Chapter - 37
Mutation

Sudden stable and inheritable changes in the genetical material of the cell of all kinds of organisms are called mutations. Mutations occur in nature consistently but our focus is only on those mutations which are effective otherwise the effect of most of mutations is very weak or they are recessive.

The term mutation was first of all used by Hugo de Vries in 1901 for those of phenotypic changes that were inheritable. He observed various differences (variations) in evening primrose Oenothra lamarckiana. In this plant suddenly seven new varieties have been which were called mutant varieties by him and this process is called mutation.

Type of Mutation : Fundamentally mutations are of two types-

1. Gene mutation
2. Chromosomal mutation

1. Gene Mutation : Replication of DNA takes place during meiosis. Usually this process completes absolutely correct or without any error but DNA is defectly formed at some times in which one or more than one pairs of nitrogen base are changed. The sequence of base pairs is the specificity of gene thus when there is change in the gene then it is called gene mutation. This change may be in structure of gene or its chemical composition because this change occurs in gene at a point or locus of a chromosome. Hence, this is also called point mutation. Because nucleotides are made of nitrogen bases hence due to their different arrangement they are also called base pair substitution mutation. The behaviour of such changed gene will be different from the original gene. Such type of free mutation occurs in nature.

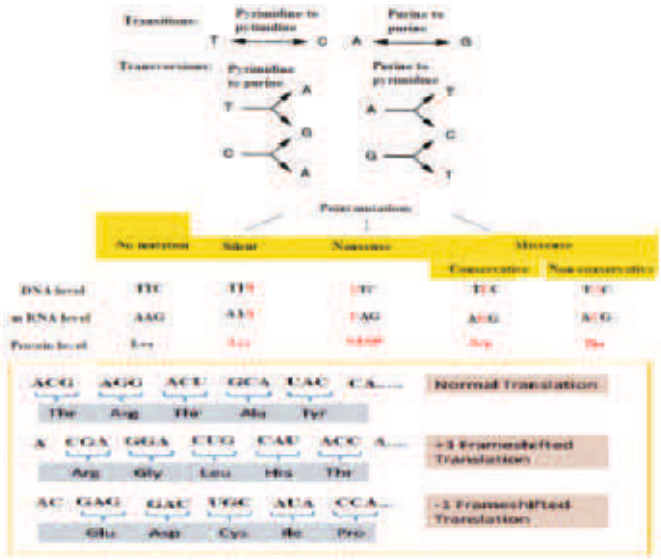

Fig. 37.1 : Variaus types of gene mutations

During the experiments scientists found that approximately out of one million fruit flies (Drosophila) mutation had already taken in 200-300 flies. Some characters of such mutant flies such as colour of eye and types of wings etc. are permanently inherited.

Mutation takes place in all livings from bacteria like microorganism to human beings. The effect of some mutations is so minute that the changes caused by them can't be seen by us. Some lethal mutation are recessive type thus till they are not in homozygous state their changes are not observed.

Gene mutations are of following type :

(I) Transition : In this type of gene mutation one purine base is replaced by another purine base or one pyramidine by another pyramidine base.

ATGCATGC → AC GCATGC

(ii) Transversion : In this type of gene mutation one purine base is replaced by another pyrimidine base or in the same way of pyrimidine base is replaced by other purine base.

ATGCATGC → ATGAATGC

(iii) Frame shift : In this gene mutation the frame of reading of complete gene and codes of genes is changed due to loss or getting of one nucleotide.

CATCACATCAT → CATATCCACATCAT

(iv) Missense : Due to the replacement of the nucleotide (base) the change in the genetic code results in addition of another amino acid. Sickle cell haemoglobin like disease can be caused by changed protein.

(v) Nonsense : If the genetic code changes in such type that in half way it becomes stop codon, then no any protein is formed.

GAAGAAGAA → GAAUAAGA

The protein synthesis stops because UAA is a nonsense codon.

(vi) Silent : In this type of gene mutation any of phenotypic is not changed by change in nucleotide.

Features of Gene Mutation :

1. Gene mutations are completely uncertain.
2. Gene mutations can take place without any predictions.
3. Any one gene can be mutated any number of times. Mutant gene can remain in wild type for some time.
4. Gene mutation is unidirectional after completing in one situation it can again mutate to its original form.
5. Almost all genes presently found in about all livings are mutant form of original gene.
6. Some mutant forms are dominant which definitely change the expression of their trait whereas some mutant forms are recessive as their expression is supressed by their dominant form.
7. Mostly the mutations are harmful to organisms because cell structure is very complex, therefore, any type of change in their gene is harmful and distructive to them.
8. Cell can die soon due to dominant gene mutation thus dominant mutations end where as recessive gene mutations can remain for longer period in cell because due to them any cell does not die.
9. Some micro dominent mutations whose effect is mixed with the cell activity they remain for a long period because inspite of mutation cell remains alive.

