posterior & lateral to vaginal opening.

Function: Secrete mucus for vaginal lubrication. Homologous to male bulbourethral glands.



Skene's glands

Around urethral opening, anterior vaginal wall.

Function: Lubrication during sexual stimulation.

Homologous to male prostate gland.

Mammary Glands

Structure:

Each breast = glandular tissue + fat.

15–20 lobes in each breast.

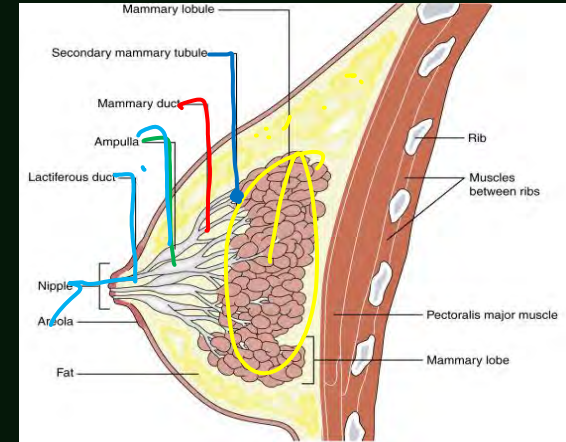
Lobe → alveoli (milk-secreting cells).

Alveoli → mammary tubules → ducts.

Ducts → widen into mammary ampulla

→ open into lactiferous ducts at nipple.

Lactocytes



What are multiple alleles? Describe multiple alleles with the help of ABO blood groups in man

1 - 2 Alleles
gene

Yellow
→ YY-Gr

Generally a gene has two alleles, one dominant and other recessive. Sometimes a gene may have more than two alleles which are called multiple alleles.

The number of **Genotypes** for multiple alleles can be known by the formula $n(n+1)/2$, where n is the number of alleles.

$$\frac{n(n+1)}{2}$$

To Find Genotypes For Multiple Alleles

$$\frac{A \ B \ O}{3} = \frac{n(n+1)}{2}$$

$$= \frac{3(3+1)}{2}$$

$$= \frac{3(4)}{2} = \frac{12}{2} = 6$$

$$A \rightarrow \begin{matrix} I^A I^A \\ I^A I^O \end{matrix} - 2$$

$$B \rightarrow \begin{matrix} I^B I^B \\ I^B I^O \end{matrix} - 2$$

$$AB \rightarrow I^A I^B - 1$$

$$O \rightarrow I^O I^O - 1$$

• The 6 genotypes are as follows:

(i) $I^A I^A$ (ii) $I^A I^O$ (iii) $I^B I^B$ (iv) $I^B I^O$ (v) $I^A I^B$ (vi) $I^O I^O$ = 6

Handwritten notes: A checkmark above (i), a checkmark above (ii), a checkmark above (iii), a checkmark above (iv), 'AB' above (v), and 'O' above (vi). Underlines are present under (i), (ii), (iii), (iv), (v), and (vi). A '1' is written below (v) and (vi).

• The four phenotypes are as follows:

(i) $I^A I^A$ and $I^A I^O$ are A group blood ✓

(ii) $I^B I^B$ and $I^B I^O$ are B group blood ✓

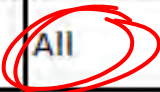
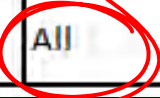
(iii) $I^A I^B$ are AB group blood ✓

(iv) $I^O I^O$ are O group blood ✓

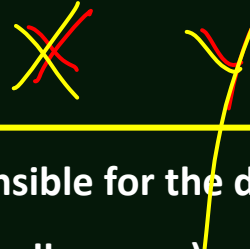
ABO Blood Groups:

The **four blood groups A, B, AB, O types** are characterized by the presence or absence of antigens on the plasma membrane of RBCs.

