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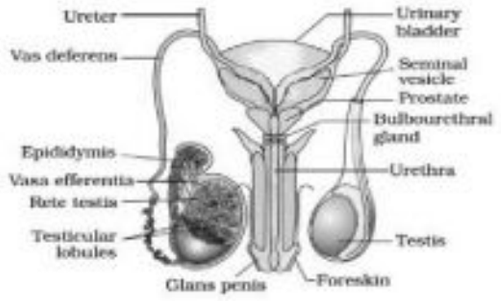
3. HUMAN REPRODUCTION

HUMAN REPRODUCTIVE SYSTEM

1. Male Reproductive System

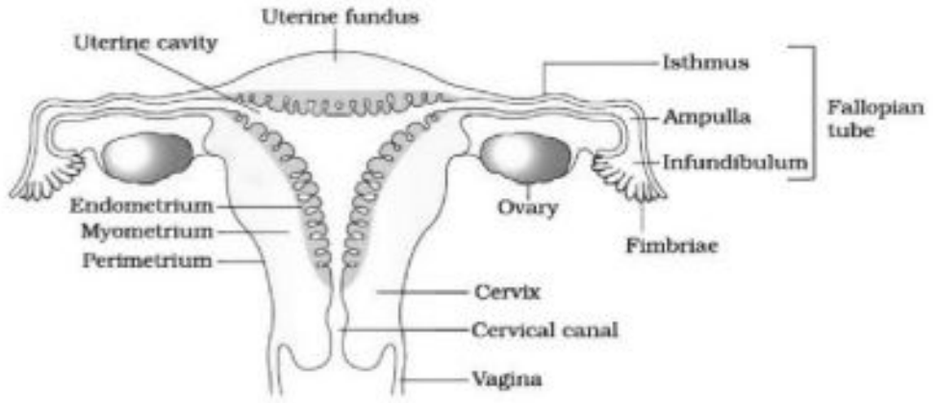


- **Testes** found in the **scrotal sac (scrotum)**.
- The low temperature (2-2.5^0 C less than the body temperature) of scrotum helps for **spermatogenesis.**
- **Seminiferous tubule** is lined internally with
  - **Male germ cells (spermatogonia):** They become sperms.
  - **Sertoli cells:** They give nutrition to the germs cells.**b.**

Duct system

- Include **rete testis, vasa efferentia, epididymis** & **vas deferens.** They conduct sperms from testis as follows:
- Seminiferous tubules → **rete testis → vasa efferentia → epididymis** (stores sperms temporarily) → **vas deferens** → join with duct of **seminal vesicle** to form **ejaculatory duct → urethra → urethral meatus.**

2. Female Reproductive System



Accessory ducts (Duct system)

Include 2 **oviducts (Fallopian tubes), a uterus** & **vagina.**

- **Oviducts:** Each oviduct (10-12 cm long) has 3 parts:
  - **Infundibulum:** Funnel-shaped opening provided with many finger-like **fimbriae.** It helps to collect the ovum.
  - **Ampulla:** Wider part.
  - **Isthmus:** Narrow part. It joins the uterus.
- **Uterus (womb):**
  The uterine wall has 3 layers:
  - **Perimetrium**: External thin membrane.
  - **Myometrium**: Middle thick layer of smooth muscle.
  - **Endometrium**: Inner glandular and vascular layer.
- **Hymen (Maiden head):** A membrane which partially cover the vaginal opening. It is often torn during the first coitus. It may also be broken by a sudden fall , active participation in some sports items etc. In some women, hymen persists after coitus. So the hymen is not a reliable indicator of virginity or sexual experience.

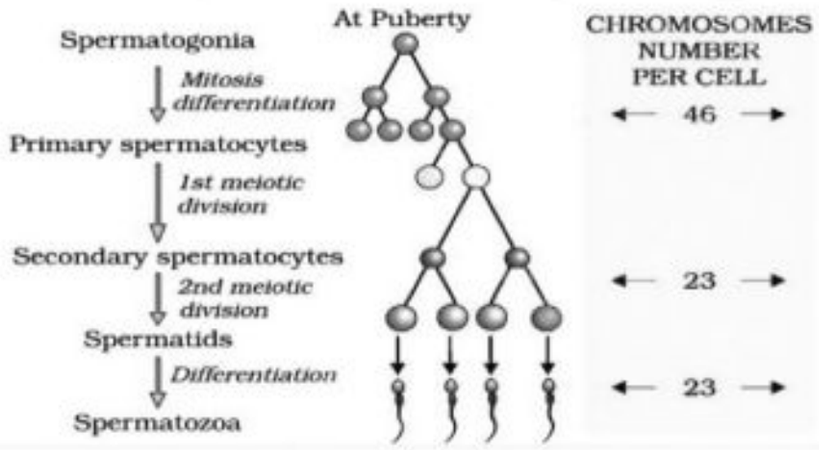
GAMETOGENESIS

- It is 2 types: **Spermatogenesis** and **Oogenesis.**

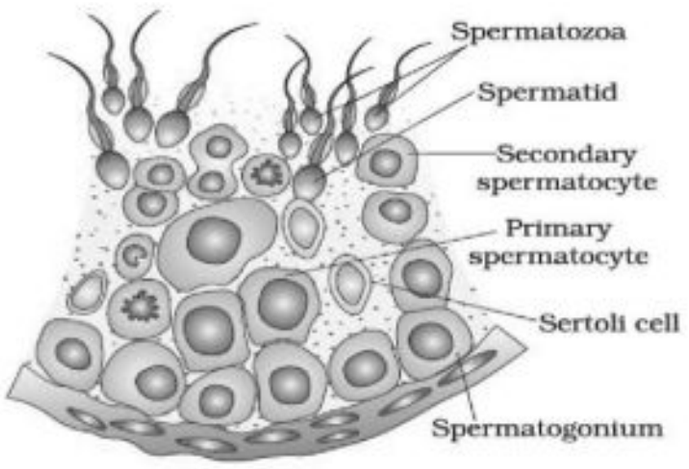
SPERMATOGENESIS

It is the formation of sperms

Schematic representation of spermatogenesis



- After spermiogenesis, sperm heads are embedded in Sertoli cells to get nourishment. Then they are released to lumen of seminiferous tubules. It is called **spermiation.**



Diagrammatic sectional view of a seminiferous tubule

Role of Hormones in Spermatogenesis

Hypothalamus

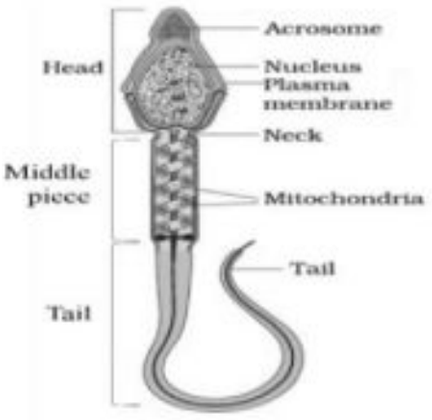
Releases GnRH

Pituitary

LH → Acts on Leydieg cells and stimulate secretion of androgens

FSH → Acts on Sertoli cells and stimulate spermiogenesis

Structure of spermatozoa (Sperm)