Factors affecting or inducing Gene Mutations:

Many factors affect the rate of gene mutations. Out of these factors some factors can be used to induce mutations artificially. For this first of all scientists Muller in 1927 performed an experiment on Drosophila by x-rays. These works performed by Muller are very significant in this field Muller was awarded Nobel Prize in 1946 for this.

Those substances or factors which cause mutation in any cell are called mutagens. Mutagens can be of the following types:

1. Normal ultra violet rays
2. α, β γ rays emitted from radioactive substances.
3. Neutron and Proton
4. Chemical mutagens, such as mustard gas group, gases of actinomycin D
5. Increased temperature increases the rate of mutation.

(2) Chromosomal Mutations or Chromosomal Aberrations :

When there is any difference in the gene arrangement of chromosome or addition of any additional gene or some genes get lost then that is called chromosomal mutation or chromosomal

aberrations. These are of two types:

(a) Numerical changes in chromosomes

(b) Structural changes in chromosomes

1. Numerical changes in chromosomes or polyploidy:

The number of chromosome is always fixed in all members of a species. In this type of mutation there is no change in the gene itself but the number of chromosomes become double, triple or many more then it is called chromosomal Mutation or polyploidy. Due to abnormality or hybridization in cell division the number of these chromosomes is changed. These changes can be of two types –

(i) Euploidy : The set of basic chromosomes present in an organism or cell is called monoploidy. If two or more than two sets of chromosomes are found in any organism then it is called euploidy. These are also of two types :

(a) Autopolyploidy : Such polyploid in which some initial sets of chromosomes are found is called autopolyploidy such as triploid (3n), tetraploid (4n) and pentaploid (5n) etc. It can be induced by colchicine alkaloid. Colchicine is extracted from corm of Colchicum autumnale. This alkaloidl breaks the spindle by making it loose by which the chromosomes do not move to the poles and their number becomes double.

(b) Allopolyploidy : In this more than two sets of chromosomes of two different species are present in any organism. The origin of these organisms is by hybridization of two different species such as *Raphanobrassica* (2n=36). Russian scientist G. D. Karpechenko obtained it in 1927 by crossing between *Raphanus sativus* (radish) (2n=18) and *Brassica oleracea* (cauliflower) (2n=18). It was completely sterile. Triticale formed by Triticum (wheat) and Secale (Rye) is the first autopolyploid crop formed by human, which is commercially produced in India and many other countries.

(ii) Aneuploidy : If non-disjunction in chromosomes takes place during chromosomal segregation in cell division resulting increase or decrease the number of one or more chromosomes in the newly formed cells then it is called aneuploidy. It is of two types-

(a) If one or more than one chromosomes are decreased then it is called hypoploidy.

(b) If one or more than one chromosomes are increased then it is called hyperploidy.

In hypoploidy if there is loss of one chromosome then it is called monosomy (2n-1) and if one pair or two chromosomes become lost from the homologous chromosomes then it is called nullisomy 2n-2. Similarly in hyperploidy one chromosome is more, then it is called Trisomy 2n+1. In human Down's syndrome or Mongolism is one of its examples. If one pair becomes more in homologous chromosomes then it is called tetrasomey (2n+2).

2. Structural changes in chromosomes :

In another type of mutation structural changes in chromosomes take place due to which main behaviour or the nature of the chromosome changes. Hence it is called Chromosomal aberration.

Chromosomal aberrations are mainly of four types :

(a) Deletion : It is considered to be the simplest type of chromosomal aberration. The shortage/loss/deficiency of a large or small acentric segment of a chromosome is called Deletion. If terminal part of any chromosome is lost then it is called terminal deletion, whereas breaking of middle or intercalary segment into two parts is called interstitial deletion i.e., the genes are deleted from lost part of that chromosome. In this type of mutation if out of the two homologous chromosomes some portion is deleted then recessive gene of other chromosome expresses its effect. If deletion takes place in both the chromosomes then result can be deadly.

