Biyani's Think Tank

Concept based notes

Biology

XII

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Preface

I am glad to present this book, especially designed to serve the needs of the students.

The book has been written keeping in mind the general weakness in understanding the fundamental concepts of the topics. The book is self-explanatory and adopts the "Teach Yourself" style. It is based on question-answer pattern. The language of book is quite easy and understandable based on scientific approach.

Any further improvement in the contents of the book by making corrections, omission and inclusion is keen to be achieved based on suggestions from the readers for which the author shall be obliged.

I acknowledge special thanks to Mr. Rajeev Biyani, *Chairman* & Dr. Sanjay Biyani, *Director* (*Acad.*) Biyani Group of Colleges, who are the backbones and main concept provider and also have been constant source of motivation throughout this Endeavour. They played an active role in coordinating the various stages of this Endeavour and spearheaded the publishing work.

I look forward to receiving valuable suggestions from professors of various educational institutions, other faculty members and students for improvement of the quality of the book. The reader may feel free to send in their comments and suggestions to the under mentioned address.

Author

Chapter 1

Sexual Reproduction in Animals

Q.1 What is reproduction ? define its type and asexual reproduction types.

Ans

Reproduction is defined as a biological process in which anorganism gives rise to young ones (offspring) similar to itself. The offspring grow, mature and in turn produce new offspring. Thus, there is a cycle of birth, growth and death. Reproduction enables the continuity of the species, generation after generation. Based on whether there is participation of one organism or two in the process of reproduction, it is of two types. When offspring is produced by a single parent with or without the involvement of gamete formation, the reproduction is **asexual.** When two parents (opposite sex) participate in the reproductive process and also involve fusion of male and female gametes, it is called **sexual reproduction**.

ASEXUAL REPRODUCTION

In this method, a single individual (parent) is capable of producing offspring. As a result, the offspring that are produced are not only identical to one another but are also exact copies of their parents. The term **clone** is used to describe such morphologically and genetically similar individuals Asexual reproduction is common among single-celled organisms, and in plants and animals with relatively simple organisations.

Cell division

In Protista and Monerans, the organism or the parent cell divides into two to give rise in these organisms **cell division** is itself a mode of reproduction.

Binary fission

Many single-celled organisms reproduce by **binary fission,** where a cell divides into two halves and each rapidly grows into an adult (e.g.,*Amoeba* ,*Paramecium*)

Budding

In yeast, the division is unequal and small **buds** are eventually gets separated and mature into new yeast organisms (cells).

Zoospores

Members of the Kingdom Fungi and simple plants such as algae reproduce through special asexual reproductive structures The most common of these structures are **zoospores** that usually are microscopic motile structures. Other common asexual reproductive structures are **conidia** (*Penicillium*), **buds** (*Hydra*) and **gemmule** in plants, the term **vegetative** reproduction is frequentlyused.

Vegetative propagation

In plants, the units of vegetative propagation such as **runner, rhizome, sucker, tuber, offset, bulb** are all capable of giving rise to new offspring . These structures are called **vegetative propagules.**

Q.2 What is sexual reproduction? Define it with its all events.

Ans

SEXUAL REPRODUCTION

Sexual reproduction involves formation of the male and female gametes, either by the same individual or by different individuals of the opposite sex. These gametes fuse to form the zygote which develops to form the new organism. It is an elaborate, complex and slow process as compared to asexual reproduction. Because of the fusion of male and female gametes, sexual reproduction results in offspring that are not identical to the parents or amongst themselves

Events in sexual reproduction :

First every organism go through the juvenile stage ,After attainment of maturity, all sexually reproducing organisms exhibit events and processes that have remarkable fundamental similarity, even though the structures associated with sexual reproduction are indeed very different. The events of sexual reproduction though elaborate and complex, follow a regular sequence. Sexual reproduction is characterised by the fusion (or fertilisation) of the male and

female gametes, the formation of zygote and embryogenesis. For convenience these sequential events may be grouped into three distinct stages namely, the **pre-fertilisation, fertilisation** and the **post-fertilisation events.**

Pre-fertilisation Events

These include all the events of sexual reproduction prior to the fusion of gametes. The two main pre-fertilisation events are **gametogenesis** and **gamete transfer.**

Gametogenes

Gametogenesis refers to the process of formation of the two types of gametes – male and female. Gametes are haploidcells. In some algae the two gametes are so similar in appearance that it is not possible to categorise the They are hence, are called **homogametes** (**isogametes)**

However, in a majority of sexually reproducing organisms the gametes produced are of two morphologically distinct types (**heterogametes).** In such organisms the male gamete is called the **antherozoid** or **sperm** and the female gamete is called the **egg** or **ovum**

Gamete Transfer

After their formation, male and female gametes must be physically brought together to facilitate fusion (fertilisation). In a majority of organisms, male gamete is motile and the female gamete is stationary. Exceptions are a few fungi and algae in which both types of gametes are motile. There is a need for a medium through which the male gametes move. In several simple plants like algae, bryophytes and pteridophytes , water is the medium through which this gamete transfer takes place. A large number of the male gametes,

however, fail to reach the female gametes. To compensate this loss of male gametes during transport, the number of male gametes produced is several thousand times the number of female gametes produced.
In seed plants, pollen grains are the carriers of male gametes and ovule have the egg. Pollen grains produced in anthers therefore, have to be transferred to the stigma before it can lead to
fertilisation . In bisexual, self-fertilising plants, e.g., peas, transfer of pollen grains to the stigma is relatively easy as anthers and stigma are located close to each other; pollen grains soon after they are shed, come in contact with the stigma. But in cross pollinating plants (including dioecious plants), aspecialised event called **pollination** facilitates transfer of pollen grains to the stigma. Pollen grains germinate on the stigma and the pollen tubes carrying the male
gametes reach the ovule and discharge male gametes near the egg. In dioecious animals, since male and female gametes are formed in different individuals, the organism must evolve a special mechanism for gamete transfer. Successful transfer and coming together of gametes is essential for the most critical event in sexual reproduction, the fertilisation.

Fertilization
The most vital event of sexual reproduction is perhaps the fusion of gametes. This process called **syngamy** results in the formation of a diploid **zygote**. The term **fertilisation** is also often used for this process. The terms syngamy and fertilisation are frequently used though , interchangeably. it has to be mentioned here that in some organisms like rotifers, honeybees and even some lizards
and birds (turkey), the female gamete undergoes development to form new organisms without fertilisation. This phenomenon is called **parthenogenesis. Where does syngamy occur?** In most aquatic organisms, such as a majority of algae and fishes as well as amphibians, syngamy occurs in the external medium (water), i.e., outside the body of the organism. This type of gametic fusion is called **external fertilisation**. Organisms exhibiting external fertilisation show great synchrony between the sexes and release a large number of gametes into the surrounding medium (water) in order to enhance the chances of syngamy. This happens in the bony fishes and frogs where a large number of offspring are produced. A major disadvantage is that the offspring are extremely vulnerable to predators threatening their survival up to adulthood. In many terrestrial organisms, belonging to fungi, higher animals such as reptiles birds, mammals and in a majority of plants (bryophytes, pteridophytes, gymnosperms and angiosperms), syngamy occurs inside the body of the organism, hence the process is called iternal **fertilisation.** In all these organisms, egg is formed inside the female body where they fuse with the male gamete. In organisms exhibiting internal fertilisation, the male gamete is motile and has to reach the egg in order to fuse with it. In these even though the number of sperms produced is very large, there is a significant reduction in the number of eggs produced. In seed plants, However, the non-motile male gametes are carried to female gamete bypollen tubes.

Post-fertilisation Events
Events in sexual reproduction after the formation of zygote are called **post-fertilisation events**.

The Zygote

Formation of the diploid zygote is universal in all sexually reproducing organisms. In organisms with external fertilisation, zygote is formed in the external medium (usually water), whereas in those exhibiting internal fertilisation, zygote is formed inside the body of the organism. Further development of the zygote depends on the type of life cycle the organism has and the environment it is exposed to. In organisms belonging to fungi and algae, zygote develops a thick wall that is resistant to dessication and damage. It undergoes a period of rest before germination. In organisms with haplontic life cycle zygote divides by meiosis to form haploid spores that grow into haploid individuals. . Every sexuallyreproducing organism, including human beings begin life as a singlecell– the zygote.

Embryogenesis

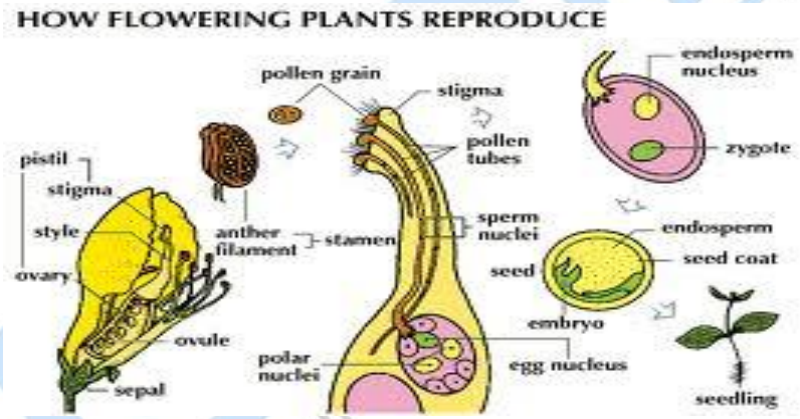
Embryogenesis refers to the process of development of **embryo** from thezygote. During embryogenesis, zygote undergoes **cell division** (mitosis) and **cell differentiation**. While cell divisions increase the number of cells in the developing embryo; cell differentiation helps groups of cells to undergo certain modifications to form specialised tissues and organs to form an organism.. Animals are categorised into **oviparous** and **viviparous** based on whether the development of the zygote take place outside the body of the female parent or inside, i.e., whether they lay fertilised/unfertilised eggs or give birth to young ones. In oviparous animals like reptiles and birds the fertilised eggs covered by hard **calcareous shell** are laid in a safe place in the **environment;** after a period of incubation young ones hatch out. On the other hand, in viviparous animals (majority of mammals including human beings), the zygote develops into a young one inside the body of the female organism. After attaining a certain stage of growth, the young ones are delivered out of the body of the female organism. Because of proper embryonic care and protection, the chances of survival of young ones is greater in viviparous organisms.

In flowering plants, the zygote is formed inside the ovule. After fertilisation the sepals, petals and stamens of the flower wither and fall off. The pistil however, remains attached to the plant. The zygote develops into the embryo and the ovules develop into the seed. The **ovary** develops into the **fruit** which develops a thick wall called **pericarp** that is protective in function . After dispersal, seeds germinate under favourable conditions to produce new plants.

