HUMAN GENETICS

Introduction

The study of transmission of characters from one generation to the other in human population is called human genetics. The principles of inheritance are applicable to man in the same way as in other animals and plants. Only during the past several decades human genetics became a flourishing discipline with increasingly sophisticated techniques for the diagnosis and treatment of many inherited conditions. The experimental study of inheritance of characters in human is not so easy as it has been in case of guinea pig, Drosophila etc. however, more than two hundred traits are reported to be inherited in human.

14.1 Chromosomes

The chromosomes are capable of self-reproduction and maintaining morphological and physiological properties through successive generations. They are capable of transmitting the contained hereditary material to the next generation. Hence these are known as '**hereditary vehicles**'. The eukaryotic chromosomes occurs in the nucleus and in certain other organelles, and are respectively called nuclear and extranuclear chromosomes. Nuclear chromosomes are long, double stranded DNA molecules of linear form and associated with proteins, separated from the cytoplasm by nuclear envelope and replicated during S phase of cell cycle, while extranuclear chromosomes are present in the mitochondria and plastid. They are short, double stranded DNA molecules of circular form and are not associated with proteins and also called prochromosomes.

(i) **Discovery of chromosomes**

Hofmeister (1848) : First observed chromosomes in microsporocytes (microspore mother cells) of *Tradescantia*.

Flemming (1879) : Observed splitting of chromosomes during cell division and coined the term, 'chromatin'.

Roux (1883) : He believed the chromosomes take part in inheritance.

W.Waldeyer (1888) : He coined the term 'chromosome'.

Benden and Boveri (1887) : They found a fixed number of chromosomes in each species.

(ii) **Kinds of chromosomes**

(a) **Viral chromosomes :** In viruses and bacteriophages a single molecule of DNA or RNA represents the viral chromosome.

(b) **Bacterial chromosomes :** In bacteria and cyanobacteria, the hereditary matter is organized into a single large, circular molecule of double stranded DNA**,** which is loosely packed in the nuclear zone. It is known as bacterial chromosome or *nucleoid*.

(c) **Eukaryotic chromosomes :** Chromosomes of eukaryotic cells are specific individualized bodies, formed of deoxyribonucleo proteins (DNA + Proteins).

(iii) **Number of chromosomes :** The number of chromosomes varies from two, the least number an organism can have, to a few hundred in different species. The number of chromosomes a species possesses has no basic significance, nor it necessarily shows relationship between two different species that have the same number.

Both dog and fowl have 78 chromosomes. Thus, it is not the number of chromosomes, but the genes in them which differentiate species. Their number also does not indicate the size or complexity of the organism. *Amoeba proteus* has 250 chromosomes and man has 46. The related species tend to have similar chromosome. Man and his nearest relatives, the apes, have chromosomes similar in size, shape and banding pattern. The least number of chromosomes are found in *Ascaris megalocephala i.e.* 2 while in a radiolarian protist (*Aulocantha*) has maximum number of chromosomes is 1600. The male of some roundworms and insects have one chromosome less than the females. For instance, the male and female roundworm *Coenorhabditis* have 11 and 12 chromosomes respectively and the male and female cockroach (*Blatta*) have 23 and 24 chromosomes respectively.

Diploid number of chromosomes in some animals

Common name	Zoological name	Chromosomes
(1) Man	*Homo sapiens*	46
(2) Gorilla	*Maccaca mulatta*	48
(3) Pig	*Sas scrofa*	40
(4) Sheep	*Ovis aries*	54
(5) Cat	*Felis maniculata*	38
(6) Dog	*Canis familiaris*	78
(7) Rat	*Rattus rattus*	42
(8) Rabbit	*Oryctolagus cuniculus*	44
(9) Honey bee	*Apis mellifera*	32, 16
(10) Mosquito	*Culex sp*	6
(11) Grasshopper	*Gryllus*	23(male), 24(female)

(iv) **Chromosome structure :** Different regions or structure recognized in chromosomes are as under

(a) **Pellicle :** It is the outer thin but doubtful covering or sheath of the chromosome.

(b) **Matrix :** Matrix or ground substance of the chromosome is made up of proteins, small quantities of RNA and lipid. It has one or two chromonemata (singular-chromonema) depending upon the state of chromosome.

(c) **Chromonemata :** They are coiled threads which form the bulk of chromosomes. A chromosome

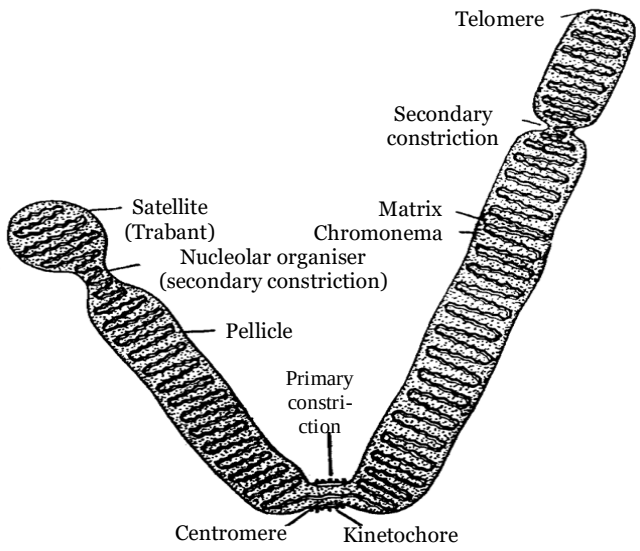


Fig : Structure of chromosome

may have one (anaphase) or two (prophase and metaphase) chromonemata. The coiled filament was called chromonema by Vejdovsky in 1912. The coils may be of the following 2 types

(1) **Paranemic coils :** When the chromonemal threads are easily separable from their coils then such coils are known as paranemic coils.

(2) **Plectonemic coils :** When the chromosomal threads remain inter-twined so intimately that they cannot be separated easily are known as plectonemic coils.

(d) **A primary Constriction and Centromere (kinetochore) :** A part of the chromosome is marked by a constriction. It is comparatively narrow than the remaining chromosome. It is known as primary constriction. The primary constriction divides the chromosome into two arms. It shows a faintly positive Feulgen reaction, indicating presence of DNA or repetitive type. This DNA is called centromeric heterochromatin.

Centromere or kinetochore lies in the region of primary constriction. The microtubules of the chromosomal spindle fibres are attached to the centromere. Therefore, centromere is associated with the chromosomal movement during cell division. Kinetochore is the outermost covering of centromere.

Type of chromosomes based on number of centromeres : Depending upon the number of centromeres, the chromosomes may be :

(1) Monocentric with one centromere.

(2) Dicentric with two centromeres, one in each chromatid.

(3) Polycentric with more than two centromeres.

(4) Acentric without centromere. Such chromosomes represent freshly broken segments of chromosomes, which do not survive for long.

(5) Diffused or non-located with indistinct throughout the length of chromosome. The microtubules of spindle fibres are attached to chromosome arms at many points. The diffused centromeres are found in insects, some algae and some groups of plants.

Types of chromosomes based on position of centromere : Based on the location of centromere the chromosomes are categorised as follows :

(1) **Telocentric :** These are rod-shaped chromosomes with centromere occupying a terminal position. One arm is very long and the other is absent.

(2) **Acrocentric :** These are rod-shaped chromosomes having subterminal centromere. One arm is very long and the other is very small.

(3) **Submetacentric :** These are J or L shaped chromosomes with centromere slightly away from the mid-point so that the two arms are unequal.

(4) **Metacentric :** These are V-shaped chromosomes in which centromere lies in the middle of chromosomes so that the two arms are almost equal.

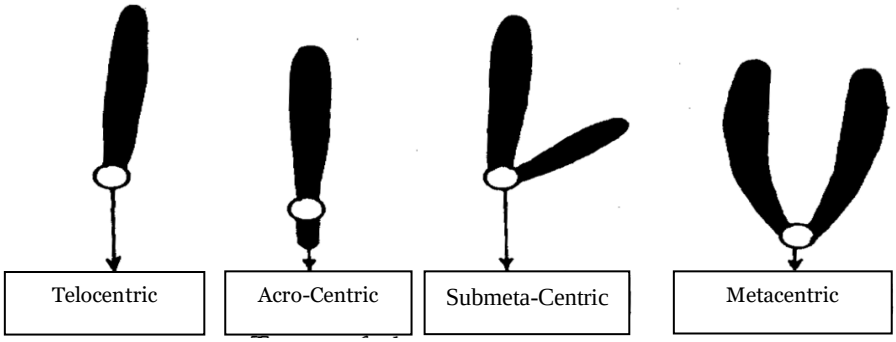


Fig : Types of chromosomes

(e) **Chromomeres :** Chromomeres are linearly arranged bead-like and compact segments described by J. Bellings. They are identified by their characteristic size and linear arrangement along a chromosome.

(f) **Secondary constriction or nucleolar organizer :** Sometimes one or both the arms of a chromosome are marked by a constriction other than the primary constriction. During interphase this area is associated with the nucleolus and is found to participate in the formation of nucleolus. It is, therefore, known as nucleolar organizer region or the secondary constriction. In certain chromosomes, the secondary constriction is (In human beings 13, 14, 15, 20 and 21 chromosome are nucleolar organizer) intimately associated with the nucleolus during interphase. It contains genes coding for 18S and 28S ribosomal RNA and is responsible for the formation of nucleolus. Therefore, it is known as nucleolar organizer region (*NOR*).

(g) **Telomeres :** The tips of the chromosomes are rounded and sealed and are called telomeres which play role in Biological clock. The terminal part of a chromosome beyond secondary constriction is called *satellite*. The chromosome with satellite is known as *sat chromosome*, which have repeated base sequence.

(h) **Chromatids :** At metaphase stage a chromosome consists of two chromatids joined at the common centromere. In the beginning of anaphase when centromere divides, the two chromatids acquire independent centromere and each one changes into a chromosome.

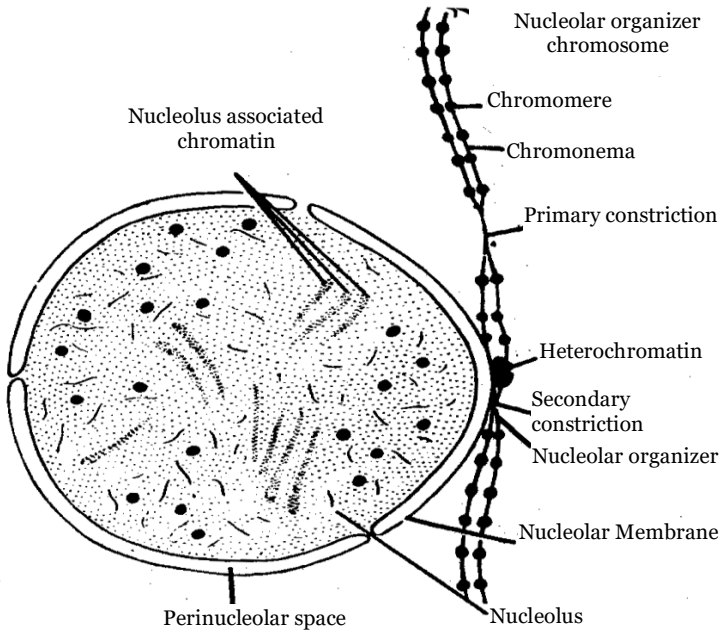


Fig : Nucleolus organizer or secondary constriction and its association with nucleolus

(v) **Molecular organisation of chromosome :** Broadly speaking there are two types of models stating the relative position of DNA and proteins in the chromosomes.

(a) **Multiple strand models :** According to several workers (Steffensen 1952, Ris 1960) a chromosome is thought to be composed of several DNA protein fibrils and atleast two chromatids form the chromosome.

(b) **Single strand models :** According to Taylor, Duprow etc. The chromosome is made up of a single DNA protein fibril. There are some popular single strand models.

(1) **Folded fibre model :** Chromosomes are made up of very fine fibrils 2 *nm* - 4 *nm* in thickness. As the diameter of DNA molecule is also 2 *nm* (20*Å*). So it is considered that a single fibril is a DNA molecule. It is also seen that chromosome is about a hundred times thicker than DNA whereas the length of DNA in chromosome is several hundred times that of the length of chromosome. So it is considered that long DNA molecule is present in folding manner which forms a famous model of chromosome called folded fibre model which given by *E.J. Dupraw* (1965).

(2) **Nucleosome model :** The most accepted model of chromosome or chromatin structure is the 'nucleosome model' proposed by Kornberg and Thomas (1974). Nucleosomes are also called *core particles or Nu-bodies*. The name nucleosome was given by *P. Outdet* et al. The nucleosome is a oblate particle of 55*Å* height and 110*Å* diameter. Woodcock (1973) observed the structure of chromatin under electron microscope. He termed each beaded structure on chromosome as nucleosome. Nucleosome is quasicylindrical structure made up of histones and DNA.

Histone are mainly of two types :

(a) **Nucleosomal histone :** These are small proteins responsible for coiling DNA into nucleosome. These are H_2A, H_2B, H_3 and H_4. Each histone protein consist of two molecule, thus the four histone proteins form a octamer. These form the inner core of nucleosome.

(b) **Linker histone :** H_1 proteins is known as linker histone that connect one core particle with another. These are present once per 200 base pairs. These are loosely associated with DNA. H_1 histone are responsible for packing of nucleosome into 30 *nm* fibre.

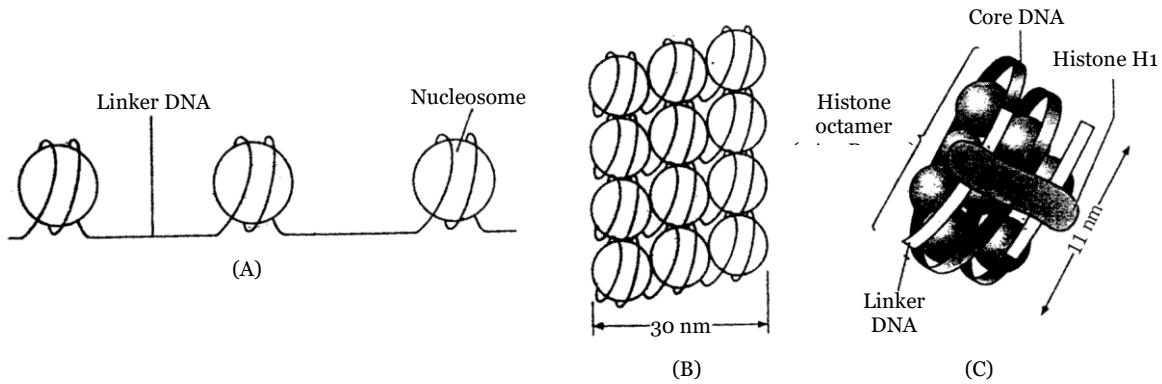


Fig : Nucleosomes : (A) 3 Nucleosomes (B) Nucleosomes coiled to form a solenoid (C) Basic structure of a nucleosome

Functions of histones : Histones in eukaryotic chromosomes serve some functions.

- These either serve as structural elements and help in coiling and packing of long DNA molecules.
- Transcription is possible only by dissolution of histones in response to certain molecular signals.

DNA in nucleosome : Nucleosome is made of core of eight molecules of histones wrapped by double helical DNA with $1\frac{3}{4}$ turns making a repeating unit. Every $1\frac{3}{4}$ turn of DNA have 146 base pairs. When H_1 protein is added the nucleotide number becomes 200. DNA which joins two nucleosome is called linker DNA or spacer DNA.

(3) **Solenoid model :** In this model the nucleosomal bead represents the first degree of coiling of DNA. It is further coiled to form a structure called solenoid (having six nucleosome per turn). It represents the second degree of coiling. The diameter of solenoid is 300*Å*. The solenoid is further coiled to form a supersolenoid of 2000-4000*Å* diameter. This represent the third degree of coiling. The supersolenoid is perhaps the unit fibre or chromonema identified under light microscopy. The solenoid model was given by Fincy and Klug 1976. Klug was awarded by nobel prize in 1982 for his work on chromosome.

(4) **Dangier-String or Radial Loop Model :** (Laemmli, 1977). Each chromosome has one or two interconnected scaffolds made of nonhistone chromosomal proteins. The scaffold bears a large number of lateral loops all over it. Both exit and entry of a lateral loop lie near each other. Each lateral loop is 30 *nm* thick fibre similar to chromatin fibre. It develops through solenoid coiling of nucleosome chain with about six nucleosomes per turn. The loops undergo folding during compaction of chromatin to form chromosome.

(vi) **Heterochromatin and Euchromatin :** Flemming (1880) named the readily stainable material in nuclei as chromatin. It is present both during *interphase* and *cell division* (as the chromosomal material). It consists of about equal parts by weight of *DNA and histones.* There are two classes of chromatin structure, heterochromatin and euchromatin.

(a) Heterochromatin or static chromatin is highly condensed and is usually transcriptionally inactive and found in the centromeres of chromosomes. Heterochromatin is of two types, (i) genetically inactive *constitutive heterochromatin* which is a permanent part of the genome, and (ii) *facultative heterochromatin* which varies in its state in different cell types and development stages.

(b) Euchromatin or dynamic chromatin is relatively extended and open. It at least has the potential of being actively transcribed. It makes up the major part of the genome, and is visible only during mitosis.

Comparison of heterochromatin and euchromatin

Heterochromatin	Euchromatin
Remains condensed throughout interphase (positive heteropycnosis) giving rise to chromocentres.	Shows normal cycle of condensation during cell division and extension during interphase.
Because of the condensed state, it stains more heavily giving rise to banding patterns or chromosomes.	Because it is less condensed, it stains less heavily (normal staining properties). Only slightly basophilic.
Found in condensed regions of the chromosome and in association with tight folding or coiling of the chromosomal fibre.	Found diffuse or less tightly coiled regions. Undergoes typical condensation-decondensation cycle.
May contain highly repetitive (satellite) *DNA* or single copy (unique) *DNA.*	Almost free of repetitive *DNA.* Contains predominantly single copy *DNA.*
Relatively inert metabolically, but does not contain a few genes.	Genetically active : Almost all the genes are located on euchromatin.
DNA is genetically inert, and *does not*	Genetically active, dispersed part of chromatin

transcribe mRNA for protein synthesis in the condensed state.	in interphase nuclei. Its DNA synthesizes *mRNA* for protein synthesis.
Late replication of DNA at the S phase of the cell cycle. Under-replication in polytene chromosomes.	Comparatively early replication of DNA during the early stage of the S phase of cell cycle.
Crossover frequency is less, because condensed regions of the chromosomal fibre cannot come close together for frequent crossover. This may help protect vital genes from the effects of crossover.	Crossover frequency is more because of the decondensed (extended) state of euchromatin.