Blood group	Antigen(s) present on the red blood cells	Antibodies present in the serum	Genotype(s)
✓ A	A A antigen	B Anti-B	AA or AO
<u>B</u>	<u>B antigen</u>	A Anti-A	BB or BO
AB	A antigen and B antigen	None	AB
O	None	Anti-A and Anti-B	OO

Blood Group	Gives to these groups	Receives from these groups
✓  O ⁻	 All	O ⁻ only
 O ⁺	AB ⁺ , A ⁺ , B ⁺ , O ⁺	O ⁻ and O ⁺
 A ⁻	AB ⁻ , AB ⁺ , A ⁺ , A ⁻	O ⁻ and A ⁻
 A ⁺	AB ⁺ and A ⁺	O ⁻ , O ⁺ , A ⁻ , A ⁺
 B ⁻	B ⁻ , B ⁺ , AB ⁻ , AB ⁺	O ⁻ and B ⁻
 B ⁺	B ⁺ and AB ⁺	O ⁻ , O ⁺ , B ⁻ , B ⁺
 AB ⁻	AB ⁻ and AB ⁺	O ⁻ , A ⁻ , B ⁻ , AB ⁻
 AB ⁺	AB ⁺ only	 All

Describe the Chromosomal theory of sex determination



In most of the animals, a pair of chromosomes are responsible for the determination of sex.

These two chromosomes are called sex chromosomes (or allosomes).

The two different chromosomes in the pair are X-chromosome & Y-chromosome.

There are two types of sex chromosomal mechanism methods:

Male Heterogametic method

Female Heterogametic method

Male Heterogametic method: In this type, the female has two 'X' chromosomes and the male has only a single 'X' chromosome.

This is of two types:

(1) XX-XO (2) XX-XY

XX-XO type:

It is found in grasshoppers.

Karyotype of female is XX and karyotype of male is XO.

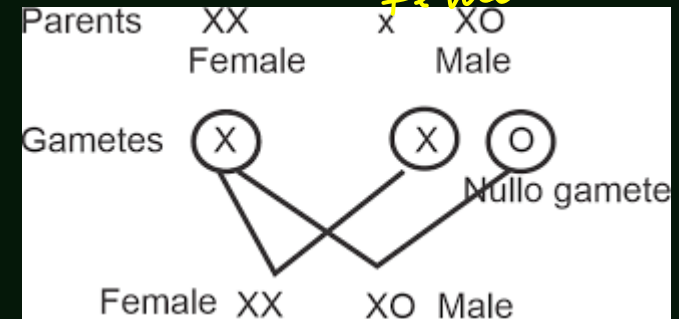
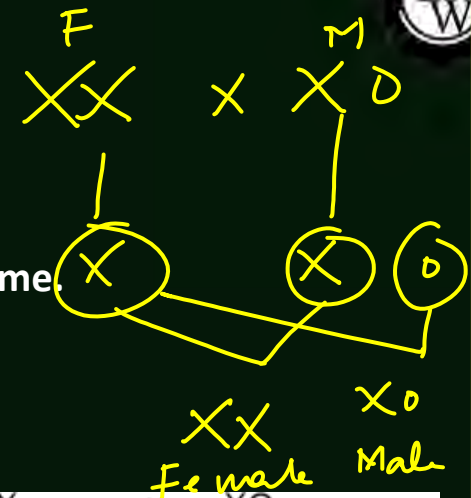
Females have two X chromosomes and males have only one X chromosome.

Unpaired X chromosome determines the sex of offspring.

The sex of the offspring is decided by the sperm that fertilizes the ovum.

If AX sperm is fertilised the child will be female.

If AO sperm is fertilised the child will be male.



XX–XY type:

It is found in human beings.

Female is homogametic with karyotype XX.

Male is heterogametic with karyotype XY.