Chapter 2
Sexual Reproduction in Flowering Plants

Q 1 **Define the structure of flower with its development** .

Ans Flower is divided into two parts **stamen –** the long and slender stalk called the **filament,** and the terminal generally bilobed structure called the **anther.** The proximal end of the filament is attached to the thalamus or the petal of the flower. The number and length of stamens are variable in flowers of different species. A typical angiosperm anther is **bilobed** with each lobe having two theca, i.e., they are **dithicous**. Often a longitudinal groove runs lengthwise separating the theca. Let us understand the various types of tissues and their organization in the transverse section of an anther . The bilobed nature of an anther is very distinct in the transverse section of the anther. The anther is a four-sided (tetragonal) structure consisting of four **microsporangia** located at the corners, two in each lobe. The microsporangia develop further and become **pollen sacs.** They extend longitudinally all through the length of an anther and are packed Swith pollen grains.



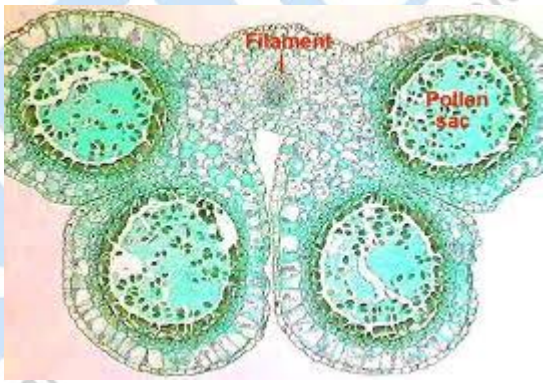
Structure of microsporangium: In a transverse section, a typical microsporangium appears near circular in outline. It is generally surrounded by four wall layers – the epidermis, endothecium, middle layers and the tapetum.

The outer three wall layers perform the function of protection and help in dehiscence of anther to release the pollen. The innermost wall layer is the **tapetum.** It nourishes the developing pollen grains. Cells of the tapetum possess dense cytoplasm and generally have more than one nucleus.
When the anther is young, a group of compactly arranged homogenous cells called the **sporogenous tissue** occupies the centre of each microsporangium.

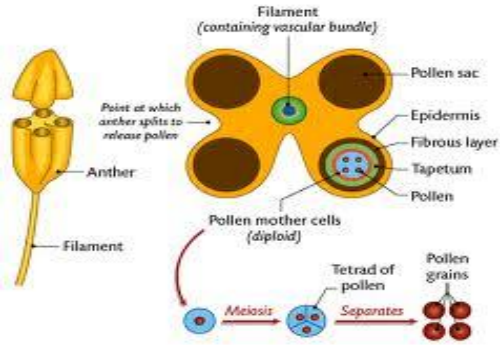
Microsporogenesis : As the anther develops, the cells of the sporogenous tissue undergo meiotic divisions to form microspore tetrads. As each cell of the sporogenous tissue is

capable of giving rise to a microspore tetrad. Each one is a potential pollen or microspore mother cell (PMC). The process of formation of microspores from a pollen mother cell through meiosis is called **microsporogenesis.** The microspores, as they are formed, are arranged in a cluster of four cells–the **microspore tetrad** . As the anthers mature and dehydrate, the microspores dissociate from each other and develop into **pollen grains .** Inside each microsporangium several thousands of microspores or pollen grains are formed that are released with the dehiscence of anther.

Pollen grain: The pollen grains represent the male gametophytes. Pollen grains are generally spherical measuring about 25-50 micrometers in diameter. It has a prominent two-layered wall. The hard outer layer called the **exine** is made up of sporopollenin which is one of the most resistant organic material known. It can withstand high temperatures and strong acids and alkali. No enzyme that degrades sporopollenin is so far known. Pollen grain exine has prominent apertures called **germ pores** where sporopollenin is absent. Pollen grains are wellpreserved as fossils because of the presence of sporopollenin. The exine exhibits a fascinating array of patterns and designs. The inner wall of the pollen grain is called the **intine.** It is a thin and continuous layer made up of cellulose and pectin. The cytoplasm of pollen grain is surrounded by a plasma membrane. When the pollen grain is mature it contains two cells, the **vegetative cell** and **generative cell .** The vegetative cell is bigger, has abundant



food reserve and a large irregularly shaped nucleus. The **generative cell** is small and floats in the cytoplasm of the vegetative cell. It is spindle shaped with dense cytoplasm and a nucleus. In over 60 per cent of angiosperms, pollen grains are shed at this 2-celled stage. In the remaining species, the generative cell divides mitotically to give rise to the two male gametes before pollen grains are shed (3-celled stage). When once they are shed, pollen grains have to land on the stigma before they lose viability if they have to bring about fertilisation.



The period for which pollen grains remain viable is highly variable and to some extent depends on the prevailing temperature and humidity. In some cereals such as rice and wheat, pollen grains lose viability within 30 minutes of their release, and in some members of Rosaceae, Leguminoseae and Solanaceae, . It is possible to store pollen grains of a large number of species for years in liquid nitrogen (-1960C). Such stored pollen can be used as pollen banks, similar to seed banks, in crop breeding programmes.

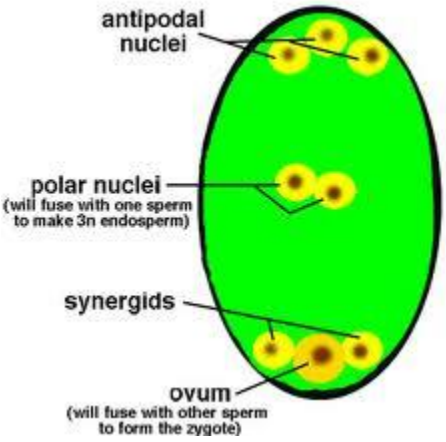
The Pistil, Megasporangium (ovule) and Embryo sac The gynoecium represents the female reproductive part of the flower. The gynoecium may consist of a single pistil (**monocarpellary**) or may have more than one pistil (**multicarpellary**). When there are more than one, the pistils may be fused together (**syncarpous**) or may be free (**apocarpous**) . Each pistil has three parts , **the stigma, style and ovary**. The **stigma** serves as a landing platform for pollen grains. The style is the elongated slender part beneath the stigma. The basal bulged part of the pistil is the **ovary.** Inside the ovary is the **ovarian cavity** (**locule)**. **The placenta** is located inside the ovarian cavity.

Arising from the placenta are the **megasporangia,** commonly called **ovules.** The number of ovules in an ovary may be one (wheat, paddy, mango) to many (papaya, water melon, orchids). **The Megasporangium (Ovule) :** Let us familiarise ourselves with the structure of a typical angiosperm ovule s. The ovule is a small structure attached to the placenta by means of a stalk called **funicle**. The body of the ovule fuses with funicle in the region called **hilum.** Thus, hilum represents the junction between ovule and funicle. Each ovule has one or two protective envelopes called **integuments.** Integuments encircle the ovule except at the tip where a small opening called the **micropyle** is organised. Opposite the micropylar end, is the **chalaza**, representing the basal part of the ovule. Enclosed within the integuments is a mass of cells called the **nucellus**. Cells of the nucellus have abundant reserve food materials. Located in the nucellus is the **embryo sac** or **female gametophyte**. An ovule generally has a single embryo sac formed from a megaspore through reduction division.

Megasporogenesis : The process of formation of megaspores from the **megaspore mother cell** is called **megasporogenesis**. Ovules generally differentiate a single megaspore mother cell (MMC) in the micropylar region of the nucellus. It is a large cell containing dense cytoplasm and a prominent nucleus. The MMC undergoes meiotic division. Meiosis results in the production of four **megaspores** .

Female gametophyte : In a majority of flowering plants, one of the megaspores is **functional** while the other three degenerate. Only the **functional megaspore** develops

into the **female gametophyte** (**embryo sac).** This method of embryo sac formation from a single megaspore is termed **monosporic** development.



The nucleus of the functional megaspore divides mitotically to form two nuclei which move to the opposite poles, forming the **2-nucleate** embryo sac. Two more sequential mitotic nuclear divisions result in the formation of the **4-nucleate** and later the **8-nucleate** stages of the embryo sac. It is of interest to note that these mitotic divisions are strictly free nuclear, that is, nuclear divisions are not followed immediately by cell wall formation. After the 8-nucleate stage, cell walls are laid down leading to the organisation of the typical **female gametophyte** or **embryo sac**. Observe the distribution of cells inside the embryo sac s. Six of the eight nuclei are surrounded by cell walls and organised into cells; the remaining two nuclei, called polar nuclei are situated below the egg apparatus in the large **central cell.**

There is a characteristic distribution of the cells within the embryo sac. Three cells are grouped together at the micropylar end and constitute the **egg apparatus**. The egg apparatus, in turn, consists of two **synergids** and one **egg cell**. The synergids have special cellular thickenings at the micropylar tip called filiform apparatus, which play an important role in guiding the pollen tubes into the synergid. Three cells are at the chalazal end and are called the **antipodals**. The large central cell, as mentioned earlier, has two polar nuclei. Thus, a typical angiosperm embryo sac, at
maturity, though **8-nucleate** is **7-celled**.

Q2 Define pollination and type of Pollination.

Ans Transfer of pollen grains (shed from the anther) to the stigma of a pistil is termed **pollination**. Flowering plants have evolved an amazing array of adaptations to achieve pollination. They make use of external agents to achieve pollination.

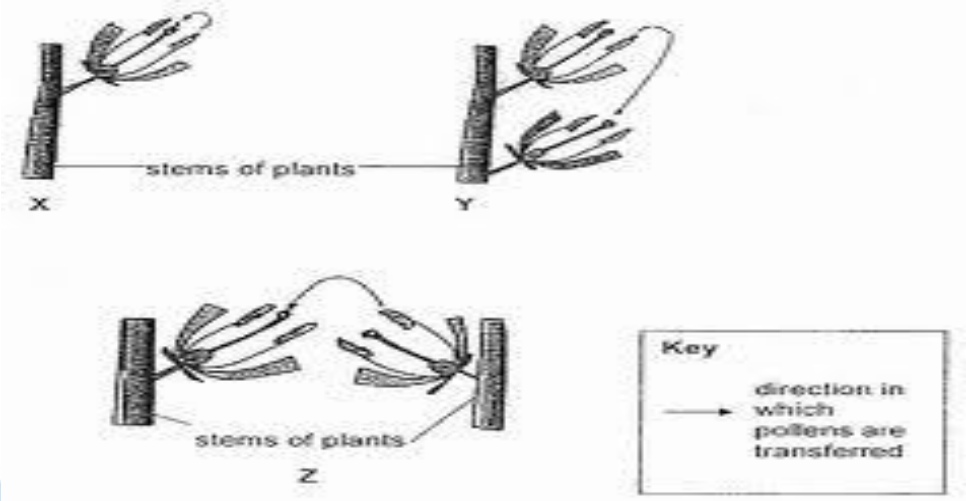
Kinds of Pollination : Depending on the source of pollen, pollination can be divided into three types.