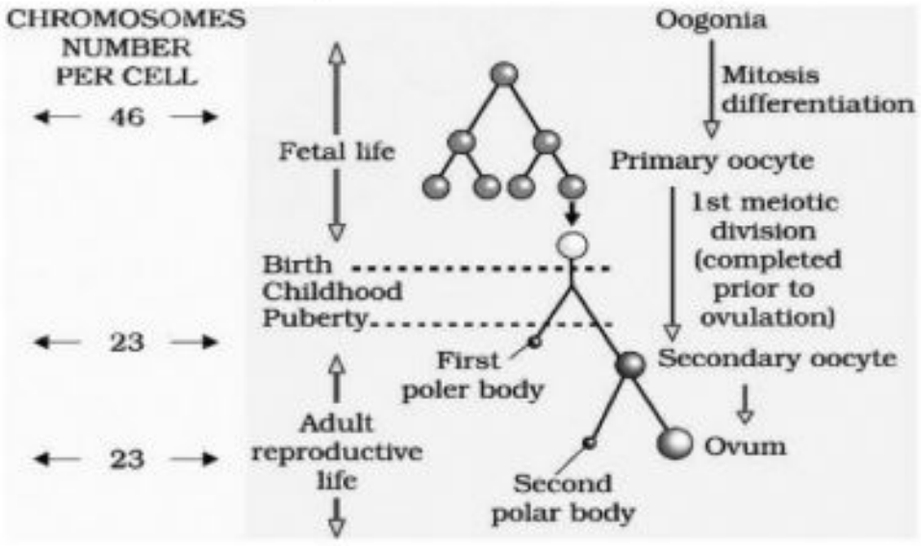


- A sperm has 3 regions:
  a. **Head:** Formed of **nucleus** and **acrosome.** Acrosome release enzymes which break the membranes of ovum and helps for fertilization
  b. **Middle piece:** Composed mitochondria , Mitochondria produce energy for the sperm motility.
  c. **Tail:** The sperm moves in fluid medium and female genital tract by the **movement** of the tail.

Oogenesis

- It is the process of formation and maturation of **ovum.**

Schematic representation of oogenesis



Differences between spermatogenesis &Ooogenesis

Spermatogenesis	Oogenesis
Formation of sperm	Formation of ovum
Occurs in testis	Occurs in Ovary
Starts at puberty	Starts at foetal life
Equal meiotic division	Unequal meiotic division
One primary sprmatocyte produces four sperm	One primary oocyte produces one ovum.

ROLE OF HORMONES IN MENSTRUAL CYCLE

Pitutary Hormones – FSH & LH

FSH- stimulates Development of ovarian follicle

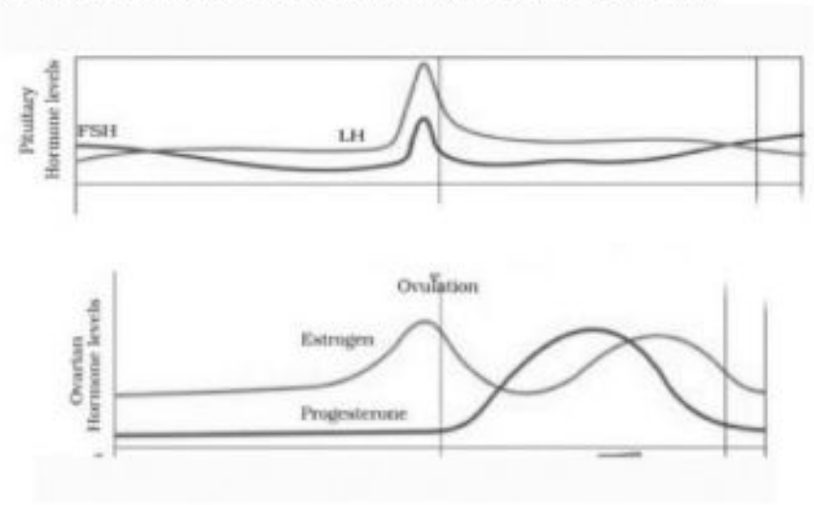
LH – Rapid secretion of LH (**LH surge**) induces rupture of Graafian follicle and thereby **ovulation** (on 14th day).

Ovarian hormones- Oestrogen& progesterone

- **Oestrogens** stimulate
  - **Proliferation** of **ruptured uterine endometrium**.
  - Suppression of FSH secretion.
  - Secretion of LH (Luteinizing hormone).

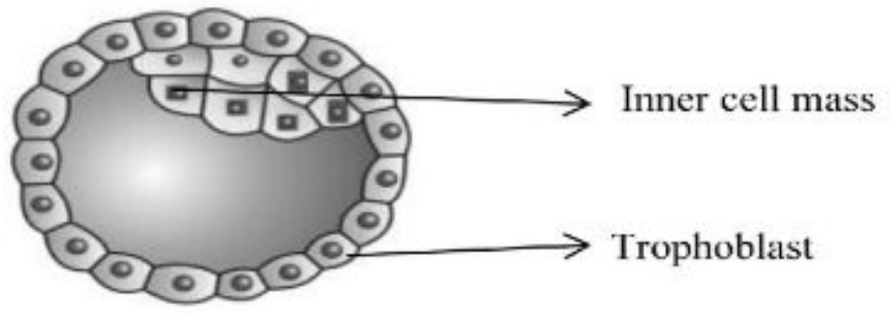
progesterone:

Makes endometrium **maximum vascular, thick** and **soft.** it maintains uterus and support pregnancy



FERTILIZATION

- Fusion of a sperm with ovum is called **fertilization.** It occurs in **Ampullary region** of fallopian tube.
- The embryo with 8-16 blastomeres is called a **morula.**
- Morula continues to divide and transforms into **blastocyst.**
- In blastocyst, blastomeres are arranged into **trophoblast** (outer layer) and an **inner cell mass** attached to trophoblast.
- The trophoblast layer gives nourishment to inner cell mass. Also, it gets attached to endometrium.
- Inner cell mass becomes embryo



(Blastocyst)

PLACENTA

placenta. It is a structural and functional unit b/w embryo

Function of Placenta

- Acts as **barrier** between the foetus and mother.
- Supply **O_2, nutrients** etc. from mother to foetus.
- Remove **CO_2 and excretory wastes** from foetus.
- Acts as an endocrine gland. It secretes **Human chorionic gonadotropin (hCG), human placental lactogen (hPL), oestrogens, progesterone** & **relaxin.**

COLOSTRUM

- The yellowish milk produced during the initial few days of lactation is called **colostrum.** It contains several antibodies essential to develop resistance for the new born babies.

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4. REPRODUCTIVE HEALTH

Reasons for population explosion

- Increased health facilities and better living conditions.
- Rapid decline in death rate, **maternal mortality rate (MMR)** and **infant mortality rate (IMR).**
- Increase in number of people in reproducible age.

CONTRACEPTIVE METHODS

Natural/Traditional methods

- Avoid chances of ovum and sperms meeting. It includes
  - **Periodic abstinence:** Avoid coitus from day **10 to 17 (fertile period)** of menstrual cycle to prevent conception.
  - **Coitus interruptus (withdrawal):** Withdraw penis from the vagina just before ejaculation to avoid insemination.
  - **Lactational amenorrhea:** It is the absence of menstrual cycle & ovulation due to intense lactation after parturition. Fully breastfeeding increases lactation.This is effective up to 6 months following parturition.
- Natural methods have **no side effect**. But chances of **failure** are **high.**

Barriers

- They prevent physical meeting of sperm & ovum. E.g.
  - **Condoms (E.g. Nirodh):** it also Protects from STD s
  - **Diaphragms, cervical caps** and **vaults:** Used in Female

Intra Uterine Devices (IUDs)

IUDs are ideal method to delay pregnancy or space children.