(vii) **Chromosome banding :** It was the technique demonstrated by **Casperson** (1968) using a fluorescent dye quinacrine mustard for the study of finer chromosomal aberrations. The development of banding techniques has made the identification of individual chromosomes easier. Each chromosome can be identified by its characteristic banding pattern. In X chromosomes the bands are large, each containing ~10^7*bp* of DNA, and could include several hundreds of genes. The different banding techniques are identified by the letters Q, G, C, R and T.

Differentiation of chromosomes by banding

Type of banding	Staining technique	Nature of bands
Q (quinacrine) banding	Chromosomes exposed to quinacrine mustard (acridine dye) which preferentially binds to AT-rich DNA. Other fluorescent dyes used are *DAPI or Hoeschst* 33258.	UV fluorescence reveals fluorescencing *Q* bands which correspond to G-bands. DNA of Q/G bands contains more closely spaced SARs, giving tighter loops (Q loops).
G (Giemsa) banding	Chromosomes treated with alkaline solution and subjected to controlled trypsin digestion before staining with Geimsa, a DNA banding chemical dye. Relatively permanent stain.	Dark bands are called G bands and pale bands are G-negative. G bands are presumed to be *AT-rich*. They are late replicating and contain highly condensed chromatin.
R (reverse) banding	Chromosomes treated with heated saline or restrictase to denature AT-rich DNA and stained with *Giemsa*. GC-specific chromomycin dyes, *e.g*, chromomycin A, olivomycin or mithracin give the same pattern.	R-banding pattern is essentially the reverse of the *G*-banding pattern. R bands are Q negative. They generally replicate in the S-phase and have less condensed chromatin.
T (telomaric) banding	Prolonged heat treatment of chromosomes before staining with	T bands are a subset of R bands which are the most inetnsely staining. They

	Giemsa or combination of dyes and fluorochromes.	are especially concentrated at the telomeres.
C (centromere) banding	Chromosomes pre-treated with sodium hydroxide or barium hydroxide and stained with Giemsa.	Preferred darkening of constitutive centromeric heterochromatin. Rest of the chromosome show Q banding pattern.

(viii) **Human karyotype and idiogram : Tjio** and **Levan** (1956) of Sweden found that human cells have 23 pairs or 46 chromosomes. 22 pairs or 44 chromosomes are autosome and the last or 23rd pairs is that of sex chromosomes, XX in females and XY in males. A set of chromosomes of an individual or species is called a karyotype. In human the 23 pairs of chromosomes in somatic cells form the karyotype. It is possible to identify individual chromosomes on the basis of the following characteristics.

(1) The total length of the chromosomes.

(2) Arm ratio.

(3) The position of the secondary constrictions and nucleolar organizers.

(4) Subdivision of the chromosome into euchromatic and heterochromatic regions.

Homologous pairs of identified chromosomes can be arranged in a series of decreasing lengths. Such an arrangement is called an idiogram. Idiogram not possible in symmetrical karyotype.

(a) **Karyotyping of human chromosomes :** Chromosomes are clearly visible only in rapidly dividing cells. Human chromosomes are studied in blood cells (WBCs), cells in bone marrow, amniotic fluid and cancerous tissues. The WBCs divide when added with phytohaemagglutinin (PHA). The division stops when colchicine is added at metaphase stage. These dividing WBCs are then treated with hypotonic saline solution. Chromosomes are now stained with stains like orcein, Giemsa dye or recent quinacrine dye. When viewed with special miscroscope in ultraviolet light the stain produces fluorescent bands on chromosomes. The chromosomes are then arranged on photographic plate for making diagram and their study. The pictorial representation of a person's chromosomes is called Karyotype.

(b) **Classification of chromosomes :** The human metaphase chromosomes were first of all classified by a conference of cytogeneticists at Denver, Colorado in 1960 and is known as the 23 pairs (46) chromosomes in human has been numbered from 1 to 23 according to their decreasing size. Patau (1960) divided the human chromosome into the following seven groups designated A to G.

Characteristics of the Chromosomes in the Human Karyotype

Group	Size	Shape	Number in set	Number in a cell
A	Large	Metacentric Submetacentric	1-3	6
B	Large	Submetacentric	4-5	4
C	Medium	Submetacentric	6-12 and X	15 male 16 female

D	Medium	Acrocentric	13-15	6
E	Small	Submetacentric	16-18	6
F	Small	Metacentric	19-20	4
G	Smallest	Acrocentric	21-22 and Y	5 male 4 female
				46

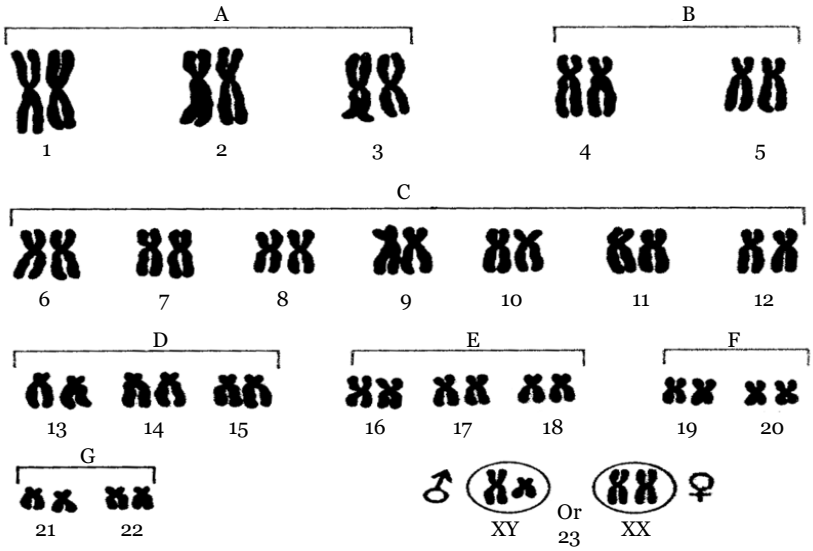


Fig : Human karyotype

- The group A consist of longest metacentric chromosomes.
- The group G consist of the shortest acrocentric chromosomes. These chromosomes have satellites that correspond to nucleolar organizers. In males, group G includes a variable Y chromosome which lacks the satellite.
- Chromsomes of group D also contains satellite.
- The X chromosomes is the member of group C and can be identified by special banding or staning methods.

(ix) **Special types of chromosomes**

(a) **Supernumerary, Accessory or B chromosomes or Satellite chromosomes or Giant lines plasmid :** In some species, chromosomes have been found that are in addition to the normal autosomes and heterosomes. These chromosomes have been called supernumerary chromosomes, accessory chromosomes or B-chromosomes, and differ from normal or A-chromosomes in the following respects.

(1) They are usually smaller than A-chromosomes.

(2) They are frequently heterochromatic and telocentric.

(3) They are genetically unnecessary, and normally do not strongly influence viability and phenotype.

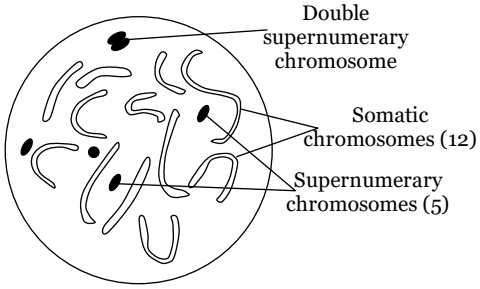


Fig : Diagram of supernumerary chromosomes of *Tradescantia*

(4) Their number may vary in different cells, tissues, individuals and populations.

(5) They are not homologous with any of the A-chromosomes and do not synapses with them.

(6) They are found more commonly in plants than in animals.

Among animals they have been reported mostly in insects and a few species of flatworms. Of the 50 species of insect in which B-chromosomes have been reported, 29 are short horned grasshoppers belonging to the family Acridiae. Usually each nucleus has one or two B-chromosomes. In *Tradescantia edwardsiana* there are 50 B-chromosomes in addition to the 12 somatic A-chromosomes.

(b) **Limited or L-chromosomes :** Limited or L-chromosomes are so called because they are limited to the germ line. They have been found in the family Sciaridae (Diptera: Insecta). The germ line cells in females have 10 chromosomes: three pairs of autosomes, a pair of X-chromosomes and a pair of L-chromosomes. Those of males have 9 chromosomes, there being only one X-chromosome. Somatic cells have 8 chromosomes in females and 7 in males, the L-chromosomes being absent. During the fifth and sixth cleavages, L-chromosomes are eliminated from nuclei destined to form somatic tissue, but retained in germ line cells. L-chromosomes differ from B-chromosomes in that they are constant in all individuals of the species having them. B-chromosomes are found only in some individuals of the species.

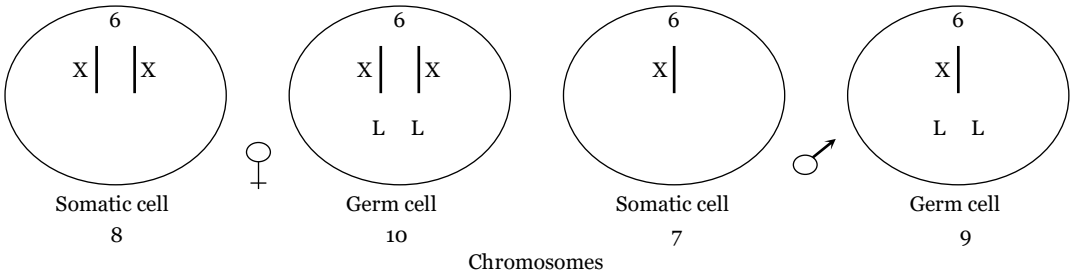


Fig : Schematic representation of the L-chromosomes in the *Sciaridae*

(c) **Minute or m-chromosomes :** Minute or m-chromosomes are so called because of their extremely small size (0.5 micron or less). They have been found in a variety of species of bryophytes, higher plants, insects of the family Coreidae (Heteroptera) and birds. They have been seen mainly during meiosis, and only occasionally during mitosis. Usually one or two chromosomes are seen, but four to five may also be present. In the moss, *Sphagnum* there are 19 large bivalents and 2 *m*- chromosomes.

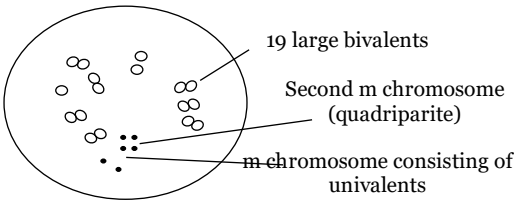


Fig : Minute or m-chromosomes

(d) **S and E-chromosomes :** S and E-chromosomes have been reported in insects in the family Cecidomyiidae (gall insects) and family Chironomidae (Diptera). In the gall insect *Miastor*, both males and females have 48 chromosomes in germ line cells. In somatic cells, however, there are only 12

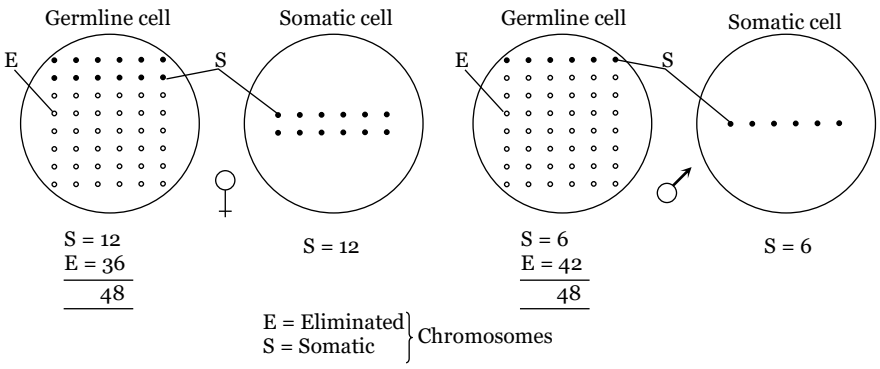


Fig : Schematic representation of the S and E chromosomes of the gall insect *Miastor*

chromosomes in females and 6 in males. Chromosomes which are present in both germ and somatic cells are called *S*-chromosomes. Those which are eliminated from somatic cells but are present in germ cells are called *E*-chromosomes. Thus in females the germ line cells have 12 S-chromosomes and 36 E-chromosomes. In male germ line cells there are 6 S-chromosomes and 42 E-chromosomes. The zygote receives half its S-chromosomes from each parent, while all the E-chromosomes are received from the female parent.

(e) **Polytene chromosome :** Polytene chromosome was described by **Kollar** (1882) and first reported by **Balbiani** (1881) in the salivary gland cells of chironomus larva. They are found in salivary glands of insects (*Drosophila*) and called as salivary gland chromosomes. These are reported in endosperm cells of embryosac by **Malik** and **Singh** (1979). Length of this chromosome may be upto 2000μm. The chromosome is formed by somatic pairs between homologous chromosomes and repeated replication or endomitosis of chromonemata. These are attached to chromocentre. It has pericentromeric heterochromatin. Polytene chromosomes show a large number of various sized intensity bands when stained. The lighter area between dark bands are called interbands. They have puffs bearing *Balbiani rings*. Balbani rings produce a number of *m*-RNA, which may remain stored temporarily in the puffs, are temporary structures. These are also occur in Malpighian tubules, rectum, gut, foot pads, fat bodies, ovarian nurse cells etc.

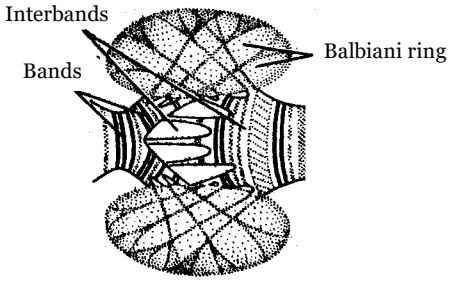


Fig : Polytene chromosome showing balbiani ring

(f) **Lampbrush chromosomes :** They are very much elongated special type of synapsed or diplotene chromosome bivalents already undergone crossing over and first observed by **Flemming** (1882). The structure of lampbrush chromosome was described by **Ruckert** (1892). The lampbrush chromosomes occur at the diplotene stage of meiotic prophase in the primary oocytes of all animal species, both vertebrates and invertebrates as in sagitta (chaetognatha), sepia (mollusca), Echinaster (Echinodermata) and in several species of insect, sharks, amphibians, reptiles, birds and mammals. Lampbrush chromosomes are also found in spermatocytes of several species, giant nucleus of acetabularia and even in plants. In urodele oocyte the length of lampbrush chromosome is upto 5900μm. These are found in pairs consisting of homologous chromosomes jointed at chiasmata (meiotic prophase-I). The chromosome has double main axis due to two elongated chromatids. Each chromosome has rows of large number of chromatid giving out lateral loops, which are uncoiled parts of chromomere with one-many transcriptional units and are involved in rapid transcription of *m*RNA meant for synthesis of yolk and other substances required for growth and development of meiocytes. Some *m*RNA produced by lampbrush chromosome is also stored as informosomes *i.e.*, *m*RNA coated by protein for producing biochemicals during the early development of embryo. Length of loop may vary between 5-100μm.

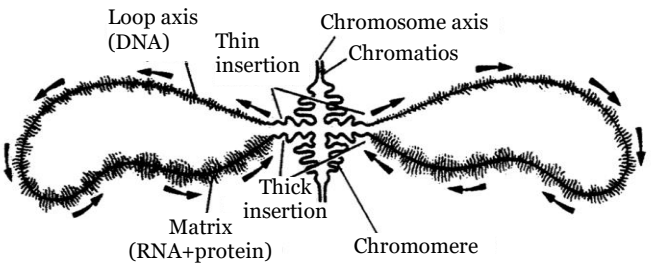


Fig : A part ot main axis with a pair of lateral loops of a lampbrush chromosome showing synthesis of RNA

Important Tips

- **Heteropyknosis :** Darkly staining property of chromatin.
- **Outdet (1975) :** Coined the term nucleosome.
- H_1, H_2A and H_2B proteins are lysine rich (H_1 is very lysine rich) while H_3 and H_4 are arginine rich polypeptide chains.
- Satellite is also called trabant.
- Chromosomes are stained by acetocarmine or acid fushsin.
- Morgan is called father of experimental genetics.
- Bateson is called father of modern genetics.
- Netil stevens (1902) discovered Y chromosomes.
- One gene one enzyme theory was given by Beadle and Tautum.
- Muton is a unit of mutation.
- Strasburger was first of all described chromosome in nucleus.
- Sum of genes in a population is called gene pool.
- One pair of genes can completely mask the expression of another pair of gene is called epistasis.
- Haploid set of chromosome is called genome.
- The frequency of an allele in an isolated population is due to genetic drift.
- Chromosomes were first seen by Hofmeister.

14.2 Multiple allelism

(i) **Mode of origin :** Genes having only two distinct alleles. If mutation occurs in the same gene but in different directions in different individuals, the population as a whole will have many different alleles of that gene. Each allele may produce a different phenotype, and various combinations of alleles produce several genotypes and phenotypes in the population.

(ii) **Characteristics**

(a) There are more than two alleles of the same genes.

(b) All multiple alleles occupy the corresponding loci in the homologous chromosomes.

(c) A chromosome or a gamete has only one allele of the group.

(d) Any one individual contains only two of the different alleles of a gene, one on each chromosome of the homologous pair carrying that gene.

(e) Multiple alleles express different alternative of a single trait.

(f) Different alleles may show codominance, dominance-recessive behaviour or incomplete dominance among themselves.

(g) Multiple alleles confirm to the Mendelian pattern of inheritance.

(iii) **Definition :** More than two alternative forms (alleles) of a gene in a population occupying the same locus on a chromosome or its homologue are known as multiple alleles.

(iv) **Examples of multiple allelism :** A well known example of a trait determined by multiple alleles is the blood groups in man and skin colour.

Blood groups in man

(a) **Blood proteins :** According to **Karl landsteiner** (1900) a Nobel prize winner, blood contains two types of proteinous substances due to which agglutinations occurs.

(1) **Agglutinogen or antigen :** It is a protein found on the cell membrane of RBC's.

(2) **Agglutinin or antibody :** This the other proteinous substance, found in the plasma of the blood.

Whenever the blood of a person receives the foreign proteins (antigen) his blood plasma starts forming the antibodies in order to neutralize the foreign antigens.