(i) ***Autogamy* :** In this type, pollination is achieved within the same flower. Transfer of pollen grains from the anther to the stigma of the same flower . In a normal flower which opens and exposes the anthers and the stigma, complete autogamy is rather rare. Autogamy in such flowers requires synchrony in pollen release and stigma receptivity and also, the anthers and the stigma should lie close to each other so that self-pollination can occur.

Some plants such as *Viola* (common pansy), *Oxalis*, and *Commelina* produce two types of flowers – **chasmogamous** flowers which are similar to flowers of other species with exposed anthers and stigma, and **cleistogamous** flowers which do not open at all . In such flowers, the anthers and stigma lie close to each other. When anthers dehisce in the flower buds, pollen grains come in contact with the stigma to effect pollination. Thus, cleistogamous flowers are invariably autogamous as there is no chance of cross-pollen landing on the stigma. Cleistogamous flowers produce assured seed-set even in the absence of pollinators.

(ii) ***Geitonogamy* –** Transfer of pollen grains from the anther to the stigma of another flower of the same plant. Although geitonogamy is functionally cross-pollination involving a pollinating agent, genetically it is similar to autogamy since the pollen grains come from the same plant.

(iii) ***Xenogamy* –** Transfer of pollen grains from anther to the stigma of a different plant . This is the only type of pollination which during pollination brings genetically different types of pollen grains to the stigma.



Q3 What is pollen pistil intraction

Ans Pollen-pistil Interaction : Pollination does not guarantee the transfer of the right type of pollen (compatible pollen of the same species as the stigma). Often, pollen of the wrong type, either from other species or from the same plant (if it is self-incompatible), also land on the stigma. The pistil has the ability to recognise the pollen, whether it is of the right type (compatible) or of the wrong type (incompatible). If it is of the right type, the pistil accepts the pollen and promotes post-pollination events that leads to fertilisation. If the pollen is of the wrong type, the pistil rejects the pollen by preventing pollen germination on the stigma or the pollen tube growth in the style. The ability of the pistil to recognise the pollen followed by its acceptance or rejection is the result of a continuous dialogue between pollen grain and the pistil. This dialogue is mediated by chemical components of the pollen interacting with those of the pistil. It is only in recent years that botanists have been able to identify some of the pollen and pistil components and the interactions leading to the recognition, followed by acceptance or rejection. As mentioned earlier, following compatible pollination, the pollen grain germinates on the

stigma to produce a pollen tube through one of the germ pores The contents of the pollen grain move into the pollen tube. Pollen tube grows through the tissues of the stigma and style and reaches the ovary in some plants, pollen grains are shed at two-celled condition (a vegetative cell and a generate cell). In such plants, the generative cell divides and forms the two male gametes during the growth of pollen tube in the stigma. In plants which shed pollen in the three-celled condition, pollen tubes carry the two male gametes from the beginning. Pollen tube, after reaching the ovary, enters the ovule through the micropyle and then enters one of the synergids through the filiform apparatus . Many recent studies have shown that filiform apparatus present at the micropylar part of the synergids guides the entry of pollen tube. All these events–from pollen deposition on the stigma until pollen tubes enter the ovule–are together referred to as pollen-pistil interaction.

Q4 What is Double Fertilisation?

Ans

After entering one of the synergids, the pollen tube releases the two male gametes into the cytoplasm of the synergid. One of the male gametes moves towards the egg cell and fuses with its nucleus thus completing the **syngamy**. This results in the formation of a diploid cell, the **zygote.** The other male gamete moves towards the two polar nuclei located in the central cell and fuses with them to produce a triploid **primary endosperm nucleus** (PEN) . As this involves the fusion of three haploid nuclei it is termed **triple fusion.** Since two types of fusions, syngamy and triple fusion take place in an embryo sac the phenomenon is termed **double fertilisation**, an event unique to flowering plants. The central cell after triple fusion becomes the **primary endosperm cell** (PEC) and develops into the **endosperm** while the zygote develops into an **embryo.** endosperm tissue. The cells of this tissue are filled with reserve food materials and are used for the nutrition of the developing embryo. In the most common type of endosperm development, the PEN undergoes successive nuclear divisions to give rise to free nuclei. This stage of endosperm development is called free-nuclear endosperm.

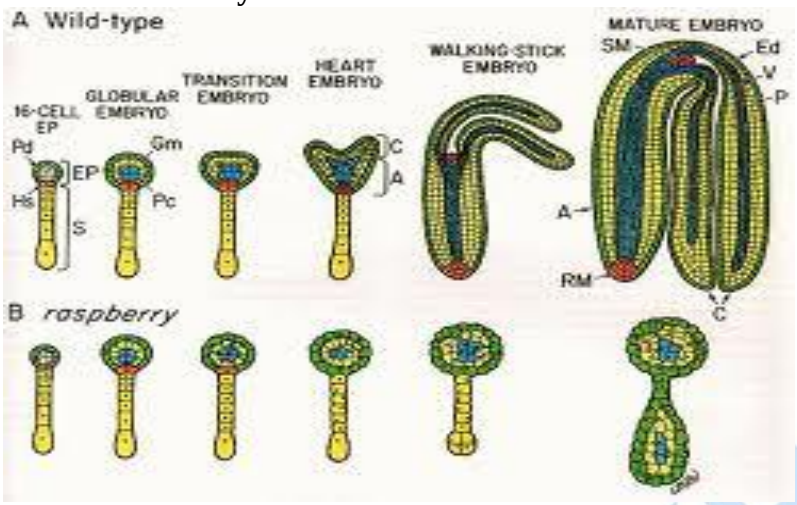
Subsequently cell wall formation occurs and the endosperm becomes cellular. The number of free nuclei formed before cellularisation varies greatly. The coconut water from tender coconut that you are familiar with, is nothing but free-nuclear endosperm (made up of thousands of nuclei) and the surrounding white kernel is the cellular endosperm. Endosperm may either be completely consumed by the developing embryo (e.g., pea, groundnut, beans) before seed maturation or it may persist in the mature seed (e.g. castor and coconut) and be used up during seed germination.

Q5 Define the Embryo development.

Ans

Embryo develops at the micropylar end of the embryo sac where The zygote is situated. Most zygotes divide only after certain amount of endosperm is formed. This is an adaptation to provide assured nutrition to the developing embryo. Though the seeds differ greatly, the early stages of embryo development **(embryogeny)** are similar in both monocotyledons and

dicotyledons. depicts the stages of embryogeny in ma dicotyledonous embryo. The zygote gives rise to the m**proembryo** and subsequently to the **globular, heart-shaped** and **mature embryo**.



A typical dicotyledonous embryo , consists of an **embryonal axis** and two **cotyledons.** The portion of embryonal axis above the level of cotyledons is the **epicotyl,** which terminates with the **plumule** or stem tip. The cylindrical portion below the level of cotyledons is **hypocotyl** that terminates at its lower end in the **radical or root tip.** The root tip is covered with a **root cap.**

Embryos of monocotyledons possess only one cotyledon. In the grass family the cotyledon is called **scutellum** that is situated towards one side (lateral) of the embryonal axis. At its lower end, the embryonal axis has the radical and root cap enclosed in an undifferentiated sheath called **coleorrhiza.** The portion of the embryonal axis above the level of attachment of scutellum is the epicotyl. Epicotyl has a shoot apex and a few leaf primordia enclosed in a hollow foliar structure, the **coleoptile.**

Chapter 3
Reproductive health

Q 1 Give the detail of BIRTH CONTROL methods.

Ans Natural methods

Natural methods work on the principle of avoiding chances of ovum and sperms meeting. **Periodic abstinence** is one such method in which the 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 or **coitus interruptus** is another method in which 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 ovulationand therefore the cycle do not occur during the period of intense lactation following parturition. Therefore, as long as the mother breast-feeds the child fully, chances of conception are almost nil. However, 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, though, of 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** (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. This can prevent conception. 'Nirodh' is a popular brand of condom for the male. Use of condoms has increased in recent years due to its additional benefit of protecting the user from contracting STDs and AIDS. Both the male and the female condoms are disposable, can be self-inserted and thereby gives privacy to the user. **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. Spermicidal creams, jellies and foams are usually used alongwith these barriers to increase their contraceptive efficiency.

IntraUterine Devices (IUDs).

Another effective and popular method is the use of **Intra Uterine Devices (IUDs)**. These devices are inserted by doctors or expert nurses in the uterus through vagina. These Intra Uterine Devices are presently available as the non-medicated IUDs (e.g., Lippes loop), copper releasing IUDs (CuT, Cu7, Multiload 375) and the hormone releasing IUDs (Progestasert, LNG-20) . 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, in addition, make the uterus unsuitable for implantation and the cervix hostile to the sperms. IUDs are ideal contraceptives for the females who want to delay pregnancy and/or space children. It is one of most widely accepted methods of contraception inIndia. **Pills** Oral administration of small doses of either progestogens or progestogen–estrogen combinations is another contraceptive method used by the females. They are used in the form of tablets and hence are popularly called the **pills**. Pills have to be taken daily for a period of 21 days starting preferably 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. 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 and are well accepted by the females. *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. 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. Administration of progestogens or progestogen-estrogen combinations or IUDs within 72 hours of coitus have been found to be very effective as emergency contraceptives as 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. Sterilisation procedure in the male is called 'vasectomy' and that in the female, 'tubectomy'. In vasectomy, a small part of the vas deferens is removed or tied up through a small incision on the scrotum whereas in tubectomy, a small part of the fallopian tube is removed or tied up through a small incision in the abdomen or through vagina. These techniques are highly effective but their reversibility is very poor.