(b) Translocation : When the broken end of any chromosome joins to the other end of the same chromosomes or joins to any other non-homologous chromosome then it is referred as translocation type aberration. In unilateral translocation the chromosomal segment from one chromosome

reaches to another chromosome but there is no exchange in both sides. In contrast of this the exchange of chromosomal segments takes place in both sides bilateral translocation. If trand location of segment is mutual in between two non-homologous chromosomes then it is called Reciprocal translocation.

Due to position effect phenotype of the offspring can be changed in this process. Sometimes the broken parts of two non-homologous chromosomes attach each other i.e., they translocate to form two homologous chromosomes.

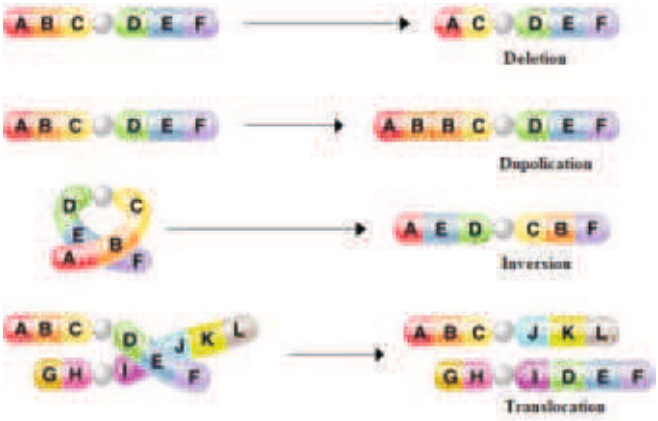

Fig. 37.2 : Types of structural changes in chromosomes

(c) Inversion : In inversion one part of chromosome is rearranged in reverse order. Chromosome is broken at first two points and this broken segment is rotated at 1800 in it and the rotated part is attached again. In this way the gene sequence is inverted. In such condition the genotype of the chromosome remains unchanged but the phenotype is changed in offspring. Because locus of gene is changed in this process, it is called a position effect. It is of two types:

1. Pericentral : In this type of inversion the centromere is present in the inverted segment.

2. Paracentral : In this type of inversion the centromere is attached outside of the inverted segment.

(d) Duplication : Sometimes any area of any chromosome repeated twice then duplication of those genes is considered. These extra parts of the chromosomes may attach to the middle portion or terminal portion of duplicated chromosome or any other chromosome (Fig. 37.2).

Genetic Code

Information for protein synthesis is present in the sequence of nucleotides in DNA. This coded information was discovered by Nirberg and Mathais Ocho in 1968.

Genetic code means the information present in the DNA molecule for synthesis of a specific protein structure which synthesizes a polypeptide chain from the stored information. That gene or DNA segment in which complete coded information for synthesis of a complete polypeptide chain (protein) is available that is called cistron.

Following are the characteristics of genetic codes:

(1) Genetic code is a triplet code : It means that "sequence of three bases in a gene that is called codon, which has information of special amino acid". The sequence of codon determines the sequence of amino acids in a protein.

(2) Genetic code is clear means a particular codon can be coded a particular acid only.

(3) Genetic code is coma less and non-overlapping. This means it can be read continuously from starting to end.

(4) Genetic code is degenerated. Different proteins of livings are formed from 20 amino acids only but one base codon is formed from 3 bases out of 4 bases present in 4 nucleotides then total codon may be 4^3=64. Thus, more than one codon code for particular amino acids. Actually you can see from the figure that the first two bases are common for one amino acid and the third base changes. It is called Wobble hypothesis.

(5) Genetic code is read from m RNA during protein synthesis.

(6) AUG codon codes for methionine amino acid and it is known as initiation codon because it is the first codon transcribed by cistron.

(7) UAA, UAG, UGA are stop codons and any one of these three codons is present at the terminal of cistron to stop protein synthesis.

(8) Genetic code is universal and same for

almost all the organisms found on the earth.

Human Genome Project: Genome project is that scientific project whose objective is to find out the complete genome sequence of any animal. Genes are keys of our life. How we appear or work is decided by the minute genes hidden in our body up to great extent. Not only had this had the genes indicate towards human history and future. DNA sequence of two different persons is different at some points. These facts compel to find out complete DNA sequence of human genome. It is believed by gene scientists that if once the structure of complete human genes is identified then on the basis of gene horoscope, the prediction of all biological events and somatic characters can be possible. Though this is not any easy task because there are thousands lakhs living cells in the human body. The large group of genes present in any cell is known as genome.