(b) **Agglutinations :** Two types of antigens are found on the surface of red blood corpuscles of man, antigen A and B. To react against these antigens two types of antibodies are found in the blood plasma which are accordingly known as antibody – *anti-A or a* and *anti-B or b.* Agglutination takes place only when *antigen A* and *antibody a* occur together or *antigen B* and *antibody b* are present in the blood. Under such condition *antibody a* reacts with *antigen A* and makes it highly sticky. Similarly *antigen B* in presence of *antibody b* become highly sticky with the result RBC's containing these antigens clump to form a bunch causing blockage of the capillaries. Agglutination in blood is therefore antigen-antibody reaction.

(c) **Types of blood groups**

(1) **ABO blood group : Landsteiner** divided human population into four groups based on the presence of antigens found in their red blood corpuscles. Each group represented a blood group. Thus there are four types of blood groups viz. A, B, AB and O. He observed that there was a reciprocal relationship between antigen and antibody according to which a person has antibodies for those antigens which he does not possess. For example a person of blood group B does not possess *antigen A* but his blood plasma has *antibody 'a'* due to which agglutination with the blood of a person with blood group A occurs. Similarly persons with blood group AB possess both the *antigens A* and *B* but their blood plasma does not possess any of the antibodies. In the same way person having blood group A does not possess *antigen B* but *antibody 'b'* is found in his blood plasma. Persons with blood group O possess none of the antigens and that is why their blood possesses both the *antibodies 'a'* and *'b'*.

Blood groups of man with antigen and antibodies

Antigen	Antibody	Type of blood group	% in society
(1) A	*Anti-B* or '*b*'	A	23.5
(2) B	*Anti-A* or '*a*'	B	34.5
(3) A, B	Absent	AB	7.5
(4) None	'a' and 'b'	O	34.5

(2) **M, N blood group : K. Landsteiner** and **A.S. Wiener** discovered that antigen M,N or both MN are also found on the surface of red blood corpuscles of human beings. No antibodies are however formed in the blood plasma for these antigens. If however, these antigens are injected into rabbit's blood, they produced such antibodies which are not found in human beings. Inheritance of such kind of blood groups is also brought about like that of A, B and AB.

In this way when blood with M group is injected in rabbit it will produce antibodies in the blood serum which will bring about agglutination with blood group M and MN but not with blood of N group. In the same way on injecting blood of N group into the rabbit it will bring about agglutination with blood group N and MN and not with blood having blood group M.

(d) **Blood transfusion :** Blood transfusion is best done in the persons of same blood group. At the same time it is possible to know in which different blood groups the blood transfusion can be made possible.

Persons with blood group AB are called **universal recipients** because both antigens A and B are found in their blood and the two antibodies 'a' and 'b' are absent. Therefore, such persons can receive blood of all the blood groups. In the same way persons who have blood group O^- are **universal donors** as they lack both the antigens and Rh^- person can donate to Rh^+ person as well as Rh^- person but Rh^+ person cannot donate blood to Rh^- person. But at the same time such persons can not be given the blood of any other blood group except blood group O because their blood possesses both the antibodies 'a' and 'b'. Persons belonging to blood group A and B contain only one antigen and one antibody against it, in their blood. Such persons can therefore receive blood either of the blood group of their own or the blood group O.

Possibilities of blood transfusion

Blood group	**Can accept from**	**Can donate to**	**Agglutination**				**Specific mention**
			A	B	AB	O	
(1) A	A, O	A, AB	No	Yes	No	Yes	
(2) B	B, O	B, AB	Yes	No	No	Yes	
(3) AB	A, B, AB, O	AB only	Yes	Yes	No	Yes	Universal recipient
(4) O	O only	A, B, AB, O	No	No	No	No	Universal donor

(e) **Blood bank :** A place where blood of different blood groups is safely stored in bottles for emergency use, is called blood bank. Blood after proper testing is stored in a sealed bottle at a definite temperature (4°-6°c) to be preserved for a definite time period.

Artificial anticoagulants are used to prevent blood clotting in the blood banks. These anticoagulants are added to the blood preserved in bottle. Such anticoagulants include sodium citrate, double oxalates (sodium and ammonium), dicumarol and EDTA (ethylene diamine tetra acetic acid). The whole blood in this way can be stored for a maximum period of 21 days.

(f) **Inheritance of blood groups :** Blood groups in human are **inheritable trait** and are inherited from parents to offsprings on the basis of Mendel's Laws. Blood group inheritance depends on genes received from parents. Genes controlling blood group in man are three instead of two and are called multiple alleles. All these three genes or alleles are located on the same locus on homologous chromosomes. A person can have only two of these three genes at a time which may be either similar or dissimilar in nature. These genes control the production of blood group/antigens in the offspring. The gene which produces antigen A is denoted by I^a, gene for antigen B by I^b and the gene for the absence of both antigens by I^o. it is customary to use the letter I (Isohaemagglutinogen) as a basic

symbol for the gene at a locus. Based on this, six genotypes are possible for four blood groups in human population.

Genotype of blood groups in man.

	Genotype	Nature of gene	Type of blood group
(1)	$I^a I^a$	Homozygous Dominant	A
(2)	$I^a I^o$	Heterozygous	A
(3)	$I^b I^b$	Homozygous Dominant	B
(4)	$I^b I^o$	Heterozygous	B
(5)	$I^a I^b$	Codominant	AB
(6)	$I^o I^o$	Homozygous Recessive	O

The alleles I^a and I^b of human blood group are said to be codominant because both are expressed in the phenotype AB. Each produces its antigen and neither checks the expression of the other. There is codominance as well as dominant recessive inheritance in the case of the alleles for the blood groups in human beings. The alleles I^a and I^b are codominant and are dominant over the allele I^o ($I^a = I^b > I^o$). The human blood groups illustrate both multiple allelism and codominance. This blood group are inherited in the simple Mendelian fashion. Thus offsprings with all four kinds of blood groups are possible. If the parents are heterozygous for blood groups A and B which is shown below.

Cross between parents heterozygous for blood group A and B

		Male (Heterozygous for blood group A)	
	Gametes	I^a	I^o
Female (Heterozygous for blood group B) I^b I^o		I^a	I^o
	I^b	$I^a I^b$	$I^b I^o$
		Group AB	Group B
	I^o	$I^a I^o$	$I^o I^o$
		Group A	Group O

If we know the blood groups of a couple the blood groups of their children can easily be predicted as shown below.

Possible blood groups of children for known blood groups of parents.

	Blood groups of parents (known)	Genotype of parents (known)	Blood groups of children	
			Possible	**Not possible**
(1)	O and O	$I^o I^o \times I^o I^o$	O	A, B, AB
(2)	O and A	$I^o I^o \times I^a I^o$	O, A	B, AB
(3)	A and A	$I^a I^o \times I^a I^o$	O, A	B, AB
(4)	O and B	$I^o I^o \times I^b I^o$	O, B	A, AB
(5)	B and B	$I^b I^o \times I^b I^o$	O, B	A, AB
(6)	A and B	$I^a I^a \times I^b I^b$ $I^a I^a \times I^b I^o$ $I^a I^o \times I^b I^o$	O, A, B, AB	None
(7)	O and AB	$I^o I^o \times I^a I^b$	A, B	O, AB
(8)	A and AB	$I^a I^o \times I^a I^b$	A, B, AB	O
(9)	B and AB	$I^b I^o \times I^a I^b$	A, B, AB	O
(10)	AB and AB	$I^a I^b \times I^a I^b$	A, B, AB	O

(g) **Significance of blood groups :** The study of blood groups is important in settling the medico-legal cases of disputed parentage because with the help of blood group of a child it can be decided as to who can be his or her genuine father, if the blood group of mother is known. It means that blood groups of the mother and a child being known, the possibilities of blood group in the father can be worked out or if blood group of child and that of father is known then that of mother can be known with the help of the table given below. Blood groups can also save an innocent from being hanged in the case of murder and can help in hanging the real culprit.

Possibilities of blood groups of other parent on the basis of blood group of child and one parent being known.

S.No.	Blood group of child (known)	Genotype of child (known)	Blood group of father or mother (known)	Blood group of other parent	
				Possible	**Not possible**
(1)	O	$I^o I^o$	O A B	A, B O, B O, A	AB
(2)	A	$I^a I^o$, $I^a I^a$	O, B	A, AB	O, B
(3)	B	$I^b I^o$, $I^b I^b$	O, A A	B, AB B, AB	O, A O, A
(4)	AB	$I^a I^b$	B AB	A, AB A, B, AB	O, B O

(h) **Rhesus or Rh factor**

(1) **Rh factor : Landsteiner** and **Weiner** (1940) discovered a different type of protein in the blood of Rhesus monkey. They called it Rh antigen or Rh factor after Rhesus monkey. When injected the blood of these monkeys into the blood of guinea pigs they noticed the formation of antibodies against the Rh antigen in the blood of guinea pigs. Formation of Rh antigen is controlled by dominant gene (R) and its absence by recipient gene (r). People having this antigen with genotype (RR or Rr) are called Rh positive (Rh^+) and those whose blood is devoid of it with genotype (rr) are Rh negative (Rh^-). About 85% human beings in Europe and 97% in India are Rh^+.

(2) **Importance of Rh factor :** Generally human blood is devoid of Rh antibodies. But it has been noticed that on transfusion of blood of a Rh^+ person to Rh^- person, the recepient develops Rh antibodies in its blood plasma. If Rh^+ blood is transfused for the second times it causes agglutination and leads to the death of Rh^- person.

(3) **Erythroblastosis foetalis :** This disease is related to the birth of a child related with Rh factor. It causes the death of the foetus within the womb or just after birth. It was studies by **Levine** together with **Landsteiner** and **Wiener**. The father of Rh affected foetus is Rh^+ and the mother is Rh^-. The child inherits the Rh^+ trait from the father. A few Rh^+ red blood corpuscles of foetus in the womb enter in the blood of the mother where they develop Rh antibodies. As mother's blood is Rh^- *i.e.* devoid of Rh antigen, it causes no harm to her. These Rh antibodies alongwith the mother's blood on reaching the foetal circulation cause clamping of foetal RBCs or agglutination reaction. The first child is some how born normal because by that time the number of antibodies in mother's blood remain lesser but they increase with successive pregnancies. Thus the foetus following the first child dies either within the womb or just after its birth. This condition is known as erythroblastosis foetalis. So a marriage between Rh^+ boy and Rh^- girl is considered biologically incompatible.

Type of biological marriage on the basis of Rh factor

Boy	Girl	Type of biological marriage
Rh^+	Rh^+	Compatible marriage
Rh^-	Rh^-	Compatible marriage
Rh^-	Rh^+	Compatible marriage
Rh^+	Rh^-	Incompatible marriage

However, there is no danger if both parents are Rh^- or mother is Rh^+ and father is Rh^-. Rh factor serum has been developed which when given to the Rh^- mother after each child birth saves the next child. This serum contains Rh antibodies which destroy the Rh antigens of foetus before they can initiate formation of Rh antibodies in the mother.

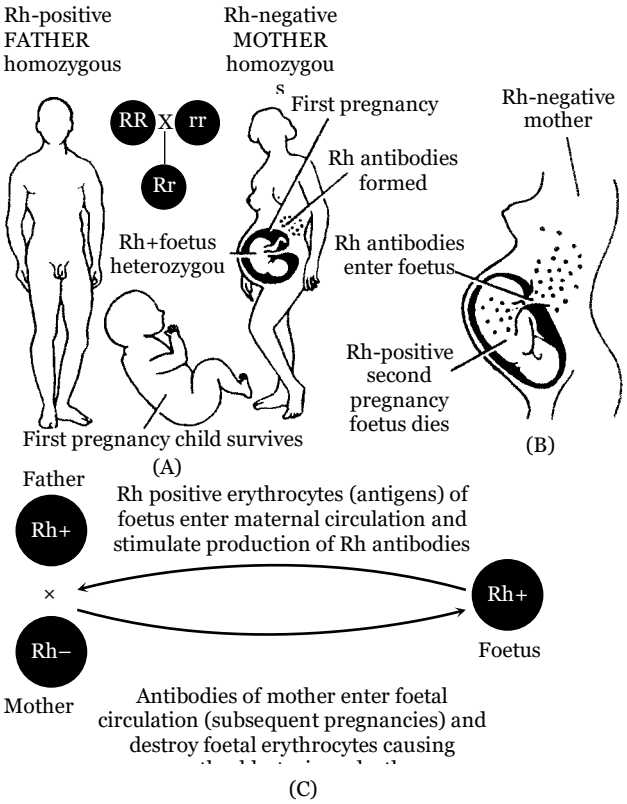


Fig : Foetal death in the womb due to erythroblastosis foetalis

(4) **Rhogam method :** It is a method of preventing erythroblastosis foetalis. In this method the Rh^- mother is given a special blood test after delivery of her Rh^+ child. If foetal Rh^+ cells are present in mother's blood. She is given injections of rhogam. Rhogam is a preparation of anti-Rh antibodies. It is obtained from immunized donors. The rhogam forms a coat around foetal RBCs in mother's blood. As a result no Rh^+ antigens are available to stimulate mother's circulation and no antibodies are formed.

(5) **Inheritance of Rh factor :** Rh factor or Rh antigen is determined by a series of four pair of multiple alleles. They are denoted as R^1, R^2, R^0, R^z, r', r", r^y and r. The alleles denoted by capital letter give rise to Rh^+ condition while those denoted by small letter to Rh^- condition. Rh^+ condition is dominant over Rh^- condition. Thus Rh^+ person may be homozygous (RR) or heterozygous (Rr) while Rh^- persons are always homozygous(rr). Hereditary trait for Rh^- factor is inherited according to Mendelian principle.

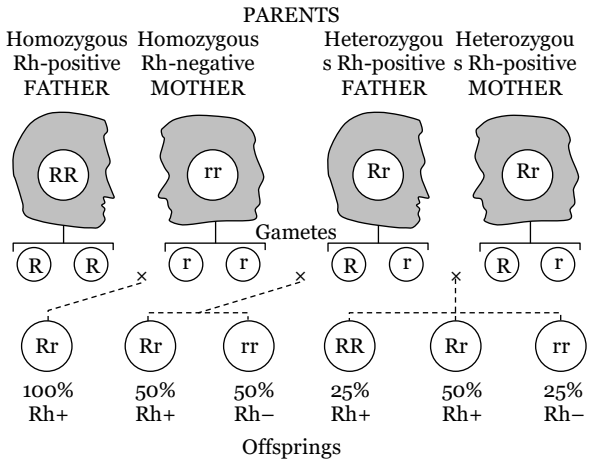


Fig : Inheritance of Rh

Important Tips

- Most common blood groups in India are B and Rh^+.
- Best recipient is AB^+.
- The AB blood group was discovered by two Landsteiner's students Von Decastello and Sturli (1902).
- Inheritance of A, B, AB and O blood types in man was discovered by Bernstein in 1925.
- A very rare h/h individual are like blood type O individuals. They are said to have the Bombay blood type.
- Rh factor was first of all reported in RBCs of Macaca rhesus (rhesus monkey) by Landsteiner and Wiener in 1940.
- Immunological incompatibility between mother and foetus sometimes results in a condition called haemolytic disease of the new born (HDN).
- HDN was earlier known as erythroblastosis foetalis.

14.3 Genetic variations

The idea of mutation first originated from the observations of a Dutch botanist **Hugo de Vries** (1880) on variations in plants of *Oenothera lamarckiana*. The mutation can be defined as sudden, stable discontinuous and inheritable variations which appear in organism due to permanent change in their genotype. Mutation is mainly of two types :

- **Spontaneous mutations :** Mutation have been occurring in nature without a known cause is called spontaneous mutation.

- **Induced mutation :** When numerous physical and chemical agents are used to increase the frequency of mutations, they are called induced mutations.

(i) **Gene mutations :** Gene or point mutations are stable changes in genes *i.e.* DNA chain. Many times a change in a gene or nucleotide pair does not produce detectable mutation. Thus the point or gene mutation mean the process by which new alleles of a gene are produced. The gene mutation are of following types

(a) **Tautomerism :** The changed pairing qualities of the bases (pairing of purine with purine and pyrimidine with pyrimidine) are due to phenomenon called tautomerism.

Tautomeres are the alternate forms of bases and are produced by rearrangements of electrons and proton in the molecules.

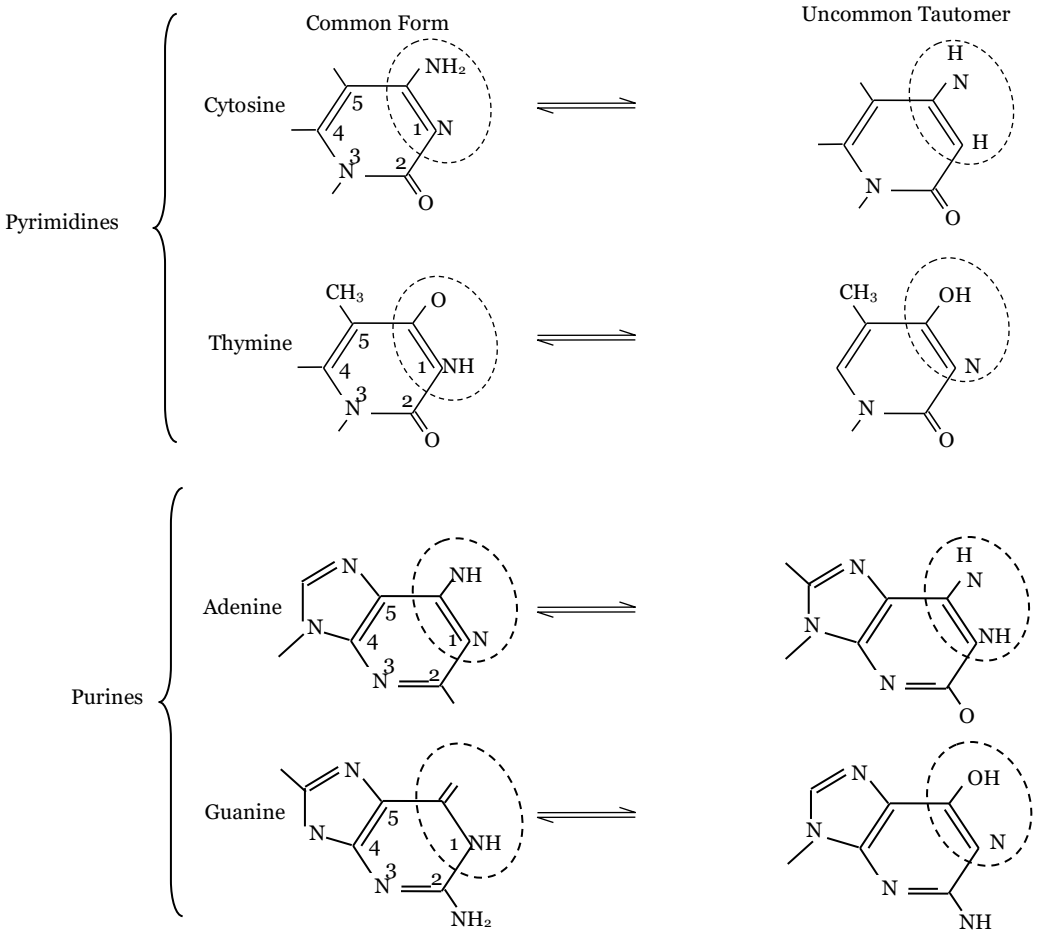
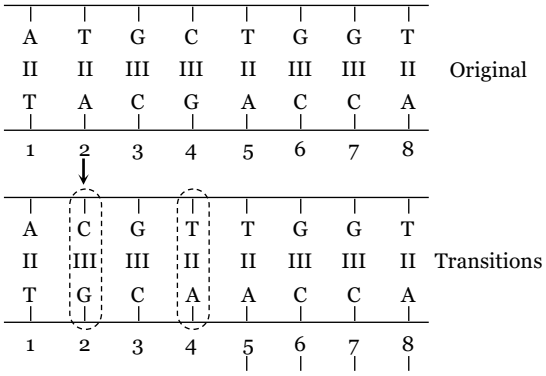


Fig : Few tautomers

Tautomerism is caused by certain chemical mutagens. In the next replication purines pair with pyrimidines and the base pair is altered at a particular locus. The uncommon forms are unstable and at the next replication, cycle revert back to their normal forms.