Q 2 Ans **How we cope with infertility 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. However, where such corrections 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) followed by **embryo transfer** (ET) is one of such methods. 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 (with upto 8 blastomeres) could then be transferred into the fallopian tube (ZIFT–**zygote intra fallopian transfer**) and embryos with more than 8 blastomeres, into the uterus (IUT **– intra uterine transfer**), to complete its further development. Embryos formed by **in-vivo fertilisation** (fusion of gametes within the female) also could be used

for such transfer to assist those females who cannot conceive. Transfer of an ovum collected from a donor into the fallopian tube (GIFT – **gamete intra fallopian transfer**) of another female who cannot produce one, but can provide suitable environment for fertilisation and further development is another method attempted. **Intra cytoplasmic sperm injection** (ICSI) is another specialised procedure to form an embryo in the laboratory in which a sperm is directly injected into the ovum. 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.

Chapter 4
Principles of inheritance And variation

Q1 Give the detail of Contrasting Traits Studied by Mendel .

Ans Seven Contrasting Traits are:

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

Q 2 Show the Diagrammatic representation of monohybrid cross.

Ans

Monohybrid cross: In this example, both organisms have the genotype *Bb*. They can produce gametes that contain either the *B* or *b* allele. (It is conventional in genetics to use capital letters to indicate dominant alleles and lower-case letters to indicate recessive alleles.) The probability of an individual offspring having the genotype *BB* is 25%, *Bb* is 50%, and *bb* is 25%.

		Maternal	
		B	**b**
Paternal	**B**	BB	Bb
	b	Bb	bb

It is important to note that Punnett squares give probabilities only for *genotypes*, not *phenotypes*. The way in which the *B* and *b* alleles interact with each other to affect the appearance of the offspring depends on how the gene products (proteins) interact (see Mendelian inheritance). For classical dominant/recessive genes, like that which determines whether a rat has black hair (*B*) or white hair (*b*), the dominant allele will mask the recessive one. Thus in the example above 75% of the offspring will be black (*BB* or *Bb*) while only 25% will be white (*bb*). The ratio of the phenotypes is 3:1, typical for a monohybrid cro

Q3 **Show the** Diagrammatic representation of dihybrid cross.

Ans **Dihybrid cross:**

More complicated crosses can be made by looking at two or more genes. The Punnett square only works, however, if the genes are independent of each other, which means that having a particular allele of gene X does not imply having a particular allele of gene Y.

The following example illustrates a dihybrid cross between two heterozygous pea plants. *R* represents the dominant allele for shape (round), while *r* represents the recessive allele (wrinkled). *Y* represents the dominant allele for color (yellow), while *y* represents the recessive allele (green). If each plant has the genotype *RrYy*, and since the alleles for shape and color genes are independent, then they can produce four types of gametes with all possible combinations: *RY*, *Ry*, *rY* and *ry*.

	RY	**Ry**	**rY**	**ry**
RY	RRYY	RRYy	RrYY	RrYy
Ry	RRYy	RRyy	RrYy	Rryy
rY	RrYY	RrYy	rrYY	rrYy
ry	RrYy	Rryy	rrYy	rryy

Since dominant traits mask recessive traits, there are nine combinations that have the phenotype round yellow, three that are round green, three that are wrinkled yellow and one that is wrinkled green. The ratio 9:3:3:1 is typical for a dihybrid cross.

Q4 **Define test cross.**

Ans In genetics, a **test cross**, first introduced by Gregor Mendel, is used to determine if an individual exhibiting a dominant trait is homozygous or heterozygous for that trait. More simply test crosses determine the genotype of an individual with a dominant phenotype.

Test crosses involve breeding the individual in question with another individual that expresses a recessive version of the same trait. If all offspring display the dominant phenotype, the individual in question is homozygous dominant; if the offspring display both dominant and recessive phenotypes, then the individual is heterozygous.

In some sources, the "test cross" is defined as being a type of backcross between the recessive homozygote and F1 generation or F1 generation crossed with recessive parent is said to be a **Test cross**.

If the individual being tested produces *any* recessive offspring (except in cases of incomplete penetrance) its genotype is heterozygous. If all the offspring are phenotypically dominant, its genotype is homozygous

Q5 Define the laws of mendel.

Ans 1 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).

The law of dominance is used to explain the expression of only one of the parental characters in a monohybrid cross in the F1 and the expression of both in the F2. It also explains the proportion of 3:1 obtained at the F2.

2 Law of Segregation

This law is based on the fact that the alleles do not show any blending and that both the characters are recovered as such in the F2 generation though one of these is not seen at the F1 stage. Though the parents contain two alleles during gamete formation, the factors or alleles of a pair segregate from each other such that a gamete receives only one of the two factors. Of course, a homozygous parent produces all gametes that are similar while a heterozygous one produces two kinds of gametes each having one allele with equal proportion.

Q6 Define Incomplete Dominance.

***Ans* Incomplete and semi-dominance**

Complete dominance occurs when the phenotype of the heterozygote is completely indistinguishable from that of the dominant homozygote. This is frequently not the case. Incomplete dominance occurs when the phenotype of the heterozygous genotype is an intermediate of the phenotypes of the homozygous genotypes. For example, the snapdragon flower color is either homozygous for red or white. When the red homozygous flower is paired with the white homozygous flower, the result yields a pink snapdragon flower. The pink snapdragon is the result of incomplete dominance.

Q7 Define Co-dominance Codominance.

Ans

Co-dominance occurs when the contributions of both alleles are visible in the phenotype. In the **ABO** example, the $\boldsymbol{I^A}$ and $\boldsymbol{I^B}$ alleles are co-dominant in producing the **AB** blood group phenotype, in which both **A**-type and **B**-type antigens are made. Another example occurs at the locus for the Beta-globin component of hemoglobin, where the three molecular phenotypes of $\mathbf{Hb^A/Hb^A}$, $\mathbf{Hb^A/Hb^S}$, and $\mathbf{Hb^S/Hb^S}$ are all equally detectable by protein electrophoresis. (The medical condition produced by the heterozygous genotype is called an *incomplete dominant*, see above). For most gene loci at the molecular level, both alleles are expressed co-dominantly, because both are transcribed into RNA.

Co-dominance is different from incomplete or semi-dominance. For example, pink flowers might be the product of two alleles that produce red and white pigments that become mixed (co-dominance on the pigment level, no dominance on the color level), or the result of one allele that produces the usual amount of red pigment and another non-functional allele that produces no pigment, so as to produce a dilute, intermediate pink color (no dominance at either level).

Q 8 Explanation of the concept of dominance:

Ans **Dominance** in genetics is a relationship between two variant forms (alleles) of a single gene, in which one allele masks the expression of the other in influencing some trait. In the simplest case, if a gene exists in two allelic forms (**A** & **B**), three combinations of alleles (genotypes) are possible: **AA**, **AB**, and **BB**. If **AB** individuals (heterozygotes) show the same form of the trait (phenotype) as **AA** individuals (homozygotes), and **BB** homozygotes show an alternative phenotype, allele **A** is said to *dominate* or *be dominant to* allele **B**, and **B** is said to *be recessive to* **A**.

By convention, dominant alleles are written in uppercase letters, and recessive alleles in lowercase letters. In this example, allele ***B*** is replaced by ***a***. Then, **A** is *dominant* to **a** (and **a** is *recessive* to **A**), the **AA** and **Aa** genotypes have the same phenotype, and the **aa** genotype has a different phenotype

Q9 Define Law of Independent Assortment

Ans In the dihybrid cross , the phenotypes round, yellow; wrinkled, yellow; round, green and wrinkled, green appeared in the ratio 9:3:3:1. Such a ratio was observed for several pairs of characters that Mendel studied.The ratio of 9:3:3:1 can be derived as a combination series of 3 yellow: 1 green, with 3 round : 1 wrinkled. This derivation can be written as follows:

(3 Round : 1 Wrinkled) (3 Yellow : 1 Green) = 9 Round, Yellow : 3 Wrinkled, Yellow: 3 Round, Green : 1 Wrinkled, Green

Based upon such observations on **dihybrid crosses** (crosses between plants differing in two traits) Mendel proposed a second set of generalizations that we call Mendel's Law of Independent Assortment. The law states that 'when two pairs of traits are combined in a hybrid, segregation of one pair of characters is independent of the other pair of characters'.

The Punnett square can be effectively used to understand the independent segregation of the two pairs of genes during meiosis and the production of eggs and pollen in the F1 **RrYy** plant. Consider the segregation of one pair of genes **R** and **r**. Fifty per cent of the gametes have the gene **R** and the other 50 per cent have **r**. Now besides each gamete having either **R** or **r**, it should also have the allele **Y** or **y**. The important thing to remember here is that segregation of 50 per cent **R** and 50 per cent *r* is *independent* from the segregation of 50 per cent **Y** and 50 per cent **y**. Therefore, 50 per cent of the **r** bearing gamete has **Y** and the other 50 per cent has **y**. Similarly, 50 per cent of the **R** bearing gamete has *Y* and the other 50 per cent has **y**. Thus there are four genotypes of gametes (four types of pollen and four types of eggs).

The four types are **RY**, **Ry**, *rY* and **ry** each with a frequency of 25 per cent or ¼th of the total gametes produced. When you write down the four types of eggs and pollen on the

two sides of a Punnett square it is very easy to derive the composition of the zygotes that give rise to the F2 plants

Q10 Define Chromosomal Theory of Inheritance

Ans The **Boveri–Sutton chromosome theory** (also known as the **chromosome theory of inheritance** or the **Sutton-Boveri Theory**) is a fundamental unifying theory of genetics which identifies chromosomes as the carriers of genetic material It correctly explains the mechanism underlying the laws of Mendelian inheritance by identifying chromosomes with the paired factors (particles) required by Mendel's laws. It also states that chromosomes are linear structures with genes located at specific sites along the states simply that chromosomes, which are seen in all dividing cells and pass from one generation to the next, are the basis for all genetic inheritance

Q11 Define the Linkage and Recombination

Ans Morgan carried out several dihybrid crosses in *Drosophila* to study genes that were sex-linked. The crosses were similar to the dihybrid crosses carried out by Mendel in peas. For example Morgan hybridised yellow-bodied, white-eyed females to brown-bodied, red-eyed males and intercrossed their F1 progeny. He observed that the two genes did not segregate independently of each other and the F2 ratio deviated very significantly from the 9:3:3:1 ratio (expected when the two genes are independent).

Morgan and his group knew that the genes were located on the X chromosome and saw quickly that when the two genes in a dihybrid cross were situated on the same chromosome, the proportion of parental gene combinations were much higher than the non-parental type. Morgan attributed this due to the physical association or linkage of the two genes and coined the term **linkage** to describe this physical association of genes on a chromosome and the term **recombination** to describe the generation of non-parental gene combination.