Types of IUDs:

IUDs	ACTION	EXAMPLES
Non-medicated IUDs	**They retard sperm motility**	**Lippes loop.**
Copper releasing IUDs:	**Cu ions suppress motility and fertilising capacity of sperms.**	**CuT, Cu7, Multiload 375.** 
Hormone releasing IUDs	**They make the uterus unsuitable for implantation**	**Progestasert, LNG-20.**

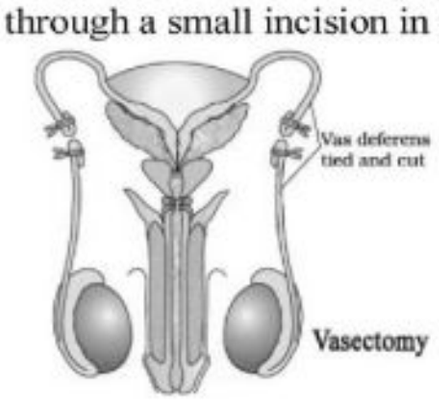
Oral contraceptives

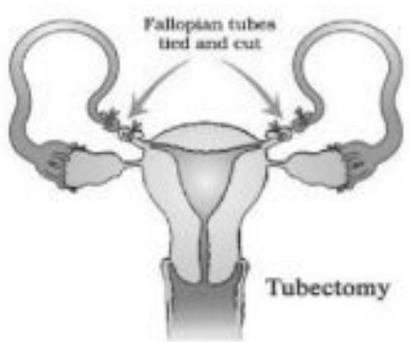
- Oral administration of **progestogens** or **progestogen–oestrogen** combinations in the form of tablets (**pills**).
- They inhibit ovulation and implantation and thicken cervical mucus to prevent entry of sperms.
- Pills are very effective with lesser side effects.
- ***Saheli:*** New oral contraceptive for the females. It is developed by **Central Drug Research Institute (CDRI, Lucknow).**

Surgical methods (sterilization)

It is very effective but reversibility is very poor.

- **Vasectomy:** Sterilization procedure in males. In this, a small part of the vas deferens is removed or tied up through a small incision on the scrotum.
- **Tubectomy:** Sterilization procedure in females. In this, a small part of the fallopian tube is removed or tied up through a small incision in the abdomen or through vagina.





MEDICAL TERMINATION OF PREGNANCY (MTP)

- Intentional or voluntary termination of pregnancy before full term is called **MTP** or **induced abortion.**

Importance of MTP

- To avoid unwanted pregnancies due to casual intercourse or failure of the contraceptive used during coitus or rapes.
- It is essential in cases where continuation of pregnancy could be harmful to the mother or to the foetus or both.

Amniocentesis: In this, some amniotic fluid of the foetus is taken to analyse the foetal cells & dissolved substances. It is used to test the presence of genetic disorders, survivability of the foetus etc.

SEXUALLY TRANSMITTED DISEASES- EXAMPLES & PREVENTION

- Diseases transmitted through sexual intercourse .
  E.g. Gonorrhoea,
- syphilis,
- genital herpes,
- chlamydiasis,
- genital warts,
- trichomoniasis,
- hepatitis-B & AIDS.

- **Prevention:**
  - Avoid sex with unknown partners/multiple partners.
  - Always use condoms during coitus.
  - In case of doubt, go to a qualified doctor for early detection and get complete treatment.

INFERTILITY & ASSISTED REPRODUCTIVE TECHNOLOGIES

- **Infertility** is the inability to produce children .
- The reasons for this may be physical, congenital, diseases, drugs, immunological or even psychological.
- The technologies used to correct the infertility problems are called **Assisted Reproductive Technologies (ART).** Some of them are given below:

1. *In vitro* fertilisation (IVF) or Test tube baby programme

In this method, ova from the wife/donor and sperms from the husband/donor are collected and are induced to form zygote under simulated conditions in the laboratory. This is followed by **Embryo transfer (ET).**

ET is 2 types:

- **Zygote Intra Fallopian Transfer (ZIFT):** Transfer of zygote or early embryo (with up to 8 blastomeres) into fallopian tube.
- **Intra Uterine Transfer (IUT):** Transfer of embryo with more than 8 blastomeres into the uterus.

2. Gamete Intra Fallopian Transfer (GIFT)

Transfer of an ovum from a donor into the fallopian tube of another female who cannot produce ovum, but can provide suitable environment for fertilization and development.

3. Intra cytoplasmic sperm injection (ICSI)

It is a laboratory procedure in which a single sperm (from male partner) is injected directly into an egg (from female partner). After fertilization, the embryo is implanted into the woman's uterus.

4. Artificial insemination (AI) technique

The semen collected from husband or a donor is artificially introduced into the vagina or the uterus of the female.
Artificial insemination into the uterus is known as **intra-uterine insemination (IUI)**.
This technique is useful for the male partner having inability to inseminate female or low sperm counts etc.

Problems of ART

- It needs specialized professionals & expensive instruments. So these facilities are available only in very few centres.
- Emotional, religious and social problems.

Legal adoption is a good method for couples looking for parenthood.

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5. PRINCIPLES OF INHERITANCE AND VARIATION

Gregor Mendel is the Father of genetics.

Mendel selected 7 pairs of true breeding pea(***Pisum sativum***)varieties:

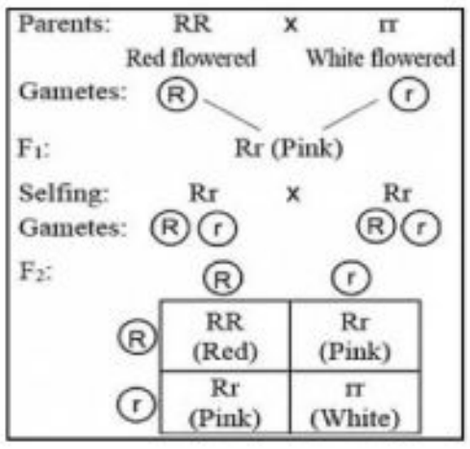
7 Characters	Contrasting Traits	
	Dominant	Recessive
1. Stem height	Tall	Dwarf
2. Flower colour	Violet	White
3. Flower position	Axial	Terminal
4. Pod shape	Inflated	Constricted
5. Pod colour	Green	Yellow
6. Seed shape	Round	Wrinkled
7. Seed colour	Yellow	Green

INHERITANCE OF ONE GENE

Monohybrid cross: A cross involving 2 plants differing in one character pair. E.g. Mendel crossed tall and dwarf pea plants to study the inheritance of one gene.

1. Incomplete Dominance

- It is an inheritance in which heterozygous offspring shows intermediate character b/w two parental characteristics.
- E.g. Flower colour in **snapdragon (dog flower or *Antirrhinum sp.*)** and ***Mirabilis jalapa* (4'O clock plant).**



Here, cross between homozygous **red** & **white** produces **pink** flowered plant. Thus phenotypic & genotypic ratios are same.

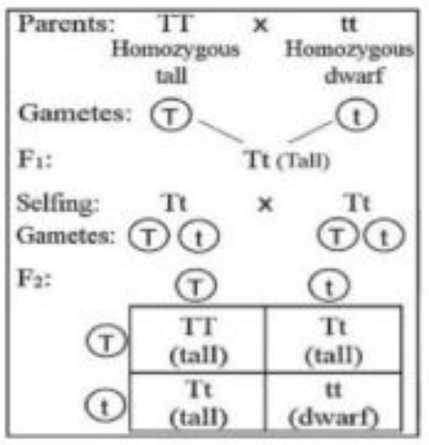
Phenotypic ratio=
1 Red: 2 Pink: 1 White

Genotypic ratio=
1 (RR): 2 (Rr): 1(rr)

- **Allele:** Alternative forms of a gene. E.g. T (tall) and t (dwarf) are two alleles of a gene for the character height.
- **Phenotype:** Physical expression of a character.
- **Genotype:** Genetic constitution of a character.

