HUMAN REPRODUCTION

Focus Area Notes -2022 Exam

MALE REPRODUCTIVE SYSTEM

Male reproductive system includes

- a pair of testes,
- accessory ducts,
- accessory glands and
- external genitalia.

TESTES

- Testes is the organ in which sperms are produced.
- It is oval in shape with a length of about 4 to 5 cm and a width of about 2 to 3 cm .

Scrotum and its Importance

- The testes are situated outside the abdominal cavity within a pouch called scrotum.
- Scrotum maintains the temperature of the testes 2- 2.5 degree celsius lower than the normal internal body temperature which is necessary for spermatogenesis.

Testicular lobules

- Each testes has about 250 compartments called testicular lobules .

Seminiferous tubules

- Each testicular lobule contains one to three highly coiled tubules called seminiferous tubules . Sperms are produced here .

Three types of cells in testes

- Spermatogonial cells (Male germ Cells)
- Sertoli cells
- Interstitial cells or Leydig cells

Spermatogonial cells (Male germ Cells)

- The cells which lines the inner surface of seminiferous tubule is called spermatogonial cells.

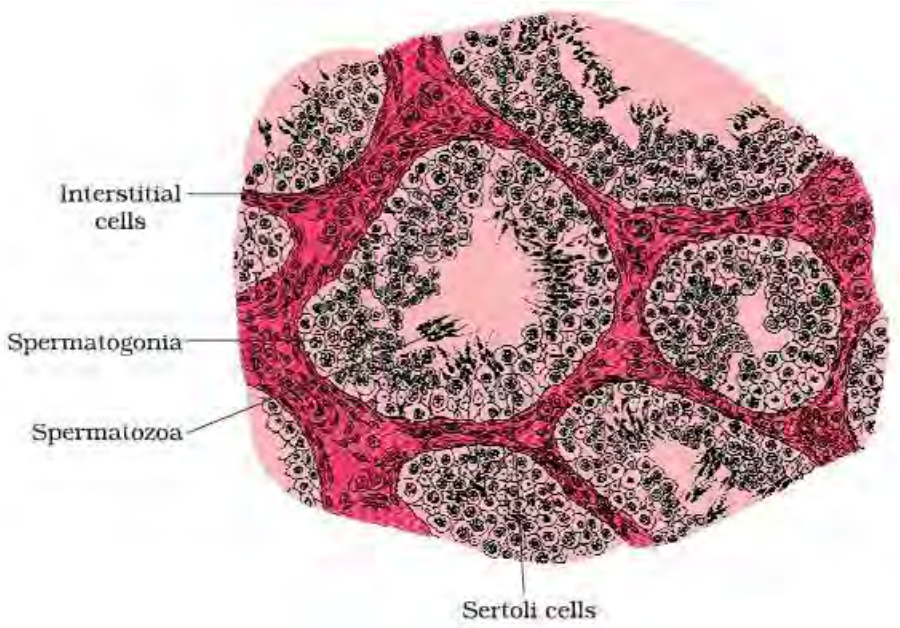
- These cells undergo meiotic division and transforms in to sperms .

Sertoli cells .

- These cells are also present in the inner surface of seminiferous tubule.
- Sertoli cells provide nutrition to the germ cells

Interstitial cells or Leydig cells

- These cells synthesise and secrete the hormones called androgens.



The male sex accessory ducts

The male sex accessory ducts include

- rete testis ,
- vasa efferentia ,
- epididymis and
- vas deferens.
- Accessory ducts store and transport the sperms from the testis to the outside through urethra.
- The urethra (ejaculatory duct) originates from the urinary bladder and extends through the penis to its external opening called urethral meatus

F**low chart of conduction of sperms through accessory ducts**

seminiferous tubules ⟶ Rete testis ⟶ Vasa efferntia ⟶ Epididymis

Vas deferens ⟶ urethra ⟶ urethral meatus

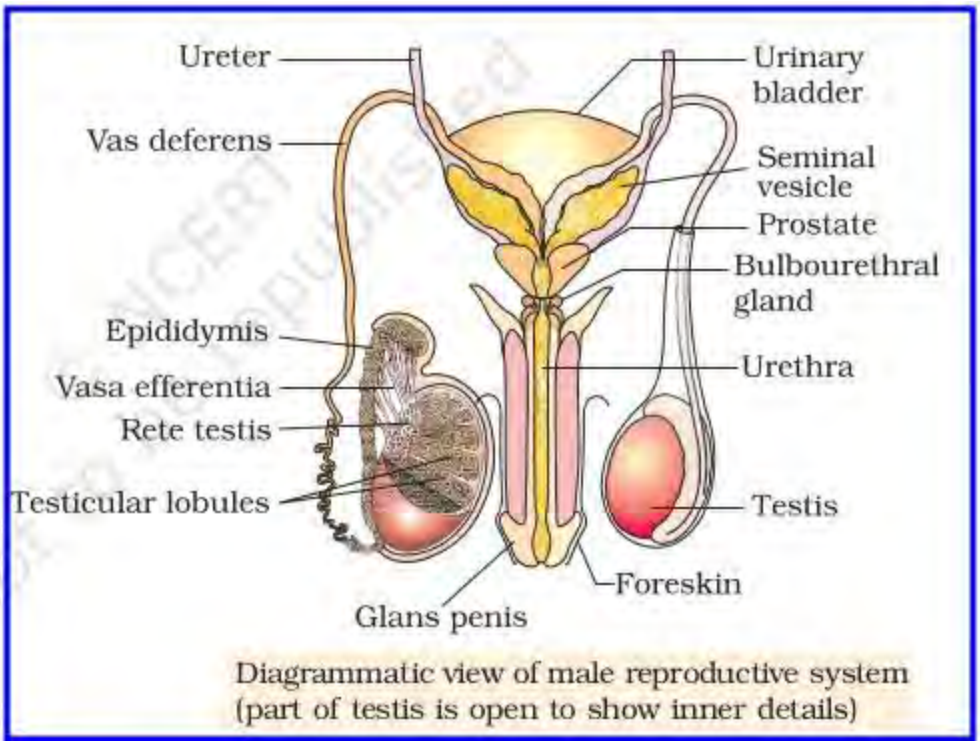
Penis

- **The penis is the male external genitalia.**
- **It is made up of special tissue that helps in erection of the penis to facilitate insemination.**
- **The enlarged end of penis called the glans penis is covered by a loose fold of skin called foreskin.**

The male sex accessory glands

The male sex accessory glands include paired

- **paired seminal vesicles ,**
- **a prostate gland**
- **paired bulbourethral glands .**
- **Secretion of these glands constitute the seminal plasma which is rich in fructose , calcium and certain enzymes .**
- **The secretion of bulbourethral glands also help in the lubrication of the penis**



Diagrammatic view of male reproductive system (part of testis is open to show inner details)

THE FEMALE REPRODUCTIVE SYSTEM

The female reproductive system consists of

- **a pair of ovaries,**
- **a pair of oviducts,**
- **uterus,**
- **cervix,**
- **vagina and the**
- **external genitalia located in pelvic region.**

OVARY

- **Ovaries are the primary female sex organs that produce the female gamete (ovum) and several steroid hormones (ovarian hormones).**
- **Each ovary is about 2 to 4 cm in length and is connected to the pelvic wall and uterus by ligaments.**
- **The outer part of the ovary is called cortex and inner medulla .**

Oviducts (Fallopian tubes)

Oviduct has the following parts

- **Fimbriae**
- **Infundibulum**
- **Ampulla**
- **Isthmus**
- **The funnel shaped part closer to the ovary is the infundibulum.**
- **The finger like projections in the edges of the infundibulum is called fimbriae.**
- **Fimbriae help in collection of the ovum after ovulation.**
- **The wider part after infundibulum of the oviduct is called ampulla.**
- **The last part of the oviduct which joins the uterus is called isthmus.**

Uterus (womb)

The uterus is single and it is also called womb.

The shape of the uterus is like an inverted pear.

It is supported by ligaments attached to the pelvic wall.

Cervix.

The uterus opens into vagina through a narrow cervix.

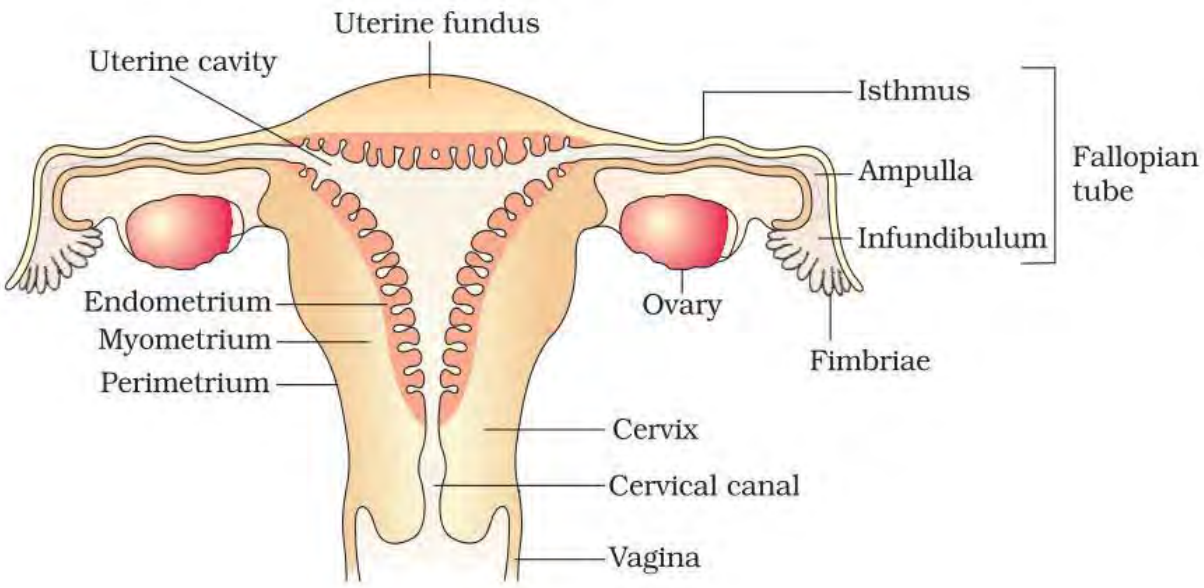
The cavity of the cervix is called cervical canal

Birth Canal

- **Cervical canal along with vagina forms the birth canal.**

Wall of Uterus

- **The wall of the uterus has three layers of tissue.**
- **Perimetrium**
- **Myometrium**
- **Endometrium**
- **Perimetrium is the external thin membrane .**
- **Myometrium is the middle thick membrane made up of smooth muscles , which exhibit strong contraction during delivery of the baby .**
- **Endometrium is the inner glandular layer that lines the uterine cavity Endometrium undergoes cyclical changes during menstrual cycle .**



FEMALE EXTERNAL GENITALIA

The female external genitalia includes

- mons pubis,
- labia majora,
- labia minora,
- hymen,
- and clitoris.
- **Mons pubis** is a cushion of fatty tissue covered by skin and pubic hair .
- **The labia majora** are fleshy folds of tissue , which extend down from the mons pubis and surround the vaginal opening .
- **The labia minora** are paired folds of tissue under the labia majora .
- **Hymen** is the thin ring of tissue partially covering the vaginal opening.
- **Clitoris** is the tiny finger like structure which lies at the upper junction of the two labia minora above the urethral opening

The presence or absence of hymen is not a reliable indicator of virginity or sexual experience. Justfy

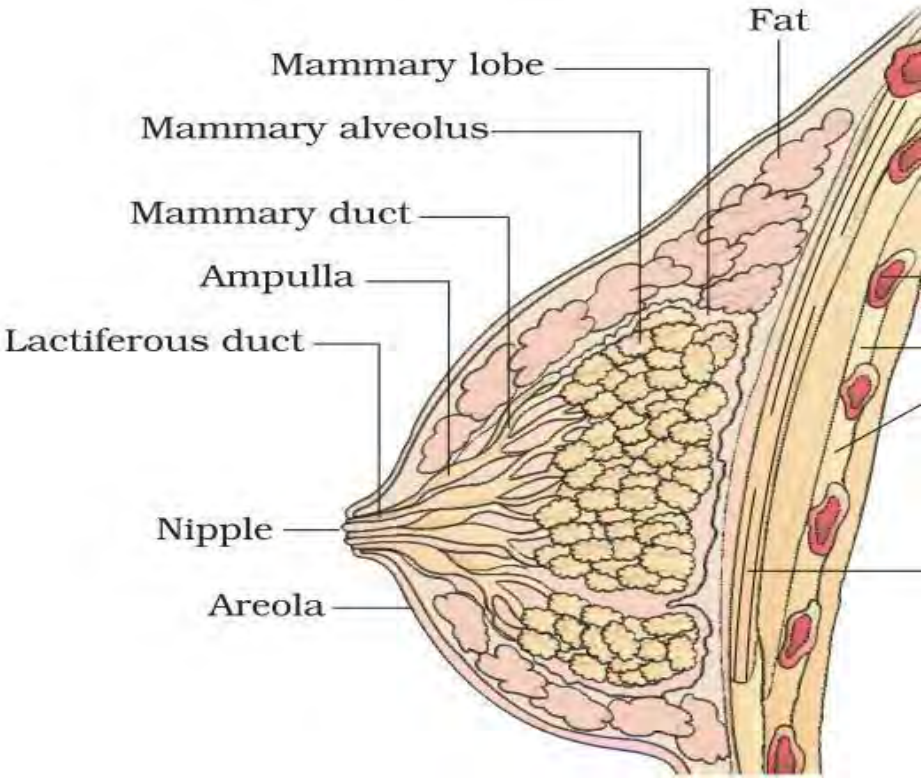
- The hymen is often torn during the first coitus (intercourse).
- It can also be broken by a sudden fall or jolt, insertion of a vaginal tampon, active participation in some sports like horseback riding, cycling, etc.
- In some women the hymen persists even after coitus.
- *So the presence or absence of hymen is not a reliable indicator of virginity or sexual experience*

MAMMARY GLANDS

- The mammary glands are paired structures (breasts) that contain glandular tissue and variable amount of fat.
- The glandular tissue of each breast is divided into **15-20 mammary lobes** containing **clusters of cells called alveoli.**
- The cells of alveoli secrete milk, which is stored in the cavities (lumens) of alveoli.
- The alveoli open into **mammary tubules.**
- The tubules of each lobe join to form a **mammary duct.**

- **Several mammary ducts join to form a wider mammary ampulla which is connected to lactiferous duct through which milk is sucked out.**

mammary lobes ⟶ mammary alveoli ⟶ mammary tubules ⟶ mammary duct ⟶ mammary ampulla ⟶ lactiferous duct



GAMETOGENESIS

- **Gametogenesis is the formation of male and female gametes .**
- **Formation of male gamete is spermatogenesis and female gamete is oogenesis.**

SPERMATOGENESIS

- **Spermatogenesis begins at puberty .**
- **The spermatogonial cells present on the inside wall of the seminiferous tubules multiply by mitotic division and increase in numbers .**
- **Each spermatogonium is diploid and contains 46 chromosomes .**

Primary spermatocytes

- **Spermatogonial cells synthesize cellular materials and stores food and grows. After growth it is known as primary spermatocytes.**

Secondary spermatocyte

- **Primary spermatocyte undergoes the first meiotic division and two haploid secondary spermatocyte are formed.(have 23 chromosomes)**

Spermatids

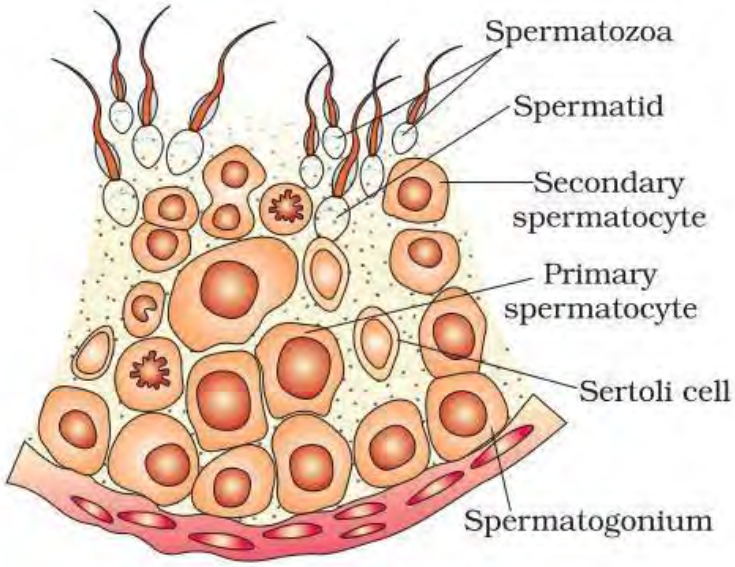
- **Secondary spermatocytes undergoes second meiotic division and four haploid spermatids are formed.**

Spermiogenesis

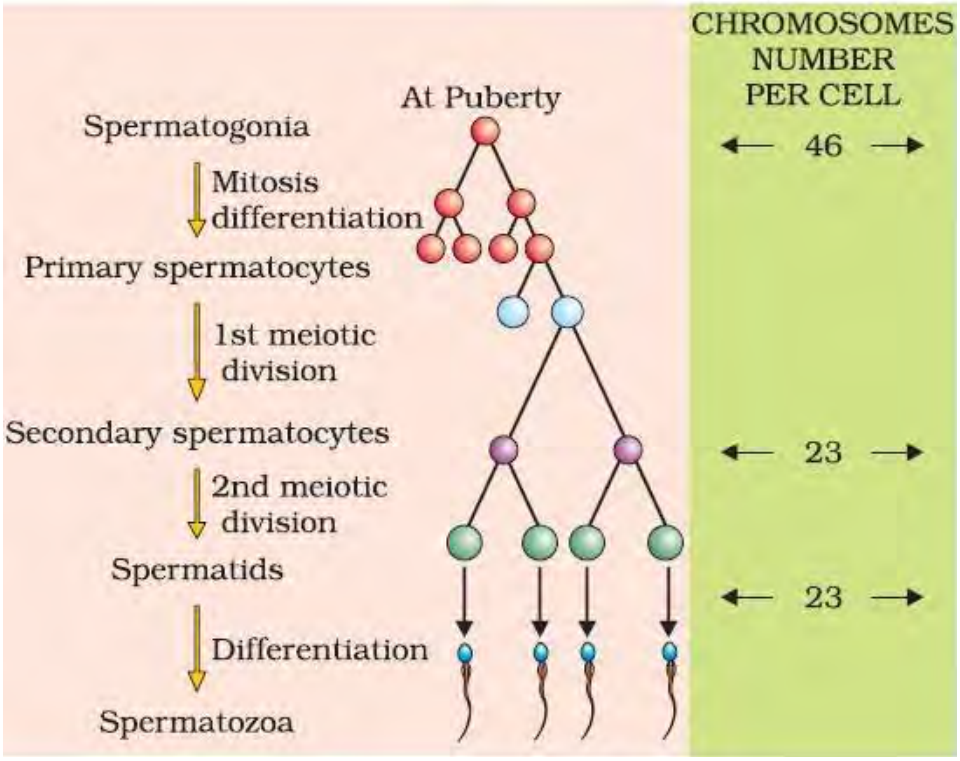
- **It is the transformation of spermatids in to functional spermatozoa or sperms.**