Morgan and his group also found that even when genes were grouped on the same chromosome, some genes were very tightly linked (showed very low recombination) while others were loosely linked (showed higher recombination) For example he found that the genes white and yellow were very tightly linked and showed only 1.3 per cent recombination while white and miniature wing showed 37.2 per cent recombination. His student Alfred Sturtevant used the frequency of recombination between gene pairs on the same chromosome as a measure of the distance between genes and ‘mapped’ their position on the chromosome.

Q12 Define the chromosomal SEX DETERMINATION.

Ans

Chromosomal determination

XX/XY sex chromosomes

XY sex-determination system

The **XX/XY sex-determination system** is the most familiar sex-determination systems, as it is found in human beings, most other mammals, as well as some insects. However, at least one monotreme, the platypus, presents a particular sex determination scheme that in some ways resembles that of the ZW sex chromosomes of birds, and also lacks the SRY gene, whereas some rodents, such as several Arvicolinae (voles and lemmings), are also noted for their unusual sex determination systems. The platypus has ten sex chromosomes; males have an XYXYXYXYXY pattern while females have ten X chromosomes. Although it is an XY system, the platypus' sex chromosomes share no homologues with eutherian sex chromosomes. Instead, homologues with eutherian sex chromosomes lie on the platypus chromosome 6, which means that the eutherian sex chromosomes were autosomes at the time that the monotremes diverged from the therian mammals (marsupials and eutherian mammals). However, homologues to the avian DMRT1 gene on platypus sex chromosomes X3 and X5 suggest that it is possible the sex-determining gene for the platypus is the same one that is involved in bird sex-determination. However, more research must be conducted in order to determine the exact sex determining gene of the platypus

In the XY sex-determination system, females have two of the same kind of sex chromosome (XX), while males have two distinct sex chromosomes (XY). Some species (including humans) have a gene SRY on the Y chromosome that determines maleness; others (such as the fruit fly) use the presence of two X chromosomes to determine femaleness. The XY sex chromosomes are different in shape and size from each other unlike the autosomes, and are termed allosomes.

XX/X0 sex determination

X0 sex-determination system

In this variant of the XY system, females have two copies of the sex chromosome (XX) but males have only one (X0). The *0* denotes the absence of a second sex chromosome. This system is observed in a number of insects, including the grasshoppers and crickets of order and in cockroaches (order Blattodea).

The nematode *C. elegans* is male with one sex chromosome (X0); with a pair of chromosomes (XX) it is a hermaphrodite.

ZW sex chromosomes

ZW sex-determination system

The **ZW sex-determination system** is found in birds and some insects and other organisms. The ZW sex-determination system is reversed compared to the XY system: females have two different kinds of chromosomes (ZW), and males have two of the same kind of chromosomes (ZZ). In the chicken, this was found to be dependent on the expression of DMRT1

Q13 Define Sex Determination in Humans

Ans It has already been mentioned that the sex determining mechanism in case of humans is XY type. Out of 23 pairs of chromosomes present, 22 pairs are exactly same in both males and females; these are the autosomes. A pair of X-chromosomes are present in the female, whereas the presence of an X and Y chromosome are determinant of the male characteristic.

During spermatogenesis among males, two types of gametes are produced. 50 per cent of the total sperm produced carry the X-chromosome and the rest 50 per cent has Y-chromosome besides the autosomes. Females, however, produce only one type of ovum with an X-chromosome. There is an equal probability of fertilisation of the ovum with the sperm carrying either X or Y chromosome. In case the ovum fertilises with a sperm carrying X-chromosome the zygote develops into a female (XX) and the fertilisation of ovum with Y-chromosome carrying sperm results into a male offspring. Thus, it is evident that it is the genetic makeup of the sperm that determines the sex of the child. It is also evident that in each pregnancy there is always 50 per cent probability of either a male or a female child.

Q14 What is Mutation?

Ans Mutation is a phenomenon which results in alteration of DNA sequences and consequently results in changes in the genotype and the phenotype of an organism. In addition to recombination, mutation is another phenomenon that leads to variation in DNA. one DNA helix runs continuously from one end to the other in each chromatid, in a highly supercoiled form.

Therefore loss (deletions) or gain (insertion/duplication) of a segment of DNA, result in alteration in chromosomes. Since genes are known to be located on chromosomes, alteration in chromosomes results in abnormalities or aberrations. Chromosomal aberrations are commonly observed in cancer cells. In addition to the above, mutation also arise due to change in a single base pair of DNA. This is known as point mutation. A classical example of such a mutation is sickle cell anemia. Deletions and insertions of base pairs of DNA, causes frame-shift mutations (see Chapter 6). The mechanism of mutation is beyond the scope of this discussion, at this level. However, there are many chemical and physical factors that induce mutations. These are referred to as mutagens. UV radiations can cause mutations in organisms – it is a mutagen.

Q15 Define the GENETIC DISORDERS

Ans Mendelian Disorders

Broadly, genetic disorders may be grouped into two categories – Mendelian disorders and Chromosomal disorders. Mendelian disorders are mainly determined by alteration or mutation in the single gene. These disorders are transmitted to the offspring on the same lines as we have studied in the principle of inheritance. The pattern of inheritance of such Mendelian disorders can be traced in a family by the pedigree analysis. Most common and prevalent Mendelian disorders are Haemophilia, Cystic fibrosis, Sickle-cell anaemia, Colour blindness, Phenylketonuria, Thalesemia, etc. It is important to mention here that such Mendelian disorders may be dominant or recessive. By pedigree analysis one can easily understand whether the trait in question is dominant or recessive. Similarly, the

trait may also be linked to the sex chromosome as in case of haemophilia. It is evident that this X-linked recessive trait shows transmission from carrier female to male progeny

Haemophilia : This sex linked recessive disease, which shows its transmission from unaffected carrier female to some of the male progeny has been widely studied. In this disease, a single protein that is a part of the cascade of proteins involved in the clotting of blood is affected. Due to this, in an affected individual a simple cut will result in non-stop bleeding. The heterozygous female (carrier) for haemophilia may transmit the disease to sons. 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.

Sickle-cell anaemia : This is an autosome linked recessive trait that can be transmitted from parents to the offspring when both the partners are carrier for the gene (or heterozygous). The disease is controlled by a single pair of allele, HbA and HbS. Out of the three possible genotypes only homozygous individuals for HbS (HbSHbS) show the diseased phenotype. Heterozygous (HbAHbS) individuals appear apparently unaffected but they are carrier of the disease as there is 50 per cent probability of transmission of the mutant gene to the progeny, thus exhibiting sickle-cell trait . 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 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 structure

Phenylketonuria : This inborn error of metabolism is also 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.

Q16 Define Chromosomal disorders

Ans The chromosomal disorders on the other hand are caused due to absence or excess or abnormal arrangement of one or more chromosomes. Failure of segregation of chromatids during cell division cycle results in the gain or loss of a chromosome(s), called **aneuploidy**. For example, Down's syndrome results in the gain of extra copy of chromosome 21. Similarly, Turner's syndrome results due to loss of an X chromosome in human females. Failure of cytokinesis after telophase stage of cell division results in an increase in a whole set of chromosomes in an organism and, this phenomenon is known as **polyploidy**. This condition is often seen in plants. The total number of chromosome of a normal human being is 46 (23 pairs). Out of these 22 pairs are autosomes and one pair of chromosomes are sex chromosome. Sometimes, though rarely, either an additional copy of a chromosome may be included in an individual or an individual may lack one of

any one pair of chromosomes. These situations are known as trisomy or monosomy of a chromosome, respectively.
Such a situation leads to very serious consequences in the individual. Down'ssyndrome, Turner's syndrome, Klinefelter's syndrome are common examples of chromosomal disorders.

Down's Syndrome : The cause of this genetic disorder is the presence of an additional copy of the chromosome number 21 (trisomy of 21). This disorder was first described by Langdon Down (1866). 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.

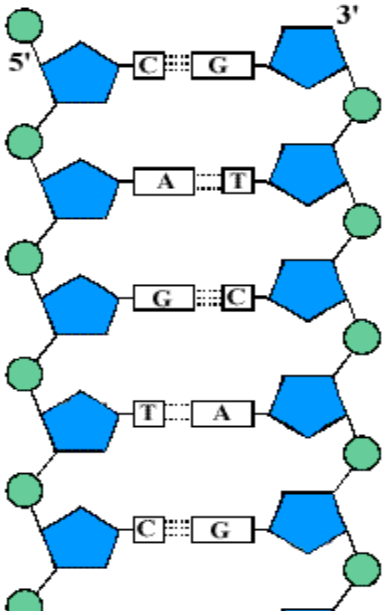
Klinefelter's Syndrome : This genetic disorder is also caused due to the presence of an additional copy of X-chromosome resulting into a karyotype of 47, XXY. Such an individual has overall masculine development , however, the feminine development (development of breast, i.e., Gynaecomastia) is also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

Turner's Syndrome : Such a disorder is caused due to the absence of one of the X chromosomes, i.e., 45 with X0, Such females are sterile as ovaries are rudimentary besides other features including lack of other secondary sexual characters .

.Chapter 5
Molecular Basis of Inheritance

Q1 Draw and label a simple diagram of the molecular structure of DNA.

Ans.



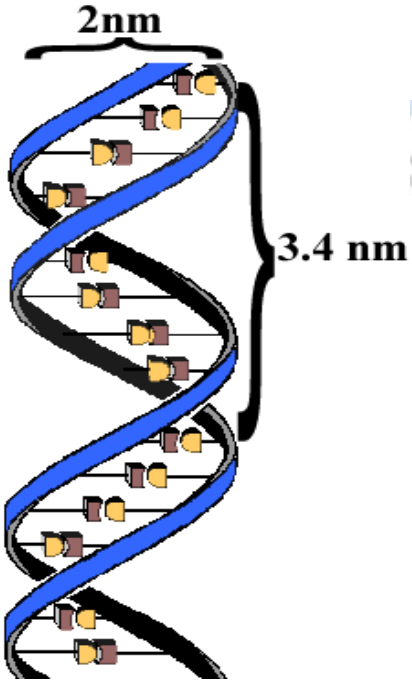
This image of DNA shows the arrangement of the two polynucleotide chains but not the helical shape which can be seen in the space filled model below.

This is image shows:

- Two polynucleotide chains.
- Two anti-parallel chains. (definition).

a)The number followed by the prime (') determined the carbon in deoxyribose free from bonding to another nucleotide.

b) Note that the two chains are in opposite directions 3' to 5' is parallel to 5' to 3' chain.