To know the human genome sequence in 1988 the Human Genome Project was started in United States of America. It was formally started in 1990. This project was started with partnership of National Institute of Health and Department of Energy. Over time it acquired worldwide about 250 laboratories of eighteen countries are involved in it. Actually it is a very ambitious, large and most expensive bio scientific project. This project is called mega project. This project was initiated under the direction of James D. Watson and later on its work was completed under the direction of Francis Collins.

Table 37.1 : Knowledge of various Genetic Codes

First Position	Second Position U				Second Position C				Second Position A				Second Position G				Third Position
	code	Amino Acid	code	Amino Acid	code	Amino Acid	code	Amino Acid	code	Amino Acid	code	Amino Acid	code	Amino Acid	code	Amino Acid	
U	UUU	phe	UCU	ser	UAU	tyr	UGU	cys	U								
	UUC		UCC		UAC		UGC		C								
	UUA	leu	UCA		UAA	STOP	UGA	STOP	A								
	UUG		UCG		UAG	STOP	UGG	trp	G								
C	CUU	leu	CCU	pro	CAU	his	CGU	arg	U								
	CUC		CCC		CAC		CGC		C								
	CUA		CCA		CAA	gln	CGA		A								
	CUG		CCG		CAG		CGG		G								
A	AUU	ile	ACU	thr	AAU	asn	AGU	ser	U								
	AUC		ACC		AAC		AGC		C								
	AUA		ACA		AAA	lys	AGA	arg	A								
	AUG	met	ACG		AAG		AGG		G								
G	GUU	val	GCU	ala	GAU	asp	GGU	gly	U								
	GUC		GCC		GAC		GGC		C								
	GUA		GCA		GAA	glu	GGA		A								
	GUG		GCG		GAG		GGG		G								

Main objectives of this project are :

1. To identify one lakh genes of human genome.
2. To determine approximately.

   3 billion 20 million bases sequences of human genome.
3. To collect above information in the date base.
4. To develop new tool for data analysis.
5. To resolve ethical, legal and social issues by the project.
6. Along with this to determine the complete

base sequence of genome of many model organisms such as bacterium E. Coli, Yeast (Saccharomyces cerevisiae), round worm (Cenorehebdited alegens), fruit fly (Drosophila) and mice (Mus musculus).

Method :

(1) Two important procedures have been used in human genome project. In first procedure all those genes were identified that expressed themselves in the form of RNA. These were called expressed sequence tag.

(2) Remaining complete genome in which coding and non-coding sequence of were in second produce, to receive their information their functions was determined. This is known as Sequence annotation.

(3) Complete DNA is too big hence their sequence cannot determine in one step. Thus for complete knowledge of DNA it is broken randomly into small segments. After it this random segment is joined to proper cloning carrier and introduced into the host cell. Each segmented DNA is amplified by this sequence of these DNA segments can be fixed easily.

(4) Mainly Bacterial Artificial Chromosome (BAC) and Yeast Artificial chromosome (YAC) are used as cloning carrier whereas bacteria and yeast cells are used as host cell.

(5) Sequence of amplified DNA segments are determined by DNA sequencer which is developed by Di Deoxy chain Termination method used for determination of DNA sequence by Fredrick Sangar.

(6) The sequenced DNA segments are arranged on the basis of overlapping. It is essential for determination of segments for sequencing.

(7) It is not possible by human to overlapping of these sequenced segments. Thus these sequenced DNA segments are queued by the computer based special program.

(8) To prepare the genetical and physical map of genome (Genome mapping) the polymorphism of identification site of restriction endonuclease and repeated DNA sequences which is called micro-satellites presence is used.

Main features of human genome :

(I) 3165.7 billion bases are found in human genome.

(ii) According to a guess approximately 80,000 to 140,000 genes are present but from human genome project it is estimated that 30,000 genes are present in human.

(iii) Each gene has overage 3,000 bases. Human has biggest gene Dystrophin in which 2.4 billion bases are found.

(iv) Single nucleotide polymorphism was identified by human genome project at about 1.4 billion places single nucleotide polymorphism is found in human. This information will provide important role in finding disease based sequence on chromosomes.

(v) Out of the discovered genes functions of about 50% genes have been identified.

(vi) Less than 2% genome is coded protein in human genome

(vii) In chromosome 1 there are maximum (2968) genes and in Y chromosomes lower most (231) genes are found.

(viii) A major part of human genome is made up of repetitive sequence.

(ix) Repetitive sequence is small expended part of DNA which sometimes can be repeated more than 100 times. These sequences have no role in relation with coding for protein synthesis. This sequence focuses on the structure, mobility and development of chromosomes.