Monohybrid phenotypic ratio:
3 Tall: 1 Dwarf = **3:1**

Monohybrid genotypic ratio:
1 Homozygous tall (TT)
2 Heterozygous tall (Tt)
1 Homozygous dwarf (tt)
= **1:2:1**



2. Co-dominance

- It is the inheritance in which both alleles of a gene are expressed in a hybrid. E.g. AB blood group in human.
- ABO blood groups are controlled by the gene **I**.
- The gene **I** has three alleles I^A, I^B & **i**.

Alleles from parent 1	Alleles from parent 2	Genotype of offspring	Blood types (phenotype)
I^A	I^A	$I^A I^A$	A
I^A	I^B	$I^A I^B$	AB
I^A	i	$I^A i$	A
I^B	**I^A**	**$I^A I^B$**	**AB**
I^B	I^B	$I^B I^B$	B
I^B	i	$I^B i$	B
i	i	ii	O

When I^A and I^B are present together, they **both express** their own types of sugars. This is due to **co-dominance.**

CHROMOSOMAL THEORY OF INHERITANCE

Proposed by **Walter Sutton** & **Theodore Boveri.**

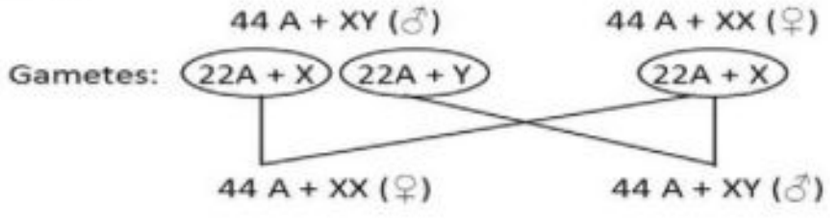
Thomas Hunt Morgan proved chromosomal theory of inheritance using fruit flies ***(Drosophila melanogaster)***.

It is the suitable material for genetic study because,

- They can grow on simple synthetic medium.
- Short generation time (life cycle: 12-14 days).
- Breeding can be done throughout the year.
- Hundreds of progenies per mating.
- Male and female flies are easily distinguishable. E.g. Male is smaller than female.
- It has many types of hereditary variations that can be seen with low power microscopes.

SEX DETERMINATION IN HUMAN BEINGS (XX-XY TYPE)

- Human has **23 pairs** of chromosomes (22 pairs of autosomes and 1 pair of sex chromosomes).
- A pair of X-chromosomes **(XX)** is present in the **female,** whereas **X and Y** chromosomes are present in **male.**
- During spermatogenesis, males produce 2 types of gametes: 50 % with X-chromosome and 50 % with Y-chromosome.
- Females produce only ovum with an X-chromosome.
- There is an equal probability of fertilization of the ovum with the sperm carrying either X or Y chromosome.
- The sperm determines whether the offspring male or female.



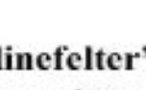
PEDIGREE ANALYSIS

- Analysis of genetic traits in several generations of a family is called **pedigree analysis**.
- In human genetics, pedigree study is utilized to trace the inheritance of a specific trait, abnormality or disease.

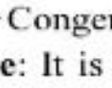
Affected individual: ■ ● ◆ Mating: □—○

Mating b/w relatives (consanguineous mating): □═○

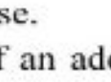
Parents above & children below



Parents with affected male child



Five unaffected offspring



Symbols used in pedigree analysis

Male: □ Female: ○ Sex unspecified: ◇

GENETIC DISORDERS

1. Mendelian Disorders

Haemophilia (Royal disease):

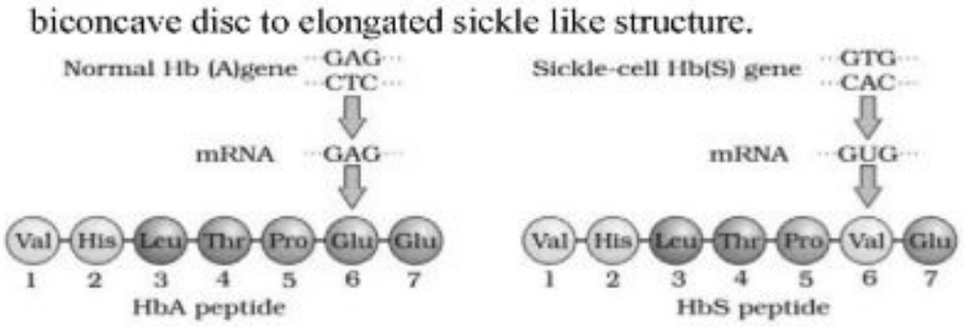
- It is a sex linked (X-linked) recessive disease.
- In this, a protein involved in the blood clotting is affected.
- A simple cut results in non-stop bleeding.
- The disease is controlled by 2 alleles, **H** & **h**. **H** is normal allele and **h** is responsible for haemophilia.
- In females, haemophilia is very rare because it happens only when mother is at least carrier and father haemophilic.
- Queen Victoria was a carrier of hemophilia. So her family pedigree shows many haemophilic descendants.

Sickle-cell anaemia:

- This is an autosome linked recessive disease.
- It can be transmitted from parents to the offspring when both the partners are carrier (heterozygous) for the gene.
- The disease is controlled by a pair of allele, Hb^A and Hb^S.

  Homozygous dominant (Hb^AHb^A): normal
  Heterozygous (Hb^AHb^S): carrier; sickle cell trait
  Homozygous recessive (Hb^SHb^S): affected

- The defect is caused by the substitution of **Glutamic acid (Glu)** by **Valine (Val)** at the **sixth position** of the **β-globin chain** of the haemoglobin (Hb).
- This is due to the single base substitution at the sixth codon of the **β-globin gene** from **GAG** to **GUG.**
- The mutant Hb molecule undergoes polymerization under low oxygen tension causing the change in shape of the RBC from biconcave disc to elongated sickle like structure.



.2. Chromosomal disorders

- **Down's syndrome:** It is the presence of an additional copy of chromosome number 21 **(trisomy of 21).**
  Genetic constitution: 45 A + XX or 45 A + XY (i.e. **47** chromosomes).
  Features:
  - ➢ They are short statured with small round head.
  - ➢ Broad flat face.
  - ➢ Furrowed big tongue and partially open mouth.
  - ➢ Many "loops" on finger tips.
  - ➢ Broad palm with characteristic palm simian crease.
  - ➢ Retarded physical, psychomotor & mental development.
  - ➢ Congenital heart disease.
- **Klinefelter's Syndrome**: It is the presence of an additional copy of X-chromosome in male (trisomy).
  Genetic constitution: 44 A + XXY (i.e. **47** chromosomes).
  Features:
  - ➢ Overall masculine development. However, the feminine development is also expressed. E.g. Development of breast **(Gynaecomastia).**
  - ➢ Sterile.
  - ➢ Mentally retarded.
- **Turner's syndrome**: This is the absence of one X chromosome in female (monosomy).
  Genetic constitution: 44 A + X0 (i.e. **45** chromosomes).
  Features:
  - ➢ Sterile, Ovaries are rudimentary.
  - ➢ Lack of other secondary sexual characters.
  - ➢ Dwarf.
  - ➢ Mentallyretarded.