Spermiation

- **After spermiogenesis sperm heads become embedded in the sertoli cells ,and are finally released from the seminiferous tubules by the process called spermiation .**

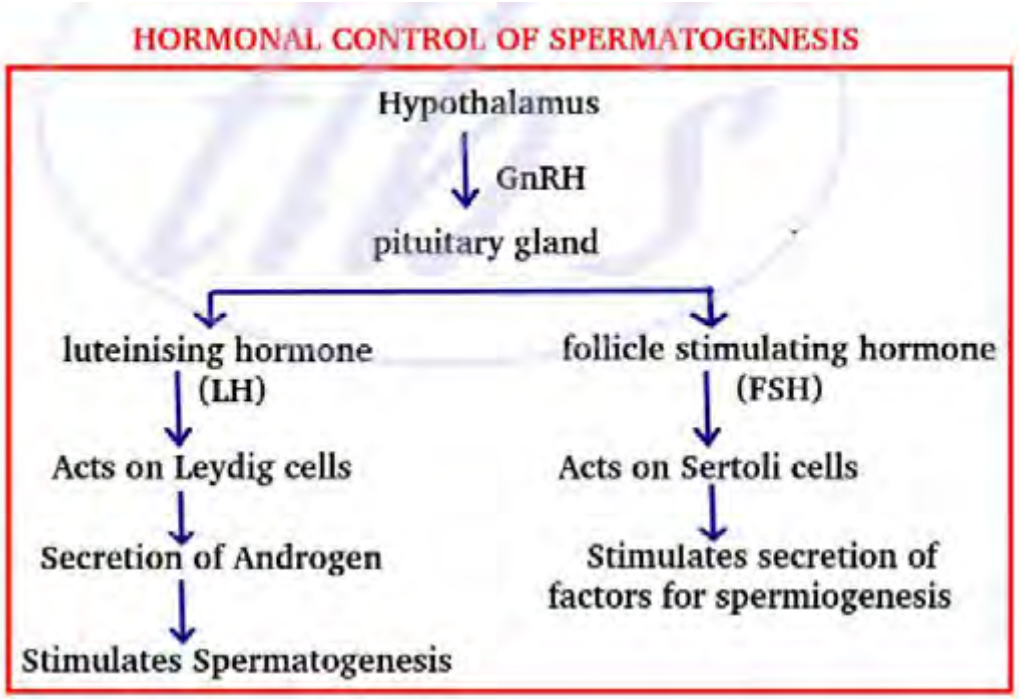


Diagrammatic sectional view of a seminiferous tubule (enlarged)



HORMONAL CONTROL OF SPERMATOGENESIS

- Spermatogenesis starts at the age of puberty due to significant increase in the secretion of gonadotropin releasing hormone (GnRH) from the hypothalamus .
- GnRH acts at the anterior pituitary and stimulate the secretion of luteinizing hormone (LH) and follicle stimulating hormone (FSH).
- LH acts at the Leydig cells and stimulate the synthesis and secretion of androgens .
- Androgens stimulate the process of spermatogenesis .
- FSH acts on sertoli cells and stimulate the secretion of some factors which help in the process of spermiogenesis.



Function of Acrosome

- The sperm head contains a cap-like structure called acrosome.
- The acrosome is filled with enzymes that help fertilisation of the ovum.

OOGENESIS

- The process of formation of a mature female gamete is called oogenesis.
- Oogenesis is initiated during the embryonic stage
- No more oogonia are formed and added after birth.

Primary oocytes

- Oogonial cells start division and enter into prophase-I of the meiotic division and get temporarily arrested at that stage, called primary oocytes.

Primary follicle

- Each primary oocytes gets surrounded by a layer of granulosa cells and is called the primary follicle.
- A large number of primary follicles degenerate during the phase from birth to puberty.
- At puberty only 60,000-80,000 primary follicles are left in each ovary.

Secondary follicles

- The primary follicles get surrounded by more layers of granulosa cells and a new theca and are called secondary follicles.

Teritiary follicles

- The secondary follicle transforms into a tertiary follicle.

Antrum

- The fluid filled cavity in tertiary follicle is called antrum.

Secondary oocyte

- The primary oocyte within the tertiary follicle grows in size and completes its first meiotic division and transforms into a large haploid secondary oocyte.
- Secondary oocyte retains bulk of the nutrient rich cytoplasm of the primary oocyte.

Polar body

- The functionless tiny cells formed during the unequal cell division of oogenesis is called polar body

Graafian follicle

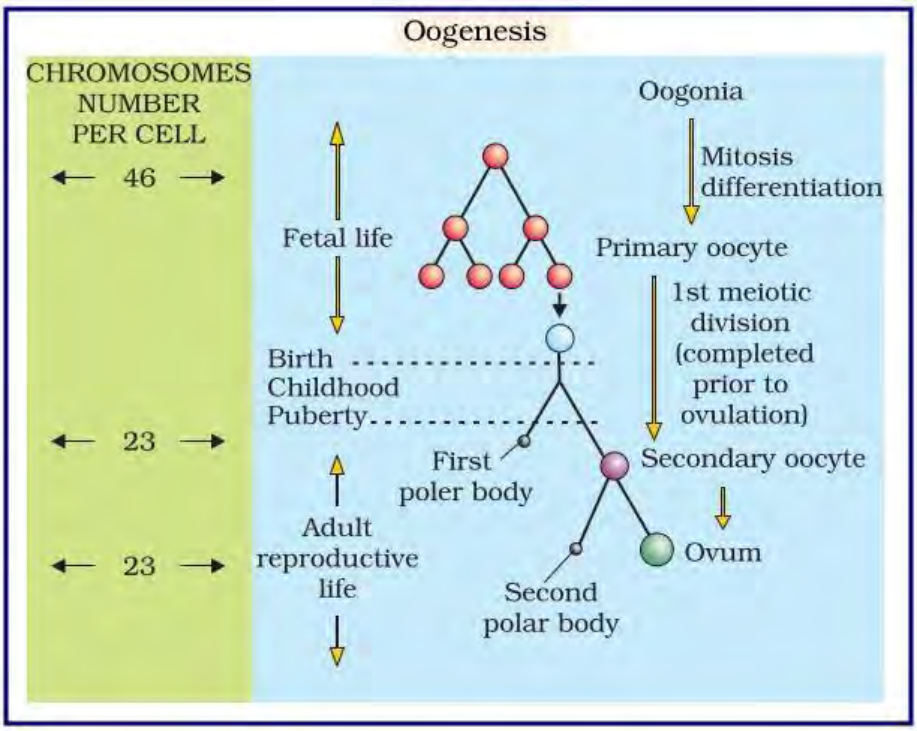
- The mature tertiary follicle is called Graafian follicle.

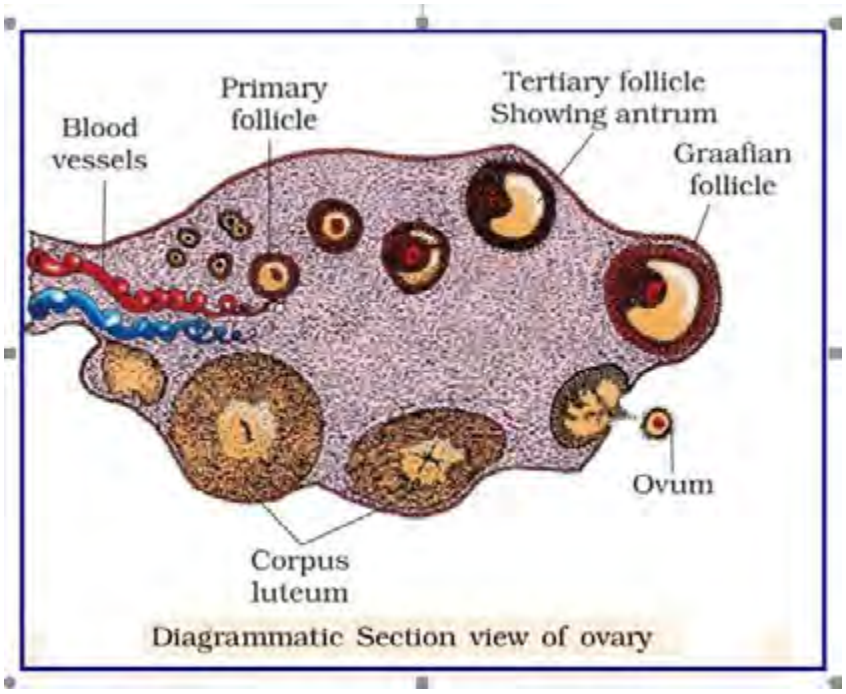
Zona pellucida

- The membrane covering secondary oocyte is called zona pellucida.

Ovulation

- The Graafian follicle ruptures to release the secondary oocyte (ovum).
- The process of release of ovum from the ovary is called ovulation





Menarche and Menopause.

- **The first menstruation begins at puberty and is called menarche.**
- **In human beings, menstrual cycles ceases around 50 years of age; that is termed as menopause.**

LH Surge

- LH and FSH secretion from pituitary increases and attains a peak level about 14 th day known as LH Surge).

Corpus luteum

- The remaining part of the Graffian follicles after ovulation transforms in to corpus luteum.
- The corpus luteum secretes large amounts of progesterone which is essential for the maintenance of the endometrium.

Fertilization

- The process of fusion of a sperm with an ovum is called fertilization .

Cleavage

- Cleavage is the repeated mitotic division of the zygote

Blastomeres

- Blastomeres are the cells formed during cleavage

Morula

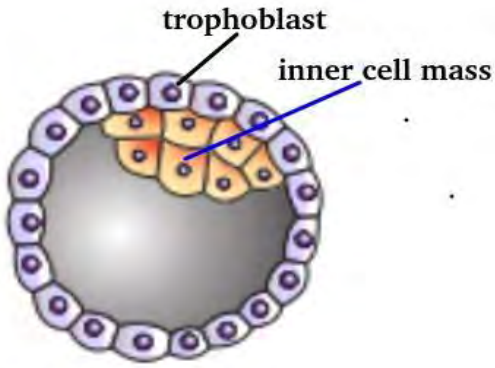
- The embryo with 8 to 16 blastomeres is called a morula

Blastocyst

- The morula continues to divide and transforms into blastocyst.
- The blastomeres in the blastocyst are arranged into an outer layer called trophoblast and an inner group of cells attached to trophoblast called the inner cell mass.

Fate of cells in blastocyst

- The trophoblast layer gets attached to the endometrium and the inner cell mass gets differentiated as the embryo.



(Blastocyst)

Placental Hormones

- **Placenta secretes**
- **Human chorionic gonadotropin (hCG),**
- **human placental lactogen (hPL),**
- **oestrogens,**
- **progesterone etc.**

Stem cells

- The inner cell mass of embryo contain certain cells called stem cells which have the potency to give rise to all the tissues and organs.

Major features of embryonic development at various months of pregnancy

Major features of embryonic development at various months of pregnancy

Month of Pregnancy	Features of embryonic development
First month	Heart sound noticed using stethoscope
Second month	develops limbs and digits
Third month /12 weeks (first trimester)	Major organ systems are formed
Fifth month	First movements of the foetus and appearance of hair on the head
Sixth month /24 weeks (second trimester)	body is covered with fine hair, eye-lids separate, and eyelashes are formed
Ninth month	Foetus is fully developed

Significance of Colostrum

- *Breast-feeding during the initial period of infant growth is recommended by doctors for bringing up a healthy baby...Why ?*
- **The milk produced during the initial few days of lactation is called colostrum which contains several antibodies absolutely essential to develop immune resistance for the new-born babies.**

REPRODUCTIVE HEALTH

Focus Area -2022 Exam

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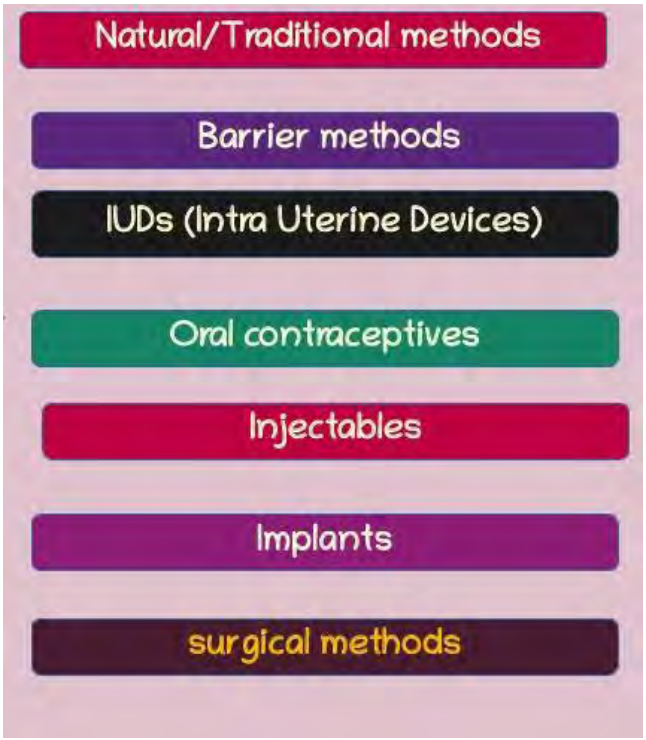
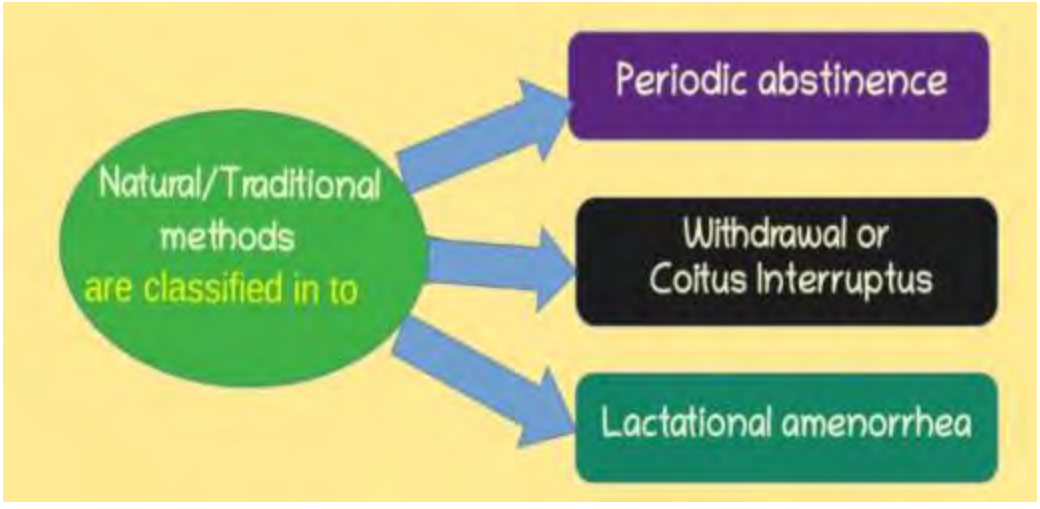
Classification of Contraceptives

- **Contraceptives can be classified into the following groups**



Natural Methods

- **Natural methods work on the principle of avoiding chances of ovum and sperms meeting.**



Periodic Abstinence

- In this method couples avoid or abstain from coitus from day 10 to 17 of the menstrual cycle when ovulation could be expected.
- As chances of fertilisation are very high during this period, it is called the fertile period.
- Therefore, by abstaining from coitus during this period, conception could be prevented.

Withdrawal/Coitus interruptus

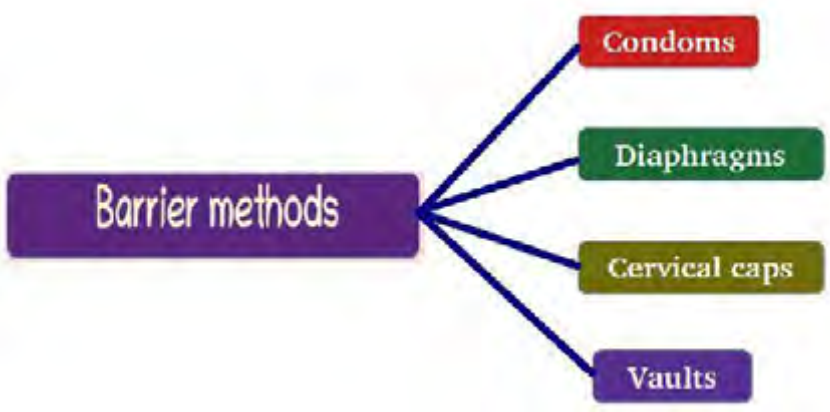
- In this method, the male partner withdraws his penis from the vagina just before ejaculation so as to avoid insemination.

Lactational amenorrhea

- Lactational amenorrhea (absence of menstruation) method is based on the fact that ovulation and therefore the cycle do not occur during the period of intense lactation following parturition.
- This method has been reported to be effective only upto a maximum period of six months following parturition.
- As no medicines or devices are used in these methods, side effects are almost nil.
- Chances of failure in this method are also high.

Barrier methods

- In barrier methods, ovum and sperms are prevented from physically meeting with the help of barriers.
- Such methods are available for both males and females.



Condoms

- Condoms are barriers made of thin rubber/latex sheath that are used to cover the penis in the male or vagina and cervix in the female, just before coitus so that the ejaculated semen would not enter into the female reproductive tract.
- 'Nirodh' is a popular brand of condom for the male.
- Condoms has the benefit of protecting the user from STIs and AIDS.