(b) **Substitutions (Replacements) :** These are gene mutations where one or more nitrogenous base pair are changed with others. It may be further of three sub types.

(1) **Transition :** In transition, a purine (adenine or guanine) or a pyrimidine (cytosine or thymine or uracil) in triplet code of DNA or *m*RNA is replaced by its type *i.e.* a purine replaces purine and pyrimidine replaces pyrimidine.



(2) **Transversion :** Transversion are substitution gene mutation in which a purine (adenine or guanine) is replaced by pyrimidine (thymine or cytosine) or vice versa.

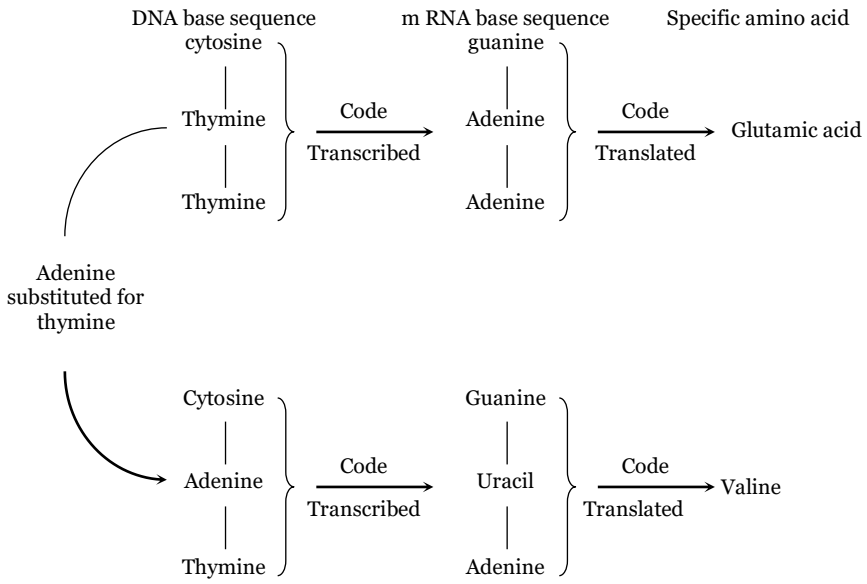


Fig : Transversion

(3) **Frame shift mutations :** In this type of mutations addition or deletion of single nitrogenous base takes place. None of the codon remains in the same original position and the reading of genetic code is shifted laterally either in the forward or backward direction.

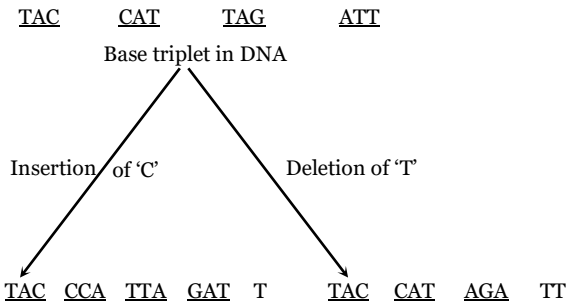


Fig : Frame shift

(ii) **Chromosomal mutation or aberrations :** A gene mutation normally alters the information conveyed by a gene, it alters the message. On the other hand, chromosomal mutation only alters the number or position of existing genes. They may involve a modification in the morphology of chromosome or a change in number of chromosomes.

Morphological aberrations of chromosomes :

(a) **Deletion or deficiency :** Sometimes a segment of chromosome break off and get lost. Deficiency generally proves lethal or semilethal.

- **Deficiency :** If a terminal segment of a chromosome is lost, it is called deficiency.
- **Deletion :** If intercalary segment is lost it is termed deletion.

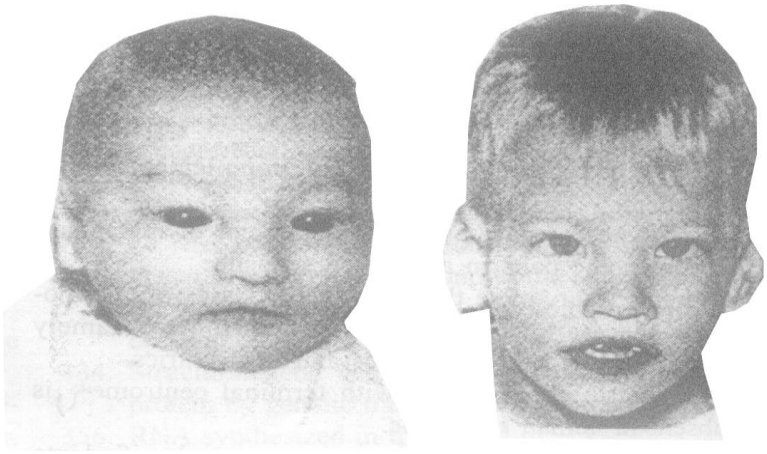
Deletions in human beings

- A missing chromosome segment is referred to either as deletion or deficiency.
- In a dilpoid organism, the deletion of a chromosome segment makes the part of genome hypoploid.

- Deletion may be associated with phenotypic effect, especially if the it is large.

(1) **Cri-du-chat Syndrome**

- A classical example of deletion is the Cri-du-chat syndrome (from the french words for "cry of the cat") in human beings discovered by **Lejeune** in 1963.
- This condition is caused by a conspicuous deletion in the short arm of one of the 5^{th} autosomes.



(A) (B)

Fig : Cat cry syndrome (A) An infant (B) An older child

- These individuals are severely impaired, mentally as well as physically; their plaintive catlike crying gives the syndrome its name.

(2) **Wolf-Hirschhorn's Syndrome**

- Wolf-Hirschhorn's syndrome is another well characterized deletion syndrome in human beings caused by a deletion of short arm of chromosome 4 (4p-).
- The phenotypic effect includes wide-spaced eyes and cleft lip.

(b) **Duplication :** In this mutation deleted chromosomal segment is attached to its normal homologous chromosome. Here a gene or many genes are repeated twice or more times in the same chromosome.

(c) **Inversion :** A piece of chromosome is removed and rejoined in reverse order. For example a chromosome with the gene order A, B, C, D, E, F, G, H is broken between B,C,D and between f and g and the centre portion turned through 180°, the resulting gene order is A, D, C, B, E, F, G, H it is of two types.

(1) **Pericentric inversion :** The centromere lies within the inverse order.

(2) **Paracentric inversion :** The centromere lies outside the inverted segment.

(d) **Translocation :** Mutual exchange (reciprocal) of the chromosome segments between non homologous chromosome. An exchange of parts between two non homologous chromosomes is called reciprocal translocation. In simple translocation a segment of one chromosome breaks and is transferred to another non-homologous chromosome.

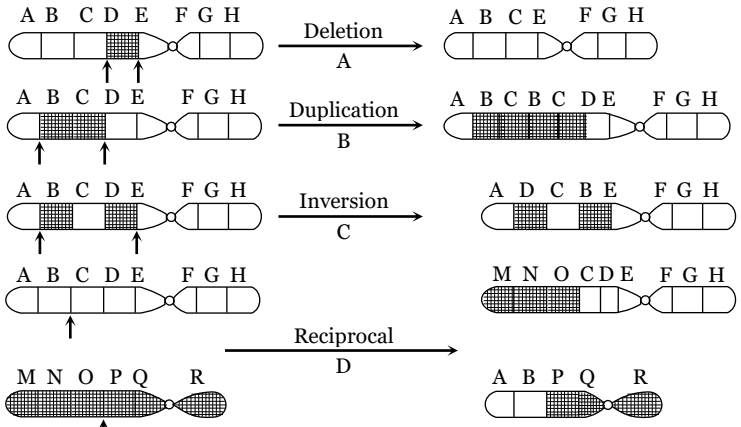


Fig : Types of chromosomal mutation

Translocations in human beings

- Certain types of cancer are associated with chromosome rearrangements.
- Two examples of tumours associated with consistent chromosome translocations are Chronic Myelogenous Leukaemia (CML) and burkitt's lymphoma.

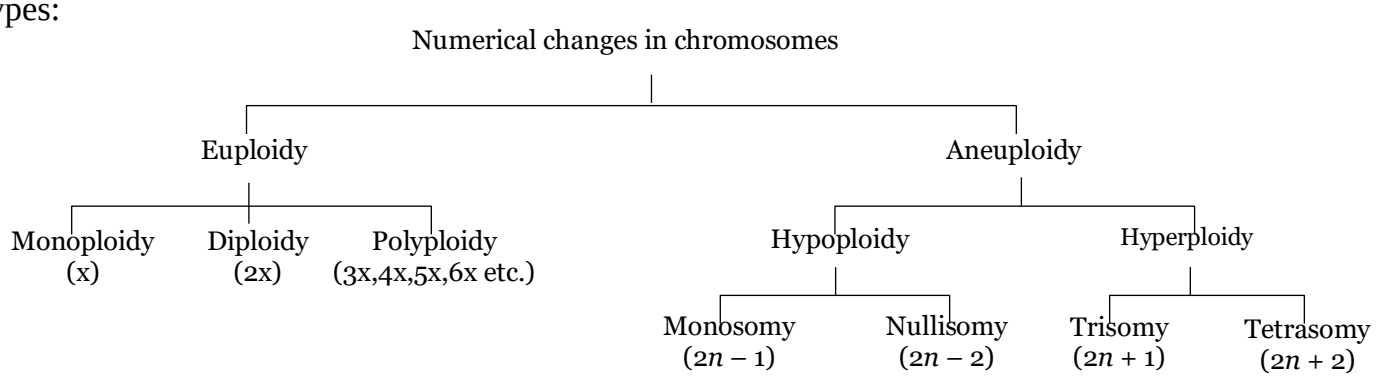
(1) **Chronic Myelogenous Leukaemia (CML)**

- Chronic myelogenous leukaemia in human beings is a fatal cancer involving uncontrolled replication of myeloblasts (stem cells of white blood cells).
- Ninety percent of CML is associated with an aberration of chromosome 22.
- This abnormal chromosome was originally discovered in the city of Philadelphia in 1959 and thus is called the 'Philadelphia chromosome'.
- Initially it was though to have a simple deletion in its long arm, however, subsequent analysis using molecular techniques has shown that the Philadelphia chromosome is actually the result of a reciprocal translocation between chromosomes 9 and 22.
- In the Philadelphia translocation, the tip of the long arm of chromosome 9 has been joined to the body of chromosome 22 and the distal portion of the long arm of chromosome 22 has been joined to the body of chromosome 9.
- CML is characterized by an excess of granular leucocytes in the blood.
- With the increase in the number of leucocytes, there is a reduction in the number of RBCs resulting in severe anaemia.

(2) **Burkitt's Lymphoma**

- Burkitt's lymphoma, a particularly common disease in Africa, is another example of a white blood cell cancer associated with reciprocal translocations.
- These translocations invariably involve chromosome 8 and one of the three chromosomes (2, 14 and 22) that carry genes encoding the polypeptides that form immunoglobulins or antibodies.
- Translocations involving chromosomes 8 and 14 are the most common.

Numerical aberrations of chromosomes : Each species has a characteristic number of chromosome. Variations or numerical changes in chromosomes (Heteroploidy) can be mainly of two types:



Different kinds of numerical changes in chromosomes
(X = basic chromosome number, 2*n* = somatic chromosome number)

(a) **Euploidy :** The somatic chromosome number in euploids is the exact multiple of basic haploid number. In euploidy an organism acquires an additional set of chromosomes over and above the diploid complement.

(1) **Monoploidy or haploidy :** Monoploids possess only one set or single basic set of chromosomes. Haploids on the other hand have half the somatic chromosome number. In diploid organisms monoploids and haploids are identical while in a tetra-or hexaploid with 4*n* or 6*n* chromosomes the haploids will possess 2*n* or 3*n* chromosome whereas its monoploid will possess only one set (*n*) of chromosome.

(2) **Diploidy :** The common chromosome number in the somatic cells of plants and animals.

(3) **Polyploidy :** Organism with more than two sets of chromosomes are known as polyploids. It may be triploid with three sets of chromosomes (3*n*) or tetraploid with four sets of chromosome (4*n*) and so on.

(b) **Aneuploidy :** Aneuploidy is the term applied for the chromosomal mutations involving only a part of a set, *i.e.*, loss (hypoploidy) or addition (hyperploidy) of one or more chromosomes. Aneuploidy may result from non disjunction of chromosome during cell division.

(1) **Monosomy :** Diploid organism that are missing one chromosome of a single pair with genomic formula 2*n* – 1. Monosomics can form two kind of gametes, (*n*) and (*n* –1).

(2) **Nullisomy :** An organism that has lost a chromsome pair is nullisomic. The result is usually lethal to diploids (2*n* – 2).

(3) **Trisomy :** Diploids which have extra chromosome represented by the chromosomal formula 2*n* + 1. One of the pairs of chromosomes has an extra member, so that a trivalent may be formed during meiotic prophase.

(4) **Tetrasomy :** In tetrasomic individual particular chromosome of the haploid set is represented four times in a diploid chromosomal complement. The general chromosomal formula for tetrasomics is 2*n* + 2 rather than 2*n* + 1+ 1. The formula 2*n* + 1 + 1 represents a double trisomic.

(iii) **Types of aneuploidy :** Aneuploidy may be of following types on the basis of chromosomes involved in non disjunction.

(a) **Aneuploidy involving non-disjunction in sex chromosomes :** This kind of aneuploidy is brought about due to non-disjunction in sex chromosomes. It may lead to following types of syndromes :

(1) **Turner's syndrome :** Such persons are monosomic for sex chromosomes *i.e.* possess only one X and no Y chromosome (XO). In other words they have chromosome number $2n - 1 = 45$. They are phenotypic females but are sterile because they have under developed reproductive organs. They are dwarf about 4 feet 10 inches and are flat chested with wide spread nipples of mammary glands which never enlarge like those in normal woman. They develop as normal female in childhood but at adolescence their ovaries remain under developed. They lack female hormone estrogen. About one out of every 5,000 female births results in Turner's syndrome.

(2) **Klinefelter's syndrome :** Since 1942, this abnormality of sex is known to geneticists and physicians. It occurs due to Trisomy of sex chromosomes which results in (XXY) sex chromosomes. Total chromosomes in such persons are $2n + 1 = 47$ in place of 46. **Klinefelter** (1942) found that testes in such male remain under developed in adulthood. They develop secondary sex characters of female like large breasts and loss of facial hair. Characters of male develop due to Y chromosome and those like female due to XX chromosomes. About one male child out of every 5,000 born, develops **Klinefelter's syndrome**.

Such children are born as a result of fertilization of abnormal eggs (XX) by normal sperms with (X) or (Y) chromosomes or by fertilization of normal eggs with (X) chromosomes by abnormal sperms with (XY) chromosome. They are sterile males mentally retarded and are **eunuchs**.

(3) **Super females and metasuper females :** Presence of extra (X) chromosomes in females shows such condition leading to (XXX, XXXX, XXXXX), having total 47, 48 or 49 chromosomes in each cell. Females with this type of aneuploidy show abnormal sexual development and mental retardation. Severeness of abnormality increases with the increase in number of (X) chromosomes.

(iv) **Criminal's syndrome (super males) :** Presence of an extra (Y) chromosome in males causes such a condition (XYY) resulting in individuals with $2n + 1 = 47$ chromosomes. They have unusual height, mentally retarded and criminal bent of mind since birth. Their genital organs are under developed. Their frequency is one in every 300 males.

(b) **Aneuploidy involving non-disjunction in autosomes :** This type of aneuploidy occurs due to trisomy of autosomes. In any particular autosomal pair, having 3 instead of normal 2 chromosomes. Such persons may be males $45 + XY = 47(2n + 1)$ or females $45 + XX = 47(2n + 1)$. On the basis of the number of the autosome pair affected by trisomy, they can be of following types.

(1) **Down's syndrome :** This autosomal abnormality is also known as Mongolian idiocy or mongolism. In Langdon Down of England (1866) studied the Mongolian idiocy and described the trisomic condition of their chromosomes. Down's syndrome, a very common congenital abnormality arises due to the failure of separation of 21^{st} pair of autosomes during meiosis. Thus an egg is produced with 24 chromosomes instead of 23. A Down's syndrome has 3 autosomes in 21st pair instead of 2. Total number of chromosomes in this case is $2n + 1\ (21^{st}) = 47$.

The affected children have a very broad fore head, short neck, flat palms without crease, stubby fingers, permanently open mouth, projecting lower jaw and a long thick extending tongue. They have low intelligence and are short heighted. They have defective heart and other organs. They are born to mothers aged 40 year and above during first pregnancy. They may survive upto 20 years under medical care.

They are called mongolian idiots because of their round, dull face and upper eyelids stretched downwards similar to mongolian race.

(2) **Edward's syndrome :** This autosomal abnormality occurs due to trisomy of eighteenth pair of autosomes in which the number of chromosomes are $2n + 1 = 47$. The child with this defect survives only about 6 months. Such children have defective nervous system, malformed ears and a receding chin.