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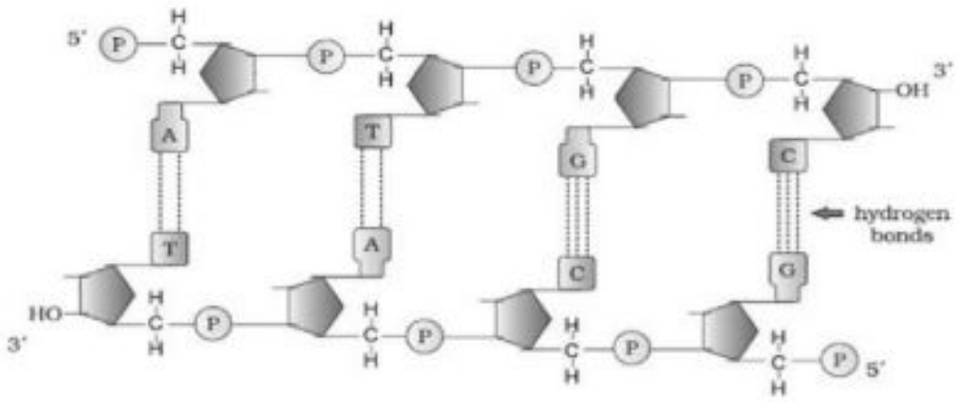
6. MOLECULAR BASIS OF INHERITANCE

THE DNA

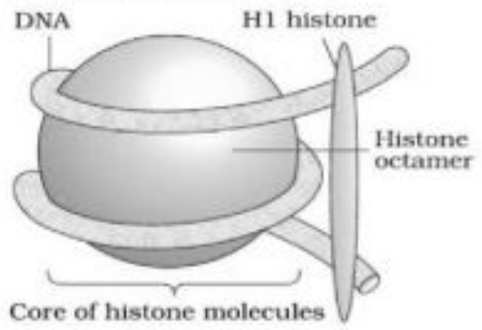
STRUCTURE OF THE DNA



- DNA is made of 2 polynucleotide chains .
- Its backbone is formed of sugar & phosphates.
- The 2 chains have **anti-parallel polarity,**
- one chain has the polarity **5'→3'** and the other has **3'→5'.**
- The bases in 2 strands are paired through **H-bonds**
  A=T (2 hydrogen bonds) C≡G (3 hydrogen bonds)
- **Erwin Chargaff's rule:** In DNA, the proportion of A is equal to T and the proportion of G is equal to C.

PACKAGING OF DNA HELIX

- positively charged, basic proteins **histones present**
- Histones are rich in ***lysines*** and ***arginines.***
- **8 histones** form **histone octamer.**
- Negatively charged DNA is wrapped around histone octamer to give **nucleosome.**



- A typical nucleosome contains **200 bp.**
- repeating unit of nucleosomes form **chromatin.**
- Chromatin has 2 forms:

Euchromatin	Heterochromatin
Loosely packed	Densely packed
Transcriptionally active	Transcriptionally inactive
Stains light	Stains Dark

THE SEARCH FOR GENETIC MATERIAL

Griffith's Transforming Principle experiment (1928)

Frederick Griffith used **mice** & ***Streptococcus pneumoniae.***

Streptococcus pneumoniae has 2 strains:

- **Smooth (S) strain (Virulent):** Has polysaccharide mucus coat. Cause pneumonia.
- **Rough (R) strain (Non-virulent):** No mucus coat. Do not cause Pneumonia.

Experiment:

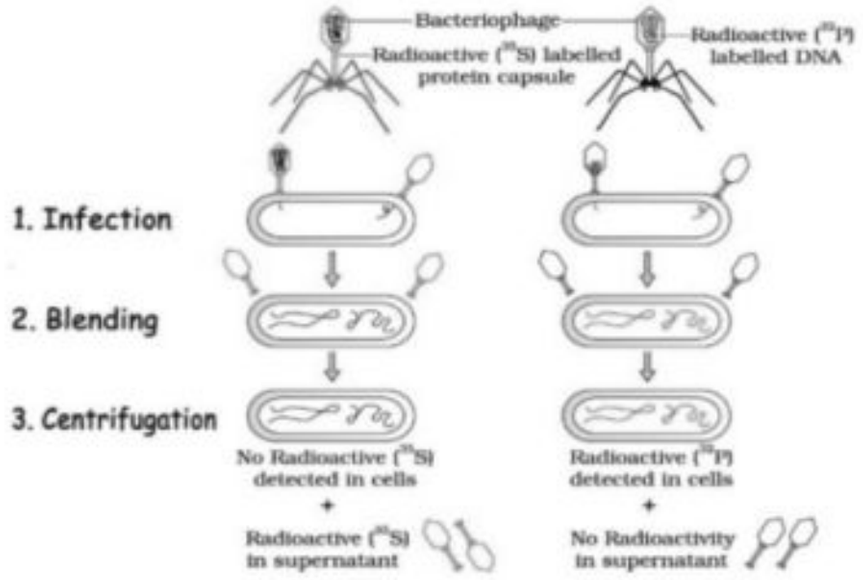
- S-strain → Inject into mice → Mice die
- R-strain → Inject into mice → Mice live
- S-strain (Heat killed) → Inject into mice → Mice live
- S-strain (Hk) + R-strain (live) → Inject into mice → Mice die

He concluded that some **'transforming principle'** transferred from heat-killed S-strain to R-strain. It enabled R-strain to synthesize smooth polysaccharide coat and become virulent. This must be due to the transfer of genetic material.

Biochemical characterization of transforming principle

- **Oswald Avery, Colin MacLeod & Maclyn McCarty** worked to determine the biochemical nature of **'transforming principle'** in Griffith's experiment.
- They purified biochemicals (proteins, DNA, RNA etc.) from heat killed S cells using suitable enzymes.
- They discovered that
  - Digestion of protein and RNA (using *Proteases* and *RNases*) did not affect transformation. It means that the transforming substance was not a protein or RNA.
  - Digestion of DNA with *DNase* inhibited transformation. It means that DNA caused transformation of R cells to S cells. It proves that DNA was the transforming principle.

Hershey-Chase Experiment (Blender Experiment)



- **Hershey & Chase** grew some bacteriophage viruses on a medium containing radioactive phosphorus

(P^{32}) and some others on medium containing radioactive sulphur (S^{35}).

- Viruses grown in P^{32} got **radioactive DNA** because only DNA contains phosphorus. Viruses grown in S^{35} got **radioactive protein** because protein contains sulphur.
- These preparations were used separately to infect ***E. coli.***
- After infection, the *E. coli* cells were gently agitated in a blender to remove the virus particles from the bacteria.
- Then the culture was centrifuged to separate lighter virus particles from heavier bacterial cells.
- Bacteria infected with viruses having radioactive DNA were radioactive. i.e., DNA had passed from the virus to bacteria. Bacteria infected with viruses having radioactive proteins were not radioactive. i.e., proteins did not enter the bacteria from the viruses. This proves that DNA is the genetic material.

CENTRAL DOGMA OF MOLECULAR BIOLOGY

- It is proposed by **Francis Crick.** It states that ***the genetic information flows from DNA → RNA → Protein.***

Central dogma

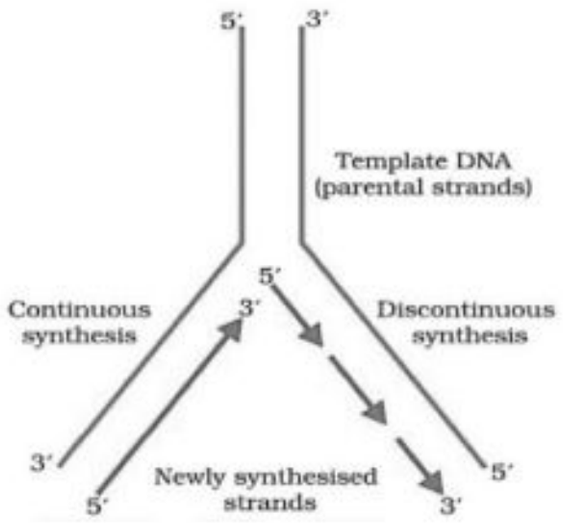
- In some viruses, flow of information is in reverse direction (from RNA to DNA). It is called **reverse transcription.**

DNA REPLICATION

- Replication is the copying of DNA from parental DNA.