- The anti-parallel chains have a uniform distance (2nm) between the outside of the two sugar phosphate backbones
- Complementary base pairs: Inside the double helix bases form one strand hydrogen bond to bases on the opposite strand but always in the following way:

a) Adenine hydrogen binds to Thymine

b) Cytosine hydrogen bonds to Guanine

The three-dimensional structure of DNA was discovered in 1953 by Watson and Crick in Cambridge, using the experimental data of Wilkins and Franklin in London, for which work they won a Nobel prize. Ms Franklin however died before the award and the Nobel Prize is never awarded posthumously.

The main features of the structure are:

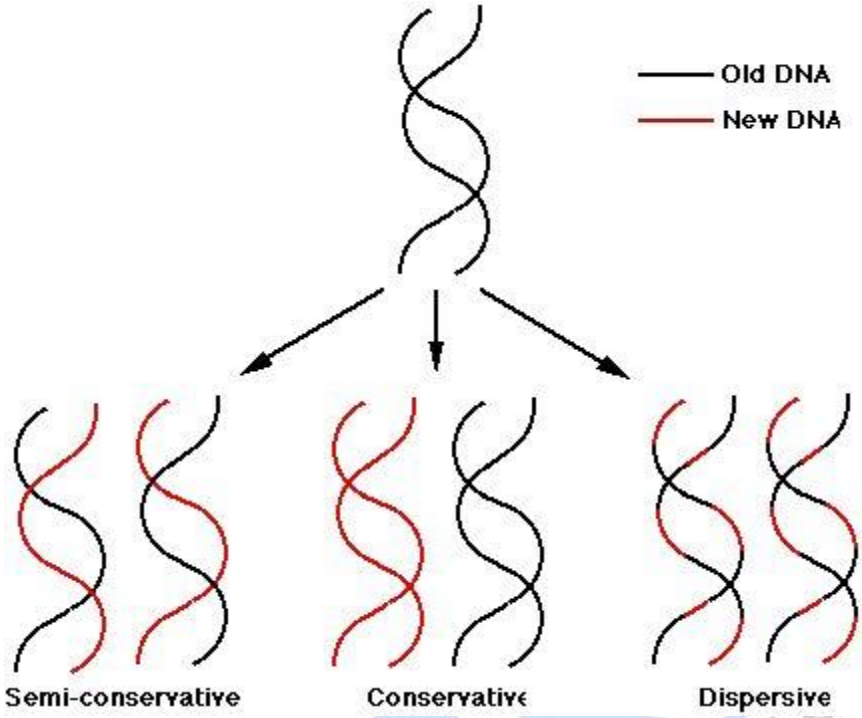
- DNA is double-stranded, so there are two polynucleotide stands alongside each other.
- The strands are antiparallel, i.e. they run in opposite directions thus 5' to 3' is parallel to 3' to 5'.
- The two strands are wound round each other to form a double helix (not a spiral, despite what some textbooks say).
- The two strands are joined together by hydrogen bonds between the bases.
- The bases therefore form base pairs, which are like rungs of a ladder.
- The base pairs are specific. A only binds to T (and T with A), and C only binds to G (and G with C).
- These are called complementary base pairs (or sometimes Watson-Crick base pairs). (A-T and G-C)
- This means that whatever the sequence of bases along one strand, the sequence of bases on the other stand must be complementary to it.

Q 2 Define the Replication.

Ans The Semiconservative Nature of DNA Replication

The Watson-Crick model of DNA structure suggested a possible mechanism for replication of DNA molecules. The nature of base pairing meant that if the two strands of a DNA molecule were separated, they could each serve as a template for the creation of a complementary strand by bringing in individual nucleotides to base pair with their complementary base on the template, and joining the new nucleotides together. Thus, each DNA molecule after replication would consist of one of the original strands plus one newly synthesized strand. This model of DNA replication is called **semiconservative**.

Semiconservative was not the only model of DNA replication, however. Other proposed models included **conservative** replication and **dispersive** replication. Conservative replication proposed that after replication, one DNA molecule consists entirely of newly synthesized DNA whereas the other molecule is entirely original DNA. Dispersive replication suggested that each DNA molecule after replication might consist of segments of new and old DNA interspersed. It would be difficult to devise a mechanism by which this latter outcome might occur, but until evidence to the contrary was produced, it had to be considered. The three possible models of DNA replication are depicted below.



The scheme suggested that the two strands would separate and act as a template for the synthesis of new complementary strands. After the completion of replication, each DNA molecule would have one mparental and one newly synthesised strand. This scheme was termed as **semiconservative** DNA replication

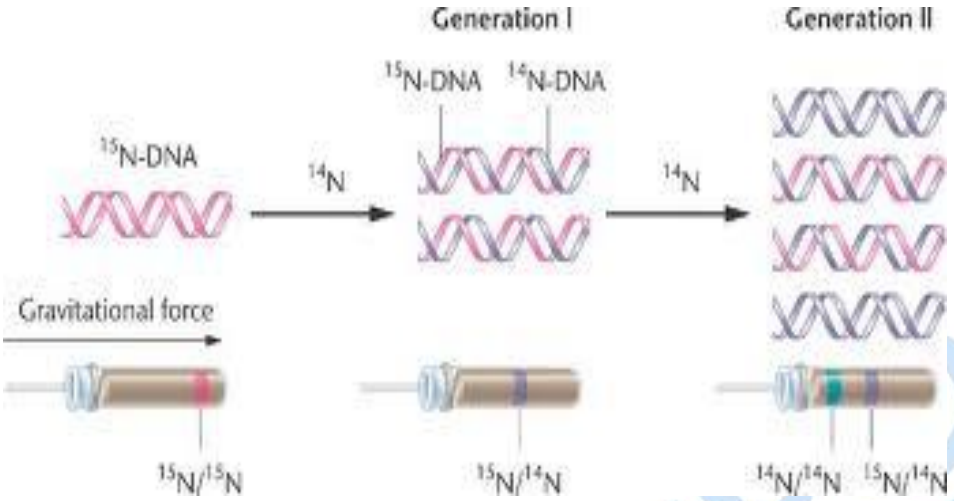
Q3 Define the The Experimental Proof of semi conservative method of DNA
Ans

To distinguish between these possibilities, Meselson and Stahl did the following experiment:

First, they grew bacteria for many generations in a growth medium containing ^{15}N. This is a heavy isotope of nitrogen (in contrast to the normal isotope, ^{14} N), which over many generations would be incorporated into all nitrogen-containing molecules of the cells, including DNA. DNA isolated from these cells could be distinguished from normal DNA because it would have a higher density.

The bacteria grown in heavy nitrogen were then transferred to growth medium containing ^{14} N for one round of replication. This lighter isotope would incorporate into any newly synthesized DNA. If semiconservative replication occurred, then each DNA molecule after replication would contain heavy nitrogen and light nitrogen, and would therefore have a density intermediate between the two. Conservative replication would produce one DNA molecule containing heavy nitrogen and one molecule containing light nitrogen, so

there would be two different densities. Dispersive replication would produce a single intermediate density, just like semiconservative.



The observed density of the DNA after one round of replication was intermediate. Replication was therefore either semiconservative or dispersive. These possibilities could be distinguished after a second round of replication. After two rounds, semiconservative replication would produce two DNA molecules containing only light nitrogen, and two DNA molecules containing one light strand and one heavy strand. Therefore there would be two different densities: light and intermediate. Two rounds of dispersive replication would produce four DNA molecules, each of which would contain mostly light nitrogen and some heavy nitrogen. There would be a single density (we'll call it 'slightly heavy'). When density of the DNA was measured after two rounds, two densities were observed: light and intermediate, indicating that DNA replication is **semiconservative**, and not dispersive or conservative

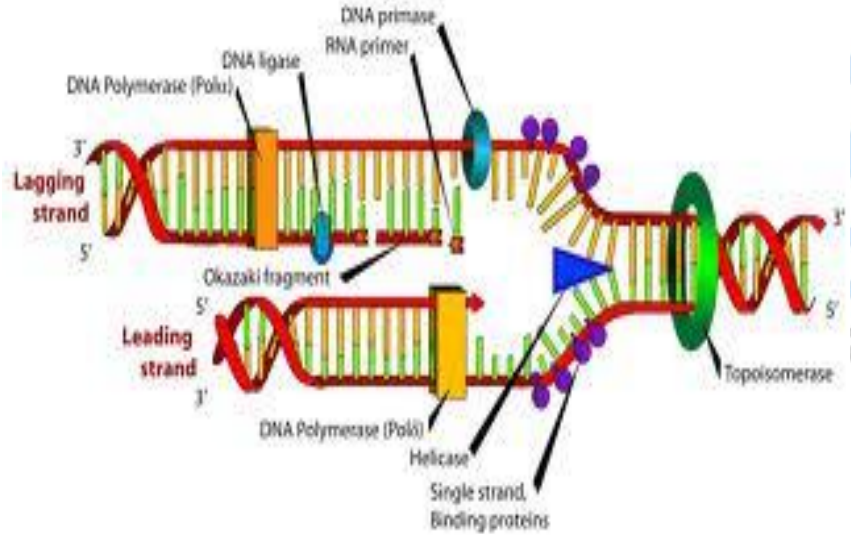
Q.4 Define the The Machinery and the Enzymes how used in replication.

Ans

In living cells, such as *E. coli*, the process of replication requires a set of catalysts (enzymes). The main enzyme is referred to as DNA-dependent **DNA polymerase**, since it uses a DNA template to catalyse the polymerisation of deoxynucleotides. These enzymes are highly efficient enzymes as they have to catalyse polymerisation of a large number of nucleotides in a very short time. *E. coli* that has only 4.6 ×106 bp (compare it with human whose diploid content is 6.6 × 109 bp), completes the process of replication within 38 minutes; that means the average rate of polymerisation has to be approximately 2000 bp per second. Not only do these polymerases have to be fast, but they also have to catalyse the reaction with high degree of accuracy. Any mistake during replication would result into mutations. Furthermore, energetically replication is a very expensive process. Deoxyribonucleoside triphosphates serve dual purposes. In addition to acting as substrates, they provide energy for polymerization reaction (the two terminal phosphates in a deoxynucleoside triphosphates are high-energy phosphates, same as in case of ATP). In addition to DNA-dependent DNA polymerases, many additional enzymes are required to complete the process of replication with high degree of accuracy. For long DNA

molecules, since the two strands of DNA cannot be separated in its entire length (due to very high energy requirement), the replication occur within a small opening of the DNA helix, referred to as **replication fork**. The DNA-dependent DNA polymerases catalyse polymerisation only in one direction,

This creates some additional complications at the replicating fork. Consequently, on one strand, the replication is **continuous**, while on the other it is **discontinuous**. The discontinuously synthesized fragments are later joined by the enzyme **DNA ligase** The DNA polymerases on their own cannot initiate the process of replication. Also the replication does not initiate randomly at any place in DNA. There is a definite region in *E. coli* DNA where the replication originates. Such regions are termed as **origin of replication**. It is because of the requirement of the origin of replication that a piece of DNA if needed to be propagated during recombinant DNA procedures, requires a vector. The vectors provide the origin of replication.