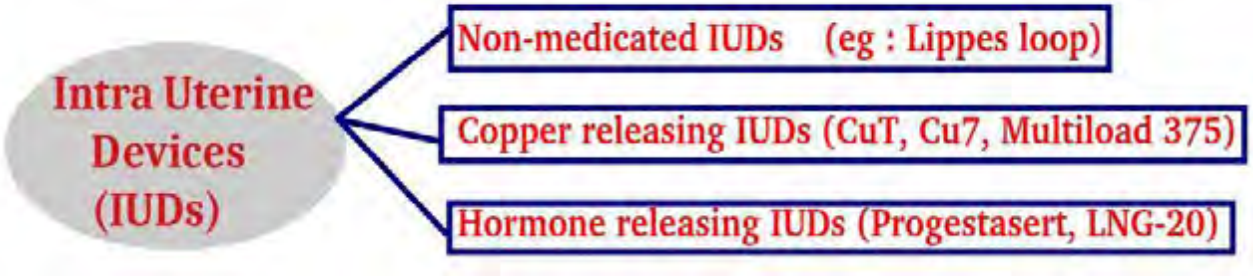
Diaphragms, cervical caps and vaults

- Diaphragms, cervical caps and vaults are also barriers made of rubber that are inserted into the female reproductive tract to cover the cervix during coitus.
- 'They prevent conception by blocking the entry of sperms through the cervix.
- They are reusable.

Intra Uterine Devices (IUDs)

- IUDs are devices inserted in the uterus through vagina with the help of doctors or expert nurses.

They are classified into



Mode of action of IUDs

- IUDs increase phagocytosis of sperms within the uterus and the Cu ions released suppress sperm motility and the fertilising capacity of sperms.

- The hormone releasing IUDs make the uterus unsuitable for implantation and the cervix hostile to the sperms.

Oral contraceptives

- Oral administration of progestogens or progestogen–oestrogen combinations in the form of tablets (pills) are called oral contraceptives.
- Pills are taken daily for 21 days starting within the first five days of menstrual cycle.
- After a gap of 7 days (during which menstruation occurs) it has to be repeated in the same pattern till the female desires to prevent conception.

Mode of action of Oral Contraceptives

- They inhibit ovulation and implantation as well as alter the quality of cervical mucus to prevent/retard entry of sperms.
- Pills are very effective with lesser side effects.

Example

- Saheli –the new oral contraceptive for the females contains a non-steroidal preparation.
- It is a 'once a week' pill with very few side effects and high contraceptive value.

injections or implants

- Progestogens alone or in combination with estrogen can also be used by females as injections or implants under the skin.
- Their mode of action is similar to that of pills and their effective periods are much longer.

Emergency Contraceptives

- Administration of progestogens or progestogen-estrogen combinations or IUDs within 72 hours of coitus have been found to be very effective as emergency contraceptives.
- They could be used to avoid possible pregnancy due to rape or casual unprotected intercourse.

Surgical methods

- Surgical methods, also called sterilisation, are generally advised for the male/female partner as a terminal method to prevent any more pregnancies.
- Surgical intervention blocks gamete transport and thereby prevent conception.

Vasectomy

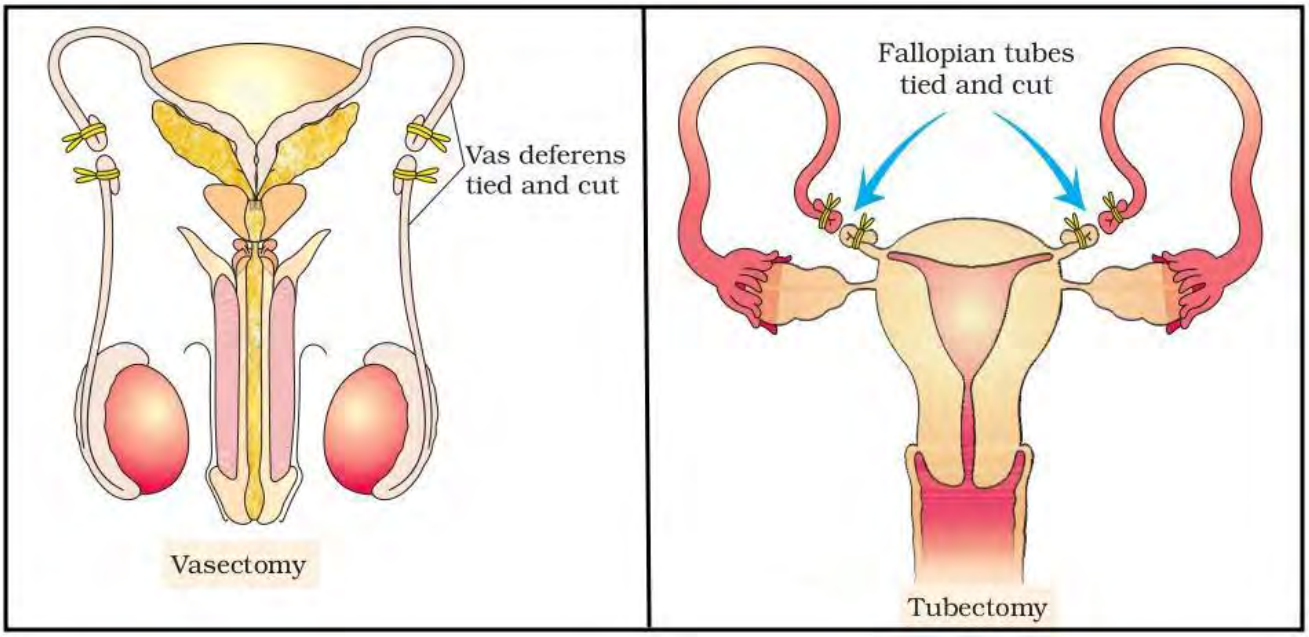
- Sterilisation procedure in male.
- In vasectomy, a small part of the vas deferens is removed or tied up through a small incision on the scrotum.

Tubectomy

- Sterilisation procedure in female.
- In tubectomy, a small part of the fallopian tube is removed or tied up through a small incision in the abdomen or through vagina.

Demerit of surgical method

These techniques are highly effective but their reversibility is very poor.



Possible ill- Effects of Contraceptives

- Nausea , Abdominal pain , Breakthrough bleeding, Irregular menstrual bleeding are some of the ill effects of contraceptives.

Sexually Transmitted Infections(STIs)

- Infections or diseases which are transmitted through sexual intercourse are collectively called
- sexually transmitted infections (STI) or
- venereal diseases (VD) or reproductive tract infections (RTI).

Examples -Sexually Transmitted Infections (STIs)

- Gonorrhoea,
- syphilis,
- genital herpes,
- chlamydiasis,
- genital warts,
- trichomoniasis,
- hepatitis-B and
- HIV infections leading to AIDS are some of the common STIs.

Mode of transmission of STIs

- Infections like hepatitis–B and HIV can be transmitted by sharing of injection needles, surgical instruments, etc., with infected persons,
- transfusion of blood, or from an infected mother to the foetus too.
- Except for hepatitis-B, genital herpes and HIV infections, other diseases are completely curable if detected early and treated properly.

Early Symptoms of STIs

- Early symptoms of most of these are minor and include
- itching,
- fluid discharge,
- slight pain, swellings, etc., in the genital region.
- Infected females may often be asymptomatic and hence, may remain undetected for long.

Later Symptoms of STIs

- Absence or less significant symptoms in the early stages of infection could lead to complications in the later stage like
- Pelvic Inflammatory Diseases (PID), abortions, still births, ectopic pregnancies, infertility or even cancer of the reproductive tract.

Preventive measures to avoid STIs

(i) Avoid sex with unknown partners/multiple partners.

(ii) Always try to use condoms during coitus.

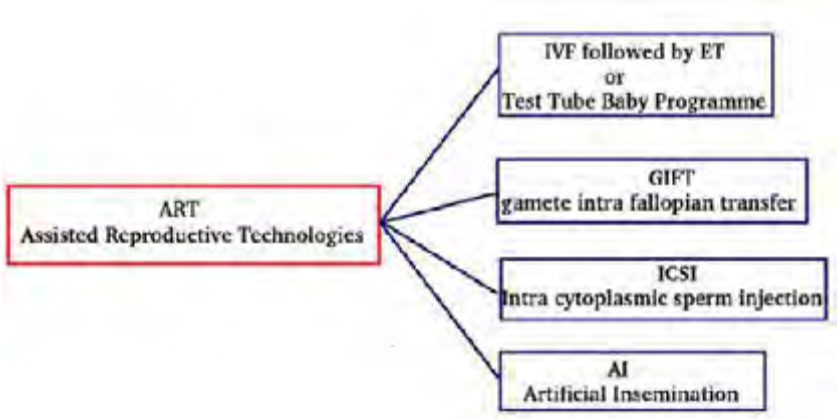
(iii) In case of doubt, one should go to a qualified doctor for early detection and get complete treatment if diagnosed with infection.

Infertility

- Inability to conceive or produce children even after 2 years of sexual co-habitation is called infertility .
- Specialised health care units (infertility clinics, etc.) could help in diagnosis and corrective treatment of some of these disorders and enable these couples to have children.

Reasons for Infertility

- The reasons for infertility may be many–physical,congenital, diseases, drugs, immunological or even psychological.
- If corrections in infertility clinics are not possible, the couples could be assisted to have children through certain special techniques commonly known as assisted reproductive technologies (ART).



In vitro fertilisation(IVF)

- Fertilisation outside the body in almost similar conditions as that in the body is known as In vitro fertilisation(IVF).
- In this method, popularly known as test tube baby programme, ova from the wife/donor (female) and sperms from the husband/donor (male) are collected and are induced to form zygote under simulated conditions in the laboratory.
- The zygote or early embryos is then transferred to fallopian tube or uterus.

ZIFT–zygote intra fallopian transfer

- The zygote or early embryos upto 8 blastomeres is transferred into the fallopian tube known as ZIFT–zygote intra fallopian transfer.

IUT – intra uterine transfer

- Early embryos with more than 8 blastomeres are transferred into the uterus known as IUT – intra uterine transfer.

In-vivo fertilisation

- Fusion of gametes within the female is known as in-vivo fertilisation.
- Embryos formed by in-vivo fertilisation is also used for embryo transfer to assist those females who cannot conceive.

GIFT – gamete intra fallopian transfer

- Transfer of an ovum collected from a donor into the fallopian tube of another female who cannot produce one, but can provide suitable environment for fertilisation and further development is GIFT – gamete intra fallopian transfer

Intra cytoplasmic sperm injection (ICSI)

- It is a method in which the sperm is directly injected into the ovum to form the zygote in the laboratory.

Artificial Insemination (AI)

- **Infertility cases either due to inability of the male partner to inseminate the female or due to very low sperm counts in the ejaculates, could be corrected by artificial insemination (AI) technique.**
- **In this technique, the semen collected either from the husband or a healthy donor is artificially introduced either into the vagina or into the uterus (IUI – intra-uterine insemination) of the female.**

PRINCIPLES OF INHERITANCE AND VARIATION

FOCUS AREA -2022 EXAM

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LAW OF DOMINANCE

(i) Characters are controlled by discrete units called factors.
(ii) Factors occur in pairs.
(iii) In a dissimilar pair of factors one member of the pair dominates (dominant) the other (recessive).

LAW OF SEGREGATION

- **This law states that during gamete formation, the two factors for a character present in an individual will separate and segregate with each other . So that a gamete receives only one of the two factors.**

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INCOMPLETE DOMINANCE

- **The type of inheritance in which the F 1 phenotype do not resemble either of the two parents but shows an intermediate character is called incomplete dominance.**

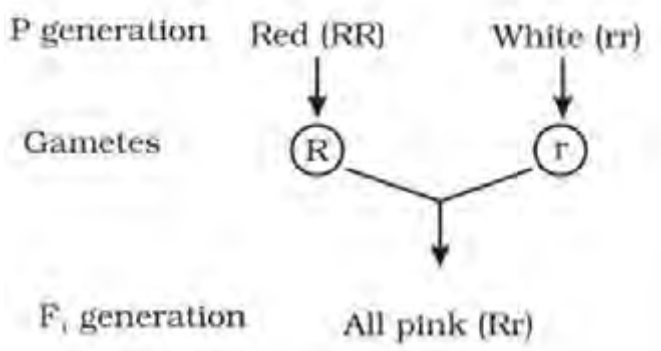
Example

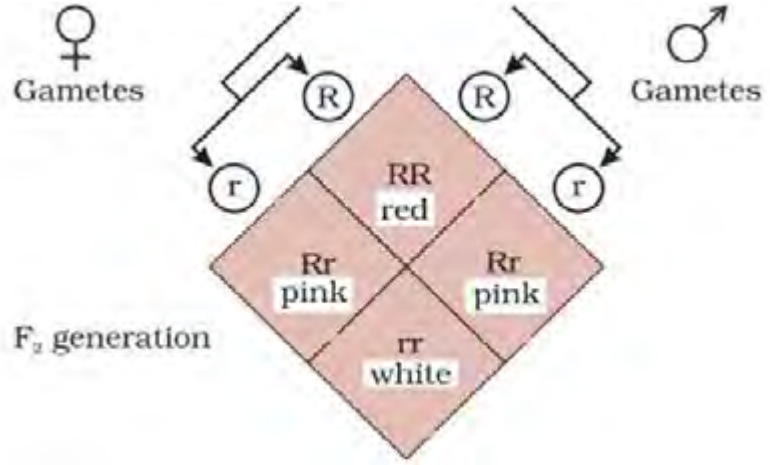
The inheritance of flower colour in the dog flower (snapdragon or Antirrhinum sp)

- **In a cross between homozygous red flowered plant (RR) with homozygous white flowered plant(rr) the F 1 (Rr) was pink instead of red.**
- **When F 1 was self fertilized the F 2 resulted the following ratio**

  1Red (RR) : 2Pink(Rr) : 1White.

- **Here the genotypic and phenotypic ratio is 1:2:1.**
- **This happens because R is not completely dominant over r. So Rr shows an intermediate pink colour**





Phenotypic ratio : 1 Red : 2 Pink : 1 White
Genotypic ratio : 1 RR : 2 Rr : 1 rr
Phenotypic ratio = Genotypic ratio
= 1 : 2 : 1

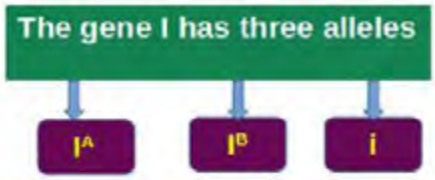
Incomplete dominance is a genetic phenomenon in which both Genotypic and phenotypic ratio are same ie, 1 : 2 : 1

CO –DOMINANCE

- **It is the phenomenon in which F 1 generation resembles both the parents.**

Eg : ABO- Blood group in human beings

- **The blood group is determined by the sugar polymers present on the plasma membrane of RBC and which is controlled by the gene I.**



Allele from Parent 1	Allele from Parent 2	Genotype of offspring	Blood types of offspring
I^A	I^A	I^AI^A	A
I^A	I^B	I^AI^B	AB
I^A	i	I^Ai	A
I^B	I^A	I^AI^B	AB
I^B	I^B	I^BI^B	B
I^B	i	I^Bi	B
i	i	ii	O

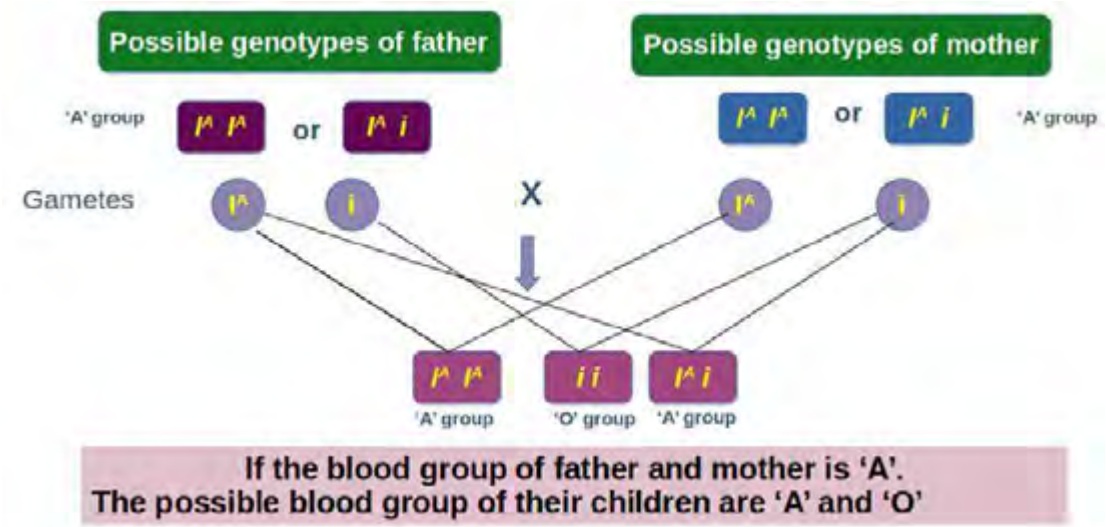
- The gene I has three alleles I^A , I^B & i.
- I^A and I^B produce a slightly different form of the sugar while allele i doesn't produce any sugar.
- When I^A and I^B are present together, they both express their own types of sugars. This is due to co-dominance.