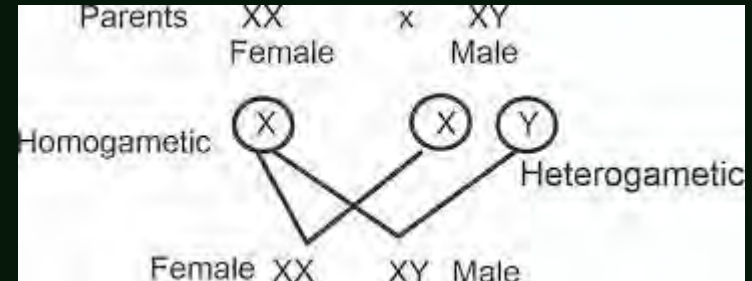
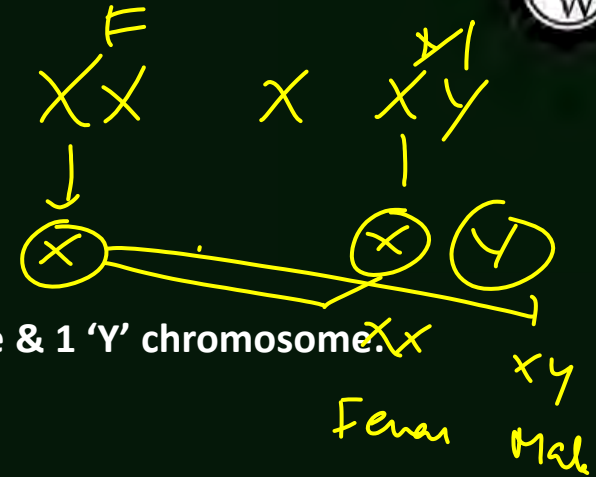
Females have 2 'X' chromosomes & males have 1 'X' chromosome & 1 'Y' chromosome.

'Y' chromosome determines the sex of offspring

The sex of the offspring is determined by the sperm that fertilizes the ovum.

If AX sperm is fertilized the child will be female

If AY sperm is fertilized the child will be male XY.



Female heterogametic Method: In this type, female produces two types of ova and male produces only one type of sperms.

This is of two types:

(1) ^{F M}
ZO-ZZ type ✓

(2) ZW-ZZ type ✓

↓
F male

ZO-ZZ type:

It is found in *Bombus* (bees).

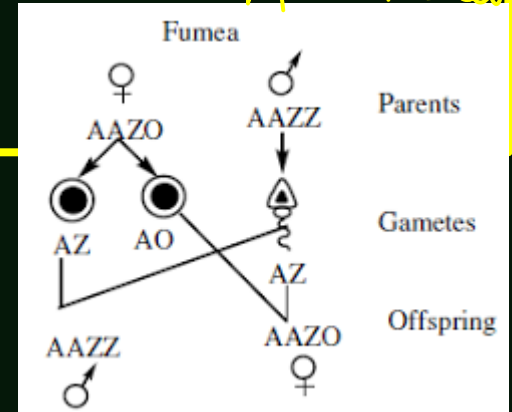
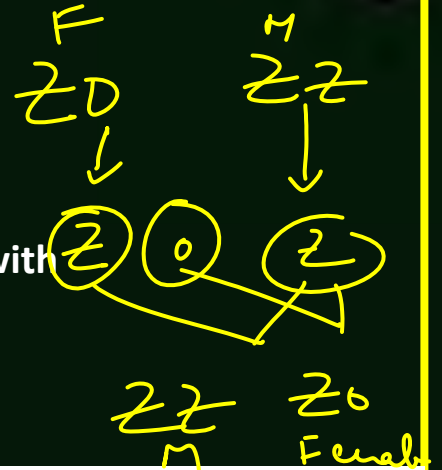
Karyo type of female is ZO and karyo type of male is ZZ.

Female is heterogametic with one Z chromosome and male is homogametic with two Z chromosomes.

The sex of the offspring is determined by the ovum that is fertilized.

If AO ovum is fertilized, it will be female. ✓

If AZ ovum is fertilized, it will be male. ✓



Female

ZW-ZZ type:

It is found in birds.