DNA Finger Printing

Human identity is properties and name. Two persons are not similar in all properties. (1) Twins also no matter how similar why not, yet they found vary. Skin colour, hair colour, colour of iris, height, voice, walking, up postures, way of talking, and standard of living etc. are such characters by which humans can be differentiate and identified.

Personal identity and distinction (2) was

needed legally. Marks of fingers vary in each human. There are ridges at different places in them method. In this way the image formed is called finger print. Actually it is a legal of human identification, which was first developed by Francis-Galton and remains prevalent. It is the gift of nature.

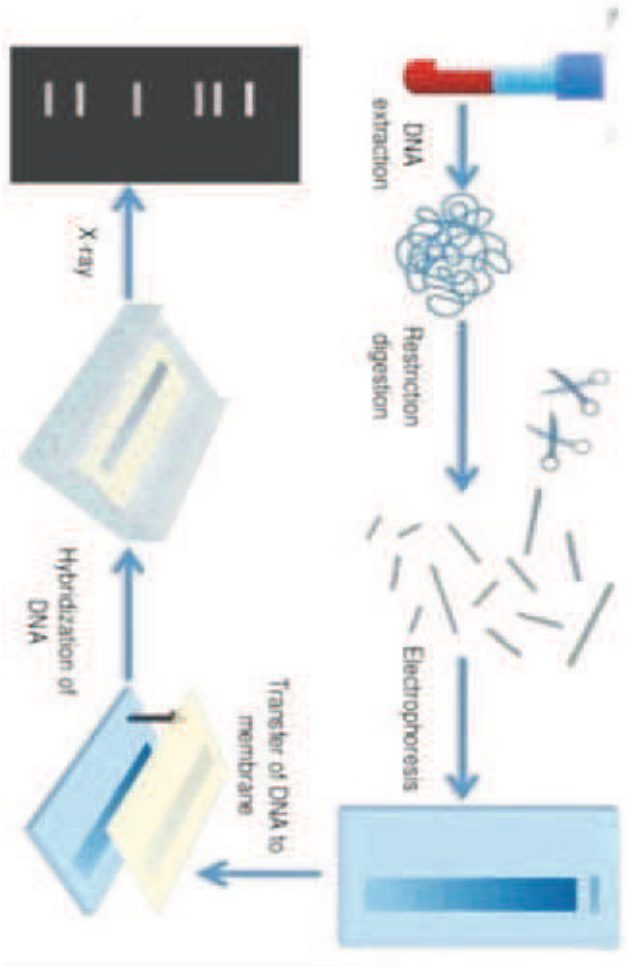

Fig. 37.3 : Technique of DNA Finger Printing

DNA in all the loving beings of the world, like in human beings is based on heredity. It is also called life formula. It is found in every micro unit of living being. Like the finger print DNA is also unique. DNA finger print is similar for each cell, tissue and organ of a person. On the basis of hidden micro variations in life formula obtained from their life parents can be identified separately from any other organism. It can't be changed by any known treatment. For DNA finger print such genes are selected which are extremely polymorphic i.e. such DNA sequence which have polymorphic alleles in human population. Thus they are different in different individuals.

British geneticist Dr. Alec Jefferys in 1984 developed the techniques of DNA finger printing. It is believed that this technique will provide signs of hereditary diseases and helped in its primary treatment.

Principal : Small nucleotide repeats are found in DNA. Their number is different in each person but these are hereditary. These are called Variable Number Tandem Repeats (VNTRs) VNTRs of two individuals may be of same lengths and at some points sequence is same but can be different at other.

As in a child six sequence repeating chromosome can be inherited from his mother and same sequence of homologous chromosome inherited from father can be repeated four times. Thus, half VNTRs of child are equal to mother and half are equal to father.

DNA finger printing techniques: DNA finger printing is completed in following steps.

(1) First of all, DNA is isolated by high speed refrigerated centrifuge from any tissue such as semen, skin cells or blood cells or follicle cells of hair.

(2) If quantity of DNA is obtained very less, it can amplified by polymerase chain reaction.

(3) After this DNA is fragmented by restriction endonuclease for analysis of Restriction Fragment Length Polymorphism (RFLP) that recognize specific locations.

(4) On the basis of molecular size these fragmented DNA segments are isolated from each other by agarose gel electrophoresis technique. These isolated DNA fragments are dyed by a fluorescent dye like ethidium bromide and viewed under ultra violet light.

(5) By the application of basic chemicals double stranded DNA is changed into single stranded DNA. This process is called DNA denaturation.