(3) **Patau's syndrome :** This is trisomy of thirteenth pair of autosomal chromosome. This trisomic condition involves numerous malformations such as harelip, clefted palate and cerebral, ocular and cardiovascular defects. Such children usually survive for about 3 months only.

(iv) **Mutagens :** Any substance or agent inducing mutation is called a mutagen. The mutagens may be broadly grouped into two classes.

(a) **Physical mutagens :** It comprise mainly radiations. Radiation has been used to induce mutations for the first time by **H.J. Muller** (1927) on animals and **L.J. Stadler** (1928) on plants. Radiation that can produce mutation is known as effective radiations which are as follows.

(1) **Ionizing (Particulate) :** α-particles, β-rays, protons and neutrons.

(2) **Ionizing (non particulate) :** X-rays, r-rays and cosmic rays.

(3) **Nonionizing :** Ultraviolet rays

(b) **Chemical mutagen :** A large number of chemicals react with the four nucleotides and modify their base-pairing capabilities. These are as follows.

(1) **Base analogues :** 5-bromodeoxyuridine (Brdu), 2-amino purine.

(2) **Chemicals modifying base-pairing**

- Hydroxylamine
- Nitrous acid
- **Alkylating agent :** Nitrogen mustard, ethyl methane sulfonate (EMS), methyl methane sulfonate (MMS) and N-methyl-N'-nitro-nitroso-guanidine (NTG).

(3) **Intercalating agents :** Proflavin and acridine orange

(v) **Genetic diseases in man :** There are many diseases in man due to gene mutations. It is either dominant or recessive. The mutated person may become incapable to produce specified enzyme, so result in inborn errors of metabolism.

(a) **Chondrodystrophic dwarfism**

(1) Chondrodystrophic dwarfism is a dominant autosomal mutation, most people are homozygous for recessive allele (c/c).

(2) The presence of one dominant C results in the premature closure of the growth areas of long bones of arms and legs, resulting in shortened and bowed arms and legs.

(b) **Huntington disease**

(1) Huntington disease is caused by a dominant gene on chromosome 4.

(2) The mutated gene causes abnormality by producing a substance that interferes with normal metabolism in the brain that leads to progressive degeneration of brain cells.

(3) The death comes ten to fifteen years after the onset of symptoms.

(c) **Neurofibromatosis**

(1) Also called "von Recklinghausen disease" caused by a dominant gene on chromosome 17.

(2) The affected individual may have ten spots on the skin which later may increase in size and number.

(3) Small benign tumours called neurofibromas may occur under the skin or in various organs.

(d) **Tay-Sachs disease**

(1) Tay-Sachs disease results from the lack of the dominant gene on chromosome 15 for the production of hexosaminidase and subsequent storage of its substrate, a fatty substance known as glycosphingolipid, in lysosomes.

(2) The patient suffers from defective vision, muscular weakness and gradual loss of all mental and physical control, death occurs by the age of three or four years.

(e) **Cystic fibrosis**

(1) The most common lethal genetic disease due to a recessive mutation on the chromosome 7.

(2) The body produces abnormal glycoprotein which interferes with salt metabolism.

(3) The mucus secreted by body becomes abnormally viscid and blocks passages in the lungs, liver and pancreas.

(f) **Alzheimer's disease**

(1) Alzheimer's disease, named after the German neurologist Alzheimer, is a degenerative brain disease characterized by memory loss, confusion, restlessness, speech disturbances, erosion of personality, judgement, and inability to perform the functions of daily living.

(2) Alzheimer's disease, a form of dementia, occurs in karyotypically normal individuals.

(3) About 5 percent of karyotypically normal individuals over age of 65 develop Alzheimer disease, and nearly 25 percent of those over age 80 do so.

(4) The brain of Alzheimer's patients show a marked loss of neurons.

(5) These patients also show an accumulation of senile plaques, which are thickened nerve cell processes (axons and dendrites) surrounding a deposit of particular type of polypeptide called amyloid β protein.

(6) In the brain of normal persons, amyloid β protein is produced and processed in a number of ways from a large number of amyloid precursor protein.

(7) The occurrence of Alzheimer's disease in people with Down's syndrome suggests that a gene or genes on chromosome 21 is involved.

(8) Genetic mapping has demonstrated that the gene for amyloid β protein is located on chromosome 21; this gene encodes an Amyloid Precursor Protein (APP) that is enzymatically cleaved to produce amyloid β proteins.

(9) According to **Bush** (2003) Alzheimer's disease is caused by a copper and zinc build up in the brain.

(g) **Marfan's syndrome**

(1) Marfan's syndrome is due to dominant mutation resulting in the production of abnormal form of connective tissues and characteristic extreme looseness of joints.

(2) The long bones of body grow longer; fingers are very long called 'spider fingers' or arachnodactyly.

(3) The lenses in eyes become displaced.

(h) **Albinism**

(1) Albinism is an autosomal recessive mutation.

(2) An albino cannot synthesize melanin which provides black colouration to skin and hair.

(3) Albinism is due to tyrosinase deficiency.

(4) The enzyme tyrosinase normally converts the amino acid tyrosine to melanin through an intermediate product **DOPA** (dihydro phenyl alanine).

(i) **Sickle-cell disease**

(1) Sickle-cell disease is a genetic disease reported from negroes due to a molecular mutation of gene Hb^A on chromosome 11 which produces the β chain of adult haemoglobin.

(2) The mutated gene Hb^S produces sickle-cell haemoglobin.

(3) The sixth amino acid in β chain of normal haemoglobin is glutamic acid.

(4) In sickle-cell haemoglobin this amino acid is replaced by valine.

(5) The children homozygous (Hb^SHb^S) produce rigid chains.

(6) When oxygen level of the blood drops below certain level, RBCs undergo sickling.

(7) Such cells do not transport oxygen efficiently; they are removed by spleen causing severe anaemia.

(8) Individuals with the Hb^AHb^A genotype are normal, those with the Hb^SHb^S genotype have sickle-cell disease, and those with the Hb^AHb^S genotypes have the sickle-cell trait.

(9) Two individuals with sickle-cell trait can produce children with all three phenotypes.

(10) Individuals of sickle-cell trait are immune to malaria.

(j) **Thalassemia**

(1) Thalassemia is a human anaemia due to an autosomal mutant gene and when this gene is present in double dose, the disease is severe thalassemia major with death occurring in childhood.

(2) Heterozygous persons show a milder disease, thalassemia minor or also called **Cooley's anaemia**.

(3) The persons suffering from thalassemia major are unable to produce β chain.

(4) Their haemoglobin contains δ chains like that of foetus which is unable to carry out normal oxygen transporting function.

(k) **Alkaptonuria**

(1) Alkaptonuria was the first of the recessive human trait discovered in 1902 by **Archibald Garrod**, 'father of physiological genetics' or 'father of biochemical genetics'.

(2) Patients of alkaptonuria excrete large amounts of homogentistic acid in urine.

(3) Such urine turns black upon exposure to light.

(4) In normal person, homogentistic acid (alkapton) is oxidized by a liver enzyme homogentistic acid oxidase to maleyl acetoacetic acid.

(l) **Phenylketonuria (PKU)**

(1) Phenylketonuria was discovered by the Norwegian physician **A. Folling** in 1934; an autosomal recessive mutation of gene on chromosome 12.

(2) PKU results when there is a deficiency of liver enzyme phenylalanine hydroxylase that converts phenylalanine into tyrosine.

(3) There is a high level phenylalanine in their blood and tissue fluids.

(4) Increased phenylalanine in the blood interferes with brain development; muscles and cartilages of the legs may be defective and the patients cannot walk properly.

(m) **Gaucher's disease**

(1) Gaucher's disease is a genetic disease associated with abnormal fat metabolism, caused by the absence of the enzyme **glucocerebrosidase** required for proper processing of lipids.

(2) Non processing of lipids results in accumulation of fatty material in spleen, liver, bone marrow and brain.

(3) The swelling of these organs occurs and patients usually die by the age of 15 years.

(n) **Galactosemia**

(1) Galactosemia is inherited as an autosomal recessive, and the affected person is unable to convert galactose to glucose.

(2) Galactosemia is due to the deficiency of the enzyme Galactose Phosphate uridyl Transferase (GPT).

(3) Milk is toxic to galactosemic infants; child usually dies at three years of age.

(o) **Taste blindness of PTC**

(1) Taste blindness of PTC is a genetic trait, not a disease, discovered by **Fox** in 1932.

(2) PTC (phenyl thiocarbamide) is a compound of nitrogen, carbon and sulphur with sour taste.

(3) About 30% people lack the ability to taste PTC which is transmitted by a dominant gene T.

(4) The genotypes TT and Tt are tasters of PTC, while tt are non-tasters or taste blind persons.

14.4 Sex determination

Fixing the sex of an individual as it begins life is called sex determination. The various genetically controlled sex-determination mechanisms have been classified into following categories

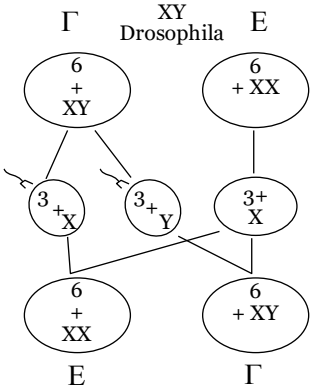
(i) **Chromosomal theory of sex determination :** The X-chromosome was first observed by German biologist, **Henking** in 1891 during the spermatogenesis in male bug and was described as X-body. The chromosome theory of sex determination was worked out by **E.B. Wilson** and **Stevens** (1902-1905). They named the X and Y chromosomes as sex-chromosomes or allosomes and other chromosomes of the cell as autosomes.

Sex chromosomes carry genes for sex. X-chromosomes carries female determining genes and Y-chromosomes has male determining genes. The number of X and Y chromosomes determines the female or male sex of the individual, Autosomes carry genes for the somatic characters. These do not have any relation with the sex.

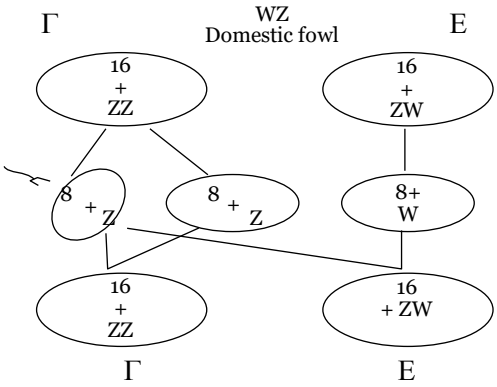
(a) **XX-XY type or Lygaeus type :** This type of sex-determining mechanism was first studied in the milk weed bug, Lygaeus turcicus by **Wilson** and **Stevens**. Therefore, it is called Lygaeus type. These are two different patterns of sex determination in Lygaeus type.

(1) **Female homogametic XX and male heterogametic XY :** The homogametic sex (XX) is female and produces ova all of one type, *i.e.* having X-chromosome. The male is heterogametic-XY and produces sperm of two types. 50% of which possess X-chromosome and other 50% Y-chromosome. This is simple XX-XY type and is found in man, *Drosophila* and certain insects.

Example : In *Drosophila* total number of chromosomes is eight, of which six are autosomes, common to both male and female. The fourth pair is of sex chromosomes. In male this is represented by XY *i.e.* Karyotype of male *Drosophila* 6+XY and in female XX *i.e.* 6+XX. Ova produced by female are all similar possessing 3+X chromosomes, whereas the sperm produced by male are 3+X and 3+Y in equal numbers.

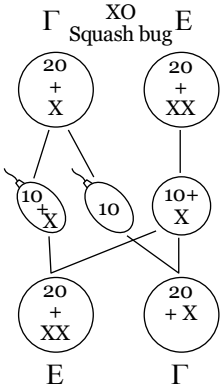


(2) **Female heterogametic and male homogametic :** In fowl, other birds and some fishes, certain moths and butterflies, the female sex is heterogametic, with X and Y chromosome often represented by Z and W and laying two types of eggs, one half with X or Z chromosome and the other half with Y or W chromosome. The male sex is homogametic having XX or ZZ chromosomes. It produces sperm all of one type.



(b) **XX-XO type or Protenor type : Mc clung** in male squash bug (*Anasa*) observed 10 pairs of chromosomes and an unpaired chromosome. Their females have eleven pairs of chromosomes (22). Thus all the eggs carry a set of eleven chromosomes but the sperm are of the two types: fifty percent with eleven chromosomes and the other fifty percent with ten chromosomes. The accessory

chromosome was X-chromosomes. Fertilization of an egg by a sperm carrying eleven chromosomes results in a female, while its fertilization by a sperm with ten chromosomes produces male. It is said to be evolved by the loss of Y-chromosome.



(c) **Haploid-diploid mechanism of sex determination :** Hymenopterous insects, such as bees, wasps, saw flies, and ants, show a unique phenomenon in which an unfertilized egg develops into a male and a fertilized egg develops into a female. Therefore, the female is diploid (2N), and the male is haploid (N). eggs are formed by meiosis and sperms by mitosis. Fertilization restores the diploid number of chromosomes in the zygote which gives rise to the female. If the egg is not fertilized, it will still develop but into a male. Thus, the sex is determined by the number of chromosomes.

In honeybee, the quality of food determines whether a diploid larva will become a fertile queen or a sterile worker female. A larva fed on royal jelly, a secretion from the mouth of nursing workers, grows into a queen, whereas a larva fed on pollen and nectar grows into a worker bee. Thus, the environment determines fertility or sterility of the bee but it does not alter the genetically determined sex. The sex ratio of the offspring in the hive is controlled by the queen. She lays more fertilized eggs that produce worker females and fewer unfertilized eggs which produce haploid males. The queen mates only once in her life time, keeps a store of sperms in the seminal receptacle, and can control fertilization of eggs by releasing or not releasing sperms.

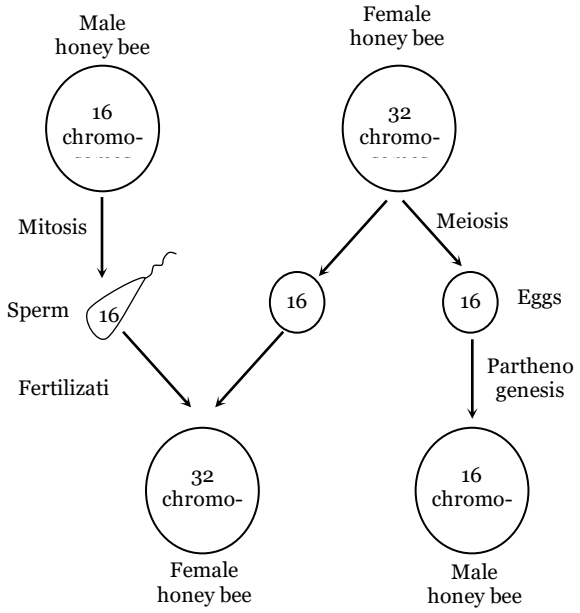


Fig : Haploid-diploid mechanism of sex determination in honeybee

Different types of chromosomal mechanisms of sex-determination in animals

S. No.	Organisms	Heterogametic sex	Gamete		Zygotes	
			Sperms	Eggs	Females	Males
(1)	*Drosophila*, man etc.	Male	X and Y	All X	XX	XY
(2)	Protenor(Bug, Grasshopper)	Male	X and O	XX	XX	XO
(3)	Birds, moths	Female	All X	X and Y	XY	XX
(4)	Fumea (a moth)	Female	All X	X and O	X	XX

(ii) **Quantitative or ratio theory of sex determination : C.B Bridges** worked out ratio theory of sex determination in *Drosophila*. According to this theory the ratio of chromosomes to autosomes is the determining factor for the sex. Single dose of X-chromosome in a diploid organism produces male, whereas 2X-chromosomes produce a female. If a complete haploid set of autosomes is designated by A then 2A : X will give rise to male and 2A : 2X to female.

(a) **Intersexes in *Drosophila* and ratio theory of sex determination :** Bridges hypothesis was supported by studies of flies arising after abnormal distribution of chromosomes on account of non-disjunction. Due to abnormal meiosis during oogenesis both the X-chromosomes fail to separate and move to one pole of meiotic spindle. Thus few eggs are formed with single autosomal genome but with 2X chromosomes, *i.e.* (AXX) and other with single autosomal genome but no sex chromosome (A). when such abnormal eggs are fertilized with normal sperm, the following result are obtained.

Results of fertilization of abnormal female gametes

AAXXY – Female
AAXXX – Super female
AAX – Sterile male
AAY – Nonviable

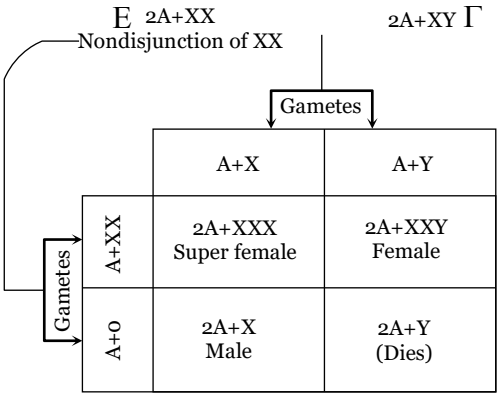


Fig : Nondisjunction of X-chromosome in a female Drosophila leading to transfer of both X-

Out of this progeny 1/4th males with no X are nonviable; the other 1/4 are without Y-chromosome and sterile. 1/4th females have an extra Y-chromosome while rest 1/4th females with 3X are super females. These are sterile with under developed sexual characteristics.

(ii) **Triploid intersexes and balance theory :** The triploid flies with (3A + 3X) are much like the normal diploid females both in appearance as well as in fertility. On mating to diploid males their progeny consisted of following types.

(1) AAAXXX – Triploid females
(2) AAXX – Dilpoid females
(3) AAXXY – Diploid females
(4) AAAXX – Intersexes
(5) AAAXXY – Intersexes
(6) AAXY – Normal males
(7) AAXXX – Super females
(8) AAAXY – Super males

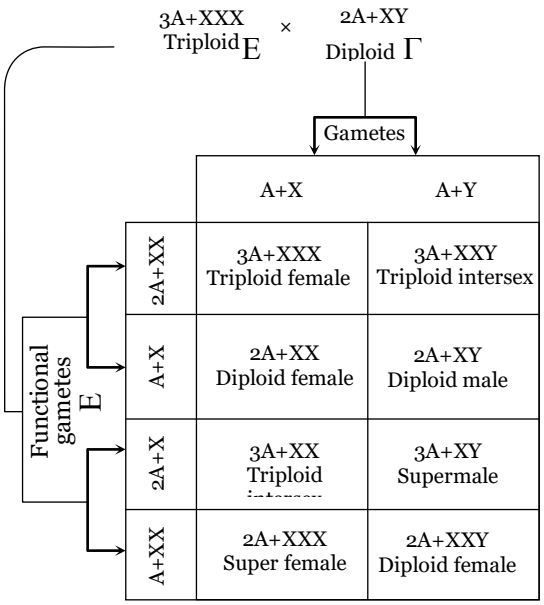


Fig : Results of a cross between triploid female and diploid male

The intersexes are sterile and intermediate between females and male, because the sex balance ratio in the intersexes comes to 2 : 3.