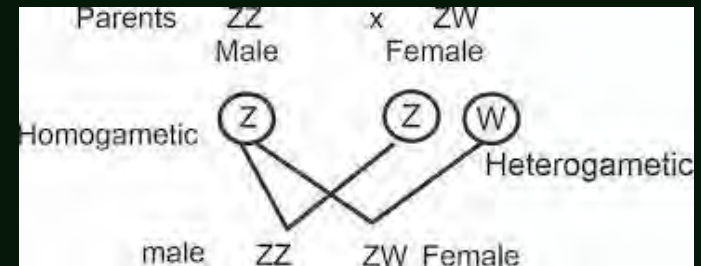
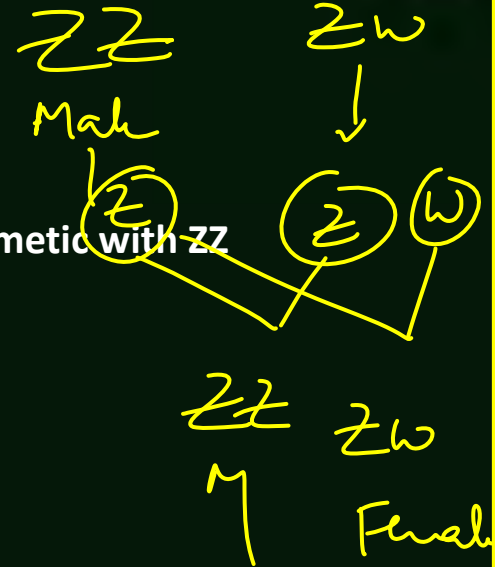
Karyo type of female is ZW and karyo type of male is ZZ.

Female is heterogametic with Z and W chromosomes and male is homogametic with ZZ chromosomes

The sex of the offspring is determined by the ovum that is fertilized.

If AZ ovum is fertilised it will be male.

If AW ovum is fertilised it will be female.



X linked

What is crisscross inheritance? Explain the inheritance of one sex linked recessive character in human beings.

Y/Y Y/y

T. H. Morgan observed sex linked inheritance in *Drosophila melanogaster*.

The X-linked recessive character present in a man is inherited to his grandson through his daughter.

This type of inheritance is called criss-cross inheritance.

Two best examples of criss-cross inheritance are Colour blindness and Haemophilia

- ✓ Every gene whether dominant or recessive is expressed phenotypically.
- ✓ Males have only one X chromosome.
- ✓ Males are more prone to X-linked recessive gene expression.
- ✓ Females have two X chromosomes. There are more than 50% of chances of having a dominant gene.
- ✓ So females are less prone to X-linked recessive gene expression.

X
M

XX
F

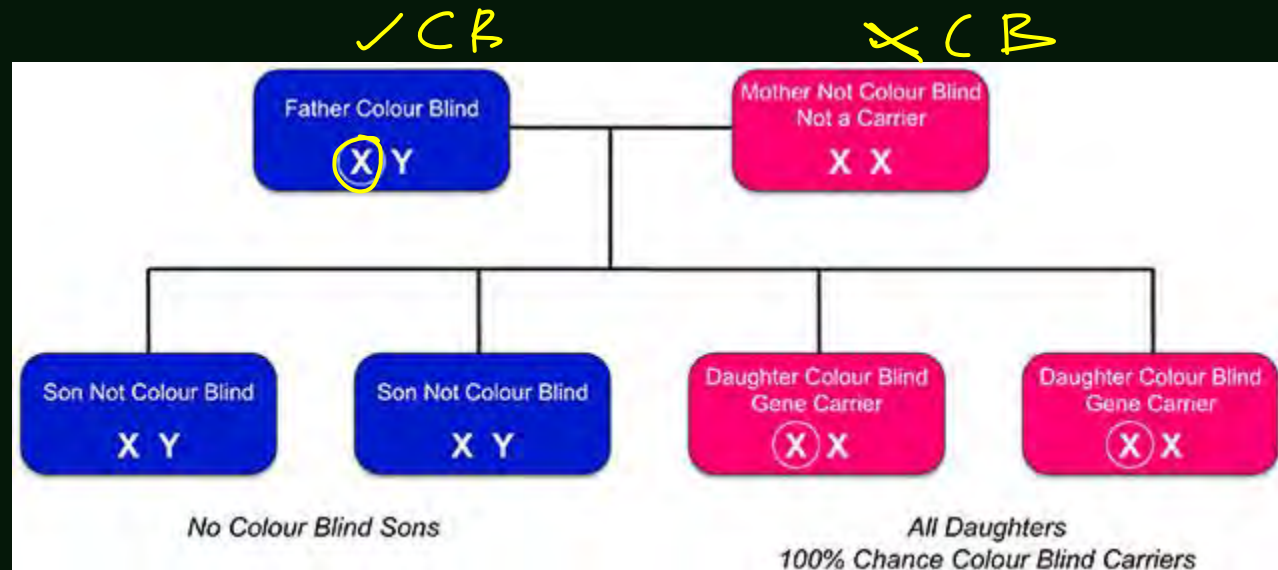
Colour Blindness:

The retina of eye is made up of rods and cone cells.