(6) This single stranded DNA is transferred from

agarose gel to nitro cellulose membrance. This process is called Southern blotting.

(7) This nitrocellulose membrane is kept with probe. The probe is radioactive synthesized segments of known segments of DNA. Probes are proper nucleotide sequence which is complementary of VNTR sequences. When these probe found on complementary sequence can hybridize with them.

(8) Now this nitrocellulose membrane with probe is exposed from X-ray. Such places where complementary bases base bonds are formed or hybridized from DNA situated on nitro cellulose membrane there dark bands are formed on X-ray film.

(9) These bands are compared with other bands for analysis with which to be compared.

Applications of DNA finger printing

(i) Investigation of criminal and family matters: In disputed paternity issues of a child DNA of parents is matched with that of child's DNA and actual parents can be identified. This technique is also used in forensic labs to identify criminals. Blood, semen, hair, remains of mutilated dead body, teeth or bone pieces found at crime spot can be positively identified. By this DNA of the accused and DNA found from the remains is compared. Thus this technique is being considered very essential in murders, rapes, heinous crime, property inheritance, divorce and Positive identity of parents in civil lawsuits etc. Because each person has the DNA of both parents hence on the basis of VNTRs present in DNA forms the basis it may be confirmed.

(ii) Medical and health check : Before conception (Pregnancy) or during pregnancy by the use of this technique hereditary diseases and congenital genetic defects can be known and frequency of these disorders can be controlled to some extent and the problem of whole humanity can be solved.

(iii) The DNA of a migrant person is compared with DNA of other person whom he calls his relative and citizen of that country in which nearest he wants to visit that country. In this way blood relatives can be detected by comparing DNA.

(iv) To understand the organic evolution relationship of different groups can be identified by comparing their DNA from this technique.

Cloning

Clone is a Greek word which means Twig (Klon=Twig). As all the branches of a tree are morphologically and genetically identical, similarly, clones are also identical to each other. The organism identical to their parent or clone can be developed by cloning. Thus clone is only such organic composition that is developed by asexual method from only parent father or mother. In this way developed clone is structurally and genetically quite identical to their parent. In nature this type of reproduction is found in many organisms but this process is not found automatically in higher organisms especially in mammals.

It is believed that twins are also a form of clone because their birth is from two cells formed by first division of fertilized egg or zygote being developed in separate isolated embryo.

Cloning is such a technique by which many cells from one cell or many genes from one gene can be formed. The cells taken from any part of the plant by which whole plant is to be formed to be cultured in special cultural medium, whole plant is obtained in this method.

Usually nuclear transfer technique is used for cloning in animals. During this technique the nucleus is removed from cell by mechanical method and it is introduced in enucleated (without nucleus) egg electrical waves are applied for its fertilization and division. electrical waves are passed. As a result of this the cell divides rapidly. In this process fully develops egg is implanted in the uterus of female and identical clone is obtained.

Scientist had found by various experiments that capability of formation of complete organism is found in egg or zygote of organism. By using this knowledge J.B. Gurdon of Oxford University performed an experiment in 1969 in which the nucleus of unfertilized egg of frog was destroyed by ultra violet radiations and nucleus of intestinal

epithelial cell of tadpole is introduced into unfertilized egg. Out of many eggs used in it, tadpoles are developed from transplanted eggs. These tadpoles were identical to their ancestors in genotype and phenotype. This nucleus transplant technique of Gurdon is still used in cloning in a modified way (Fig. 37.5).

Types of cloning

Cloning may be of two types :-

(1) Gene cloning : Formation of identical copies of a gene or chromosome or its one part is called gene cloning. This type of cloning is done for study of DNA. For this, cloning carrier or an instrument thermo cycler is used. Thermo cycler is used for polymerase chain reaction.

(2) Animal cloning : In this type of cloning complete organism is cloned. This type of cloning has three methods-

(a) Blastomere separation : Zygote is formed by the fusion of male gamete (sperm) and female gamete (ovum). Initial embryonic cells blastomeres are formed from this zygote. If each of the blastomere cells are to be separated and are allowed to develop them, then a complete embryo can be formed from each separated cell. Because the genetic material of developed embryos and adults developed from these are identical hence the embryos formed from it are clones of each other. This type of cloning is called twinning. This method is also called embryo cloning.

(b) Nuclear Transfer Technique : In this technique clone of an adult organism is formed by the process of without fertilization. In this technique the nucleus is removed from female egg and nucleus of the desired cell is implanted there. Developing embryo and adult is the copy or clone that animal from which the nucleus was taken.