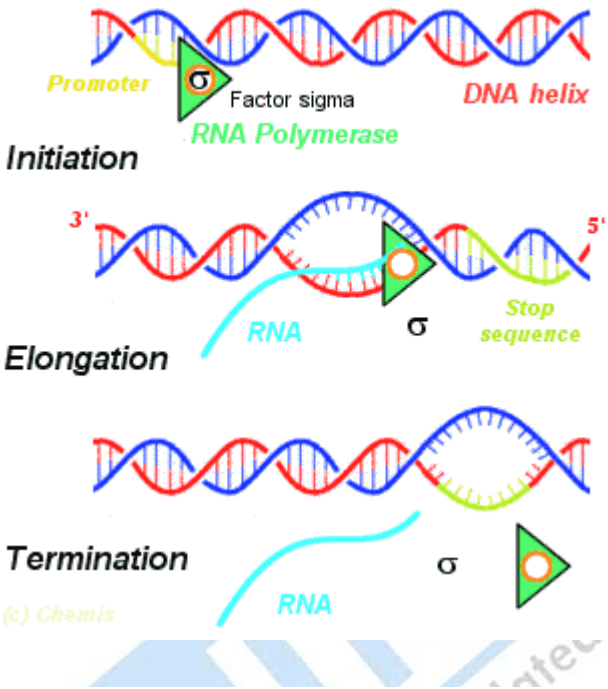


Further, not every detail of replication is understood well. In eukaryotes, the replication of DNA takes place at S-phase of the cell-cycle. The replication of DNA and cell division cycle should be highly coordinated. A failure in cell division after
DNA replication results into polyploidy(a chromosomal anomaly). You will learn the detailed nature of origin and the processes occurring at this site, in higher classes.

Q.5 Give the detail of TRANSCRIPTION

Ans DNA transcription is a process that involves the transcribing of genetic information from DNA to RNA. The transcribed DNA message is used to produce proteins. DNA is housed within the nucleus of our cells. It controls cellular activity by coding for the production of enzymes and proteins. The information in DNA is not directly converted into proteins, but must first be copied into RNA. This ensures that the information contained within the DNA does not become tainted.

TRANSCRIPTION



DNA Transcription

DNA consists of four nucleotide bases [adenine (A), guanine (G), cytosine (C) and thymine (T)] that are paired together (A-T and C-G) to give DNA its double helical shape.

There are three main steps to the process of DNA transcription.

- RNA Polymerase Binds to DNA

DNA is transcribed by an enzyme called RNA polymerase. Specific nucleotide sequences tell RNA polymerase where to begin and where to end. RNA polymerase attaches to the DNA at a specific area called the promoter region.

- Elongation

Certain proteins called transcription factors unwind the DNA strand and allow RNA polymerase to transcribe only a single strand of DNA into a single stranded RNA polymer called messenger RNA (mRNA). The strand that serves as the template is called the antisense strand. The strand that is not transcribed is called the sense strand.

Like DNA, RNA is composed of nucleotide bases. RNA however, contains the nucleotides adenine, guanine, cytosine and uricil (U). When RNA polymerase transcribes the DNA, guanine pairs with cytosine and adenine pairs with uricil.

- Termination

RNA polymerase moves along the DNA until it reaches a terminator sequence. At that point, RNA polymerase releases the mRNA polymer and detaches from the DNA.

Since proteins are constructed in the cytoplasm of the cell by a process called translation, mRNA must cross the nuclear membrane to reach the cytoplasm. Once in the cytoplasm, mRNA along with ribosome and another RNA molecule called transfer RNA, work together to produce proteins. Proteins can be manufactured in large quantities because a single DNA sequence can be transcribed by many RNA polymerase molecules at once.

Q6 What is GENETIC CODE?

Ans

The genetic code consists of 64 triplets of nucleotides. These triplets are called **codons**.With three exceptions, each codon encodes for one of the 20 amino acids used in the synthesis of proteins. That produces some redundancy in the code: most of the amino acids being encoded by more than one codon.

One codon, **AUG** serves two related functions:

- it signals the start of translation
- it codes for the incorporation of the amino acid methionine (Met) into the growing polypeptide chain

The genetic code can be expressed as either RNA codons or DNA codons. RNA codons occur in messenger RNA (**mRNA**) and are the codons that are actually "read" during the synthesis of polypeptides (the process called **translation**). But each mRNA molecule acquires its sequence of nucleotides by **transcription** from the corresponding gene. Because DNA sequencing has become so rapid and because most genes are now being discovered at the level of DNA before they are discovered as mRNA or as a protein product, it is extremely useful to have a table of codons expressed as DNA. So here are both.

Note that for each table, the left-hand column gives the first nucleotide of the codon, the 4 middle columns give the second nucleotide, and the last column gives the third nucleotide.

Q7 Give the feature of CODON .

Ans The salient features of genetic code are as follows:

(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.

(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.

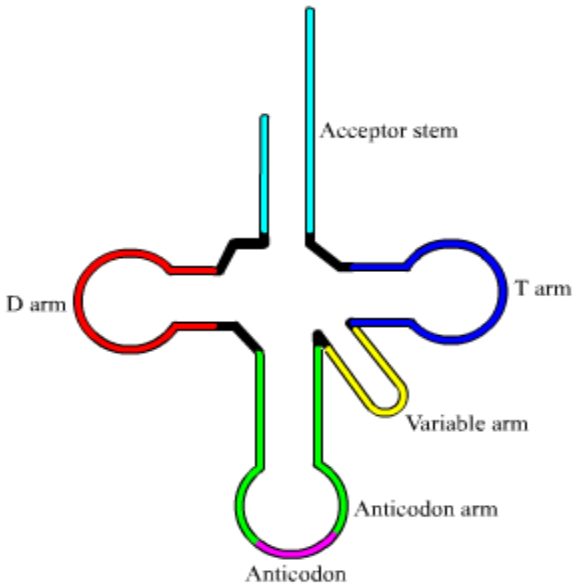
(vi) AUG has dual functions. It codes for Methionine (met) , and it also act as **initiator** codon.

Q8 Define the structure of tRNA– the Adapter Molecule.

Ans

The structure of tRNA can be decomposed into its primary structure, its secondary structure (usually visualized as the *cloverleaf structure*), and its tertiary structure (all tRNAs have a similar L-shaped 3D structure that allows them to fit into the P and A sites of the ribosome). The cloverleaf structure becomes the 3D L-shaped structure through coaxial stacking of the helices which is a common RNA Tertiary Structure motif.

1. The 5'-terminal phosphate group.
2. The acceptor stem is a 7-base pair (bp) stem made by the base pairing of the 5'-terminal nucleotide with the 3'-terminal nucleotide (which contains the CCA 3'-terminal group used to attach the amino acid). The acceptor stem may contain non-Watson-Crick base pairs.
3. The CCA tail is a cytosine-cytosine-adenine sequence at the 3' end of the tRNA molecule. This sequence is important for the recognition of tRNA by enzymes critical in translation. In prokaryotes, the CCA sequence is transcribed in some tRNA sequences. In most prokaryotic tRNAs and eukaryotic tRNAs, the CCA sequence is added during processing and therefore does not appear in the tRNA gene.
4. The D arm is a 4 bp stem ending in a loop that often contains dihydrouridine.
5. The anticodon arm is a 5-bp stem whose loop contains the anticodon.
6. The T arm is a 5 bp stem containing the sequence TΨC where Ψ is a pseudouridine.
7. Bases that have been modified, especially by methylation, occur in several positions throughout the tRNA. The first anticodon base, or wobble-position, is sometimes modified to inosine (derived from adenine), pseudouridine (derived from uracil) or lysidine (derived from cytosine).]



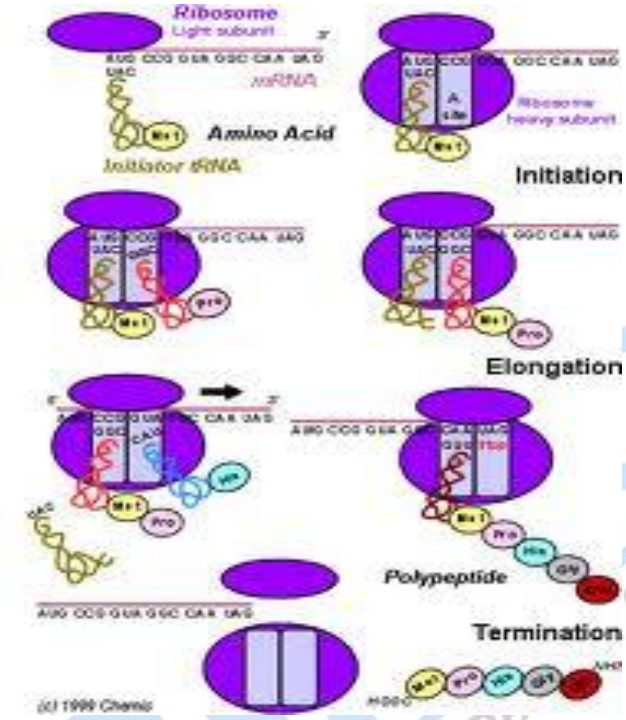
Q.9 Define the TRANSLATION.