The Machinery and Enzymes for Replication

- During replication, the 2 strands unwind and separate by breaking H-bonds.
- Unwinding of the DNA molecule at a point forms a 'Y'-shaped structure called **replication fork.**
- The separated strands act as **templates** for the synthesis of new strands.
- In presence of an enzyme, DNA dependent ***DNA polymerase,*** many nucleotides join with one another to primer strand and form a polynucleotide chain (new strand).
- During replication, one strand is formed as a continuous stretch in 5'→ 3' direction **(Continuous synthesis).** This strand is called **leading strand.**

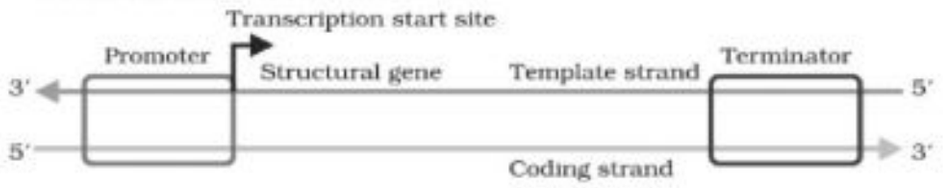


- The other strand is formed in small stretches **(Okazaki fragments)** in 5'→ 3' direction **(Discontinuous synthesis).**
- The Okazaki fragments are then joined together to form a new strand by an enzyme, ***DNA ligase***. This new strand is called **lagging strand.**

TRANSCRIPTION

Transcription Unit

- It is the segment of DNA between the sites of initiation and termination of transcription. It consists of 3 regions:
  - **A promoter:** Binding site for *RNA polymerase.*
  - **Structural gene:** The region between promoter and terminator where transcription takes place.
  - **A terminator:** The site where transcription stops. Located towards 3'-end.



Transcription unit and gene

Structural gene in a transcription unit is 2 types:

- **Monocistronic structural genes (split genes)**: It is seen in eukaryotes. Here, coding sequences ***(exons or expressed sequences)*** are interrupted by ***introns*** (intervening sequences).
  Exons appear in processed mRNA.
  Introns do not appear in processed mRNA.
- **Polycistronic structural genes**: It is seen in prokaryotes. Here, there are no split genes.

GENETIC CODE

Salient features of genetic code

- **Codon is triplet** (three-letter code).
- **61 codons code for amino acids**. 3 codons (**UAA, UAG & UGA)** do not code for any amino acids. They act as **stop codons (Termination codons or non-sense codons).**
- Genetic code is **universal.** Means Applicable to all from bacteria to human
- **No punctuations** b/w adjacent codons
- Genetic code is **non-overlapping.**
- **Degeneracy/degenerate**: some amino acids are coded by more than one codon

- Genetic code is **unambiguous** and **specific.** i.e. one codon specifies only one amino acid.
- AUG has dual functions. It codes for Methionine and acts as **initiator codon.**

REGULATION OF GENE EXPRESSION

Lac Operon in *E. coli*

- The operon controlling lactose metabolism.
- It is proposed by **Francois Jacob & Jacque Monod.**

It consists of

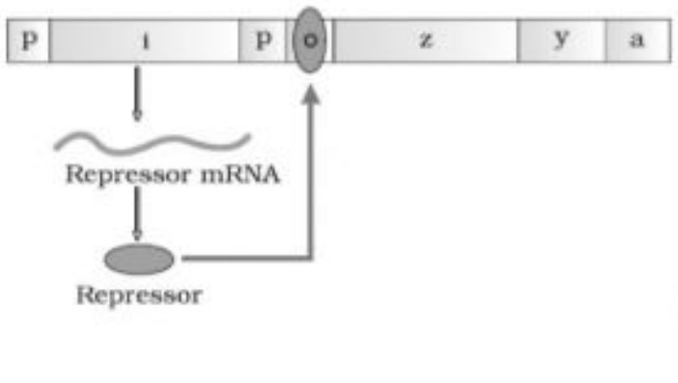
a) A regulatory or inhibitor (i) gene: Codes for repressor protein.

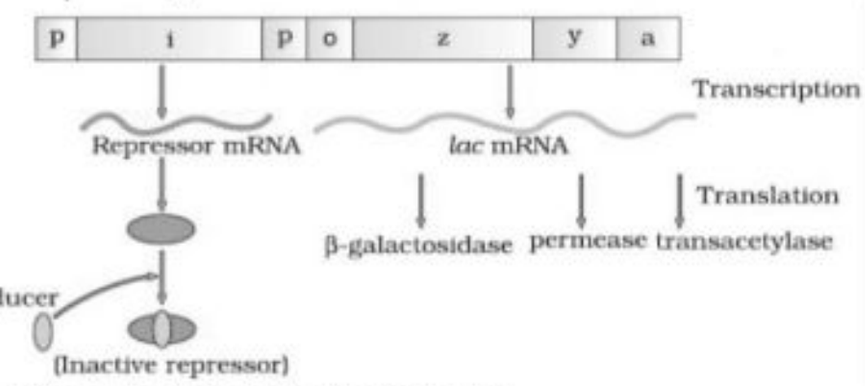
b) 3 structural genes:

i. **z gene:** Codes for ***β galactosidase.***
ii. **y gene:** Codes for ***permease.***
iii. **a gene:** Codes for a **transacetylase**

- Genes in the operon function together in the same or related metabolic pathway.
- If there is no **lactose (inducer),** lac operon remains switched off. The regulator gene synthesizes mRNA to produce **repressor protein.** This protein binds to the **operator** region and blocks *RNA polymerase* movement. So the structural genes are **not expressed.**
- If lactose or allolactose is provided in the growth medium, it is transported into ***E. coli*** cells . Lactose (inducer) binds with repressor protein. So repressor protein cannot bind to operator region. The operator region becomes free and induces the *RNA polymerase* to bind with **promoter.** Then transcription starts.
- Regulation of lac operon by repressor is called **negative regulation.**

In the absence of inducer





In the presence of inducer

HUMAN GENOME PROJECT(HGP)

- **Human Genome Project** was the first mega project for the sequencing of nucleotides and mapping of all the genes in human genome.

BAC= Bacterial Artificial Chromosomes
YAC= Yeast Artificial Chromosomes

DNA FINGERPRINTING

Steps of DNA fingerprinting (Southern Blotting Technique)

a. **Isolation** of DNA (from any cells or blood stains, semen stains, saliva, hair roots, bone, skin etc.).
b. **Digestion** of DNA by **restriction endonucleases.**
c. **Separation** of DNA fragments by **gel electrophoresis.**
d. **Transferring (blotting)** DNA fragments to synthetic membranes such as **nitrocellulose** or **nylon**.
e. **Hybridization** using radioactive labelled **VNTR probe.**
f. **Detection** of hybridized DNA by **autoradiography.**

The autoradiogram gives an image in the form of dark & light bands. It is called **DNA fingerprint.**

Application of DNA fingerprinting

- **Forensic tool** to solve paternity, rape, murder etc.
- For the diagnosis of **genetic diseases.**
- To determine **phylogenetic status** of animals.
- To determine **population and genetic diversities.**

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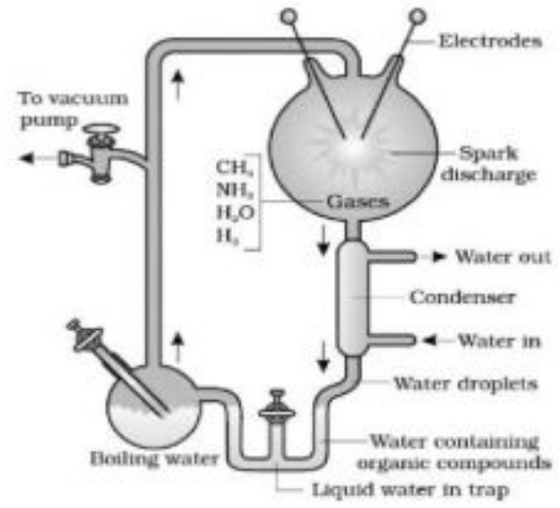
7. EVOLUTION

ORIGIN OF LIFE

Urey-Miller experiment

- **Harold Urey** & **Stanley Miller** experimentally proved theory of chemical evolution. They created a condition like that of primitive earth (i.e. high temperature, volcanic storms, reducing atmosphere with CH_4, NH_3, H_2O, H_2 etc).
- They made electric discharge in a closed flask containing CH_4, NH_3, H_2 and water vapour at 800° C. As a result, some amino acids are formed.