Dr. Ian Wilmut and co-workers were developed "Dolly" named sheep in 1996 in Roslin Institute, Scotland by using this technique.

Mechanism of cloning

'Dolly' Sheep was developed by following method :

Dr. Wilmut removed a cell from the mammary glands of a 6 year old sheep. It was called donor strain. This cell was reprogrammed by gradually decreasing its nutrition means the original form of that cell was destroyed. After this unfertilized egg is taken from ovary of another sheep its nucleus is removed. In this manner enucleated egg was obtained. This sheep was called egg donor sheep.

After this the nucleus of mammary gland cell was combined with the enucleated egg cell by electrical stimulations. This transplant cell was kept in culture medium for about one week to grow. By division that transplanted cell was transformed into a blastocyst stage in a week. In this stage this blastocyst was implanted in the uterus of a third adult sheep. This sheep is called foster mother. After gestation period of about 5 months world's first mammal clone was born on 5th July, 1996 at 4.00 pm which was called Dolly.

This complete process can be explained by given fig. 37.4.

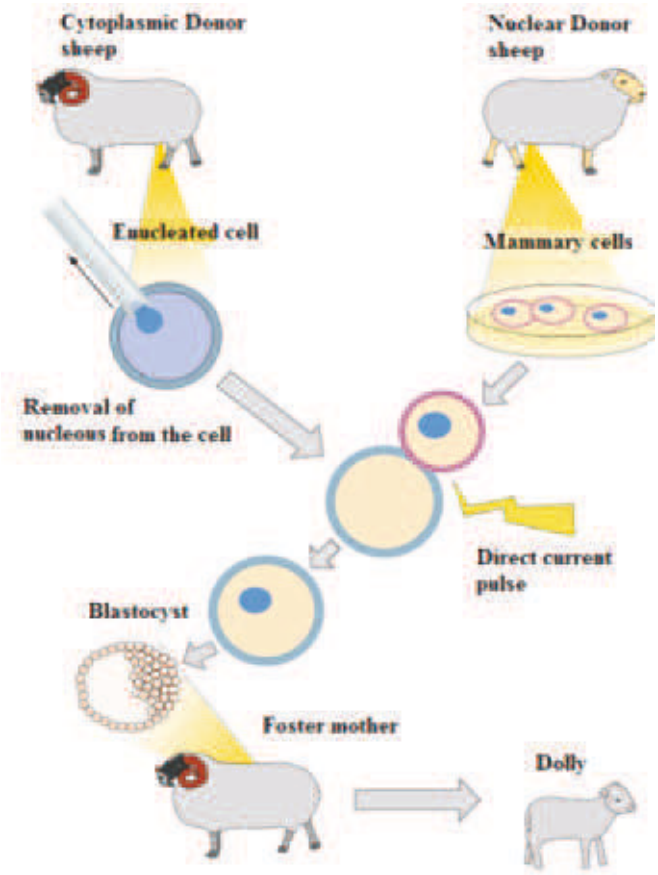

Fig. 37.4 : Method of formation of Dolly Sheep

Fig. 37.5 : Gurdon's experiment

Noori-Pashmina clone goat : World's first clone Pashmina goat was born on 09 March, 2012 in Kashmir under a research project conducting under joint collaboration of Sher-e-Kashmir Agriculture University for Science and Technology and National Dairy Research Institute, Karnal. This project was sponsored by World Bank. This clone goat was named 'Noori'. This name is derived from Arabic language which means- Light. Pashmina sheep is a species found in cold hilly regions whose wool is extremely expensive.

(c) Honolulu technique : This technique was developed by Teruhiko Wakayama in 1998 under the direction of Rujo Yanagimachi in University of Howaii. In this technique donor and recipient cells are not fused but nucleus of donor cell is implanted into the egg cell like Gurdon's technique. Besides this, the nucleus of a cell which is in G0 and G1 phase of cell cycle is used before cloning. Due to this it is no need to change into resting stage. He announced to clone about 50 mice by this technique. The first cloned mica of them was 'Cumulus'. This mouse was cloned for three generations. It is proved from this that there is no any abnormality is developed.

Application of cloning :

(1) On damaging any organs of human or in many diseases such as- leukemia, damaged liver, Parkinson, Alzimer, diabetes, heart and renal diseases, organ transplantation is a useful treatment. For this, organs can be designed by cloning to provide organ accessible. This is called therapeutic cloning.