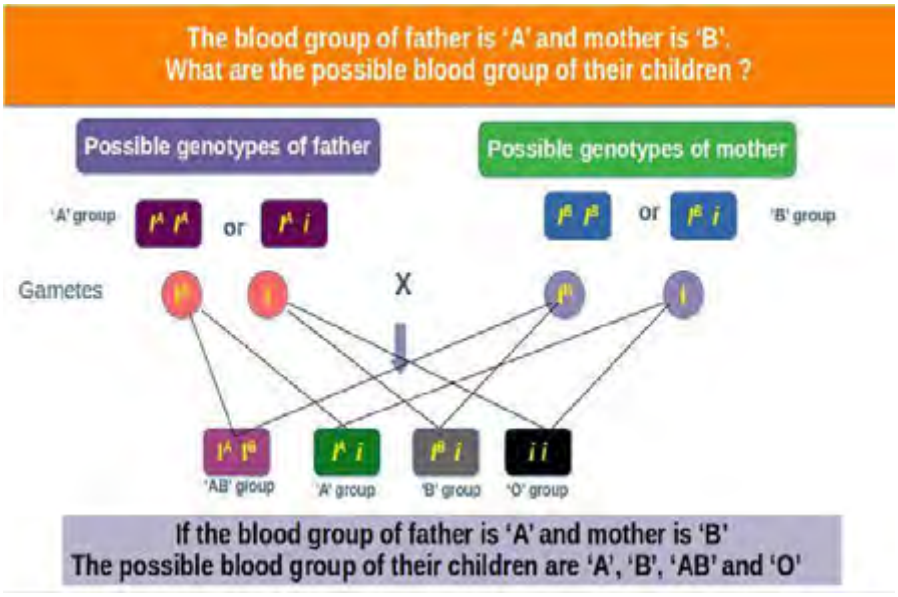
Multiple alleles

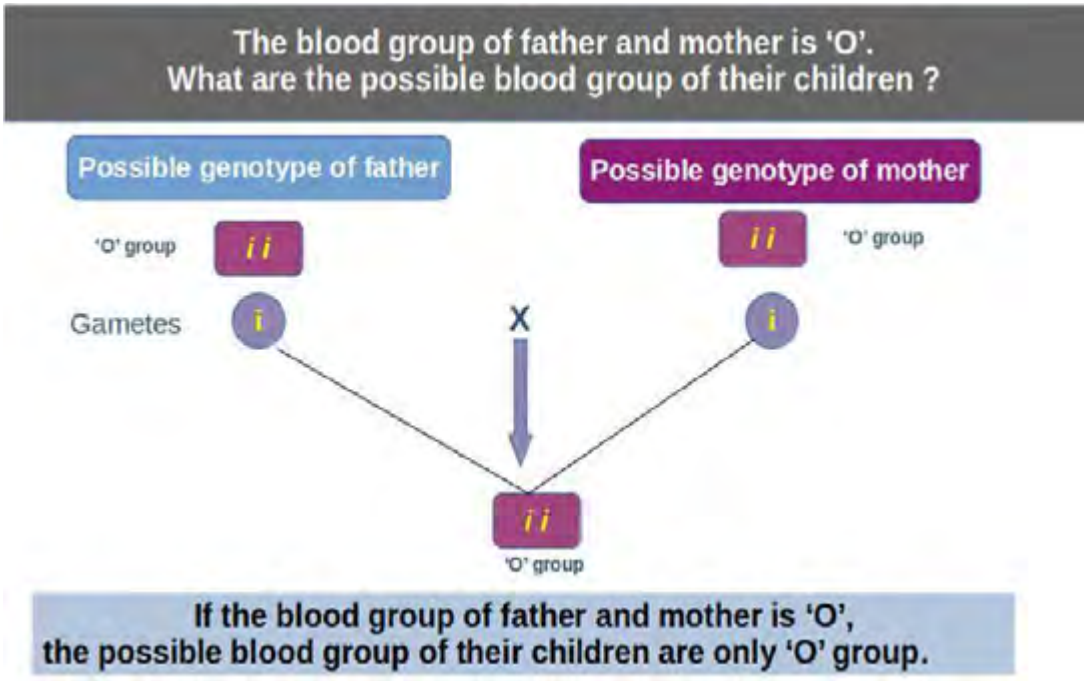
- More than two alleles of a gene are called multiple alleles and the phenomenon is called multiple allelesim.
- ABO-Blood group in man is an excellent example for multiple allelism . Here three alleles determine the same character.
- In an individual only two alleles can be present.

I^AI^A I^Ai I^BI^B I^Bi I^AI^B ii

Multiple alleles can be found only when population studies are made.







A single gene product may produce more than one effect. Substantiate the statement.

Eg : Starch synthesis in pea seeds - controlled by one gene

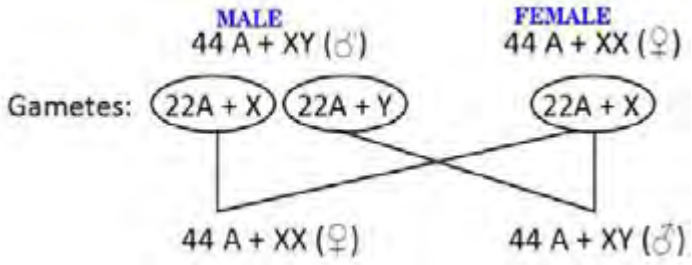
- Starch synthesis in pea is controlled by two alleles (B and b).
- Starch is synthesised effectively by BB homozygotes and therefore, large starch grains are produced.
- In bb homozygotes have lesser efficiency in starch synthesis and produce smaller starch grains..
- After maturation of the seeds, BB seeds are round and the bb seeds are wrinkled.
- Heterozygotes produce round seeds, and so B seems to be the dominant allele.
- But, the starch grains produced are of intermediate size in Bb seeds.
- So if starch grain size is considered as the phenotype, then from this angle, the alleles show incomplete dominance

SEX DETERMINATION IN MAN

XX-XY Mechanism

- The chromosome pattern in female is XX and that in the male is XY.
- Females are homo gametic with only X chromosomes and males are hetero gametic and produce gametes with either X or Y chromosomes .

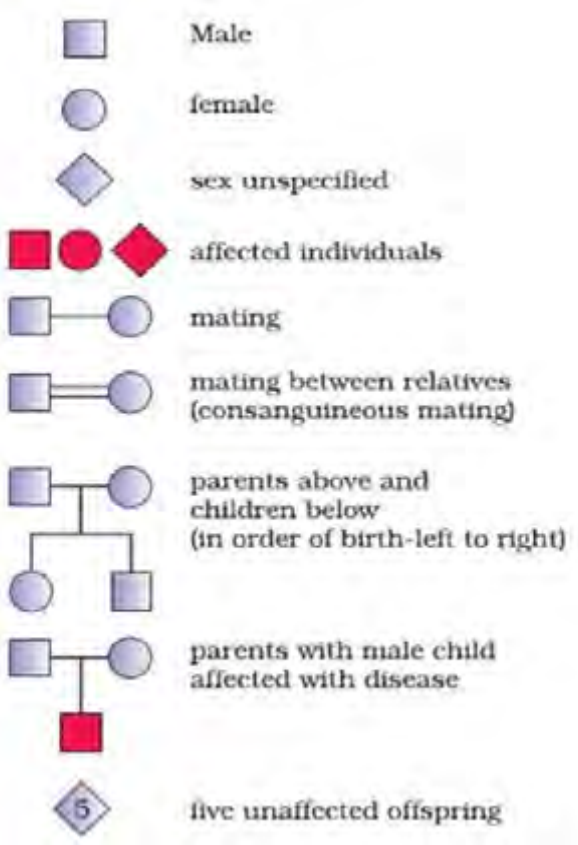
- **The XY zygote will be formed if the Y chromosome carrying male gamete fuses with the X gamete of female, and the child thus formed is male .**
- **Female child will born from XX zygote if the X gamete of female fuses with the X gamete of male .**

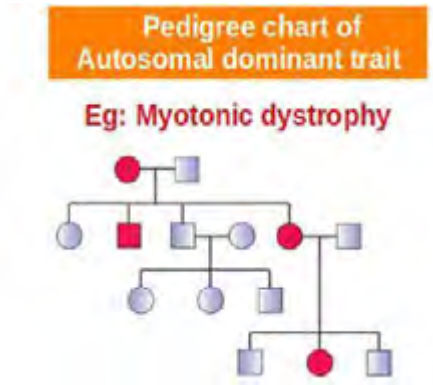


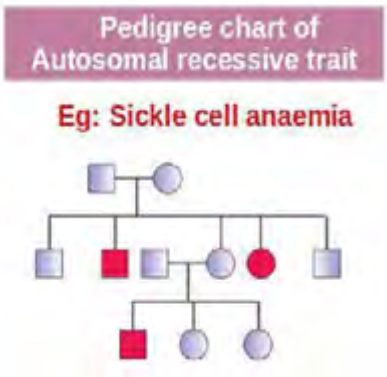
Pedigree Analysis

Analysing the inheritance of the character through several previous generations in a family is known as pedigree analysis.

- **In the pedigree analysis the inheritance of a particular trait is represented in the family tree over generations.**
- **pedigree study provides a strong tool, which is utilised to trace the inheritance of a specific trait, abnormality or disease.**

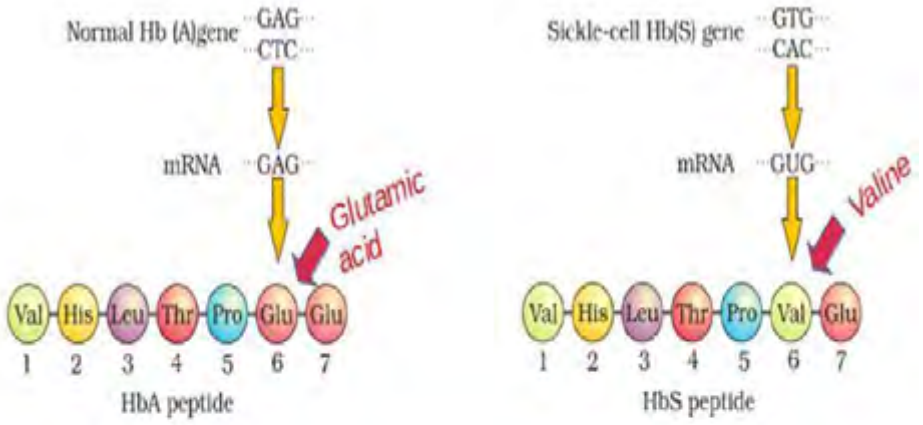


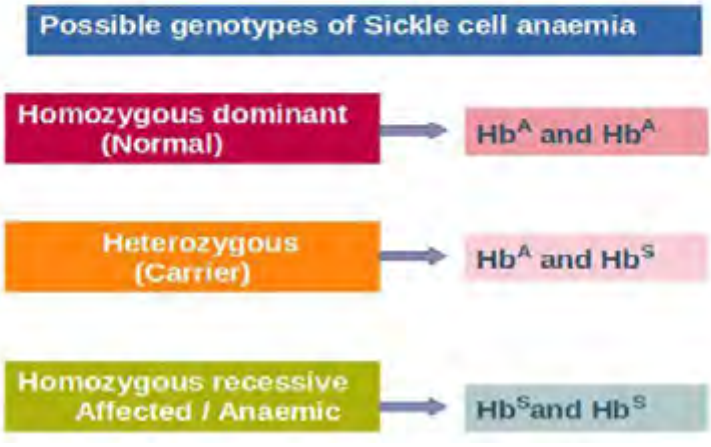




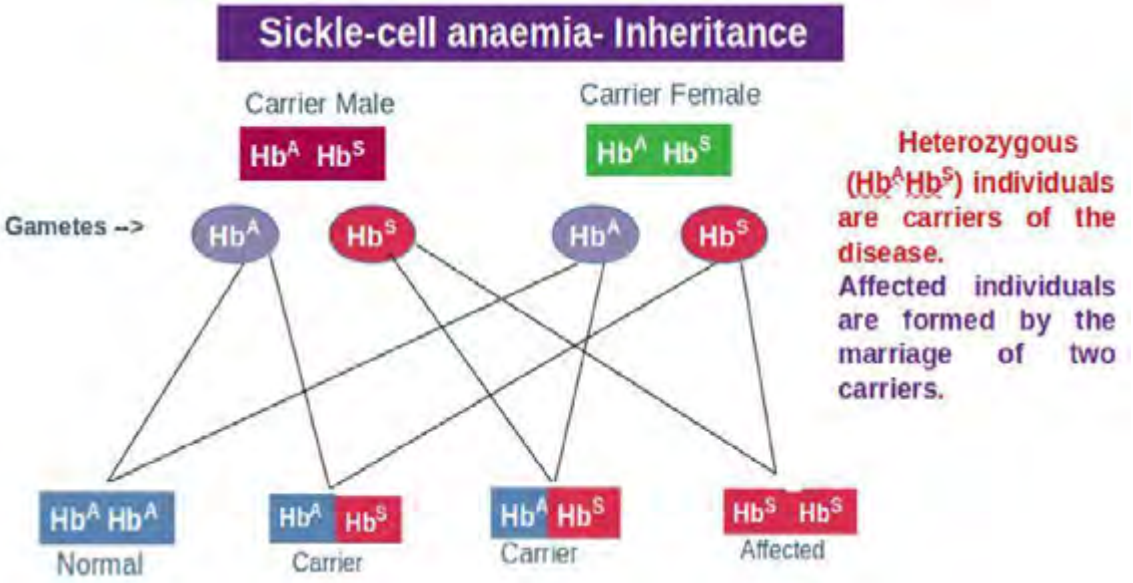
Sickle-cell anaemia

- Sickle cell anaemia is an autosome linked recessive trait.
- The gene for Sickle cell anaemia occurs in chromosome 11.
- The defect is caused by the substitution of Glutamic acid (Glu) by Valine (Val) at the sixth position of the beta globin chain of the haemoglobin molecule.
- The substitution of amino acid in the globin protein results due to the single base substitution at the sixth codon of the beta globin gene from GAG to GUG.



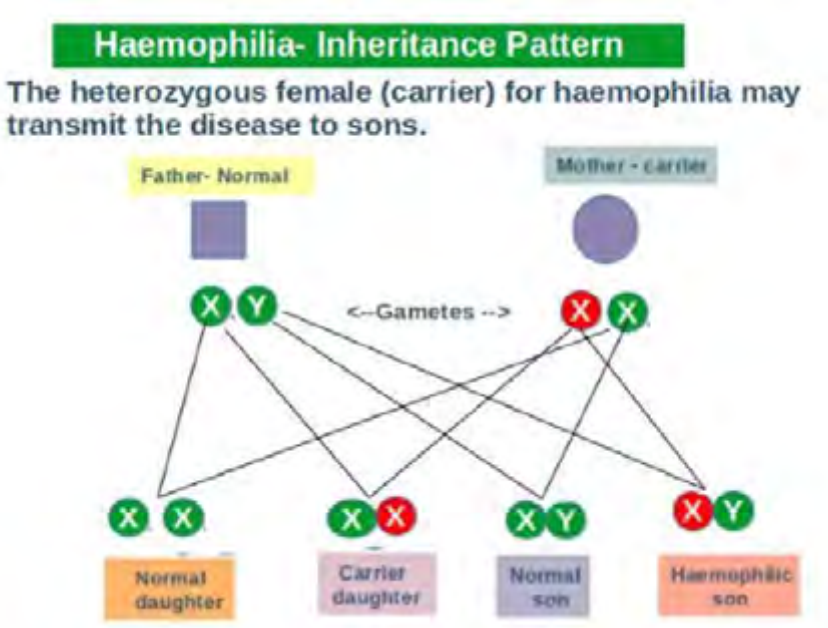


- The disease is controlled by a single pair of allele, Hb^A and Hb^S .
- Homozygous individuals Hb^S Hb^S show the diseased phenotype.
- Heterozygous Hb^A and Hb^S individuals are carrier of the disease.
- The mutant haemoglobin molecule undergoes polymerisation under low oxygen tension causing the change in the shape of the RBC from biconcave disc to elongated sickle like.



Haemophilia

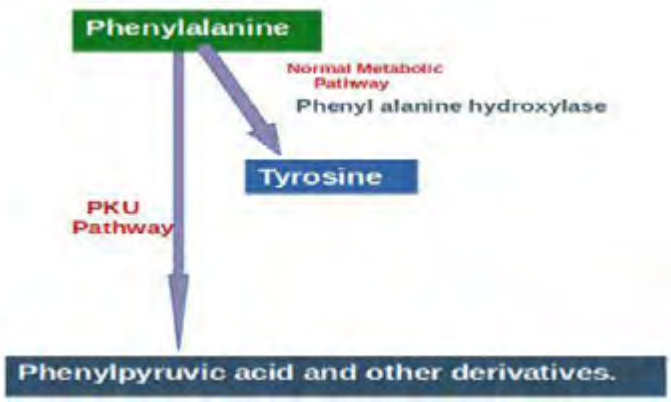
- It is a sex-linked recessive disorder due to the mutation of genes present in X chromosome (X-linked disease)
- The affected person may die due to minor injuries because of excessive bleeding.
- The genes responsible for haemophilia are located on the X-chromosomes.
- The Y-chromosome carries no genes for this trait.