EVIDENCES FOR EVOLUTION

a. Homologous organs

- **Homologous organs** are the organs having fundamentally **similar structure and origin but different functions**. This phenomenon is called **Homology.**
- E.g. Human hand, Whale's flippers, Bat's wing & Cheetah's foot. These forelimbs have different functions but similar anatomical structures such as bones (e.g. humerus, radius, ulna, carpals, metacarpals & phalanges).
- **Homology in plants:** E.g. Thorns of *Bougainvillea* and tendrils of *Cucurbita*.
- The origin of homologous organs is due to ***Divergent evolution.*** It is the evolution by which **related species** become **less similar** to survive and adapt in different environmental condition.
- Homology indicates common ancestry.

b. Analogous organs

These are the organs having **similar function but different structure & origin.** This phenomenon is called **Analogy.** E.g.

- **Wings of insects** (formed of a thin flap of chitin) and **wings of birds** (modified forelimbs).
- **Eyes of Octopus** (retina from skin) and **mammals** (retina from embryonic brain).
- **Flipper of Penguins** and **Dolphins**.
- **Sweet potato** (modified root) & **Potato** (modified stem).
  - Origin of analogous organs is due to ***Convergent evolution.*** It is the evolution by which **unrelated species** become more **similar** to survive and adapt in similar environmental condition.

Evidences for evolution by natural selection:

Industrial melanism: In England, before industrialization, there were more white-winged moths on trees than dark winged (melanised) moths. After industrialization, more dark-winged moths and less white winged moths were developed.

Reason:

Before industrialization: There was white lichens covered the trees. In that background, white winged moths survived but dark winged moths were picked out by predators.

After industrialization: The tree trunks became dark due to industrial smoke and soot. No growth of lichens. So white winged moths did not survive because the predators identified them easily. Dark winged moth survived because of suitable dark background.

HARDY-WEINBERG PRINCIPLE

- It states that *allele frequencies in a population are stable and is constant from generation to generation.*
- E.g. Consider, in a diploid, **p** & **q** are the frequencies of alleles **A & a** respectively.

Frequency of AA = p^2 Frequency of aa = q^2

Frequency of Aa = 2pq Hence $\mathbf{p^2 + 2pq + q^2 = 1}$

Factors affecting Hardy-Weinberg equilibrium

a. **Gene migration:** Gene flow from one population to another.

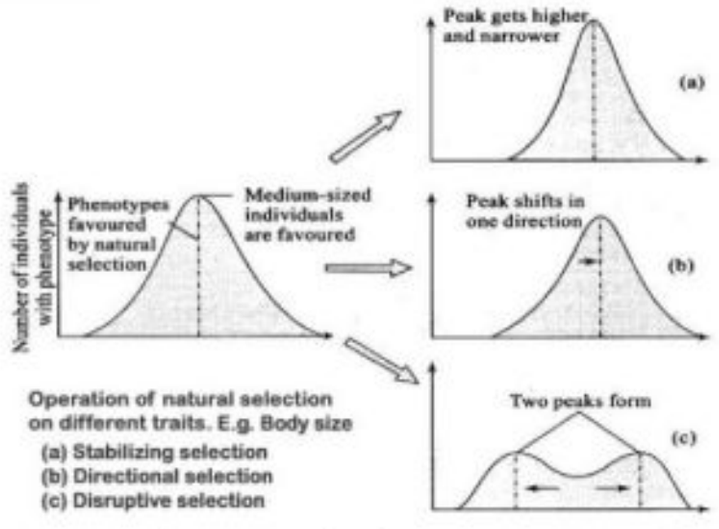
b. **Genetic drift:** Gene flow by chance causing change in frequency.

c. **Mutation:** It results in formation of new phenotypes.

d. **Genetic recombination:** Reshuffling of gene combinations during crossing over resulting in genetic variation.

e. **Natural selection:** It is 3 types.

- **Stabilizing selection:** Here, more individuals acquire mean character value and variation is reduced.
- **Directional selection:** Individuals of one extreme (value other than mean character value) are more favoured.
- **Disruptive selection:** Individuals of both extremes are more favoured.



ORIGIN AND EVOLUTION OF MAN

Dryopithecus

↓

Ramapithecus

↓

Australopithecines

↓

Homo habilis

↓

Homo erectus

↓

Homo neanderthalensis

↓

Homo sapiens

8. HUMAN HEALTH AND DISEASES

COMMON INFECTIOUS DISEASES IN MAN

BACTERIAL DISEASES

a. **Typhoid:** Pathogen is ***Salmonella typhi.***
   - **Mode of transmission:** It enters small intestine through food & water and migrates to other organs via blood.
   - **Symptoms:** Sustained high fever (39°-40° C), headache, weakness, stomach pain, constipation & loss of appetite. Intestinal perforation and death may occur.

   Widal test is used for confirmation of the disease.

PROTOZOAN DISEASES

a. **Malaria:** Pathogen is ***Plasmodium sp.*** *(P. vivax, P. malariae & P. falciparum).*

   Most serious (Malignant) malaria is caused by *P. falciparum.*
   - **Mode of transmission:** By female *Anopheles* mosquito.
   - **Symptoms:** Haemozoin (toxin released by *Plasmodium)* causes chill and high fever recurring every 3-4 days.

HUMAN IMMUNE SYSTEM

IMMUNITY

It is 2 types: Innate and Acquired.

1. Innate (inborn) immunity

- It is the *non-specific* immunity present at the time of birth.
- It includes 4 types of **Barriers:**

a. **Physical barriers:** Prevents entry of microbes. E.g. ***Skin***, ***Mucus coating*** of the respiratory, gastro-intestinal and urino-genital tracts. Mucus traps microbes.

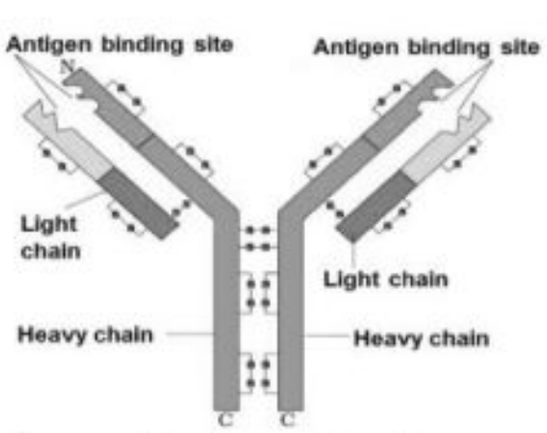
b. **Physiological barriers:** They prevent microbial growth. E.g. gastric HCl, saliva, tear etc.

c. **Cellular barriers: Phagocytes** like *WBC* [*Polymorpho-nuclear leukocytes (PMNL)* or *neutrophils, monocytes* and natural killer *lymphocytes], macrophages* etc.

d. **Cytokine barriers:** Virus infected cells secrete a cytokine protein called ***interferon.*** It protects non-infected cells from further viral infection.