(2) Farm animals can be improved by this process. In this process useful gene from another animal or human can be transferred in farm animals. These animals are called transgenic animals. Transgenic animals are developed by cloning for obtaining more milk wool and meat.

(3) Many such heredity diseases that have no any treatment till now their treatment can be possible by cloning and gene engineering.

(4) Insect resistant, high productive crops and high salt tolerant crops can be produced by cloning.

Important Points

1. Total genes present in a set of haploid chromosomes are inherited as a unit through gametes from one generation to another, are called genome. Study of genome is called genome science.
2. The arrangement of gene is changed due to change in the structure of a chromosome is called chromosomal aberration.
3. Deletion, duplication, inversion and translocation are methods of chromosomal aberration.
4. The damage/loss of any segment of chromosome is called deletion.
5. Under reverse heredity any special gene is first identified and then its phenotype is studied.
6. Chromosomal aberration in which a segment of a gene is repeated more than one time is called duplication.
7. The rotation of a group of gene at 1800 within

the chromosome and reuniting is called inversion. Inversion is of two types- pericentic and paracentic inversion.

8. Chromosomal aberration in which rearrangement between segments of non-homologous chromosomes is called translocation.
9. There are two types of numerical changes in the chromosomes: (i) Euploidy and (ii) Aneuploloidy.
10. If two or more than two sets of primary chromosomes are present in any organism then it is called euploidy.
11. If in an organism from its diploid (2n) number addition or deletion of one or more chromosomes takes place then it is called aneuploidy.
12. Human Genome project was started in 1990 for the aim of identity and determine base pair sequence of all the genes of human.
13. The first director of human genome project was James D. Watson.
14. Sudden changes in chromosomes are known as chromosomal mutations.
15. If there is a change in one nitrogenous base pair then it is known as point or gene mutation.
16. Short nucleotides repetitive are found in DNA. Its number is different in each person but these are inherited. These are known as VNTRs (Variable Number Tandem Repeats).
17. The organisms identical to their parent or clone are developed by cloning.

Practice Questions

Multiple choice Questions -

1. Genome is :
   (a) Total number of genes present on haploid chromosome set.
   (b) Total number of chromosomes present on haploid chromosome set
   (c) Total number of chromosomes present on diploid chromosome set
   (d) Total number of genes found on the chromosomes of zygote
2. The number of nucleotide pairs in human is about
   (a) Three lakhs (b) Thirty lakhs
   (c) Three crore (d) Three billion
3. What is the basis of DNA finger printing?
   (a) Formation of exact replication by DNA
   (b) Taking impression of fingers with the help of DNA
   (c) DNA sequencing image of any two individuals is not identical
   (d) None of the above
4. 'Dolly' sheep was obtained by which method?
   (a) By normal hybridization
   (b) By normal reproduction method
   (c) By cloning
   (d) By tissue culture
5. What is in mutation?
   (a) Temporary change in the genetic material of the cell
   (b) Permanent and inheritable change in the genetic material of the cell
   (c) Any change in the cytoplasm of the cell
   (d) Any type of variation
6. When adenine is replaced by guanine in a mutation process then it is called
   (a) Frame shift mutation (b) Transcription
   (c) Transition (d) Transversion
7. Genetic code has:
   (a) 3 bases, 64 codons
   (b) 3 bases, 18 codons
   (c) 2 bases, 32 codons
   (d) 2 bases, 64 codons
8. Polyploidy can be artificially developed by-
   (a) Colchicine (b) X-rays
   (c) Gama rays (d) None of above

Very short Answer Questions-

1. Which international agencies were initiated the human genome project?
2. What is VNTRs?
3. What is called the stage in which the number of chromosomes changes?
4. What is non-disjunction?
5. What is mutagen?
6. What is probe?
7. Which scientist discover Honolulu Technique and where?

Short Answer Questions-

1. Define gene mutation.
2. What is duplication?
3. What is point mutation?
4. What do you understand by genome?
5. What do you understand by euploidy?
6. What is silent mutation?
7. What is embryo cloning or twinning technique?
8. What is Wobble hypothesis?

Essay type Questions-

1. What is mutation? Write its characteristics.
2. Describe the structural changes in chromosomes in detail.
3. Write an essay on 'Human Genome project in detail.
4. Explain in detail about DNA finger printing technique and explain its applications.
5. What is the meaning of cloning? How was developed the first animal clone of the world? Give its detail.
6. Describe in detail the numerical changes of chromosomes.

Answer Key-

1 (a) 2 (d) 3 (c) 4 (c) 5 (b) 6 (c) 7 (a) 8 (a)