- **The possibility of a female becoming a haemophilic is extremely rare because mother of such a female has to be at least carrier and the father should be haemophilic (unviable in the later stage of life).**
- **The family pedigree of Queen Victoria shows a number of haemophilic descendents as she was a carrier of the disease.**

Phenylketonuria

- **This is an inborn error of metabolism.**
- **It is inherited as the autosomal recessive trait.**
- **The affected individual lacks an enzyme that converts the amino acid phenylalanine into tyrosine.**
- **As a result of this phenylalanine is accumulated and converted into phenylpyruvic acid and other derivatives.**
- **Accumulation of these in brain results in mental retardation.**
- **These are also excreted through urine because of its poor absorption by kidney.**



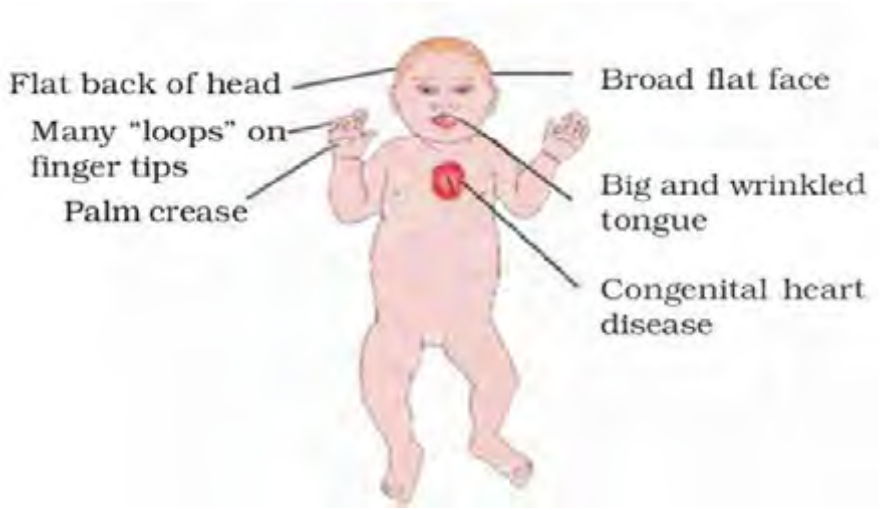
Chromosomal Disorders

Down's Syndrome

- **This disorder was first described by Langdon Down (1866).**
- **The cause of this genetic disorder is the presence of an additional copy of the chromosome number 21.**

The affected individual is

- **short statured with small round head,**
- **furrowed tongue and partially open mouth**
- **Palm is broad with characteristic palm crease.**
- **Physical, psychomotor and mental development is retarded**



Klinefelter's Syndrome

- **This genetic disorder is caused due to the presence of an additional copy of X-chromosome resulting into a karyotype of 47, XXY.**
- **Such an individual has overall masculine development, however, the feminine development (development of breast, i.e., Gynaecomastia) is also express.**
- **Such individuals are sterile.**

TURNER'S SYNDROME

- **Turner's Syndrome is caused due to the absence of one of the X chromosomes, i.e., 45 with X0.**
- **Such females are sterile with rudimentary ovaries**
- **They lack of other secondary sexual characters.**

Name of Disorder	Karyotype	Symptoms
Down's Syndrome	45 A+XX / 45A +XY (trisomy of chromosome No. 21)	• Short statured with small round head • furrowed tongue and partially open mouth • Broad Palm with characteristic palm crease. • Retardation in physical, psychomotor and mental development.
Klinefelter's Syndrome	44 A +XXY (trisomy of sex chromosome)	• Overall masculine development • Development of breast (Gynaecomastia) is expressed. • Sterile individuals.
Turner's Syndrome	44 A +XO (Monosomy of sex chromosome)	• Sterile females. • Rudimentary ovaries • lack of secondary sexual characters

MOLECULAR BASIS OF INHERITANCE

FOCUS AREA -HSE EXAM 2022

The salient features of the Double-helix structure of DNA

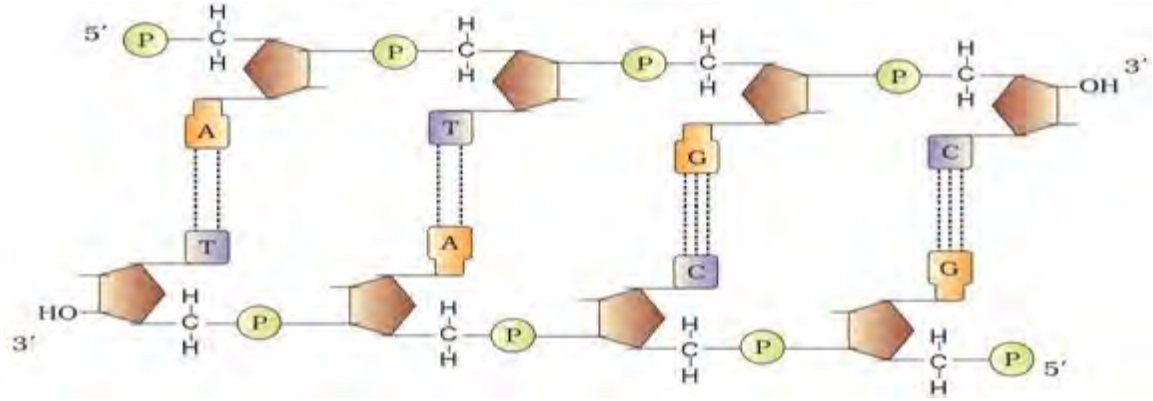
i) DNA is made of two polynucleotide chains, where the backbone is constituted by sugar-phosphate, and the bases project inside.

ii) The two chains have anti-parallel polarity. 0ne chain has the polarity 5'->3', the other has 3'->5'.

iii) The bases in two strands are paired through hydrogen bond (H-bonds) forming base pairs (bp). Adenine forms two hydrogen bonds with Thymine .Similarly, Guanine is bonded with Cytosine with three H-bonds.

iv) The two chains are coiled in a right-handed fashion. The pitch of the helix is 3.4 nm and there are roughly 10 bp in each turn. Consequently, the distance between a bp in a helix is approximately equal to 0.34 nm.

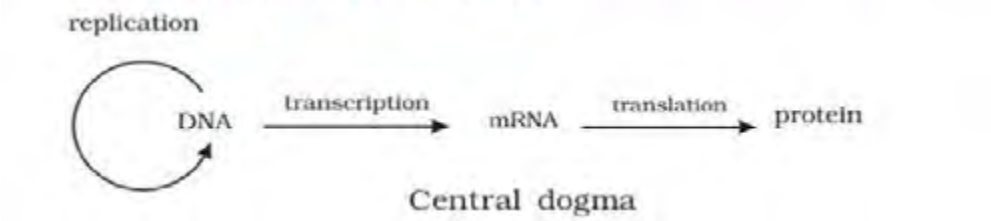
v) The plane of one base pair stacks over the other in double helix. This, in addition to H-bonds, confers stability of the helical structure



Central dogma in molecular biology

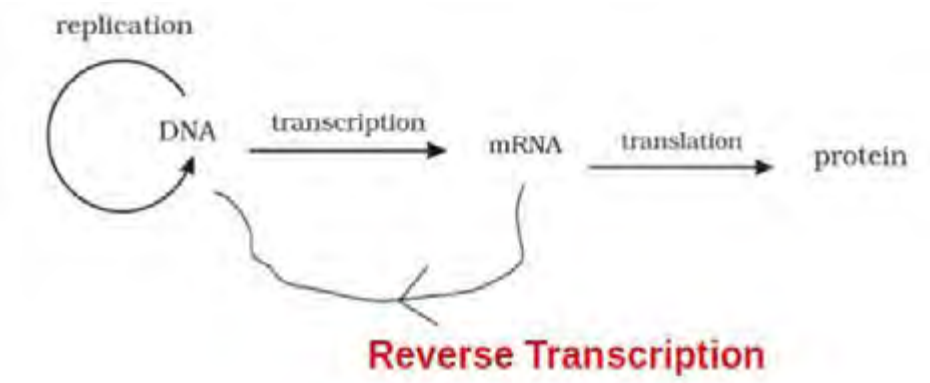
Francis Crick proposed the Central dogma in molecular biology, which states that the genetic information flows from

DNA-->RNA-->Protein.



Central dogma

Reverse Transcription



In some viruses the flow of information is in reverse direction, that is,from RNA to DNA. It is known as Reverse transcription.

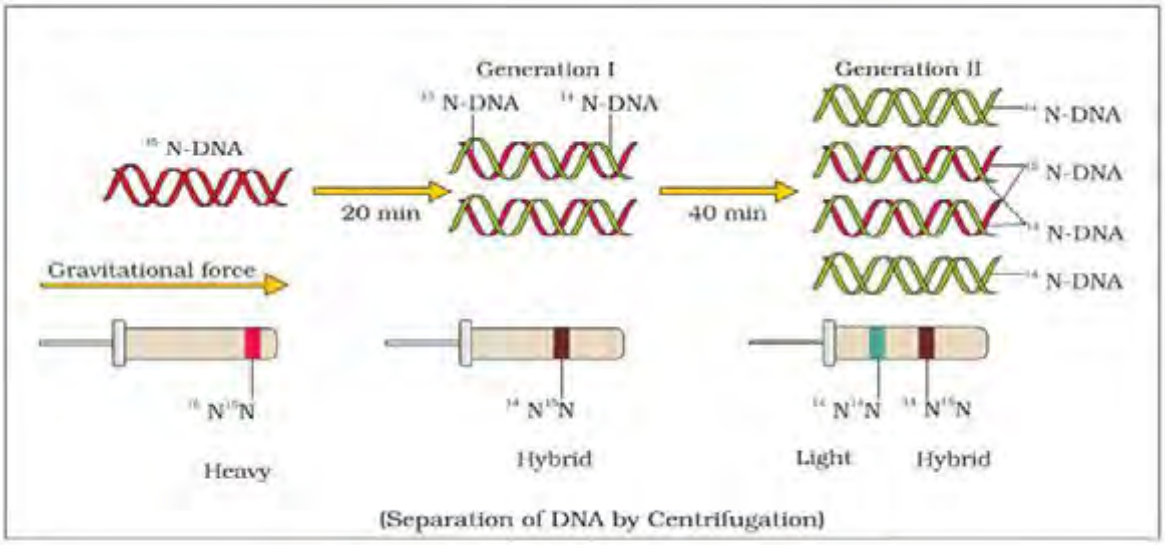
REPLICATION OF DNA

- Watson and Crick proposed Semiconserve mode of DNA replication.
- In semiconservative replication, the newly formed DNA has one old strand and one new strand.
- The two strands separate and act as template for the synthesis of new complementary strands.
- After the completion of replication, each DNA molecule would have one parental and one newly synthesized strand.

Meselson and Stahl's Experiment

- **Mathew Meselson and Franklin Stahl** demonstrated the semiconservative mode of DNA replication in E.coli(1958).
- They grew E. coli in a medium containing $^{15}NH_4Cl$ as the only nitrogen source for many generations.
- The result was that ^{15}N was incorporated into newly synthesised DNA
- Bacterium thus formed became heavier and the DNA is called heavier DNA
- This heavy DNA molecule is then subjected to centrifugation in a Cesium Chloride (CsCl).
- Due to higher density ^{15}N DNA deposited at the bottom of the test tube.
- Then they transferred the cells into a medium with normal $^{14}NH_4Cl$ (^{14}N medium) .

- Then the DNA was extracted from the culture after one generation(20 minutes) after the transfer from ^{15}N to ^{14}N medium.
- Then subjected to centrifugation and found that it had a hybrid or intermediate density between ^{15}N DNA and ^{14}N DNA.
- The DNA that was extracted from the culture after (after 40 minutes) two generation after the transfer from ^{15}N to ^{14}N medium had composed of equal amounts of this hybrid DNA and of 'light' DNA.
- The experiment proved that the two strands of DNA separates at the time of replication.
- After the completion of replication each DNA molecule would have one parental and one newly synthesised strand.



Messelson and Stahl's Experiment

- This scheme of DNA replication is termed as Semiconservative Replication
- Taylor and colleagues in 1958 proved that the DNA in chromosomes also replicate Semiconservatively.
- They conducted experiments in *Vicia faba* (faba beans) by using radioactive thymidine.

The Machinery and the Enzymes for DNA Replication

The main enzyme of DNA replication is DNA-dependent DNA Polymerase

DNA Polymerase

- This is one of the fastest enzyme which catalyses the polymerization of nucleotides with greatest accuracy.

- Any defect in replication leads to mutation.
- In E.coli, 4.6x10 6 bp completes replication with 38 mts, ie, average rate is 2000 bp per second.

Deoxyribonucleotide triphosphates

- Deoxyribonucleotide triphosphates provide energy for replication.
- Deoxyribonucleoside triphosphates serve dual purposes.
- It act as substrates and also provide energy for polymerisation reaction

Replication fork

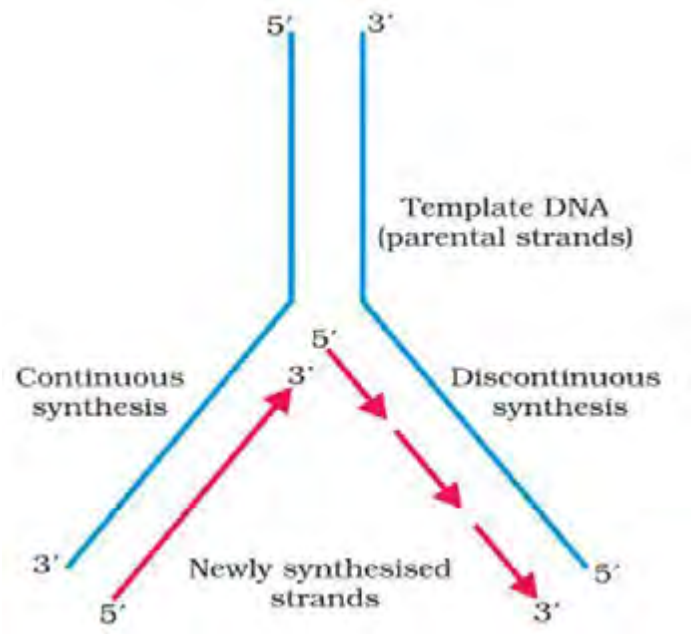
- For long DNA molecules, the two strands of DNA cannot be separated in its entire length due to very high energy requirement.
- In such DNAs replication occur within a small opening of the DNA helix known as replication fork.

Leading strand

- The DNA dependent DNA Polymerases catalyses polymerization in 5`->3` direction only.
- In 5`->3` direction , (the template with polarity 3`-> 5`) the replication is continuous and it is known as leading strand.

Lagging strand

- In the other strand, template with polarity 5`->3` , the replication is discontinuous and is called lagging strand.
- Discontinuously synthesized DNA fragments are called okazaki fragments.



DNA ligase

- **The discontinuously synthesized okazaki fragments are joined by the enzyme called DNA ligase.**

Origin of Replication

- **The definite regions in which replication originates are known as origin of replication.**

TRANSCRIPTION

- **The process of copying genetic information from one strand of the DNA into RNA is called transcription.**
- **In transcription only a segment of DNA and only one of the strands is copied into RNA.**

Transcription Unit

A transcription unit in DNA is defined primarily by the three regions in the DNA

(i) A Promoter

(ii) The Structural gene

(iii) A Terminator

- ***The two DNA strands have opposite polarity and the DNA-dependent RNA polymerase catalyse the polymerisation of RNA in only one direction, that is, 5' →3' direction.***
- **The strand that has the polarity 3'→5' acts as a template, and is referred to as template strand.**
- **The other strand which has the polarity (5'→3') and the sequence same as RNA is referred to as coding strand.**

write the sequence of RNA transcribed from this DNA?

- 3′-ATGCATGCATGCATGCATGCATGC-5′ Template Strand
  5′-TACGTACGTACGTACGTACGTACG-3′ Coding Strand

Answer

3′-ATGCATGCATGCATGCATGCATGC-5′ Template Strand

5′-uACGuACGuACGuACGuACGuACG-3′ mRNA Strand

Promoter

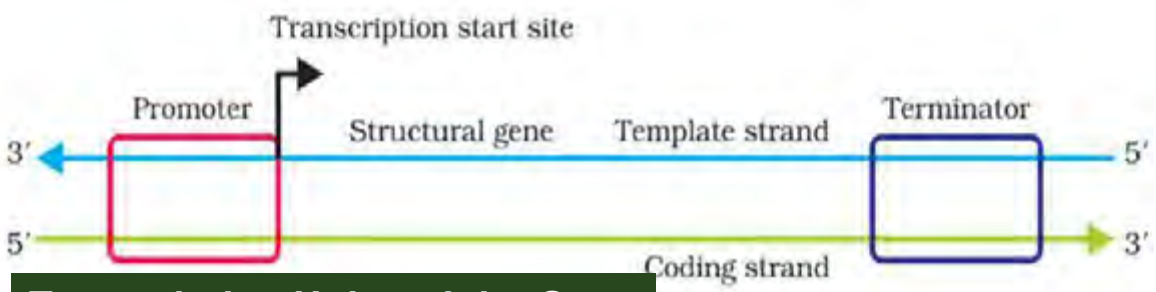
Promoter is a DNA sequence that provides binding site for RNA polymerase.

It is located towards the 5` end(upstream) of the structural gene.

The presence of a promoter in a transcription unit defines the template and coding strands.

Terminator

The terminator is located towards 3' -end (downstream) of the coding strand and it usually defines the end of the process of transcription.



Transcription Unit and the Gene

Cistron

- ***Cistron is a segment of DNA coding for a polypeptide.***

Monocistronic unit

- **The structural unit in transcription unit of eukaryotes are monocistronic.**
- **Monocistronic structural genes have interrupted coding sequences. Ie, the genes are split or the genes with coding sequences and genes with non-coding sequences.**

Exons

- **The coding sequence or expressed sequences of cistron are called exons.**

Introns

The non coding sequences of cistrons are called introns or intervening sequence. **They do not appear in the mature or processed RNA.**

Polycistronic unit

This is the structural transcription unit of bacteria or prokaryotes

GENETIC CODE

- It was George Gamow, a physicist who argued that since there are only 4 bases and if they have to code for 20 amino acids, the code should constitute a combination of bases.
- He suggested that in order to code for all 20 amino acids, the code should be made up of three nucleotides.
- This would generate 64 codons (4^3 i.e., 4x4x4= 64) for coding 20 amino acids.
- Providing proof that the codon was a triplet, was a difficult task.
- Har Gobind Khorana developed a chemical method for synthesising RNA molecules with defined combinations of bases.
- Marshall Nirenberg's cell-free system for protein synthesis finally helped to understand the genetic code.
- Severo Ochoa enzyme (polynucleotide phosphorylase) was also helpful in polymerising RNA with defined sequences (enzymatic synthesis of RNA).
- The sequence of nucleotides (nitrogen bases) in the m RNA which contains the information for protein synthesis is known as genetic code.
- The genetic information is written in a coded language in the form of triplet codons (3 letter code).
- The sequence of three bases determining a single amino acid is called codon.

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

The salient features of genetic code

(i) The codon is triplet.

61 codons code for amino acids and 3 codons do not code for any amino acids, hence they function as stop codons.

Stop codons
UAA
UAG
UGA

(ii) One codon codes for only one amino acid, hence, it is unambiguous and specific.

(iii) Some amino acids are coded by more than one codon, hence the code is degenerate.

(iv) The codon is read in mRNA in a contiguous fashion.

There are no punctuations.

(v) The code is nearly universal: for example, from bacteria to human UUU would code for Phenylalanine (phe). Some exceptions to this rule have been found in mitochondrial codons, and in some protozoans.

(vi) AUG has dual functions. It codes for Methionine (met) , and it also act as initiator codon.

REGULATION OF GENE EXPRESSION

In eukaryotes, the gene expression is regulated at the following levels

(i) transcriptional level (formation of primary transcript),

(ii) processing level (regulation of splicing),

(iii) transport of mRNA from nucleus to the cytoplasm,

(iv) translational level.