The cone cells are responsible for colour vision.

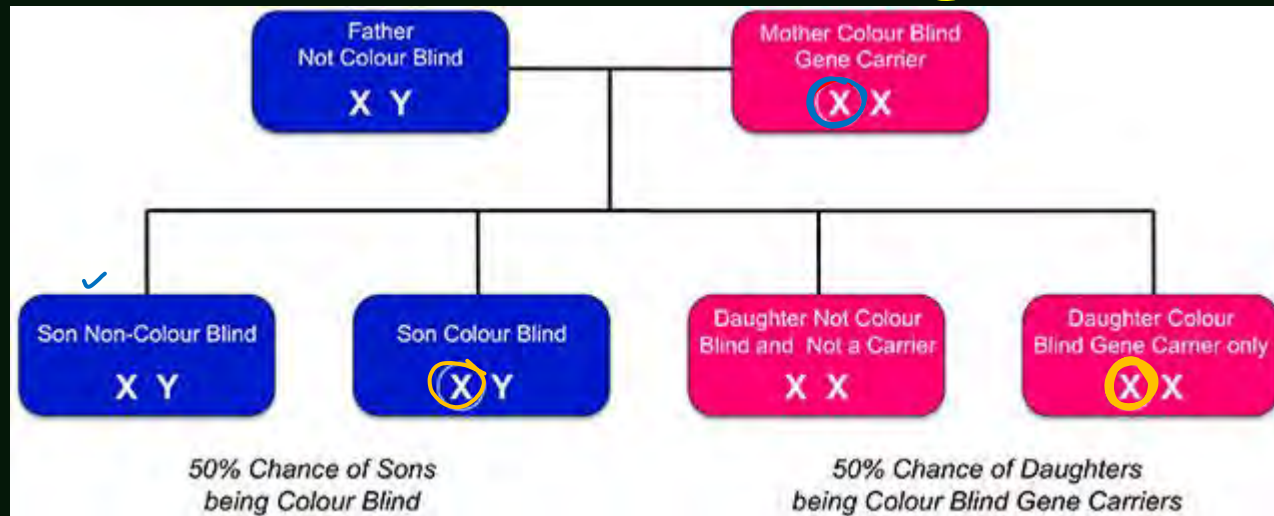
A recessive gene present on X chromosome causes colour blindness (Red–green).

When a woman with normal vision marries a 'colour-blind' man all the daughter will be the carriers. with normal vision. Hence all the sons have normal vision.



When the carrier daughter marries a man with normal colour vision, 50% of sons gets colour blindness. All others will have normal vision, of which 50% of daughters are carriers.