(2) **Gyandromorphs in Dorsophila and ratio theory of sex determination :** In *Drosophila* occasionally flies are obtained in which a part of the body exhibits female characters and the other part exhibits male characters. Such flies are known as **gynandromorphs**. These are formed due to misdivision of chromosomes and start as female with 2A+2X-chromosomes. One of the X-chromosomes is lost during the division of the cell with the result that one of the daughter cells possesses 2A+2X chromosomes and the other 2A+X. If this event happens during first zygotic division, two blastomeres with unequal number of X-chromosomes are formed. The blastomere with 2A+2X-chromosomes develops into female half, while the second blastomere with 2A+X chromosomes produces male half and the resultant fly is a bilateral gynandromorph. The occurrence of gynandromorphs clearly indicates that the number of X-chromosomes determines the sex of the individual.

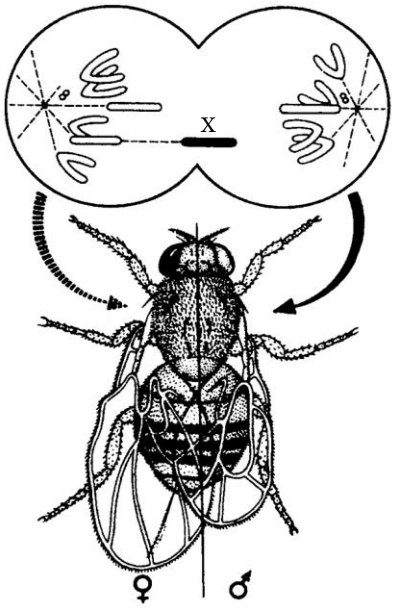


Fig : Gynandromorph of Drosophila in which right half is male and left half is female

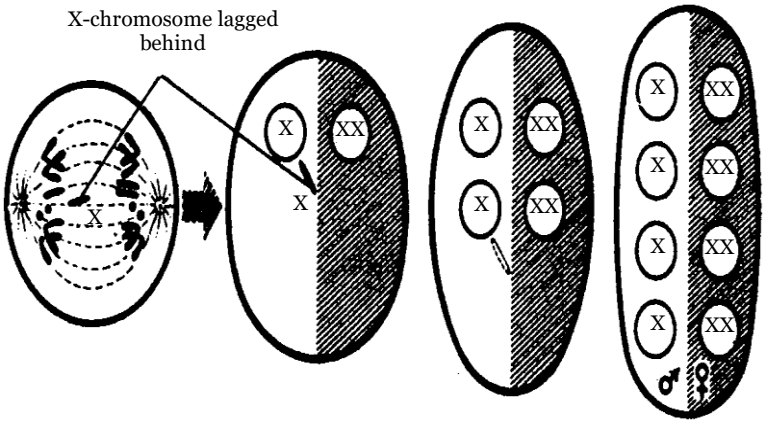


Fig : Diagram to show origin of gynandromorphs

(iii) **Genic balance theory :** Based upon the observations of ratio theory **Bridges** put forward genic balance theory in which he suggested that every individual whether male or female possesses in its genotype genes for both male and female characteristics. Which sex will actually develop is decided by the preponderance of that type of genes.

According to the genic balance theory of Bridges in *Drosophila melanogaster*, sex is determined by the ratio of the X-chromosomes and the set of autosomes. The *Y*-chromosomes play no part in sex determination it only governs male fertility. The XO flies are male, but sterile. Sex is governed by the ratio of the number of X chromosomes to sets of autosomes. The table given below indicates how the ratio of X/A help to determine the sex.

Ratio of X-chromosome to autosomes and the corresponding phenotype in *Drosophila*

S. No.	Sex	Number of X-chromosomes	Number of autosomal set	Sex index X/A ratio
(1)	Super female	XXX (3)	AA (2)	3/2 = 1.5
(2)	Normal female Tetraploid Triploid Diploid Haploid	XXXX (4) XXX (3) XX (2) X (1)	AAAA (4) AAA (3) AA (2) A (1)	4/4 = 1.0 3/3 = 1.0 2/2 = 1.0 1/1 = 1.0
(3)	Intersex	XX (2)	AAA (3)	2/3 = 0.66
(4)	Normal male	X (1)	AA (2)	1/2 = 0.50
(5)	Super male	X (1)	AAA (3)	1/3 = 0.33

Genes for maleness are carried on the autosomes, those for femaleness on the X-chromosomes. The sex index ratio of female is 1.0 while in males is 0.50. If X/A ratio is greater than 1.0 produces super females (meta females) and less than 0.50 produces super males. The X/A ratio lesser than 1.0 but greater than 0.5 (for example 0.66) result in intersexes. The degree of femaleness is greater where the X/A ratio is closer to 1.0 and the degree of maleness is greater where that ratio is closer to 0.5.

Human sex determination : The genic balance theory of sex determination is not universally accepted. Unlike *Drosophila* X : A does not influence sex determination. The key to sex determination in humans is the SRY (for sex region on the Y) gene located on the short arm of the Y-chromosome. In the male, the testis-determining factor (TDF) is produced by SRY on the Y-chromosome. TDF induces the medulla of the embryonic gonads to develop into testes. In the absence of SRY on Y, no TDF is produced. The lack of TDF allows the cortex of the embryonic gonads to develop into ovaries.

(iv) **Hormonal theory of sex determination :** The sex determination theories of chromosomes and genic balance successfully apply to the lower animals but in higher vertebrates and under certain conditions in invertebrates, the embryo develops some characters of the opposite sex together with the characters of its own sex-chromosome. It means, the sex changes under specific circumstances. This is due to the hormones secreted by the gonads of that animal.

(a) **Free martinism :** The influence of hormones on sex determination comes from free-martins often found in cattles. LILLIE and others found that where twins of opposite sex (one male and other female) are born, the male is normal but female is sterile with many male characteristics. Such sterile females are known as free martins.

The scientific explanation for the formation of free martins is the effect of hormones of the male sex on the female. In cattle the foetal membranes of the twins are fused in such a manner that they have a common circulation of blood. The female hormone is produced at a slightly later stage in the development and guides its development towards female side. But since the twins have a common circulation and blood passes from one twin into the body of other twin, the male hormone which is produced slightly in advance of female hormone, enters the body of female twin and before the female hormone onsets the development of female characteristics it is already differentiated in the guidance of male hormone. As a result the developing female is sterile.

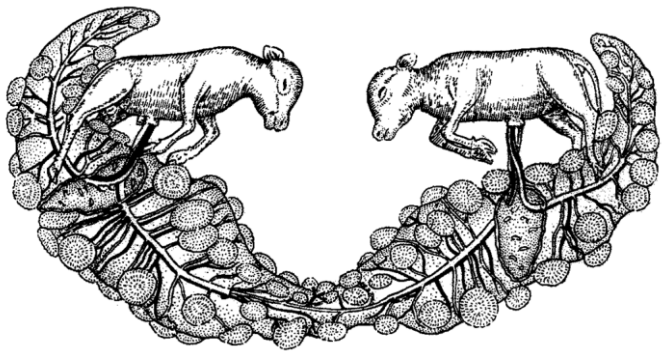


Fig : Free martins in cattle

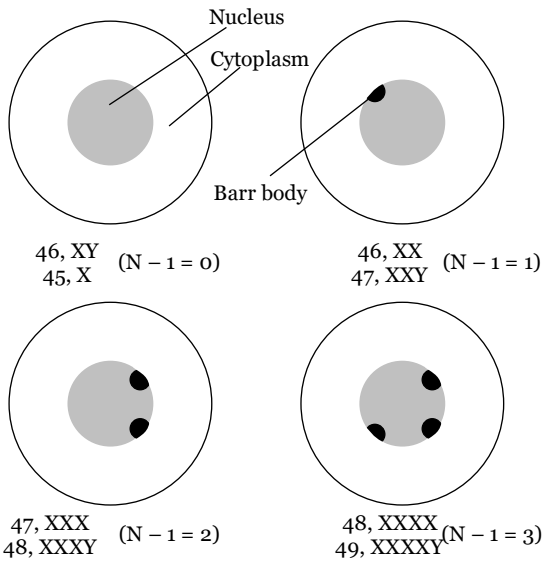


Fig : Chromosome variations and Barr body

(v) **Environmental theory of sex determination :** In some animals, there is environmental determination of sex.

(a) In *Bonellia*, a marine worm, the swimming larva has no sex. If it settles down alone, it develops into a large (2.5 *cm*) female. If it lands on or near an existing female proboscis, a chemical secreted from her proboscis causes the larva to develop into a tiny (1.3 *mm*) male. Male lives as a parasite in the uterus of the female.

(b) In turtles, a temperature below 28°C produces more males, above 33°C produces more females, and between 28°C to 33°C produces males and females in equal proportion, while in crocodile male sex is predominant at high temperature.

(vi) **Barr body in sex determination : Murray Barr** (1949), a geneticist noticed a small body in the nucleus of the nerve cells of female cats which stained heavily with nuclear stains. Further investigations showed that not only nerve cells, but many other cells from female cats only, had these

bodies, now known as sex chromatin or Barr bodies. It was soon learnt that such bodies can be found in females of many mammals including human. In women the Barr body lies against the nuclear membrane like a round disc in the neutrophil blood cells, skin cells, nerve cells, cells of mucous membrane, cells of lining in vagina and urethra. They are absent in man. These bodies are thus named after the discover **Barr**.

Barr bodies are used to determine the sex of unborn human embryos. In this technique called **amniocentesis** sample of the amniotic fluid is examined for Barr bodies. The sex is determined by the presence or absence of Barr bodies in epithelial cells of embryo present in the amniotic fluid sample. Studies from the cells of aborted embryos show that Barr bodies can be distinguished at about 15 or 16^{th} day after conception that means several weeks before the formation of gonads. Whereas sex of embryo is determined soon after fertilization, sex differentiation can be noticed in third week stage of pregnancy.

Mary Lyon hypothesis : According to the British geneticist **Mary Lyon** (1961), one of the two X-chromosomes of a normal female becomes heterochromatic and appears as Barr body. This inactivation of one of the two X-chromosomes of a normal female is the dosage compensation or Lyon's hypothesis.

It is estimated that number of Barr bodies is one less from the total number of X chromosomes present in embryo. Therefore, Barr bodies are also used to decide the genic constitution of such persons who have irregular number of sex chromosomes. More than one X chromosome in such persons is transformed into Barr bodies.

S. No.	Individual	No. of X chromosome	No. of Barr body (X – 1)
(1)	Normal woman	XX	2–1 = 1 (one barr body)
(2)	Women with Turner's syndrome	XO	1–1 = 0 (no barr body)
(3)	Super female	XXX	3–1 = 2 (two barr bodies)
(4)	Man	XY	1–1 = 0 (no barr body)
(5)	Man with Klinefelter's syndrome	XXY	2–1 = 1 (one barr body)

Sex can also be distinguished by studies of simple blood smears. The neutrophils, the most common of the white blood corpuscles, have a nucleus divided into two or three lobes. Female neutrophils showing a small drumstick extending out from one of the nuclear lobes, is a definite indication of the female chromosome component in the cells.

Important Tips

- Goldschmidt brought forward the quantitative theory of sex..
- The term "gynandromorphism" was introduce by Goldschmidt in 1915.
- Drumstick is the sex chromatin present in the neutrophil (Polymorphonuclear leucocyte) of 3 to 5% cells in females, but not in males.
- Y chromatin (Y body) can be identified as bright spot by staining cells with acridine dyes.
- First X-linked gene was discovered by T.H. Morgan (1910) for white eye mutation.

- Pedigree of colour blindness was first described by Horner (1876).
- It is also called bleeder's disease, first studied by John Cotto in 1803.
- Duchenne Muscular Dystrophy (DMD) is the disease which is characterized by a progressive weakness and loss of muscle.
- Inheritance of beard in a man is sex-limited.
- In melandrium (Garden flower) the sex determination type is XX-XY.

14.5 SEX LINKED INHERITANCE

Sex chromosomes of some animals and man besides having genes for sex character also possess gene for non sexual (somatic) characters. These genes for non sexual characters being linked with sex chromosomes are carried with them from one generation to the other. Such non-sexual (somatic) characters linked with sex chromosomes are called sex linked characters or traits, genes for such characters are called sex linked genes and the inheritance of such characters is called sex linked inheritance. The concept of sex-linked inheritance was introduced by **THOMAS H. MORGAN** in 1910, while working on *Drosophila melanogaster*.

The sex chromosomes in man and *Drosophila* are almost same in structure. The X and Y chromosomes, although different (non-homologous) in shape, size and structure, have atleast some similar (homologous) part which is known as homologous segment and the remaining part as non-homologous or differential segment. Genes for sex linked characters occur in both segments of X and Y chromosomes. Many sex linked characters (About 120) are found in man. Such characters are mostly recessive.

(i) **Types of sex linked inheritance**

(a) **Diandric sex linked or X linked traits :** Genes for these characters are located on non-homologous segment of X chromosome. Alleles of these genes do not occur on Y chromosome. Genes of such characters are transferred from father to his daughter and from his daughter to her sons in F_2 generation. This is known as Cris-cross inheritance. As the genes for most sex linked characters are located in X chromosome, they are called X-linked characters *e.g.* colour blindness and haemophilia in man and eye colour in *Drosophila*.

(1) **Sex linked inheritance in Drosophila :** *Drosophila melanogaster* has XX and XY sex chromosomes in the female

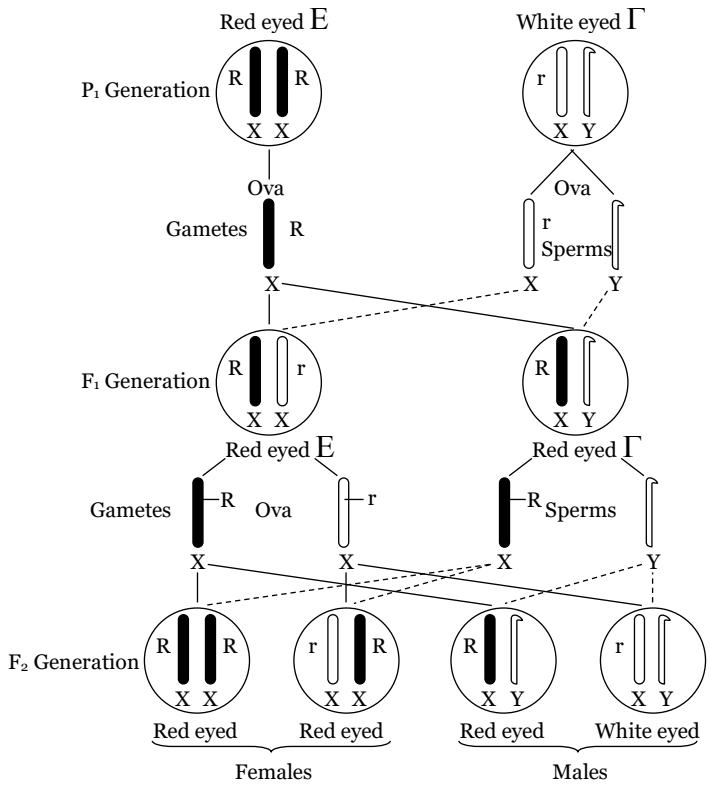


Fig : Sex-linked inheritance in *Drosophila melanogaster*. Note the transmission of sex chromosomes carrying eye colour gene R and r in a cross between red-eyed female and white-eyed male

and male respectively. Its eye colour is sex linked.

Allele of the eye colour gene is located in the X chromosome, and there is no corresponding allele in the Y chromosome. The male expresses a sex-linked recessive trait even if it has a single gene for it, whereas the female expresses such a trait only if it has two genes for it. The normal eye colour is red and is dominant over the mutant white eye colour. The following crosses illustrate the inheritance of X-linked eye colour in *Drosophila*.

(i) **Red-eyed female × White-eyed male :** If a homozygous red-eyed female fly is mated with a **hemizygous** (having a single allele for a trait) white-eyed male fly, all the F_1 flies, irrespective of their sex, are red eyed. When the red-eyed male and female flies of F_1 are intercrossed (equivalent to self pollination in peas), the F_2 flies are in the ratio of 2 red-eyed females to 1 red-eyed male to 1 white-eyed male. Thus, the red-eyed and white-eyed flies are in the ratio of 3 : 1 in F_2 generation (Mendelian monohybrid ratio).

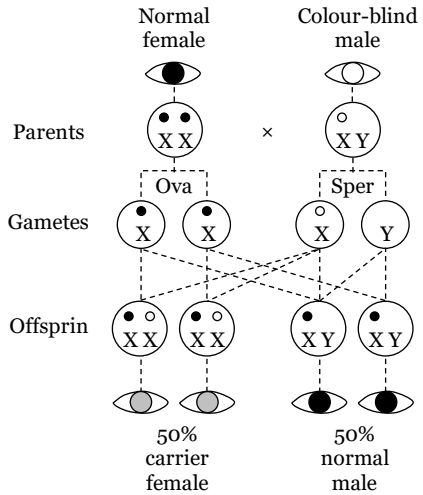
If X^R represents a gene for red eye and X^r that for white eye colour, the above cross may be diagramed as follows. The above cross shows that a recessive X-linked trait follows criss-cross inheritance, *i.e.*, transmission from the father to the grandsons through the daughters. The latter are called carriers because they have a trait but do not express it.

(2) **Sex linked inheritance in man**. Colour blindness and Haemophila are the two main sex linked or X-linked disease are found in man.

(i) **Colour blindness :** Person unable to distinguish certain colours are called colour blind. Several types of colour blindness are known but the most common one is 'red-green colour blindness'. It has been described by **HORNER** (1876).