Ans

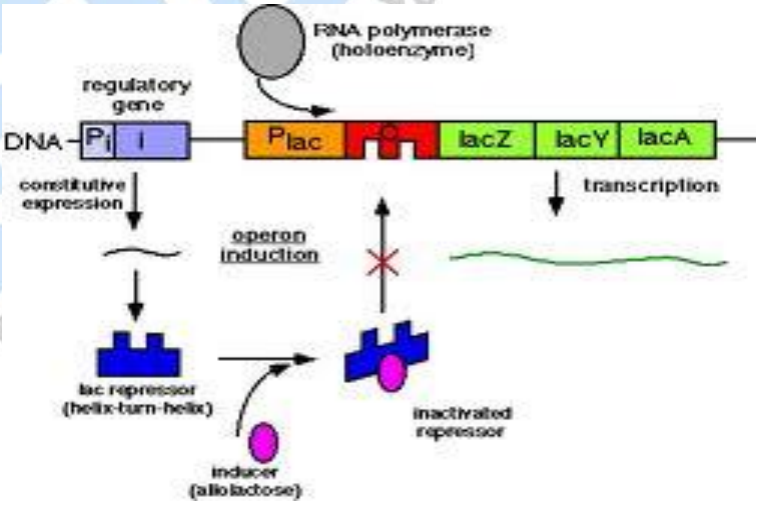
Translation refers to the process of polymerisation of amino acids to form a polypeptide . The order and sequence of amino acids are defined by the sequence of bases in the mRNA. The amino acids are joined by a bond which is known as a peptide bond. Formation of a peptide bond requires energy. Therefore, in the first phase itself amino acids are activated in the presence of ATP and linked to their cognate tRNA– a process commonly called as **charging of tRNA** or **aminoacylation of tRNA** to be more specific. If two such charged tRNAs are brought close enough, the formation of peptide bond between them would be favoured energetically. The presence of a catalyst would enhance the rate of peptide bond formation. The cellular factory responsible for synthesising proteins is the ribosome. The ribosome consists of structural RNAs and about 80 different proteins. In its inactive state, it exists as two subunits; a large subunit and a small subunit. When the small subunit encounters an mRNA, the process of translation of the mRNA to protein begins. There are two sites in the large subunit, for subsequent amino acids to bind to and thus, be close enough to each other for the formation of a peptide bond. The ribosome also acts as a catalyst (23S rRNA in bacteria is the enzyme- ribozyme) for the formation of peptide bond.

A translational unit in mRNA is the sequence of RNA that is flanked by the start codon (AUG) and the stop codon and codes for a polypeptide. An mRNA also has some additional sequences that are not translated and are referred as **untranslated regions** (**UTR**). The UTRs are present at both 5′ -end (before start codon) and at 3′ -end (after stop codon). They are required for efficient translation process. For initiation, the ribosome binds to the mRNA at the start codon (AUG) that is recognised only by the initiator tRNA. The ribosome proceeds to the elongation phase of protein synthesis. During this stage, complexes composed of an amino acid linked to tRNA, sequentially bind to the appropriate codon in mRNA by forming complementary base pairs with the tRNA anticodon. The ribosome moves from codon to codon along the mRNA. Amino acids are added one by one, translated into Polypeptide sequences dictated by DNA and

represented by mRNA. At the end, a **release factor** binds to the stop codon, terminating translation and releasing the complete polypeptide from the ribosome.



Q10 Define the The *Lac* operon.



The ***lac* operon** is an operon required for the transport and metabolism of lactose in *Escherichia coli* and some other enteric bacteria. It consists of three adjacent structural

genes, lacZ, lacY and lacA. The *lac* operon is regulated by several factors including the availability of glucose and of lactose. Gene regulation of the *lac* operon was the first complex genetic regulatory mechanism to be elucidated and is one of the foremost examples of prokaryotic gene regulation In its natural environment, the *lac* operon allows for the effective digestion of lactose. The cell can use lactose as an energy source by producing the enzyme β-galactosidase to digest that lactose into glucose and galactose. However, it would be inefficient to produce enzymes when there is no lactose available, or if there is a more readily-available energy source available such as glucose. The *lac* operon uses a two-part control mechanism to ensure that the cell expends energy producing β-galactosidase, β-galactoside permease and thiogalactoside transacetylase (also known as galactoside O-acetyltransferase) only when necessary. It achieves this with the lac repressor, which halts production in the absence of lactose, and the Catabolite activator protein (CAP), which assists in production in the absence of glucose. This dual control mechanism causes the sequential utilization of glucose and lactose in two distinct growth phases, known as diauxie. Similar diauxic growth patterns have been observed in bacterial growth on mixtures of other sugars as well, such as mixtures of glucose and arabinose, etc. The genetic control mechanisms underlying such diauxic growth patterns are known as *xyl* operon and ara operon, etc.

Q11 Give the goals of human genome project.

Ans. Some of the important goals of HGP were as follows:

(i) Identify all the approximately 20,000-25,000 genes in human DNA;
(ii) Determine the sequences of the 3 billion chemical base pairs that make up human DNA;
(iiii) Store this information in databases;
(iv) Improve tools for data analysis;
(v) Transfer related technologies to other sectors, such as industries;
(vi) Address the ethical, legal, and social issues (ELSI) that may arise from the project.

The Human Genome Project was a 13-year project coordinated by the U.S. Department of Energy and the National Institute of Health. During the early years of the HGP, the Wellcome Trust (U.K.) became a major partner; additional contributions came from Japan, France, Germany, China and others. The project was completed in 2003. Knowledge about the effects of DNA variations among individuals can lead to revolutionary new ways to diagnose, treat and someday prevent the thousands of disorders that affect human beings. Besides providing clues to understanding human biology, learning about non-human organisms DNA sequences can lead to an understanding of their natural capabilities that can be applied toward solving challenges in health care, agriculture, energy production, environmental remediation. Many non-human model organisms, such as bacteria, yeast, *Caenorhabditis elegans* (a free living non-pathogenic nematode), *Drosophila* (the fruit fly), plants (rice and *Arabidopsis*), etc., have also been sequenced.

Methodologies : The methods involved two major approaches. One approach focused on identifying all the genes that expressed as RNA (referred to as **Expressed Sequence**

Tags (ESTs). The other took the blind approach of simply sequencing the whole set of genome that contained all the coding and non-coding sequence, and later assigning different regions in the sequence with functions (a term referred to as **Sequence Annotation**). For sequencing, the total DNA from a cell is isolated and converted into random fragments of relatively smaller sizes (recall DNA is a very long polymer, and there are technical limitations in sequencing very long pieces of DNA) and cloned in suitable host using specialised vectors. The cloning resulted into amplification of each piece of DNA fragment so that it subsequently could be sequenced with ease. The commonly used hosts were bacteria and yeast, and the vectors were called as **BAC** (bacterial artificial chromosomes), and **YAC** (yeast artificial chromosomes).

The fragments were sequenced using automated DNA sequencers that worked on the principle of a method developed by Frederick Sanger. (Remember, Sanger is also credited for developing method for determination of amino acid sequences in proteins). These sequences were then arranged based on some overlapping regions present in them. This required generation of overlapping fragments for sequencing. Alignment of these sequences was humanly not possible. Therefore, specialized computer based programs were developed (Figure 6.15). These sequences were subsequently annotated and were assigned to each chromosome. The sequence of chromosome 1 was completed only in May 2006 (this was the last of the 24 human chromosomes – 22 autosomes and X and Y to be sequenced). Another challenging task was assigning the genetic and physical maps on the genome. This was generated using information on polymorphism of restriction endonuclease recognition sites, and some repetitive DNA sequences known as microsatellites (one of the applications of polymorphism in repetitive DNA sequences shall be explained in next section of DNA fingerprinting).

Q.12 Give the Salient Features of Human Genome.

Ans Some of the salient observations drawn from human genome project are as follows:

(i) The human genome contains 3164.7 million nucleotide bases.

(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 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. This information promises to revolutionise the processes of

finding chromosomal locations for disease-associated sequences and tracing human history.

Q13 Write a short note on DNA FINGERPRINTING.

Ans DNA fingerprinting involves identifying differences in some specific regions in DNA sequence called as **repetitive DNA**, because in these sequences, a small stretch of DNA is repeated many times. These repetitive DNA are separated from bulk genomic DNA as different peaks during density gradient centrifugation. The bulk DNA forms a major peak and the other small peaks are referred to as **satellite DNA**. Depending on base composition (A : T rich or G:C rich), length of segment, and number of repetitive units, the satellite DNA is classified into many categories, such as micro-satellites, mini-satellites etc. These sequences normally do not code for any proteins, but they form a large portion of human genome. These sequence show high degree of polymorphism and form the basis of DNA fingerprinting. Since 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. Further, as the polymorphisms are inheritable from parents to children, DNA fingerprinting is the basis of paternity testing, in case of disputes. As polymorphism in DNA sequence is the basis of genetic mapping of human genome as well as of DNA fingerprinting, it is essential that we understand what DNA polymorphism means in simple terms.

Polymorphism (variation at genetic level) arises due to mutations. New mutations may arise in an individual either in somatic cells or in the germ cells (cells that generate gametes in sexually reproducing organisms). If a germ cell mutation does not seriously impair individual's ability to have offspring who can transmit the mutation, it can spread to the other members of population (through sexual reproduction). Allelic sequence variation has traditionally been described as a DNA polymorphism if more than one variant (allele) at a locus occurs in human population with a frequency greater than 0.01. In simple terms, if an **inheritable mutation** is observed in a population at high frequency, it is referred to as **DNA polymorphism**. The probability of such variation to be observed in no coding DNA sequence would be higher as mutations in these sequences may not have any immediate effect/impact in an individual's reproductive ability. These mutations keep on accumulating generation after generation, and form one of the basis of variability/polymorphism. There is a variety of different types of polymorphisms ranging from single nucleotide change to very large scale changes.

The technique of DNA Fingerprinting was initially developed by Alec Jeffreys. He used a satellite DNA as probe that shows very high degree of polymorphism. It was called as **Variable Number of Tandem Repeats** (VNTR). The technique, as used earlier, involved Southern blot hybridisation using radiolabelled VNTR as a probe. It included

(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. A schematic representation of DNA fingerprinting is shown in Figure 6.16.

The VNTR belongs to a class of satellite DNA referred to as mini-satellite. A small DNA sequence is arranged tandemly in many copy numbers. The copy number varies from chromosome to chromosome in an individual. The numbers of repeat show very high degree of polymorphism. As a result the size of VNTR varies in size from 0.1 to 20 kb. Consequently, after hybridisation with VNTR probe, the autoradiogram gives many bands of differing sizes. These bands give a characteristic pattern for an individual DNA . It differs from individual to individual in a population except in the case of monozygotic (identical) twins. The sensitivity of the technique has been increased by use of polymerase chain reaction (PCR–you will study about it in have been shown to contain different copy number of VNTR. For the sake of understanding different colour schemes have been used to trace the origin of each band in the gel. The two alleles (paternal and maternal) of a chromosome also contain different copy numbers of VNTR. It is clear that the banding pattern of DNA