- **The gene expression is controlled by metabolic, physiological or environmental conditions.**

Example

- **The enzyme beta-galactosidase is synthesized by E.coli to catalyse the hydrolysis of lactose into galactose and glucose.**

- **If the bacteria are living in medium devoid of lactose, they need not require the particular enzyme.**
- **The development and differentiation of embryo into adult organisms are also a result of the co-ordinated activities of several set of genes and their expressions.**

Operon

- **Francois Jacob and Jacque Monod were the first to explain regulation of gene expression at transcriptional level.**
- **A metabolic reaction is controlled by a set of genes is referred to as operon.**

Examples of Operon

- **lac operon, trp operon,ara operon,his operon, val operon**
- **In lac operon (lac referes to lactose), a polycistronic structural gene is regulated by a common promoter gene and regulatory genes.**

The Lac operon

- **Lactose is the substrate for the enzyme beta-galactosidase and it regulates switching on and off of the operon.**
- **Hence, it is termed as inducer.**

Components of Lac operon

Regulatory gene (i gene)

- **Regulatory gene produces the repressor protein.**

Promoter gene (P)

- **It is the region in which RNA Polymerase binds**

Operator gene (O)

- **It is the region in which repressor protein binds.**

Structural gene(z, y,a)

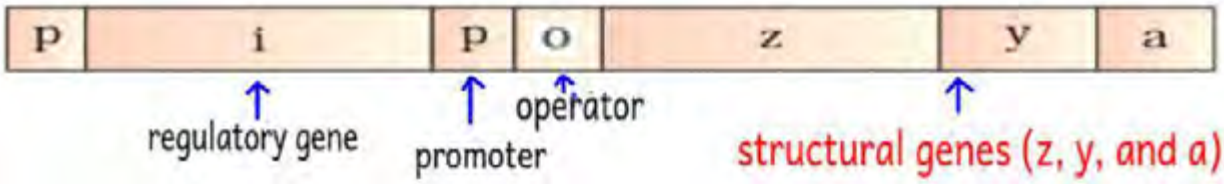
- **Structural gene transcribe into lac mRNA and it is translated into lac proteins:-**

β -galactosidase

- **z gene codes for beta-galactosidase (β -gal),**
  It converts the disaccharide, lactose into galactose and glucose.
- **The y gene codes for permease**

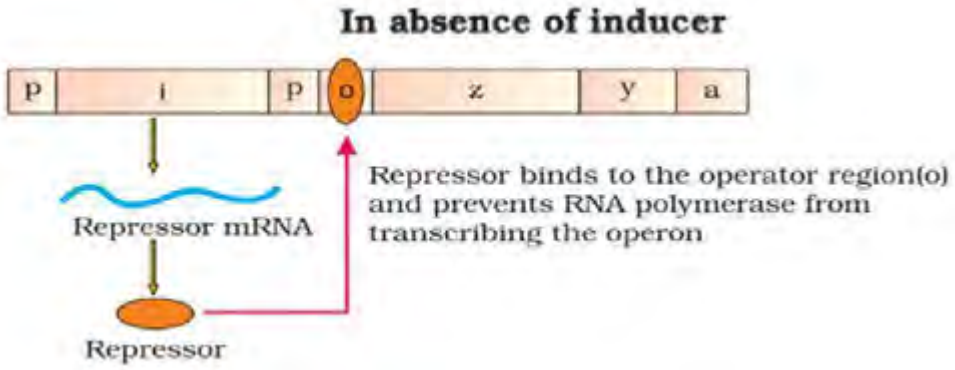
Permease increases permeability of the of the cell to β -galactosides.

- **The a gene encodes a transacetylase.**



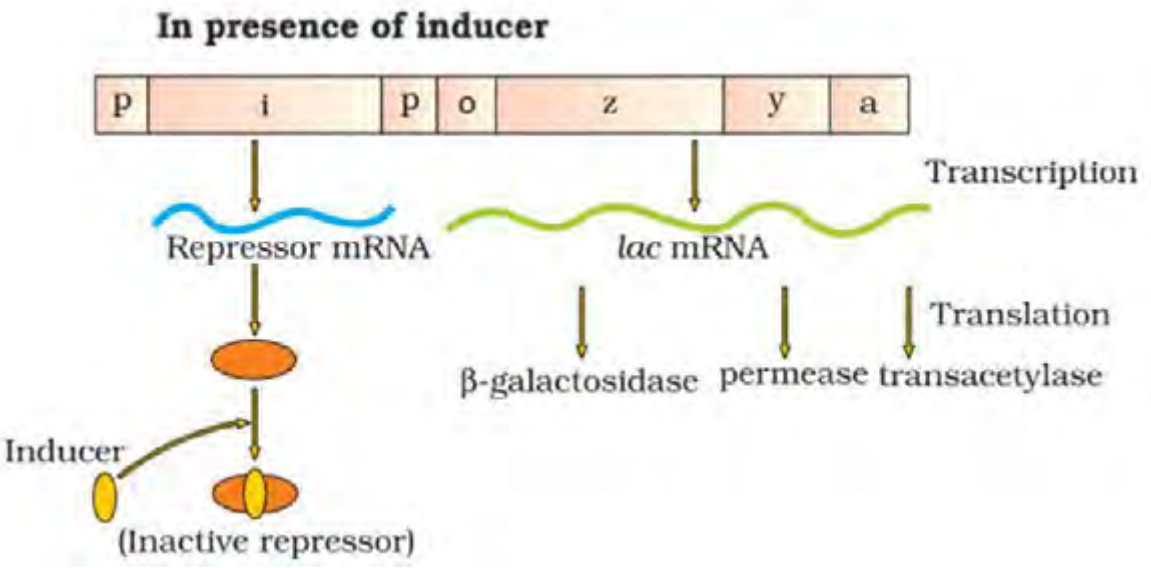
Lac operon (In absence of inducer)

- **Regulatory gene transcribes into repressor mRNA.**
- **Repressor mRNA translates into repressor protein.**
- **Repressor protein binds to the Operator region.**
- **This binding prevents the RNA Polymerase binding to the Promoter region.**
- **Thus the transcription of the structural genes is prevented.**



Lac operon (In presence of inducer)

- Regulatory gene transcribes into repressor mRNA.
- Repressor mRNA translates into repressor protein.
- inducer, such as lactose or allolactose binds with repressor protein and it becomes inactive.
- Inactive repressor could not bind to operator. So RNA Polymerase binds to Promoter region.
- RNA Polymerase transcribes lac mRNA
- Thus the transcription of the structural genes takes place.



HUMAN GENOME PROJECT (HGP)

- For sequencing, the total DNA from a cell is isolated and converted into random fragments of relatively smaller sizes.
- Cloned in suitable host using specialised vectors.

BAC & YAC

- If bacteria is used as vector then it is called as BAC (bacterial artificial chromosomes).
- If we use Yeast as vector then it is called YAC (yeast artificial chromosomes)

Salient Features of Human Genome

(i) The human genome contains 3164.7million bp.

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

(iii) The total number of genes is estimated at 30,000– much lower than previous estimates of 80,000 to 1,40,000 genes.

Almost all (99.9 per cent) nucleotide bases are exactly the same in all people.

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

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

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

(vii) Repetitive sequences are stretches of DNA sequences that are repeated many times, sometimes hundred to thousand times.

They are thought to have no direct coding functions, but they shed light on chromosome structure, dynamics and evolution.

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

(ix) Scientists have identified about 1.4 million locations where single-base DNA differences (SNPs – single nucleotide polymorphism, pronounced as 'snips') occur in humans.

DNA FINGERPRINTING

STEPS IN DNA FINGERPRINTING

(i) isolation of DNA,

(ii) digestion of DNA by restriction endonucleases,

(iii) separation of DNA fragments by electrophoresis,

(iv) transferring (blotting) of separated DNA fragments to synthetic membranes, such as nitrocellulose or nylon,

(v) hybridisation using labelled VNTR probe, and

(vi) detection of hybridised DNA fragments by autoradiography.

Applications of DNA Fingerprinting

- DNA from every tissue (such as blood, hair-follicle, skin, bone, saliva, sperm etc.), from an individual show the same degree of polymorphism, they become very useful identification tool in forensic applications.
- It is used as a powerful forensic tool to solve the problems of paternity, rape, murder etc.
- It is used in the diagnosis of genetic diseases.
- It is used in the determination of phylogenetic status of animals etc.

EVOLUTION

FOCUS AREA -HSE 2022 EXAM

EVIDENCES FOR EVOLUTION

1. Paleontological evidence

Fossils are remains of hard parts of life-forms found in rocks.

- Different-aged rock sediments contain fossils of different life-forms who probably died during the formation of the particular sediment.
- A study of fossils in different sedimentary layers indicates the geological period in which they existed.

Evidences from Comparative anatomy and morphology

Eg : Homologous organs

"Anatomically similar structures performing dissimilar functions are called Homologus organs"

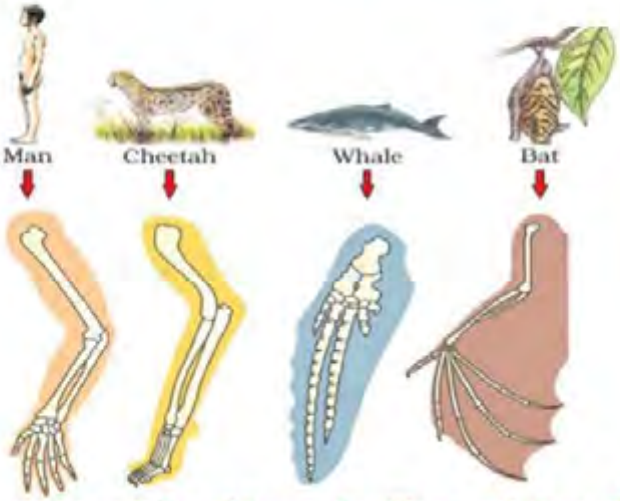
Examples of homologous organs

- Forelimbs of mammals
- Thorn and tendrils of Bougainvillea and Cucurbita
- vertebrate hearts or brain

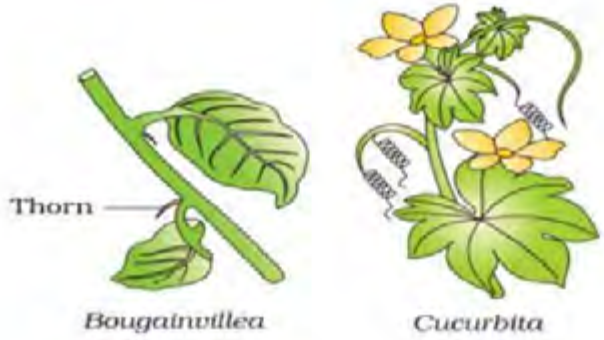
Forelimbs of mammals

- Whales, bats, Cheetah and human (all mammals) share similarities in the pattern of bones of forelimbs.
- Though these forelimbs perform different functions in these animals, they have similar anatomical structure –
- all of them have humerus, radius, ulna, carpals, metacarpals and phalanges in their forelimbs.
- Hence, in these animals, the same structure developed along different directions due to adaptations to different needs.

- This is divergent evolution and these structures are homologous.
- Homology indicates common ancestry.



- Thorn and tendrils of Bougainvillea and Cucurbita



Analogous organs

"Anatomically dissimilar structures performing similar functions are called Analogus organs."

Examples of Analogus organs

- Wings of butterfly and of birds
- eye of the octopus and of mammals
- flippers of Penguins and Dolphins
- Sweet potato (root modification) and potato (stem modification)

Homologous organs	Analogous organs
Anatomically similar structures performing dissimilar functions	Anatomically dissimilar structures performing similar functions
Homologous structures are as a result of **Divergent Evolution**	Analogous structures are as a result of **Convergent Evolution**
same structure developed along different directions due to adaptations to different needs.	similar habitat resulted in selection of similar adaptive features in different groups of organisms but toward the same function.

INDUSTRIAL MELANISM

- Industrial melanism clearly indicates the process of evolution by natural selection.
- In 1850s, i.e., before industrialisation, in England there are two types of moths, White-winged moths and dark-winged or melanised moths.
- The white-winged moths rest on the tree trunk containing grey lichens
- The colour of the lichen and the colour of the moths were similar and the colour of the moth easily blended with the dull background of tree trunk.
- So they escaped from the enemies from predation
- While the black coloured ones were easily spotted and eaten by the enemies.
- In 1920, With the advent of industrial revolution, large scale burning of coal became common and the soot and smoke produced by the factories got deposited on tree trunks and made them black.
- As a result the white-winged moths become more visible than the black moths and were preyed by predators.
- Thus the number of grey moths decreased considerably.

- This understanding is supported by the fact that in areas where industrialisation did not occur e.g., in rural areas, the count of melanic moths was low.
- This showed that in a mixed population, those that can better-adapt, survive and increase in population size.

Evolution by Anthropogenic action

Evolution due to the action of human beings is called evolution by anthropogenic action.

Examples :

- The excess use of herbicides, pesticides, etc., has resulted in selection of resistant varieties in a much lesser time scale.
- Resistance of antibiotics and drugs by microbes/ cells appearing in a time scale of months or years.

HARDY -WEINBERG PRINCIPLE

- The genetic structure of a non-evolving population is described by Hardy Weinberg law proposed by G.H. Hardy and W. Weinberg independently in 1908.
- This Principle states that in a large population, the gene frequencies of various kinds of genes remain constant generation after generation.
- That is ,the gene pool remains a constant.
  (Gene pool is the total genes and their alleles in a population)
- This is called genetic equilibrium.
- In a population the sum total of all the allelic frequencies is 1.
- Individual frequencies, for example, can be named p, q, etc.
- In a diploid, p and q represent the frequency of allele 'A' and allele 'a'

In a diploid organism
Frequency of allele A = p
Frequency of allele a = q
Thus frequency of alleles AA = p^2
Frequency of alleles aa = q^2
Frequency of alleles Aa = 2pq
Hence $p^2 + 2pq + q^2 = 1$

- This is a binomial expansion of $(p+q)^2$.
- When frequency measured, differs from expected values, the difference indicates the extent of evolutionary change.
- The change of frequency of alleles in a population would then be due to evolution.

Factors affecting Hardy-Weinberg equilibrium

Gene migration or Gene flow

Genetic drift

Mutation

Genetic recombination

Natural selection

Gene migration or Gene flow

- When migration of a section of population to another place and population occurs, gene frequencies change in the original as well as in the new population.
- New genes/alleles are added to the new population and these are lost from the old population.
- There would be a gene flow if this gene migration, happens multiple times.

Genetic drift

- The random changes in the allele frequency occurring by chance in small populations are called genetic drift.

- Sometimes the change in allele frequency is so different in the new sample of population that they become a different species.
- The original drifted population becomes founders and the effect is called founder effect.

Mutation

- Advantageous mutations when selected will result in observation of new phenotypes.
- Over few generations, this would result in Speciation.

Genetic recombination

- It is the reshuffling of gene combinations during crossing over resulting in genetic variation.

Natural selection

- Natural selection is a process in which heritable variations enabling better survival are enabled to reproduce and leave greater number of progeny.

Natural selection can lead to

Stabilisation Selection

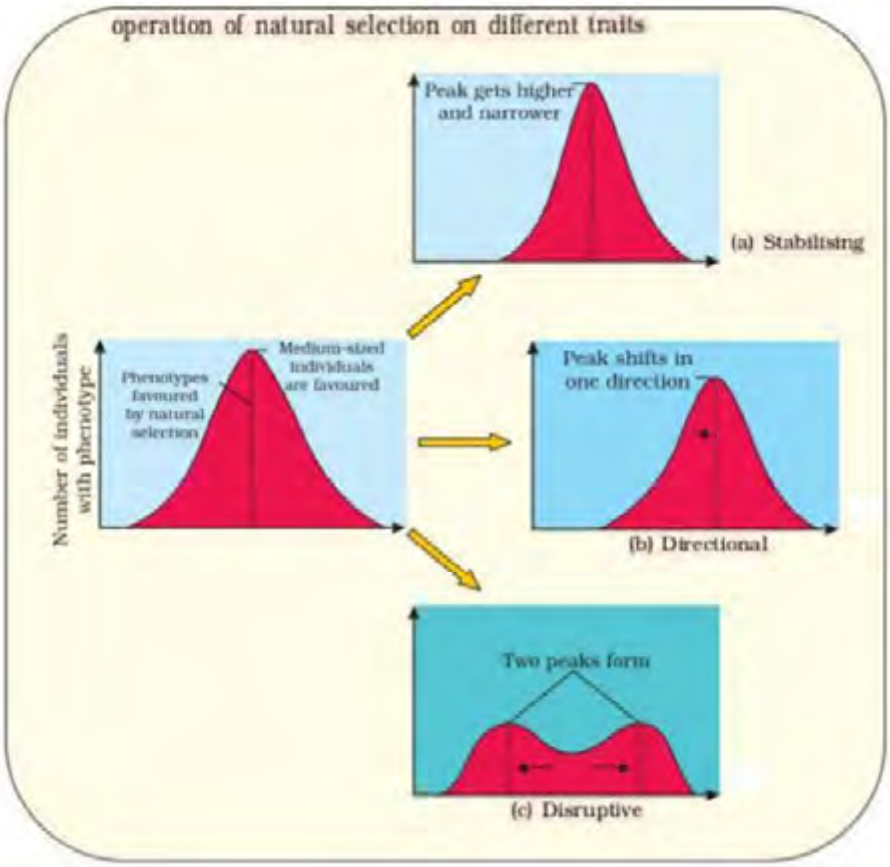
- in this selection more individuals acquire mean character value and peaks get higher and narrower.

Directional change

- in this selection more individuals acquire value other than the mean character value and peak shifts in one direction.

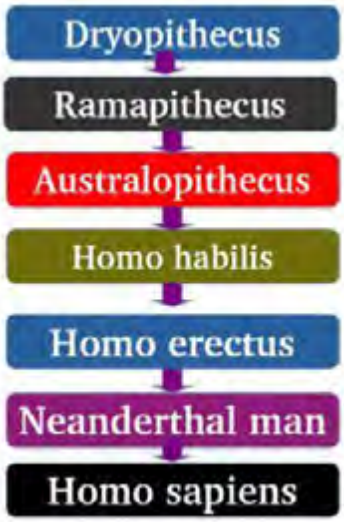
Disruptive Selection

- more individuals acquire peripheral character value at both ends of the distribution curve and more than one peaks are formed.



ORIGIN AND EVOLUTION OF MAN

- The gradual evolution of man from ape is fully supported by fossils.
- For convenience, the fossils are arranged in the order of apes, ape-man and primitive man. They are the following.



Dryopithecus and Ramapithecus

- **About 15 mya, primates called** Dryopithecus and Ramapithecus were existing.
- They were hairy
- walked like gorillas and chimpanzees.
- Ramapithecus was more man-like
- while Dryopithecus was more ape-like.