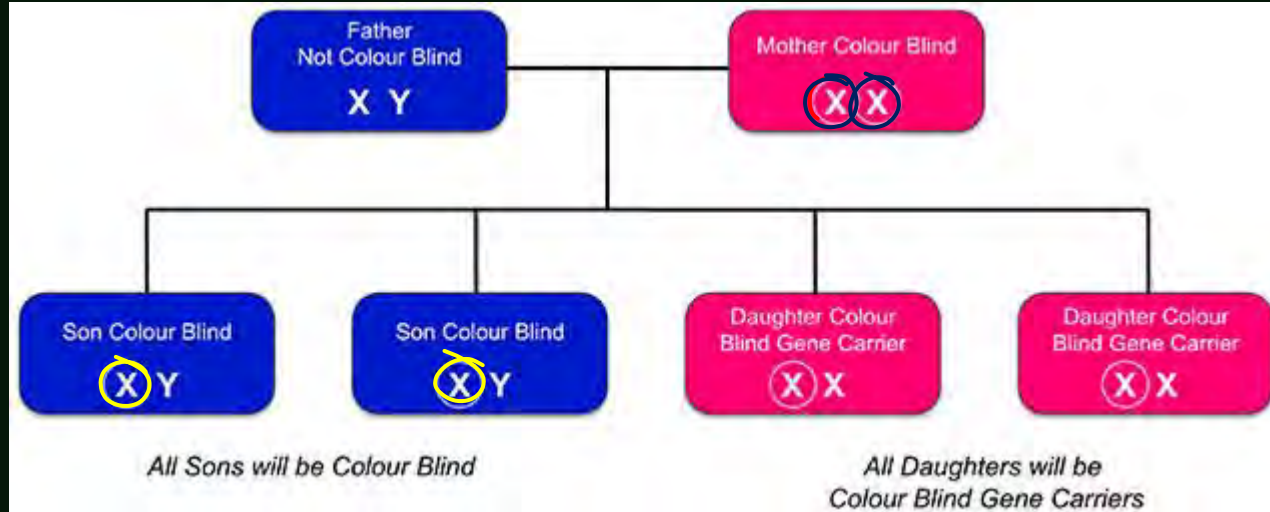
50%
Sons
CB



Carrier CB Mother

50%
Carrier

- When mother is colour blind and father has normal vision, all their sons inherit colour blindness from mother. All the daughters will be carriers because they get a dominant gene from father and recessive gene from mother.



100%
Sons
affected
CB

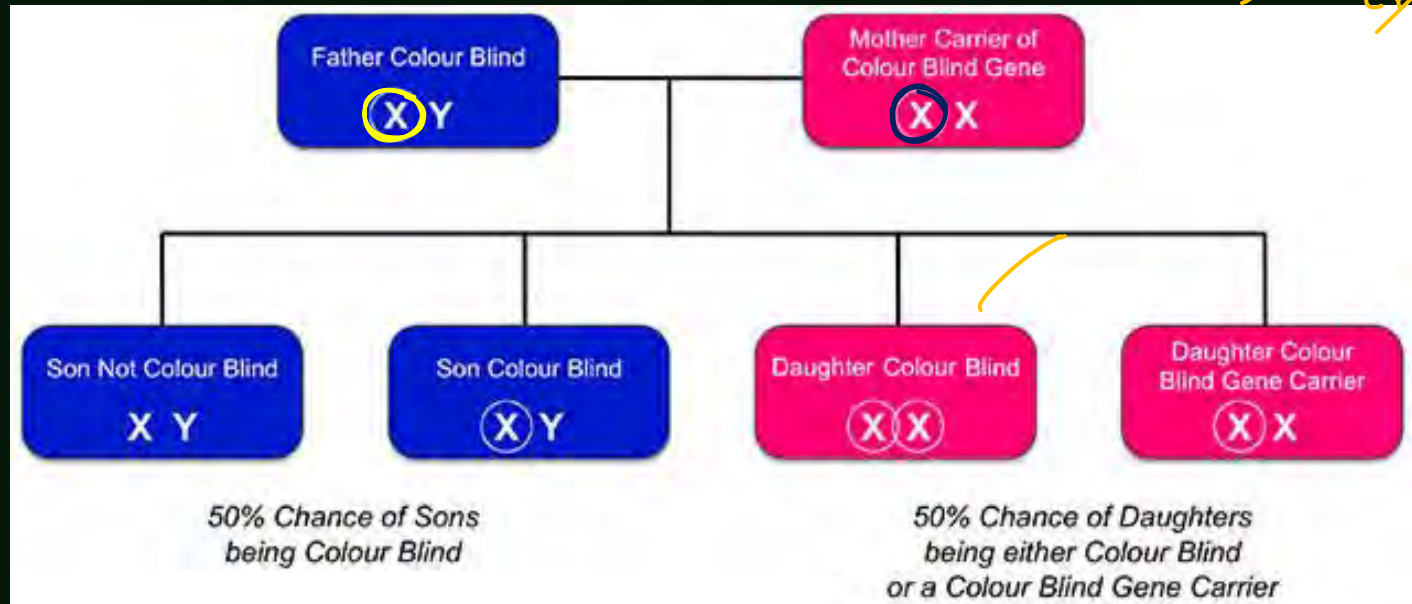
100%
Daughters
Carriers

- When mother is a carrier and father is colour blind, 50% of daughters and 50% of sons get colour blindness.