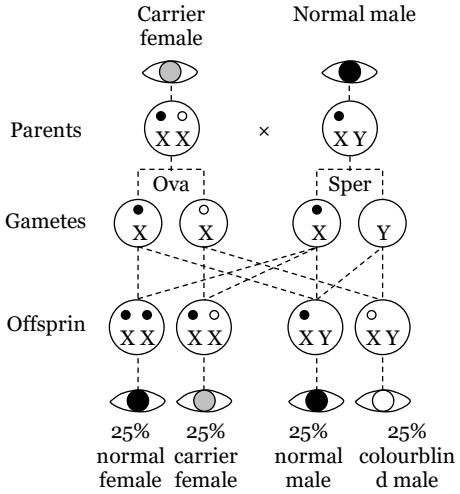
The red blindness is called protanopia and the green blindness deutoranopia. X-chromosome possesses a normal gene which control the formation of colour sensitive cells in the retina. Its recessive allele fails to do its job properly and results in colour blindness. These alleles are present in X chromosome is evidenced by the following results.

(1) If a normal female is married to a colour blind man.



Results : All her sons and daughter have normal colour vision, but all daughters are carrier.

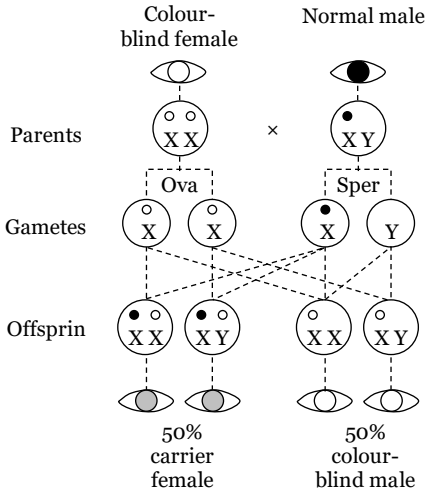
(2) But when her daughter (carrier) are married to man with normal colour vision man.



Result : Some colour blind sons are formed.

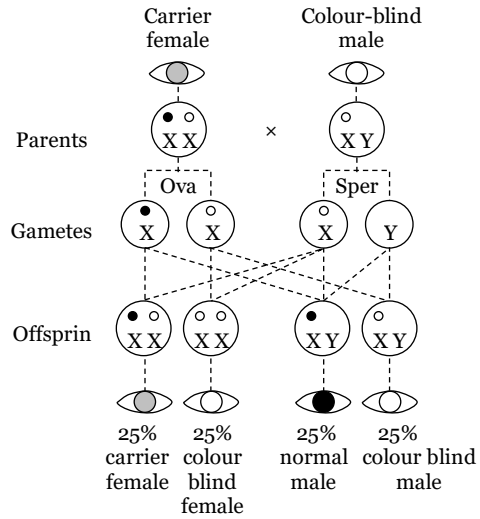
Conclusion : It means that a woman with normal colour vision whose father is colour blind gives birth to children, of which about half of the sons are colour blind and other half are normal.

(3) If a colour blind woman is married to a normal man.



Result : All her sons are colour blind whereas all the daughter have normal colour vision.

(4) But when these daughters having normal colour vision (Heterozygous) are married to colour blind man.



Result : The colourblind grandsons and grand daughters are produced with almost equal number of normal grandsons and grand daughters.

Conclusion : It means that a colour blind woman has sons all colour blind and daughters all with normal vision and a colour blind woman always has a colour blind father and her mother is a carrier.

Inheritance of colourblindness

PARENTS				**OFFSPRINGS**			
Female		**Male**		**Daughters**		**Sons**	
Genotype	**Phenotype**	**Genotype**	**Phenotype**	**Genotype**	**Phenotype**	**Genotype**	**Phenotype**
XX	Normal	X^cY	Colourblind	XX^c	Carrier	XY	Normal
XX^c	Carrier	XY	Normal	(i) XX (ii) XX^c	Normal Carrier	XY X^cY	Normal Colourblind
XX^c	Carrier	X^cY	Colourblind	(i) XX^c (ii) X^cX^c	Carrier Colourblind	XY X^cY	Normal Colourblind
X^cX^c	Colourblind	XY	Normal	X^cX	Carrier	X^cY	Colourblind

The above results could easily be explained with the assumption that colour vision is sex linked character and its gene is present on X-chromosome, Y-chromosome lacks its allele. Always male receives its X-chromosome from mother (through ovum) and Y-chromosome from father (through sperm), whereas the female receives one X-chromosome from each parent (through ovum and sperm). From the above result following conclusions may be drawn.

(1) Colour blindness is more common in males than in females.

(2) Two recessive genes are needed for the expression of colour blindness in female, whereas only one gene gains expression in male.

(3) Males are never carriers.

(4) Colour blind women always have colour blind fathers and always produce colour blind sons.

(5) Colourblind women produce colour blind daughters only when their husbands are colour blind.

(6) Women with normal colour vision, whose fathers are colour blind, produce normal and colour blind sons in approximately equal proportion.

(ii) **Haemophilia :** In haemophilia the blood fails to clot when exposed to air and even a small skin injury results in continuous bleeding and can lead to death from loss of blood.

It is also called bleeder's disease, first studied by **John Cotto** in 1803. The most famous pedigree of haemophilia was discovered by **Haldane** in the royal families of Europe. The pedigree started from Queen Victoria in the last century. In a patient of haemophilia blood is deficient due to lack necessary substrate, thromboplastin. It is of two types.

(a) **Haemophilia-A :** Characterized by lack of antihaemophilic globulin (Factor VIII). About four-fifths of the cases of haemophilia are of this type.

(b) **Haemophilia-B :** 'Christmas disease' (after the family in which it was first described in detail) results from a defect in Plasma Thromboplastic Component (PTC or Factor IX).

Like colour blindness, haemophilia is a well known disorder which is sex-linked recessive condition. The recessive X-linked gene for haemophilia shows characteristic Criss-cross inheritance like the gene for colour blindness. Its single gene in man results in disease haemophilia, whereas a woman needs two such genes for the same.

(iii) **Defective enamel :** It is a dominant X-linked trait and is inherited through a dominant X-linked gene. As X-chromosome is present in both man and woman, it is expressed in both the sexes. However, such persons have defective enamel on teeth like grey or brown unlike pure white enamel in a normal man.

Another example of dominant X-linked gene is the dimpled cheeks. Dimple may occur on one or both the cheeks.

(b) **Holandric or Y-linked traits :** Genes for these characters are located on non-homologous segment of Y chromosome. Alleles of these genes do not occur on X chromosome. Such characters are inherited straight from father to son or male to male *e.g.* hypertrichosis of ears in man.

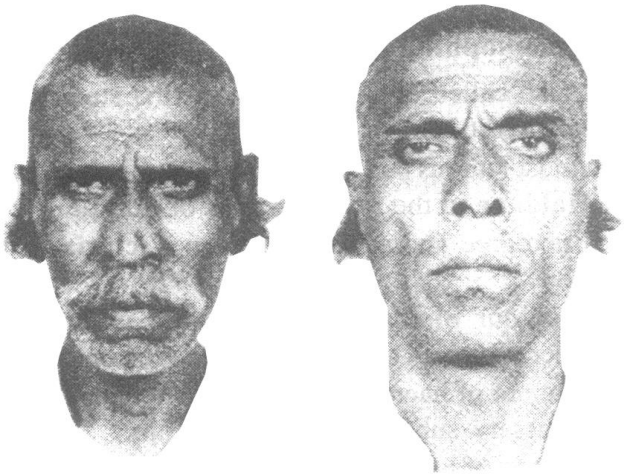


Fig : 'Hairy ears', an inheritance by holandric gene

(1) **Hypertrichosis of ears :** This is a condition in which excessive amount of large hair grow on the pinna in man. It is sex-linked trait controlled by a gene present on the non-homologous segment of the Y-chromosome. Hence its inheritrance is called **holandric inheritance** and it appears only in man. It passes directly from father to son.

(c) **XY-linked inheritance :** The genes which occur in homologous sections of X and Y-chromosomes are called XY-linked genes and they have inheritance like the autosomal genes.

Example of XY-linked genes are those of the inheritance of following

(1) **Xeroderma pigmentosa**, a skin disease characterized by the pigment patches and cancerous growth on the body.

(2) **Nephritis**, a kidney disease.

(ii) **Sex-influenced traits :** The autosomal traits in which the dominant expression depends on the sex hormones of the individual are called sex-influenced traits. These traits differ from the sex limited traits which are expressed in only one sex. It has following examples.

(1) **Baldness in man :** Baldness in humans is the best example of sex-influenced traits. This trait is due to a single mutant gene but the expression of the heterozygous is different in man and woman. This is a hereditary character controlled by sex-influenced gene which is dominant in men and recessive in women. The difference in expressions may be caused by varying amounts of male and female sex hormones. If autosome dominant gene 'B' is regarded to inherit the baldness, the homozygous (BB) dominant condition will cause baldness in man as well as women. This gene for baldness acts recessively in woman when present in heterozygous (Bb) condition, the baldness develops in males only because under such condition the phenotype expression (baldness) is influenced by androgen hormone secreted by man. A heterozygous female is normal. A homozygous recessive condition (bb) does not allow baldness to develop either in male or female.

Phenotypic expression of genotype for baldness

Genotype	**Phenotype**	
	Men	**Women**
B/B	Bald	Bald
B/b	Bald	Non-bald
b/b	Non-bald	Non-bald

The different phenotypes in men and women shown in above table are sex-influenced characters and also called sex-controlled traits.

The progeny that would be obtained from the marriage of heterozygous (B/b) man and woman for baldness have been shown below.

Progeny resulting from the marriage of bald men and non-bald women both heterozygous

P_1 Male gametes Female gametes	**Women (B/b) (non-bald) B and b**	**Man (B/b) Bald B and b**
	B	**b**
B	B/B bald male Bald female	B/b bald male Non-bald female
b	B/b bald male Non-bald female	b/b non-bald male Non-bald female

(2) **Length of index finger :** It is another example of sex-influenced trait in man. It is controlled by a gene which is dominant in male and recessive in the female. When the hand is placed on white board the tip of the fourth finger or ring finger just touches a horizontal line, it is seen that index or second finger does not reach this line in many cases. In some persons index finger extends beyond this horizontal line as shown in figure. The short index finger is inherited as a dominant trait in men and as a recessive condition in women.

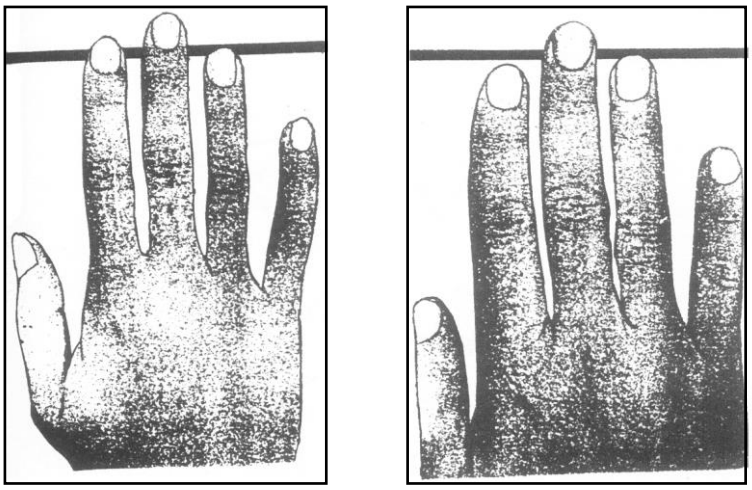


Fig : Sex influenced inheritance of length of index finger

(iii) **Sex limited traits :** Traits or characters which develop only in one sex are called sex-limited characters. They are produced and controlled by the genes which may be located on autosomes in only one sex. Such genes are responsible for secondary sexual characters as well as primary sexual characters. They are inherited according to Mendel's laws.

Sex-limited traits in man : Beard is produced by sex-limited genes in man, which does not develop in woman. Breast development is normally limited to woman. In case of abnormalities of hormonal secretions facial hair may develop in woman and a faminine breast development may occur in man. It means that expression of sex-limited characteristics in vertebrates depend upon the secretion of sex hormones. For example genes for deep masculine voice, masculine body, masculature in man will express themselves only in the presence of male hormone. Genes for faminine voice and faminine musculature on the other hand express themselves in the absence of male hormone and will not require the secretion of female hormone. Similarly, breast development in woman requires the presence of female hormone rather than mere absence of the male hormone. It can be concluded that certain sex-limited characteristics are expressed in the absence of certain hormones and other express only in the presence of sex-hormones.

14.6 Pedigree analysis

Inheritance of hundreds of characteristics such as polydactyly, haemophilia, colour blindness, attached ear lobes and tongue rolling, generation after generation in particular families of man have been studied. In order to conduct such study, a standard method has been used to represent the family pedigree in a concise, easily understood form so that one can visualize the entire pedigree (family history) at a glance of the chart.

(i) **Pedigree chart and symbols :** It is customary to represent men by squares and women by circles in a chart for study of pedigree analysis. Marriage is indicated by a connecting horizontal line and the children by attachment to a vertical line extending downward from the horizontal line. Individuals having particular characters to be studied are denoted by solid squares or circles while those not having them are indicated by outlines only. Twins are denoted by bifurcating vertical lines.

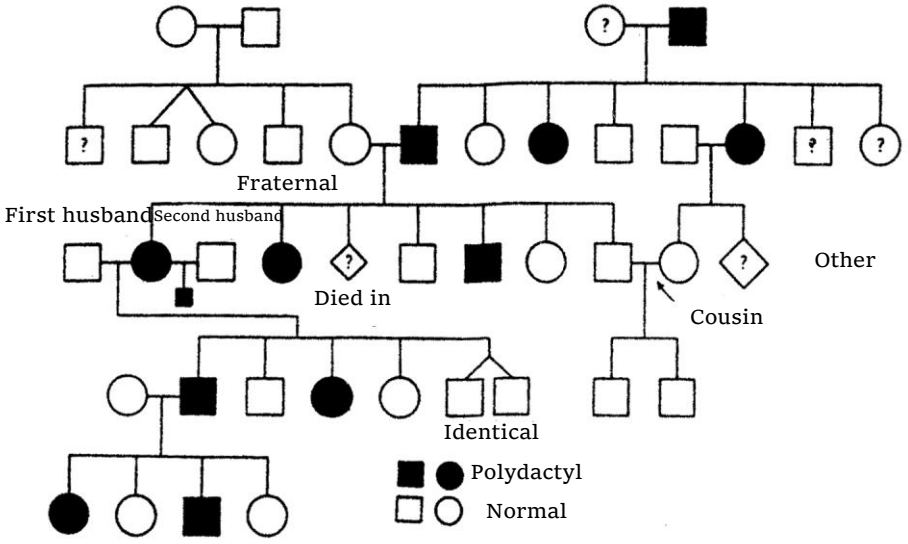


Fig : Commonly used symbols in pedigree chart

In such a pedigree analysis a person who is the beginner of the family history is called proband. It is called propositus, if male and poposita, if female. The children of such parents are known as sibs or siblings. So a family is constituted by such parents and their siblings. Sometimes, a very large family is formed as a result of interconnected marriages. Such a circle of large persons interconnected is called Kindred.

In order to study pedigree analysis we have taken some of the important case histories as follows :

(a) **Polydactyly :** The pedigree of this trait has become standard usage among the geneticists and it helps us to understand the process of transmission of this trait.

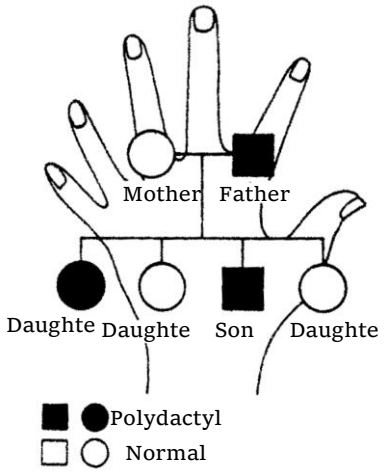


Fig : Pedigree of polydactyly

This inheritable trait was discovered when a woman brought her young daughter to a doctor for examination as she had an extra finger on one hand and an extra toe on one foot. On investigation it was found that child's father had this characters (though his extra finger had been removed surgically) and that her brother also had the character. The other two children of this family had normal number of fingers and toes. This type of inheritance is typical of characters which are known as dominant.

(b) **Attached ear lobes :** This is a recessive type of inheritance and is inherited in a different way.

(1) Two parents with free ear lobes produced two children with attached ear lobe in a family of five children.

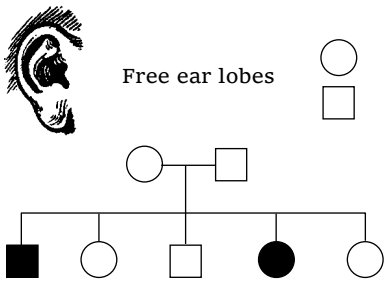


Fig : Pedigree of free ear lobes

(2) In another family both parents had attached ear lobes but all the four of their children had this trait of attached ear lobes.

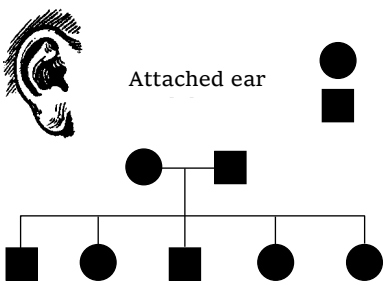


Fig : Pedigree of attached ear lobes

(c) **Tongue rolling :** Some persons are capable of rolling their tongue while others are not gifted with this power. A couple both of whom are tongue roller have two out of these children as tongue rollers.

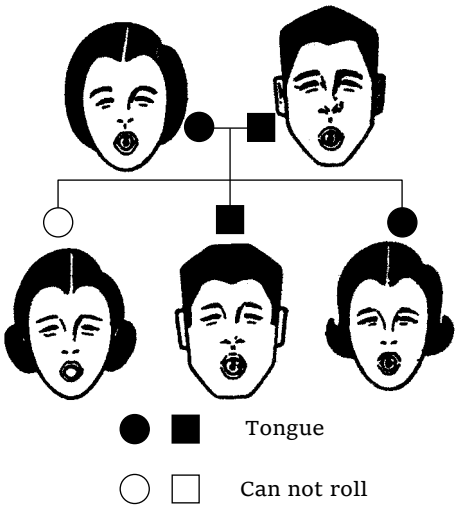


Fig : Pedigree of tongue rolling

(d) **Crooked little fingers :** This is a family pedigree of a human family where crooked little fingers are inherited through a simple dominant gene. In this pedigree a woman had two sons one of which had crooked little finger. Her husband also had same type of defective fingers. On further survey of her husbands, family it was found that her husband's sister and mother both had crooked little fingers, as well as his grandfather also possesses this trait. The characteristic also appeared in more distant relatives.