2. Acquired (adaptive) immunity

- It is *pathogen specific* immunity developed during lifetime.



Structure of an antibody molecule:

An antibody has **4 polypeptide chains:** 2 **light chains** and 2 **heavy chains (H_2L_2).**

AIDS (Acquired Immuno Deficiency Syndrome)

- It is caused by **HIV (Human Immunodeficiency Virus),**

Transmission:

- Sexual contact with infected person.
- Transfusion of contaminated blood & blood products.
- Sharing of infected needles.
- From infected mother to her child through placenta.

Diagnosis:

- **ELISA test** (Enzyme-linked immuno-sorbent Assay).

Prevention of AIDS:

- **"Don't die of ignorance":** Educate people about AIDS through organisations like **National AIDS Control Organisation (NACO), Non-Governmental Organisations (NGOs), WHO** etc. WHO started the following programmes:
- Make blood (from blood banks) safe from HIV.
- Use disposable needles and syringes.
- Advocate safe sex and free distribution of condoms.
- Control drug abuse.
- Regular check-ups for HIV in susceptible population.

CANCER

- Cancer is an abnormal and uncontrolled multiplication of cells resulting in the formation of tumour (masses of cells).

Types of Tumours

- **Benign tumours:** Confined to the place of its origin. They do not spread to other parts. Cause little damage.
- **Malignant tumours:** Mass of proliferating cells **(neoplastic or tumour cells)** that grow rapidly, invade and damage the surrounding normal tissues. Due to active division and growth, they starve normal cells by competing for nutrients.

Treatment of cancer

- **Radiotherapy**
- **Chemotherapy**
- **Immunotherapy**
- **Surgery**

Effects of Drug/alcohol abuse

- Reckless behaviour, vandalism and violence.
- Coma and death due to respiratory failure, heart failure or cerebral haemorrhage.
- Drugs mixed with alcohol may cause death.
- Damage of nervous system and liver cirrhosis.
- Mental and social distress to family and friends.
- Social problems like stealing and spread of infectious diseases (e.g. AIDS, hepatitis B).
- Use of drugs and alcohol by pregnant woman affect the foetus (Foetal alcohol syndrome or FAS).
- Loss of sexual drive and necrospermia.
- Misuse of drugs by athletes (e.g. narcotic analgesics, anabolic steroids, diuretics & certain hormones to increase muscle strength and bulk and to promote aggressiveness).

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10. MICROBES IN HUMAN WELFARE

MICROBES IN HOUSEHOLD PRODUCT

- **Lactobacillus or Lactic acid bacteria (LAB):**
  - It converts milk to curd by producing acids that coagulate and partially digest the milk proteins.
  - Fresh milk can be converted to curd by adding some curd containing LAB. It also increases vitamin B_{12} in curd.
  - In stomach, LAB helps to check pathogens.

MICROBES IN INDUSTRIAL PRODUCTS

Chemicals, enzymes & other bioactive molecules

1. **Organic acids:** Acid producer microbes include
   Aspergillus niger (a fungus) : Citric acid
   Acetobacter aceti (a bacterium) : Acetic acid
   Clostridium butylicum (a bacterium) : Butyric acid
   Lactobacillus (a bacterium) : Lactic acid
2. **Alcohol:** Yeast (*S. cerevisiae*) is used to produce ethanol.
3. **Enzymes:**
   - **Lipases:** Used in detergent formulations. Help to remove oily stains from the laundry.
   - **Pectinases & Proteases:** To clarify bottled juices.
   - **Streptokinase:** Produced by ***Streptococcus.*** Used as a **'clot buster'** to remove clots from the blood vessels of patients who have **myocardial infarction.**
4. **Cyclosporine A:** Produced by ***Trichoderma polysporum*** (fungus). Used as an **immunosuppressive agent** in organ transplant patients.
5. **Statins:** Produced by ***Monascus purpureus*** (a yeast). Used as **blood-cholesterol lowering agents**. It inhibits the enzymes responsible for synthesis of cholesterol.

MICROBES AS BIO CONTROL AGENTS

Microbial biocontrol agents

- ***Bacillus thuringiensis (Bt):*** To control butterfly caterpillar.
  The dried spores of Bt (available in sachets) are mixed with water and sprayed on to vulnerable plants such as brassicas and fruit trees. These are eaten by the caterpillar. In their gut, the toxin is released and the larvae get killed.
  The scientists have introduced *B. thuringiensis* toxin genes into plants. E.g. Bt cotton.
- ***Trichoderma sp*** (fungus): These are free livings present in the root ecosystems. They control several plant pathogens.

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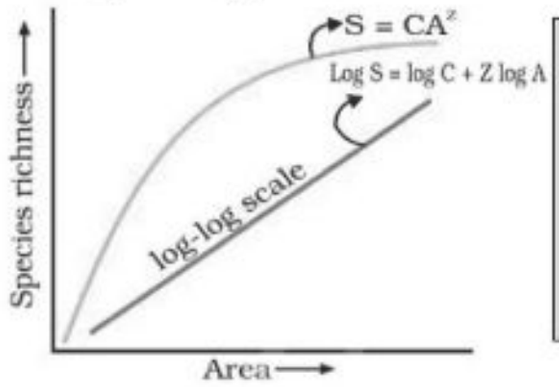
15. BIODIVERSITY AND CONSERVATION

Species- Area relationship

According to the study of **Alexander von Humboldt** in South American jungles, within a region, species richness increases with increasing explored area, but only up to a limit.

Relation between species richness and area gives a **rectangular hyperbola.**



$S = CA^z$

Where,
S= Species richness
A= Area
C= Y-intercept
Z= slope of the line
(regression co-

IMPORTANCE OF SPECIES DIVERSITY

- **'Rivet popper hypothesis':** It is an analogy used to understand the importance of biodiversity.
  It is proposed by Stanford ecologist **Paul Ehrlich.**
  In an airplane **(ecosystem),** all parts are joined with many rivets **(species).** If passengers pop a rivet (extinction of a species), it may not affect flight safety **(functioning of the ecosystem).** But as more and more rivets are removed, the plane becomes dangerously weak. Loss of rivets on the wings (**key species** that drive major ecosystem functions) is more dangerous than loss of a few rivets on the seats or windows.

LOSS OF BIODIVERSITY

Causes of Biodiversity losses ('The Evil Quartet')

1. **Habitat loss and fragmentation:** Most important cause.
   - E.g. Tropical rain forests (loss from 14% to 6%).
   - Thousands of hectares of rain forests are being lost within hours.
   - **The Amazon rain forest** is being cut for cultivating soya beans or for conversion of grass lands for cattle.
   - Fragmentation badly affects animals requiring large territories and migratory animals.
2. **Over-exploitation: Stellar's sea cow, Passenger pigeon** etc. extinct due to over exploitation.
3. **Alien species invasions:** Alien species cause decline or extinction of **indigenous species.** E.g.
   - **Nile Perch** introduced in **Lake Victoria (East Africa)** caused extinction of more than 200 species of **cichlid fish.**
4. **Co-extinction:** When a species becomes extinct, the species associated with it also extinct. E.g.
   - Extinction of the **parasites** when the **host** is extinct.

Biodiversity conservation

BIO DIVERSITY CONSERVATION

2 types: ***In situ*** **(on site)** and ***Ex situ*** **(off site).**

a. *In situ* conservation (on site)

It is the conservation of genetic resources within natural or human-made ecosystems in which they occur. E.g. **National Parks, Sanctuaries, Biosphere reserves, cultural landscapes, natural monuments etc.**

b. *Ex situ* conservation (off site)

It is the conservation of organisms outside their habitats.
E.g. genetic resource centres, zoological parks, wildlife safari parks, botanical gardens, gene banks,cryopreservation etc.

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