Australopithecus

- Lived before 2 mya in east African grass lands.
- They ate fruit
- hunted with stone weapons.

Homo habilis

- Lived before 1.6 to 2 mya.
- Brain capacity is 650-800 cc.
- Made stone weapons and hunted animals.

Homo erectus

- Fossils discovered in Java in 1891.
- Lived before 1.5 mya. They ate meat
- Brain is large with a capacity of 900 cc.

Neanderthal man

- Lived near east and central Africa between 1 lakh to 40,000 years ago.
- They used hides to protect their body and buried their dead.
- Brain capacity is 1400 cc.

Homosapiens

- During ice age between 75,000 – 10,000 years ago modern man arose.
- They arose in Africa and moved across continents and developed into distinct races.
- Pre historic cave art developed about 18,000 years ago.
- Agriculture came around 10,000 years back.

HUMAN HEALTH AND DISEASES

FOCUS AREA -HSE EXAM 2022

COMMON DISEASES IN HUMANS

BACTERIAL DISEASES

Include

- Typhoid, Pneumonia, Dysentery, Plague, Diphtheria etc.

Typhoid fever

- It is a disease of alimentary canal caused by the bacterium called *Salmonella typhi.*
- The bacterium enters the body through contaminated food and water and immigrates to other organs through blood.

Common symptoms of typhoid

include

- sustained fever (39-40 0 C),
- weakness,
- stomach pain,
- constipation,
- headache,
- loss of appetite etc.
- In severe case death may occur due to intestinal perforations.

Widal test

It is an antigen- antibody reaction test used for the diagnosis of typhoid fever.

Pneumonia

It is a lung disease caused by bacteria called

- *Streptococcus pneumoniae and*
- *Haemophilus influenzae.*

- In this infection, the alveoli get filled with fluid leading to serious respiratory problems.
- Fever,
- chills,
- cough and headache are the symptoms.
- In extreme cases, the lips and finger nails turn blue.

Transmission

- occurs through the droplets of infected persons.
- It may also get transmitted by sharing of utensils with an infected person.

VIRAL DISEASES

Include

- common cold,
- chicken pox, measles etc.

Common cold

- It is a common viral disease of nose and respiratory passage, caused by Rhino viruses.

symptoms

- Nasal congestion and discharge,
- sore throat,
- cough,
- headache,
- tiredness etc. Which usually last for 3-7 days are the symptoms.

Transmission

- occurs through the droplets resulting from cough or sneeze of infected person or through the contaminated objects like pens, books, cups, keyboards etc.

PROTOZOAN DISEASES

Include malaria, amoebiasis etc.

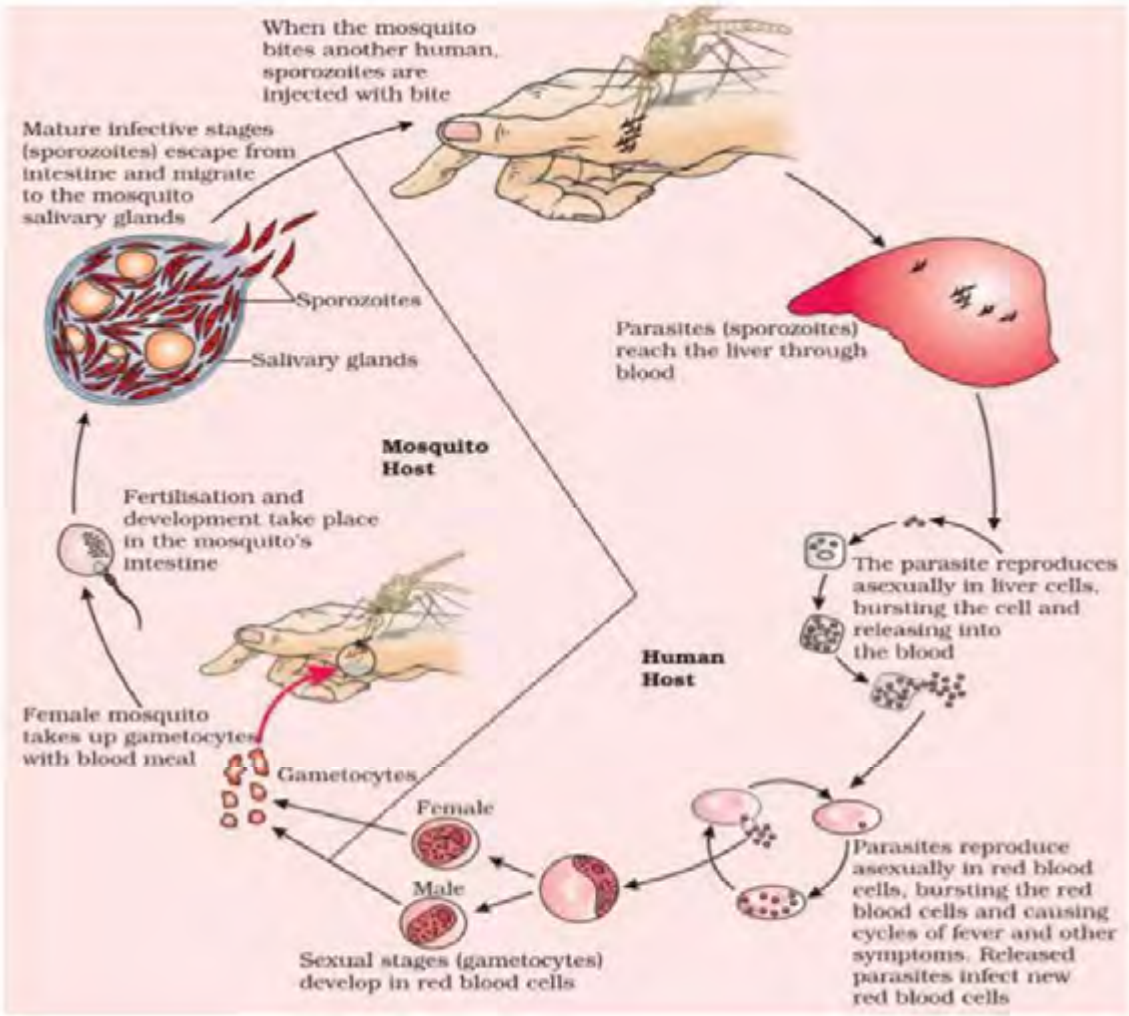
Malaria

- It is a protozoan disease caused by different species of plasmodium such as
- *Plasmodium vivax,*
- *Plasmodium malariae,*
- *Plasmodium ovale and*
- *Plasmodium falciparum.*
- Of these malaria caused by Plasmodium falciparum is the most dangerous and even fatal.

Life cycle

- Plasmodium is a **digenetic parasite** which requires two hosts for the completion of its life.
- **Man is the primary host and Anopheles mosquito is the secondary host or vector.**
- Plasmodium enters the body as **sporozoites** (infectious form) through the bite of infected anopheles mosquito.
- Sporozoites undergo multiplication in the liver cells and then attack the RBC resulting in their rupture.

- The rupture of RBC is associated with the release of toxic substance known as **haemozoin,** which is responsible for chill and recurrent fever every 3-4 days.
- When an infected mosquito bites an infected person, these parasites enter the body of mosquito for further development.
- The parasites undergo multiplication within the body of mosquito and form sporozoites that are stored in the salivary glands.
- When these mosquitoes bite a healthy person, the sporozoites are introduced into his body and the above cycle is repeated.



AMOEBIASIS

- It is a protozoan disease of large intestine caused by *Entamoeba histolytica.*
- It is also known as amoebic dysentery.
- It is transmitted through contaminated food and water by faecal matters or it is associated with unhygienic condition.
- House flies play a vital role in the transmission of amoebiasis.
- Symptoms include abdominal pain and cramps, stools with excess mucous and blood coats.

HELMINTH DISEASES

- Include **ascariasis, filariasis etc.**

Ascariasis

- It is also called worm trouble caused by helminth parasite known as *Ascaris lumbricoides.*
- Transmission of this occurs through contaminated food and water.
- Symptoms include internal bleeding, muscular pain, fever, anaemia and blockage of intestinal passage.
- Ascaris is a monogenetic parasite.
- The eggs of the parasite reach along with the faeces of infected person.

Elephantiasis

- It is also called **filariasis** caused by a helminth known as *Wucheraria bancrofti.*
- Usually it affects the lymphatic vessels and causes their inflammation.
- The affected parts are limbs, genital organs, mammary glands etc.
- When it affects the lymphatic vessels or lower limbs, resulting in the enlargement of limb and is called elephantiasis.
- Wucheraria is a digenetic parasite.
- Man is the primary host and female mosquito belonging to Culex is the vector.
- Transmission occurs when an infected mosquito bites a healthy person.

Diseases caused by fungi

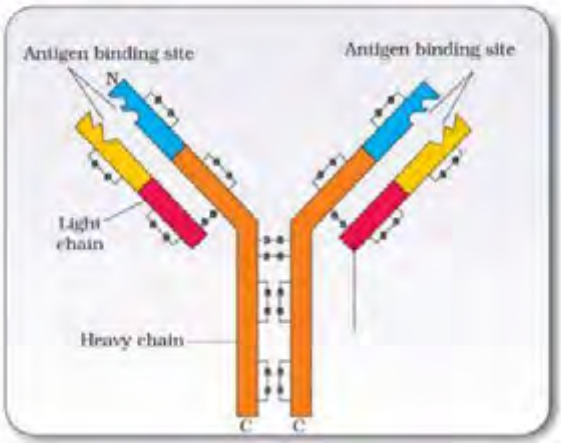
Many fungi belong to the genera

- *Microsporam,*
- *Trichophyton and*
- *Epidermophyton* are responsible for **ring worm.**
- The main symptoms include dry scaly lesions on the skin, nails and scalp.
- Usually scaly lesions are associated with itching.
- Heat and moister in the skin folds such as those in the groin, between toes etc. Help the fungi to grow.
- Transmission mainly occurs by using the clothes, comb etc. of infected persons.

Structure of an Antibody Molecule

- Each antibody molecule has four peptide chains,
- two small chains called light chains and two longer chain called heavy chains.
- Hence, an antibody is represented as H_2L_2
- Different types of antibodies are produced in our body.

  IgA, IgM,IgE, IgG are some of them.



AIDS

- The word AIDS stands for *Acquired Immuno Deficiency Syndrome.*
- It is a disease in which the body lost its acquired immunity.
- AIDS is caused by the *Human Immuno Deficiency Virus* (HIV), a member of a group of viruses called retroviruses.
- Retroviruses have an envelope enclosing the RNA genome

Transmission of HIV-infection

(a) sexual contact with infected person,

(b) by transfusion of contaminated blood and blood products,

(c) by sharing infected needles as in the case of intravenous drug abusers and

(d) from infected mother to her child through placenta.

It is not reasonable to isolate the AIDS patients from family and society.Justify the statement.

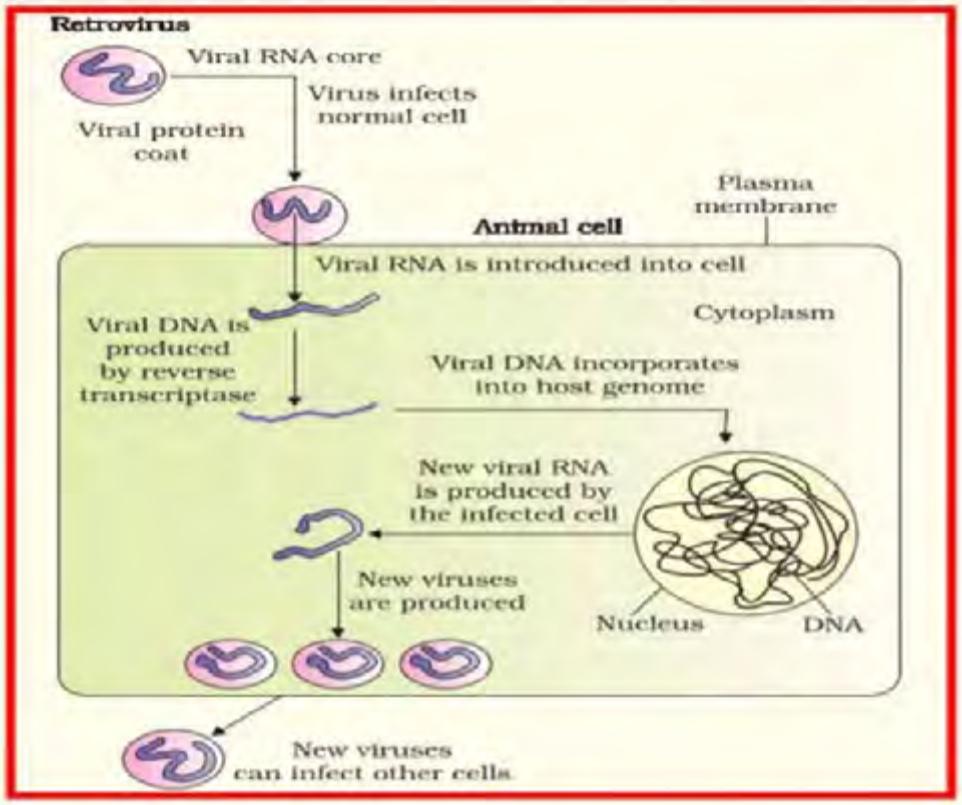
- HIV/AIDS is not spread by mere touch or physical contact.
- It spread only through body fluids.
- So it is not reasonable to isolate the AIDS patient from family and society.

There is always a time lag between infection and appearance of symptoms. The period may vary from a few months to many years (usually 5-10 years).

Replication of Retrovirus

- After getting into the body, the virus enters into the macrophages and converts its RNA genome into DNA with the help of reverse transcriptase enzyme.
- The viral DNA gets incorporated into host cell's DNA and directs the infected cells to produce more virus particles. i.e, the infected macrophages act like a HIV factory.
- Simultaneously, HIV enters into helper T-lymphocytes (T_H), replicate and produces more viruses.
- Then these viruses are released into the blood and attack other T-lymphocytes.

- This will lead to the decrease in number of T-lymphocytes and the patient begins to show the symptoms such as fever, diarrhoea, weight loss etc.
- Subsequently, the immune system weakens and becomes more prone to infections of bacteria like Mycobacterium, viruses, fungi and even parasites like Toxoplasma.
- Finally he is unable to protect himself.



ELISA- Test

- A widely used diagnostic test for AIDS is **Enzyme Linked *Immuno-Sorbent Assay (ELISA)*.**

Treatment

- Treatment of AIDS with anti-retroviral drugs is only partially effective.
- They can only prolong the life of the patient but cannot prevent death, which is inevitable.

Prevention of AIDS

- No treatment is available for AIDS. Prevention is the best option.
- In our country the National AIDS Control Organisation (NACO) and other non-governmental organisation (NGOs) are doing a lot to educate people about AIDS.
- WHO has started a number of programmes to prevent the spreading of HIV infection.
- *Making blood (from blood banks) safe from HIV,*
- *ensuring the use of only disposable needles and syringes in public and private hospitals and clinics,*
- *free distribution of condoms,*
- *controlling drug abuse,*
- *advocating safe sex and promoting regular check-ups for HIV in susceptible* populations.

CANCER

- *It is a disease characterized by the* uncontrolled growth of cells resulting in the formation of tumour.
- Normal cells show a property called contact inhibition by virtue of which contact with other cells inhibits their uncontrolled growth.
- Cancer cells appears to have lost this property.
- As a result of this, cancerous cells just continue to divide giving rise to masses of cells called tumors.

Tumors are of two types:

Benign and Malignant.

1) Benign tumour

- The tumours which is confined to the place of its origin is called benign tumour.
- These tumours do not migrate but grow in size.

2) Malignant tumour

- These tumour cells grow very rapidly, invading and damaging the surrounding normal cells.
- The cells from this tumours are carried to other parts of the body through blood and lymph and where they form secondary growth.

Metastasis

- The spread of cancerous cells to other parts of the body is called metastasis.

Treatment of cancer

Surgery

- *Surgical removal of tumour* mass is one of the easiest approaches in the treatment of cancer.
- But it is not applicable in all types of cancer.

Radiation therapy

- *Treatment of cancer with radiations* like X-rays, gamma rays or electron beams is called radiation therapy.
- In this method tumour cells are irradiated lethally, taking proper care of the normal tissues surrounding the tumour mass.

Chemotherapy

- Treatment of cancer with anticancer drugs to kill cancer cells is known as chemotherapy.
- Majority of drugs have side effects like hair loss, anaemia etc

Most cancers are treated by combination of surgery, radiotherapy and chemotherapy.

Tumour cells are not detected and destroyed by immune system. So the patients are given substances called alpha interferon which activates their immune system and help in destroying the tumour.

Effects of Drug/Alcohol Abuse

- Excessive doses of drugs may lead to coma and death due to respiratory failure, heart failure or cerebral hemorrhage.
- A combination of drugs or their intake along with alcohol generally results in overdosing and even deaths.
- Sometimes abuser may turn to stealing for getting money to buy drugs/alcohol.
- Those who take drugs intravenously, likely to acquire infections like AIDS and hepatitis B.
- The chronic use of alcohol and drugs damage nervous system and liver (cirrhosis).
- The use of drugs/alcohol during pregnancy adversely affects the foetus.

Warning Signs of Drug and Alcohol Abuse

- The most common warning signs of drug and alcohol abuse among youth include
- drop in academic performance,
- unexplained absence from school/college,
- lack of interest in personal hygiene,
- withdrawal,
- isolation,
- depression,
- fatigue, aggressive and rebellious behaviour,
- deteriorating relationships with family and friends,
- loss of interest in hobbies,
- change in sleeping and eating habits,
- fluctuations in weight, appetite, etc

Misuse of Drugs by Sports persons

- Certain sports persons misuse drugs to enhance their performance.
- They (mis)use narcotic analgesics,
- anabolic steroids,
- diuretics and

- certain hormones to increase muscle strength and and to promote aggressiveness and as a result increase athletic performance.

Side-effects of anabolic steroids in females

The side-effects of the use of anabolic steroids in females include

- masculinisation (features like males),
- increased aggressiveness,
- mood swings,
- depression,
- abnormal menstrual cycles,
- excessive hair growth on the face and body,
- enlargement of clitoris,
- deepening of voice.