50/
Sons
CB

50/ Daughters
CB

50/



Write an essay on common genetic disorders.

Common genetic disorders: These disorders are associated with genes or chromosomes.

They are divided into two categories.

Mendelian disorders ✓

Chromosomal disorders ✓

Haemophilia:

- i) It is a bleeder's disease. ✓
- ii) Blood doesn't clot easily when a wound occurs accidentally. ✓
- iii) Haemophilia A is due to deficiency of clotting factor VIII.
- iv) Haemophilia B is due to deficiency of clotting factor IX.
- v) Haemophilia is dangerous to women and rarely occurs in them because it needs two recessive genes in females. It is common in men.

HA - VIII
HB - IX

Sickle cell anaemia:

It is an autosomal recessive metabolic disorder caused by a defective gene on chromosome 11.

It is expressed in homozygous condition.

Symptoms: Physical weakness, pain, organ damage due to internal bleeding and even paralysis.



Phenyl Ketounria:

It is autosomal metabolic disorder due to the gene located on 12th chromosome. ✓

Accumulation of phenyl ketone causes mental retardation..

PA

Absence

(or)

Mutation of

PAH

Cystic fibrosis:

It is **autosomal recessive disorder**. The affected gene is present on the **long arm chromosome of 7** that influences salt and water movement.

Symptoms:

- i) Increased sodium and chloride in sweat.
- ii) The mucus become viscous and sticky.
- iii) Mucus builds up in lungs, pancreas, alimentary canal etc.,
- iv) The patient may die before reaching age of five if untreated.

CFTR

Thalassemia is an autosomal recessive genetic disorder of haemoglobin synthesis in which the production of globin chains is reduced or absent, leading to anaemia.

Chromosomal disorders: ✓

These disorders are caused either by addition or deletion of a chromosome or accidental exchange of chromosomal segments (non-disjunction).

When a chromosome is deleted from a pair, it is monosomy.

When a chromosome is added to a pair, it is trisomy..

$$2n = 46 \quad 47 \quad \times \quad XY = 46$$

$$2n+1 = 47$$

Disorder	Chromosomal Change	Karyotype / Pair
Klinefelter's syndrome ✓	Trisomy (sex chromosome)	47, XXY (23rd pair, male)
Turner's syndrome ✓	Monosomy (sex chromosome)	<u>45</u> , XO (23rd pair, female)
<u>Down syndrome</u>	Trisomy	21st pair ✓
Edwards syndrome	Trisomy	18th pair ✓
Patau syndrome	Trisomy	13th pair ✓

Write an essay on DNA Finger Printing



Point	Details
Definition	DNA fingerprinting is a <u>method for identifying individuals by the particular structure of their DNA</u>
Also called	DNA typing
Basis	Differences in specific DNA sequences VNTR
DNA source	Blood, saliva, hair roots, semen, tissues, even burned tissues and bones
Similarity	More than 99% of DNA is identical in all humans
Uniqueness	No two people have the same DNA (except identical twins)
Key sequences	Variable Number Tandem Repeats (VNTRs)
Developed by	Jeffreys (myoglobin gene studies)

Step Name	Description
<u>Obtaining DNA</u>	DNA is isolated from <u>blood</u> , <u>saliva</u> , <u>hair roots</u> , <u>semen</u> etc.; copies made by cloning or amplification
Fragmentation of DNA	DNA cut into fragments using restriction endonucleases
Separation	DNA fragments separated by agarose gel electrophoresis; smaller fragments move faster
Denaturation	DNA strands separated using alkaline chemicals
Blotting ✓	DNA transferred to nylon membrane by Southern blotting
Probe binding	Radioactive probes bind to complementary DNA sequences
Hybridisation	Excess probe washed off after binding
Exposure to film	Autoradiography produces banding pattern (DNA fingerprint)

Forensic analysis	Identification of criminals
Medico-legal cases	Accurate paternity and maternity testing
Pedigree analysis	Study of inheritance across generations
Taxonomy	Study of phylogenetic relationships
Wildlife conservation	Maintaining DNA records of endangered species
Anthropological studies	Tracing origin and migration of humans

Thank

You