Side-effects of anabolic steroids in males

- In males it includes acne, increased aggressiveness, mood swings, depression, reduction of size of the testicles, decreased sperm production, potential for kidney and liver dysfunction, breast enlargement, premature baldness, enlargement of the prostate gland.

Prevention and control

- Generally the teenagers and youngsters fall in the habits of smoking, drinking or taking drug.
- One of the most effective measures to save them is to identify the situations that may push an adolescent towards the use of drugs, alcohol etc.

Following are some of the measures useful for prevention and control of alcohol and drug abuse

(i) Avoid undue peer pressure -

- Every child has his/her own choice and personality, which should be respected and nurtured.
- A child should not be pushed unduly to perform beyond his/ her threshold limits; be it studies, sports or other activities.

(ii) Education and counselling -

- Educating and counselling him/ her to face problems and stresses, and to accept disappointments and failures as a part of life.
- It would also be worthwhile to channelise the child's energy into healthy pursuits like sports, reading, music, yoga and other extracurricular activities.

(iii) Seeking help from parents and peers -

- Help from parents and peers should be sought immediately so that they can guide appropriately.
- Help may even be sought from close and trusted friends.

(iv) Looking for danger signs -

- Alert parents and teachers need to look for and identify the danger signs discussed above.
- Even friends, if they find someone using drugs or alcohol, should not hesitate to bring this to the notice of parents or teacher in the best interests of the person concerned.

(v) Seeking professional and medical help -

- A lot of help is available in the form of highly qualified psychologists, psychiatrists, and de- addiction and rehabilitation programmes to help individuals who have unfortunately got in the quagmire of drug/alcohol abuse.

MICROBES IN HUMAN WELFARE

Focus Area-2022 Exam

MICROBES IN HOUSEHOLD PRODUCTS

- We use microbes or products derived from them everyday.
- A common example is the production of curd from milk.
- Micro-organisms such as *Lactobacillus* and others commonly called lactic acid bacteria (LAB) grow in milk and convert it to curd.
- Curd improves its nutritional quality by increasing vitamin B12 .
- In our stomach, LAB play very beneficial role in checking disease-causing microbes.
- The dough, which is used for making foods such as dosa and idli is also fermented by bacteria.
- The puffed-up appearance of dough is due to the production of CO2 gas.
- The dough, which is used for making bread, is fermented using baker's yeast (Saccharomyces cerevisiae).
- 'Toddy', a traditional drink of some parts of southern India is made by fermenting sap from palms.
- The large holes in 'Swiss cheese' are due to production of a large amount of CO2 by a bacterium named *Propionibacterium sharmanii.*
- The 'Roquefort cheese' are ripened by growing a specific fungi on them, which gives them a particular flavour.

Chemicals, Enzymes and other Bioactive Molecules

Examples of acid producers

- Aspergillus niger (a fungus) produces citric acid,
- *Acetobacter aceti* (a bacterium) produces acetic acid;
- *Clostridium butylicum* (a bacterium) produces butyric acid and
- *Lactobacillus* (a bacterium) produces lactic acid.
- Yeast (*Saccharomyces cerevisiae*) is used for commercial production of ethanol.

Microbes used for production of enzymes

- **Lipases** are used in detergent formulations and are helpful in removing oily stains from the laundry.
- **Pectinases and Proteases** are used for the preparation of bottled juices.

Streptokinase

- **Streptokinase** produced by the bacterium ***Streptococcus*** and modified by genetic engineering is used as a 'clot buster' for removing clots from the blood vessels of patients who have undergone myocardial infarction leading to heart attack.

Cyclosporin A

- The bioactive molecule, **Cyclosporin A**, is used as an immunosuppressive agent in organ-transplant patients, is produced by the fungus ***Trichoderma polysporum.***

Statins

- **Statins produced by the yeast *Monascus purpureus*** have been commercialised as blood-cholesterol lowering agents.
- It acts by competitively inhibiting the enzyme responsible for synthesis of cholesterol.

Organisms	Scientific name	Product	Uses
Bacteria	*Streptococcus*	Streptokinase	clot buster in myocardial infarction
Yeast	*Monascus purpureus*	Statins	blood-cholesterol lowering agent
Fungus	*Trichoderma polysporum*	Cyclosporin A	immunosuppressive agent in organ-transplantation

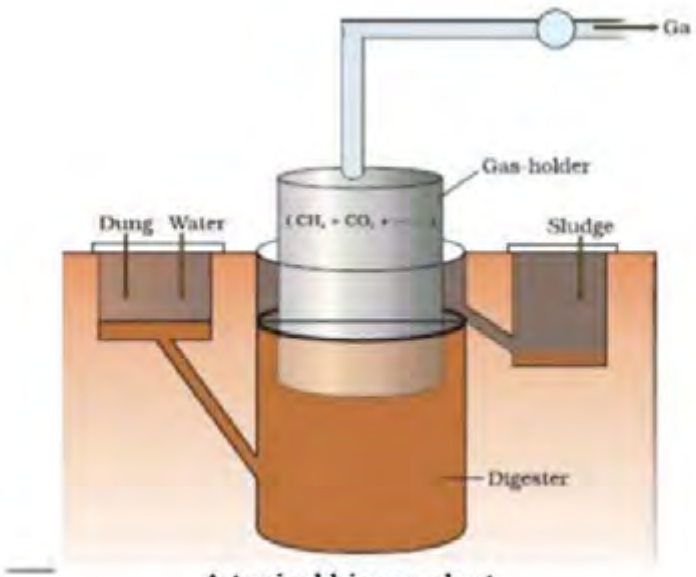
Organisms	Scientific name	Bioactive Product
Bacteria	*Lacto bacillus*	Lactic acid
Bacteria	*Acetobacter aceti*	Acetic acid
Bacteria	*Clostridium butylicum*	Butric acid
Bacteria	*Streptococcus*	Streptokinase
Yeast	*Monascus purpureus*	Statins
Yeast	*Saccharomyces cerevisiae*	Ethanol
Fungus	*Aspergillus niger*	Citric acid
Fungus	*Trichoderma polysporum*	Cyclosporin A

MICROBES IN PRODUCTION OF BIOGAS

- Biogas is a mixture of gases such as methane, CO_2 and H_2S. The methane is the predominant gas.
- It is produced by the microbial activity and is used as a source of energy.
- The bacteria grow anaerobically on cellulose, produce large amount of methane along with hydrogen and CO_2.
- These bacteria are collectively called **methanogens.**
- One of the common metahanogen is Methanobacterium.
- They are present in the sewage and also in the rumen of cattle where a large amount of cellulosic food is present.
- These bacteria help in the breakdown of cellulose and thus play a vital role in digestion.
- Thus the excreta of cattle, commonly called gobar can be used for the production of biogas and hence called **gobar gas.**

BIOGAS PLANT

- It consists of a concrete tank that measures 10-15 feet deep.
- Bio wastes are collected and slurry of dung is fed in the tank.
- A floating cover is placed over the slurry, which keeps on rising as the gas is produced in the tank due to the microbial activity.
- The plant has an outlet, which is connected to a pipe to supply the gas to the nearby houses.
- The spent slurry is collected through another outlet and may be used as fertilizer.
- The biogas produced is used for cooking and lighting.
- The technology of biogas production was developed in India by Indian Agricultural Institute (IARI) and Khadi and Village Industries Commission (KVIC).



A typical biogas plant

MICROBES AS BIOCONTROL AGENTS

Example of microbial biocontrol agents- *Bacillus thuringiensis*

- An example of microbial biocontrol agents that can be introduced in order to control butterfly caterpillars is the bacteria ***Bacillus thuringiensis*** (often written as Bt).
- These are available in sachets as dried spores which are mixed with water and sprayed onto vulnerable plants such as brassicas and fruit trees, where these are eaten by the insect larvae.
- In the gut of the larvae, the toxin is released and the larvae get killed.
- The bacterial disease will kill the caterpillars, but leave other insects unharmed.
- Because of the development of methods of genetic engineering in the last decade or so, the scientists have introduced **B. Thuringiensis toxin genes** into plants.
- Such plants are resistant to attack by insect pests.
- **Bt-cotton is one such example**, which is being cultivated in some states of our country.

Trichoderma

- **Trichoderma species are free-living fungi that are very common in the root ecosystems.**
- **They are effective biocontrol agents of several plant pathogens.**

MICROBES AS BIOFERTILIZERS

- Biofertilizers are organisms that enrich the nutrient quality of the soil.
- The main source of biofertilizers is bacteria, fungi and cyanobacteria.
- **Rhizobium**, found in the roots of leguminous plants fixes atmospheric nitrogen into organic forms which is used by the plant as nutrient.
- **Azospirillum and Azobacter**, the free living bacteria are also able to fix atmospheric nitrogen.
- Certain fungi such as **mycorrhiza** form symbiotic association with plants.
- The fungal symbiont helps the plants to absorb phosphorus from the soil.
- The other benefits with this association are resistance to root-borne pathogens, tolerance to salinity and drought.
- Moreover the growth of the plant is accelerated by this association.
- Cyanobacteria such as Anabaena, Nostoc, Oscillatoria etc. living in the terrestrial and aquatic habitats can also fix atmospheric nitrogen.
- In the paddy fields, cyanobacteria serve as an important biofertilizer.
- The biofertilizers enrich the soil nutrients and it reduces the use of chemical fertilizers.

BIODEVRSITY AND CONSERVATION

Focus area-2022 Exam

Components of biodiversity

- **Genetic diversity,**
- **species diversity and**
- **ecological diversity** are the three important components of biodiversity

1. Genetic diversity

- The variation of genes within the species is known as genetic diversity.

Examples

a) The medicinal plant *Rauwolfia vomitoria* growing in different Himalayan ranges produce different varieties of active chemical *reserpine.*

b) India has more than 50,000 genetically different strains of rice and 1000 varieties of mango.

2. Species diversity

- The occurrence of different verity of species within a region is called species diversity.

Example;-

The Western Ghats have greater number of amphibian species than Eastern Ghats.

3. Ecological diversity

- Diversity at the level of community and ecosystem is known as ecological diversity.

Example;-

- India has greater ecosystem diversity in deserts, rain forests, mangroves, coral reefs, wet lands, estuaries and alpine meadows than the country like Norway.

PATTERNS OF BIODIVERSITY

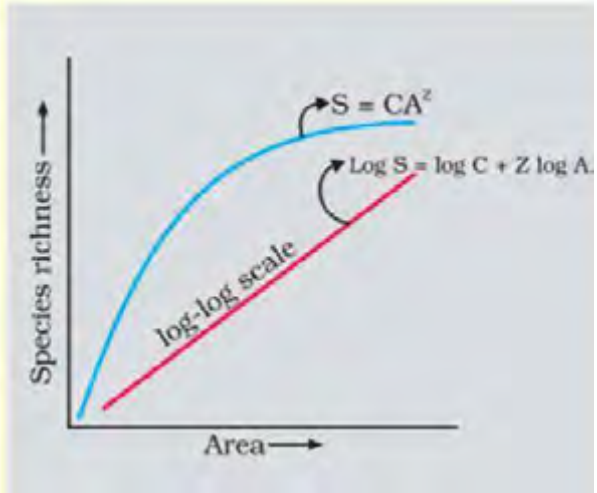
Two patterns of biodiversity are present

I. **Lattitudinal gradients**

II. **Species area relationship**

Explain species area relationship in biodiversity?

- **The great German Naturalist and Geographer Alexander Von Humboldt observed that within a region species richness increased with increasing explored area, but only up to a limit.**
- **The relation between species richness and area for a wide variety of taxa such as angiosperm plants, birds, bats, freshwater etc turns out to be a rectangular hyperbola.**



- **On a logarithmic scale, the relationship is a straight line described by the equation,**

Log S = log C + Z log A

Where, S = species richness

C = Y intercept

Z = slope of the line (regression coefficient)

A = Area

What is the significance of the slope of regression coefficient in a species area relationship?

- **Ecologists have discovered that the value of Z lies in the range of 0.1 to 0.2 regardless of the taxonomic group or the region.**
- **But, if we analyse the species area relationship among very large areas like the entire continents, we will find the slope of the line to be much steeper; that is Z values in the range of 0.6 to 1.2.**
- **For example, for frugivorous (fruit eating) birds and mammals in the tropical forest of different continents, the slope is found to be 1.15.**

VALUE OF Z LINE

Any taxonomic group /region	Between 0.1 and 0.2
Very large area like a continent	0.6 to 1.2
Frugivorous birds and mammals in tropical rainforest	1.15

LOSS OF BIODIVERSITY

What are the causes of biodiversity losses or species losses in a geographical region ?

OR

what *are "The Evil Quartlet"of biodiversity losses?*

- **The extinction species and consequent loss of biodiversity are due to mainly four major reasons. They are known by the sobriquet *"The Evil Quartlet"***

1. **Habitat loss and fragmentation**
2. **Over exploitation**
3. **Alien species invasions**
4. **Co-extinctions**

1. Habitat loss and fragmentation

- It is due to cutting down of trees, ploughing a grassland, filling a wetland or burning a forest.
- The best example is the tropical rain forest. Once the forest covered the 14% of earth's land. Now it covers only 6%.
- The Amazonian rain forest, the ***lungs of the planet,*** harbouring millions of species is being cut for cultivating soya beans or for conversion to grasslands for raising beef cattle.
- The fragmentation of the forest i.e., broken down into small fragments due to human activities also led to the disappearance of many species.

2. Over - exploitation

Humans have always depended on nature for food and shelter, but when 'need' turns to 'greed', it leads to over -exploitation of natural resources.

Example

- The extinction of Steller's sea cow and passenger pigeon were due to over exploitation by humans.

3. Alien species invasions

- *(Introduction of exotic species is one of the major threats to biodiversity. Substantiate?)*

- New species entering a geographical region are known as exotic or alien species.
- Introduction of alien species to a new habitat may adversely affect the native species. Some time it may leads to the extinction of the species.

Examples

- The Nile perch introduced into Lake Victoria in east Africa led to the extinction of 200 species cichlid fish in the lake.

- **Carrot grass (Parthenium), Lantana and water hyacinth (eicchornia) is also alien invasive species that destroyed the environment.**
- **The introduction of African cat fish *Clarias gariepinus* for aquaculture purpose is posing a threat to the native cat fishes.**

4. Co-extinctions

- **When a species extinct, the animal or plant associated with it also extinct. This is known as co-extinction.**

Examples

- **The extinction of a host fish leads to the extinction of parasites associated with it**
- **In co evolved plant-pollinator mutualism extinction of one leads to the extinction of the other.**

BIODIVERSITY CONSERVATION

How do we conserve biodiversity?

Conservation of biodiversity can be done by two ways.

1. ***In situ conservation(on site)***
2. ***Ex situ conservation(off site)***

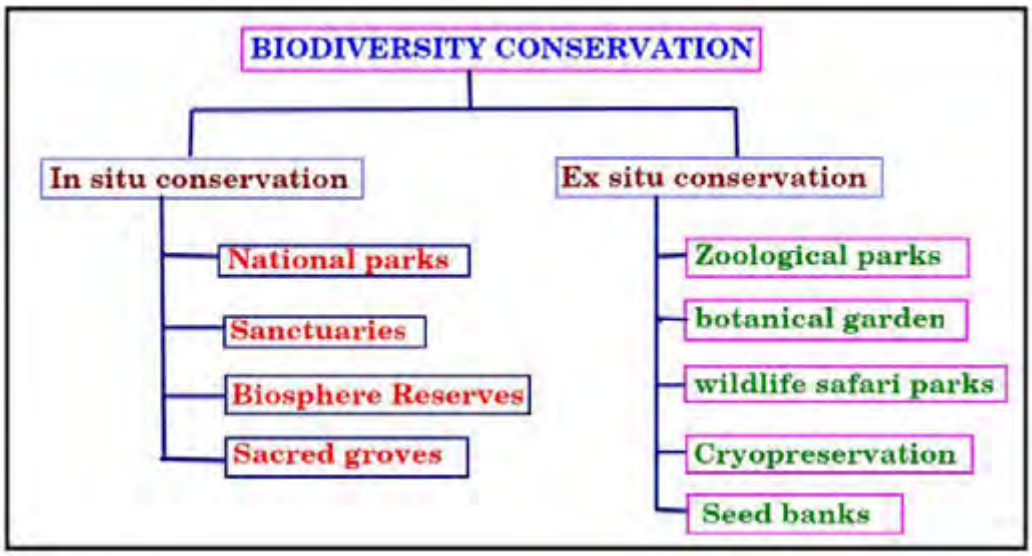
1. In situ conservation

- **The conservation of genetic resources through their maintenance within natural or even human made ecosystem in which they occur is called in situ conservation.**
- **'biodiversity hotspots' are the regions with very high levels of species richness and high degree of endemism.**
- **The species confined to a region and not found anywhere else are known as endemic species.**
- **There are 34 biodiversity hotspots in the world.**

- **Three of these hotspots – Western Ghats and Sri Lanka, Indo-Burma and Himalaya – cover our country's exceptionally high biodiversity regions.**
- **In India, ecologically unique and biodiversity-rich regions are legally protected as biosphere reserves, national parks and sanctuaries.**
- **India now has 14 biosphere reserves, 90 national parks and 448 wildlife sanctuaries.**
- **Sacred groves are forests protected by tribal communities, which are free from all disturbances. Tribal give religious sanctity to the forest. The flora and fauna is well protected here.**
- **Sacred groves are found in Meghalaya, Rajasthan, Western Ghat regions of Karnataka and Maharashtra and Madhya Pradesh.**
- **In Meghalaya, the sacred groves are the last refuges for a large number of rare and threatened plants.**

2. Ex situ conservation

- **In this approach, threatened animals and plants are taken out from their natural habitat and placed in special setting where they can be protected and given special care.**
- **Zoological parks, botanical gardens and wildlife safari parks, Cryopreservation, Seed banks etc are examples for Ex situ conservation.**
- **There are many animals that have become extinct in the wild but continue to be maintained in zoological parks.**
- **Cryopreservation techniques are used for preserving gametes of threatened species in viable and fertile condition for long periods.**
- **Seed banks are used for preserving seeds of different genetic strains of commercially important plants for long periods.**


BIODIVERSITY CONSERVATION
In situ conservation
National parks
Sanctuaries
Biosphere Reserves
Sacred groves
Ex situ conservation
Zoological parks
botanical garden
wildlife safari parks
Cryopreservation
Seed banks