Fig : Inheritance of crooked little fingers (dominant trait)

14.7 Twins

Twins : Two birth occurring at the same time in human are called twins, they are of peculiar genetic interest. The hereditary basis of a number of human traits has been established by the study of twins. There are 3 kinds of twins.

(i) **Identical or monozygotic twins :** Identical twins are formed when one sperm fertilizes one egg to form a single zygote. As a result of separation of two daughter cells or blastomeres after the first cleavage, each of the cell develops into a separate individual. Such individuals are called **identical twins**. Since they develop from a single zygote, they are called **monozygotic twins**. They have the same genotype and phenotype and are of same sex. Differences if any, may be due to different environmental conditions.

(ii) **Siamese twins or conjoint twins :** Like monozygotic twins, siamese twins also originate from one zygote but the daughter cells formed as a result of first cleavage fail to separate completely and they remain joined at some point. They grow into two individuals joined together. Thus the two individuals called **conjoint twins** remain attached at one or more parts of the body. They were first studied in the country Siam, hence called Siamese twins. Siamese twins usually do not survive after birth although a few cases of their survival are well known. They are always of the same sex, same genotype and phenotype.

(iii) **Fraternal twins :** They are dizygotic twins formed from the two eggs fertilized by two sperms separately but at the same time. They may be both males, both females or one male and one female. They may have different genotypic constitution and different phenotype. Thus fraternal twins develop in same environment with different constitution but are the members of same age. They resemble each other just like any two brothers and sisters. Although they may be of same sex but due to different hereditary traits, they may carry congenital variations.

Among the twins fraternal twins are most common and Siamese twins are most rare.

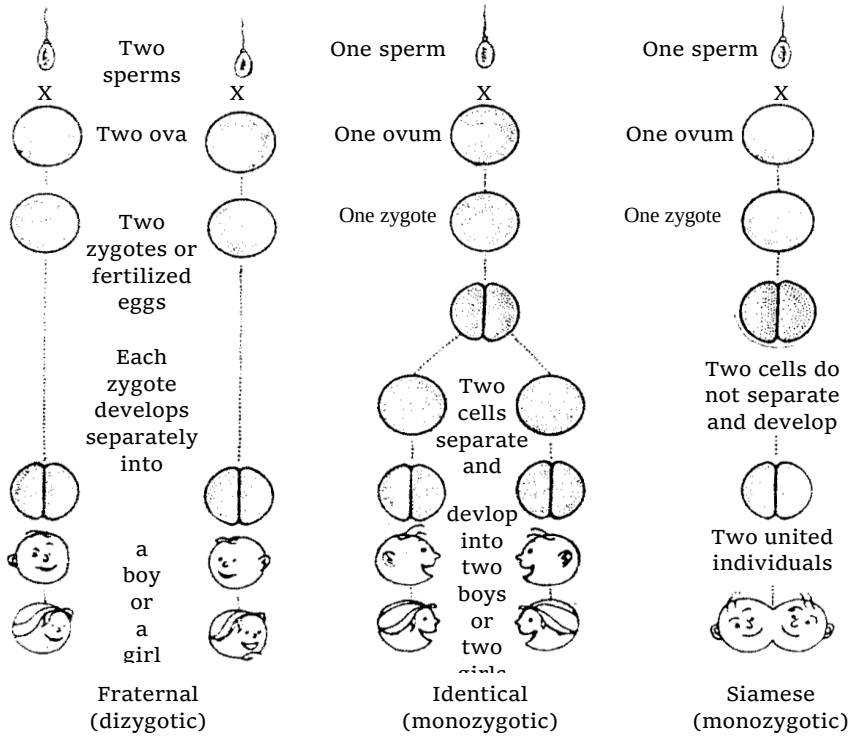


Fig : Human twins : formation and types

14.8 Eugenics, Euthenics and Euphenics

(i) **Eugenics :** The term eugenics (Gr. Eugenes, well born) was coined by British scientist **Sir Francis Galton** in 1883. Galton is called 'Father of eugenics' as this branch has been started by him.

Eugenics is the branch of science which deals with improvement of human race genetically. This aspect of human betterment aims to improve the human germplasm by encouraging the inheritance of best characteristics so that defective characters may be eliminated. Eugenics attempts to attain its objective bilaterally by suggesting a number of 'do's and 'don'ts' to improve the human gene pool. The 'don'ts' are meant to check inheritance of the poor or undesirable germplasm, while the do's aim at perpetuating desirable germplasm to be inherited. By this method aim of improvement of human race may be achieved by two ways :

(a) **Positive eugenics :** In this approach of eugenics the future generations are improved by encouraging the inheritance of better traits. Following methods may be adopted to achieve this.

(1) **Planned marriages :** The selection of mate for marriage should be made on the basis of better traits rather than on the basis of dowry, caste or religion etc. will give rise to the progeny with better traits.

(2) **Perevention of loss of good germplasm :** Many intelligent, specialists, educationists and politicians with better traits should be encouraged getting marriage at early stage and practice polygamy may contribute for more and more utilization of good germplasm.

(3) **Medical engineering :** To destroy the unwanted germplasm or such genes before their expression **Liederberg** (1963) put forth a novel idea of medical engineering.

(4) **Germinal choice or eutelegenesis :** In human beings artificial fertilization or insemination is a biological process. **Muller** (1963) put forward the idea of production of children of high mental qualities and good traits by artificial fertilization of a woman of high quality traits with the sperms of desired best man.

(5) **Genetic counselling :** Production of healthy progeny should be the endeavour of man to ensure a better future for humanity. Genetic counselling can make a significant contribution in this direction. It can provide much needed relief for families with history of genetic diseases. Genetic counselling is even useful after marriage. A Rh^- woman with Rh^+ partner when aware of the implications in the second child, can go for suitable medical aid well in advance.

(b) **Negative eugenics :** This is a negative aspect of improving mankind by restricting the transmission of poor and defective germplasm. This restriction can be brought about in the following ways.

(1) **Segregation :** Persons with serious abnormal hereditary defects like feeble mindedness, epilepsy, leucoderma, criminals, immorals and stupid people, should be isolated *i.e.* should not be permitted to mingle and marry with normal, intelligent persons.

(2) **Restriction on blood marriages :** Marriage between close relative like cousins tend to bring together the recessive alleles in homozygous condition and can be expressed in haemophilia, albinism and colour blindness.

(3) **Sterilization :** The most effective method to stop the persons with defective germplasm to produce offsprings is the sterilization. This prevents the transmission of undesirable traits in man and woman by vasectomy and tubectomy respectively.

(ii) **Euthenics :** Euthenics is the improvement of human race by improving the environmental conditions, *i.e.*, by subjecting them to better nutrition better unpolluted ecological conditions, better education and sufficient amount of medical facilities.

(a) **Better education :** Education is one of the surest agents which can provide better humanity. The conditions of surroundings *i.e.* the immediate environment of an individual has a great bearing upon personality of the person. The society which an individual chooses determines to some extent, his character. Medical facilities are largely responsible for maintenance of sound health. Employment conditions determine the degree of fulfillment of the individual's basic requirements and hence it influences his entire outlook. Euthenics attempts to provide the best of education, the healthiest of surroundings, the finest of societies, full medical facilities and rewarding employment conditions.

(b) **Subsidization of superior students :** Euthenics requires that a best student be selected and be provided opportunities for his multifaceted development. Students of no definite class and group may be equally intelligent. A few are most intelligent, some are average, still others are below average and some are dull or feeble minded or idiots. A definite scale to measure the mental ability has been prescribed which is known as intelligence quotient.

Intelligence quotient (IQ) : The ratio between actual (chronological) age and mental age multiplied with 100 is known as I.Q. Intelligence quotient is the mental competence in relation to chronological age in man. It can be denoted by following formula.

$$\text{I.Q.} = \frac{\text{Mental age}}{\text{Actual age}} \times 100$$

By applying this formula we can easily calculate the IQ, such as if a 10 year child has mental age 14, his IQ will be

$$\text{I.Q.} = \frac{14}{10} \times 100 = 140$$

On the basis of different levels of I.Q. persons are classified as follows.

S. No.	I.Q.	Person
(1)	0 – 24	Idiot
(2)	25 – 49	Imbecile
(3)	50 – 69	Moron
(4)	70 – 79	Dull
(5)	80 – 89	Ordinary
(6)	90 – 109	Average
(7)	110 – 119	Superior
(8)	120 – 139	Most superior
(9)	140 or more	Genius

(iii) **Euphenics :** The study of born defectives and their treatment is called euphenics. The term euphenics was given by **A.C. Pai** (1974) for symptomatic treatment of human genetic disease especially in born errors of metabolism. Following methods can be employed as euphenic measures.

(a) **Amniocentesis :** It is a test to detect genetic diseases as well as the sex of embryo during development in mother's womb. Amniotic fluid is tested and if embryo has genetic disease the embryo can be aborted.

(b) **Infusion of missing enzyme :** Genetic physiological diseases occur due to lack of particular enzymes. Infusion of such missing enzyme may help in treatment of such disease.

(c) **Genetic engineering :** Treatment of the gene controlling genetic disease by genetic surgery and genetic engineering is also helpful in euthenics.

14.9 GENETIC ENGINEERING

(i) **Recombinant DNA technology**

(a) **Definition :** Genetic engineering, a kind of biotechnology, is the latest branch in applied genetics dealing the alteration of the genetic make up of cells by deliberate and artificial means. Genetic engineering involves transfer or replacement of genes, so also known as recombination DNA technology or gene splicing.

(b) **Tools of genetic engineering :** Two enzymes used in genetic engineering are restriction endonuclease and ligases. R.E. is used to cut the plasmid as well as the foreign DNA molecules of specific points while ligase is used to seal gaps or to join bits of DNA.

The ability to clone and sequence essentially any gene or other DNA sequence of interest from any species depends on a special class of enzymes called restriction endonucleases. Restriction endonucleases are also called as molecular scissors or 'chemical scalpels'. Restriction endonucleases cleave DNA molecules only at specific nucleotide sequence called restriction sites. The first restriction enzyme identified from a bacterial strain is designated I, the second II and so on, thus, restriction endonuclease EcoRI is produced by Escherichia coli strain RY 13. Restriction enzyme called EcoRI recognizes the sequence $\begin{matrix} G\downarrow A\,A\,T\,T\,C \\ C\,T\,T\,A\,A\uparrow G \end{matrix}$ and cleaves the DNA between G and A on both strands. Restriction nucleases make staggered cuts; that is, they cleave the two strands of a double helix at different joints and blunt ended fragments; that is, they cut both strands at same place.

Characteristics of some restriction endonucleases

Enzyme name	Pronunciation	Organism in which enzyme is found	Recognition sequence and position of cut
Bam HI	"bam-H-one"	*Bacillus amyloliquefaciens* H	5′ G↓GAT C C 3′ 3′ C C TAG↑ G 5′
Bgl II	"bagel-two"	*Bacillus globigi*	A↓G A T C T T C T A G↑A
Eco RI	"echo-R-one"	*E. coli* RY13	G↓ A A T T C C T T A A↑G
Hae II	"hay-two"	*Haemophilus aegyptius*	R G C G C↓Y

			Y↑C G C G R
Hind III	"hin-D-three"	*Haemophilus influenzae* Rd	A↓ A G C T T T T C G A↑ A
Pst I	"P-S-T-one"	*Providencia stuartii*	C T G C A↓G G↑ A C G T C
Sma I	"sma-one"	*Serratia marcescens*	C C C ↓ G G G G G G ↑ C C C
Hae III	"hay-three"	*Haemophilus aegyptius*	G G ↓ C C C C ↑ G G
Hha I	"ha-ha-one"	*Haemophilus hemolyticus*	G C G ↓C C↑G C G
Hpa II	"hepa-two"	*Haemophilus parainfluenzae*	C↓ C G G G G C↑C

(c) **Steps of recombinant DNA technology**

(1) Isolating a useful DNA segment from the donor organism.

(2) Splicing it into a suitable vector under conditions to ensure that each vector receives no more than one DNA fragment.

(3) Producing of multiple copies of his recombinant DNA.

(4) Inserting this altered DNA into a recipient organism.

(5) Screening of the transformed cells.

(d) **Vectors :** Vector in genetic engineering is usually a DNA segment used as a carrier for transferring selected DNA into living cells. Which are as follows

(1) **Plasmid :** Plasmid are extrachromosomal, closed circular double stranded molecules of DNA present in most eukaryotes. All plasmid carry replicons pieces of DNA that have the genetic information required to replicate. Plasmid pBR 322 was one of the first widely used cloning vectors, it contain both ampicillin and tetracycline resistance genes.

(2) **Phage :** It is constructed from the phage λ chromosomes and acts as bacteriophage cloning vectors.

(3) **Cosmid :** The hybrids between plasmid and the phage λ chromosome give rise to cosmid vectors.

Beside all these there are artificial chromosomes like

BACs (Bacterial Artificial chromosomes)

YACs (Yeast Artificial chromosomes)

MACs (Mammalian Artificial chromosomes) are very efficient vectors for eukaryotic gene transfers.

(e) **Application of recombinant DNA technology :** The technique of recombinant DNA can be employed in the following ways.

(1) It can be used to elucidate molecular events in the biological process such as cellular differentiation and ageing. The same can be used for making gene maps with precision.

(2) In biochemical and pharmaceutical industry, by engineering genes, useful chemical compounds can be produced cheaply and efficiently which is shown in table.

Applications of recombinant DNA products

Medically useful recombinant products	Applications
Human insulin	Treatment of insulin-dependent diabetes
Human growth hormone	Replacement of missing hormone in short stature people
Calcitonin	Treatment of rickets
Chronic gonadotropin	Treatment of infertility
Blood clotting factor VIII/IX	Replacement of clotting factor missing in patients with Haemophilia A/B
Tissue plasminogen activator	Dissolving blood clots after heart attacks and strokes
Erythropoitin	Stimulation of the formation of erythrocytes (RBCs) for patients suffering from anaemia during kidney dialysis or side effects of AIDS patients treated by drugs
Platelet derived growth factor	Stimulation of wound healing
Interferon	Treatment of pathogenic viral infections, cancer

Interleukins	Enhancement of action of immune system
Vaccines	Prevention of infectious diseases such as hepatitis B, herpes, influenza, pertussis, meningitis, etc.

(ii) **Cloning :** Cloning is the process of producing many identical organisms or clones. In this process nucleus of ovum (n) is removed and replaced by nucleus of diploid cell of same organism. Now the egg with 2*n* nucleus is transferred to the uterus of mother to have normal pregnancy and delivers clone of itself.

Examples of organism cloning

(1) Cloning of sheep was done by **Dr. Ian Wilmut** (1995) of Roslin Institute, Edinberg U.K. and normal healthy lamb (DOLLY) was born in Feb, 1996. This lamb was exactly similar to her mother.

(2) The first cloned calves George and Charlie were born in January 1998.

(3) ANDI was the world's first genetically altered primate produced by inserting a jelly fish gene into the embryo of a rhesus monkey.

(4) Scientist at Scotland cloned POLLY and MOLLY. Unlike Dolly, polly and molly were transgenic (they carried human protein gene) polly and molly were born in july 1997.

(5) **Brigitte Boissliar**, a 46-year old french chemist announced the creation of the world's first cloned human boby nicknamed "Eve" (December 2002).

(iii) **Polymerase chain reaction (PCR) :** It was developed by **Kary Mullis** in 1983 and won Nobel prize in 1993. PCR is a method for amplifying a specific piece of DNA molecule without the requirement for time-consuming cloning procedure. This process require Target DNA, a heat stable DNA polymerase, which work at optimum temperature of 70°C usually Taq DNA and four types of nucleotides with small single stranded strands of DNA of about 20 nucleotide called primers, produce multiple copy of desired DNA.

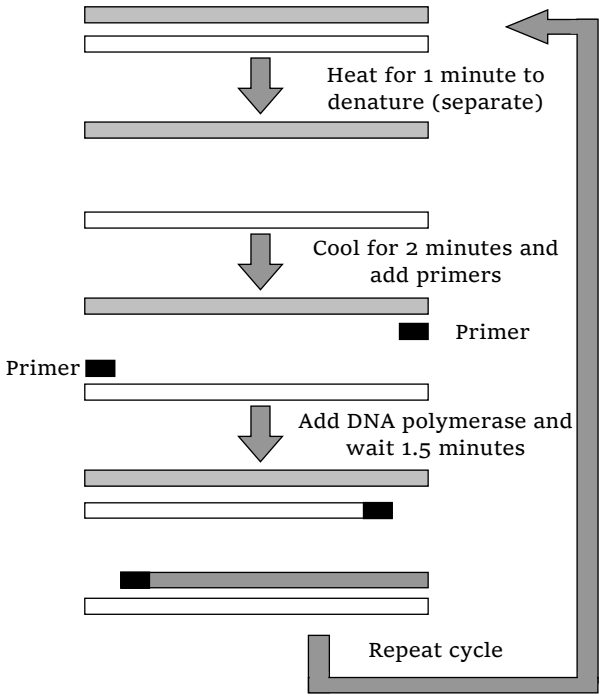


Fig : DNA amplification by PCR

(iv) **Gene libraries and gene banks**

(a) **Gene libraries :** A gene library is a collection of gene clones that contains all the DNA present in some source. If the original source of the DNA was original DNA from a living organism, then the library seek to include clones of all that DNA, it is called a genomic gene library. Gene libraries can also be created by using RNA.

(b) **cDNA :** If a gene library is created by enzymatic copying of RNA by reverse transcriptase (RNA-dependent DNA polymerase), it would be called c-DNA library. c-DNA stands for complimentary DNA or copy DNA. c-DNA is made to use PCR to amplify an RNA. PCR does not work on RNA, so one can copy it to DNA using reverse transcriptase and then PCR amplify the c-DNA; this is called RT-PCR (reverse transcriptase PCR).

(c) **Gene bank :** A gene bank is repository of clones of known DNA fragments, genes, gene maps, seeds, spores, frozen sperms or eggs or embryos. These are stored for possible use in genetic engineering and breeding experiment where species have become extinct.

(v) **DNA finger printing : Alec Jeffreys et al** (1985) developed the procedure of genetic analysis and forensic medicine, called DNA finger printing. It is individual specific DNA identification which is made possible by the finding that no two people are likely to have the same number of copies of repetitive DNA sequences of the regions. It is also known as DNA profiling. The chromosomes of

every human cell contain scattered through their DNA short, highly repeated 15 nucleotide segments called “mini-satellites” or variable-number Tandem Repeat (VNTR).

(a) **Technique for DNA fingerprinting**

- Only a small amount tissues like blood or semen or skin cells or the hair root follicle is needed for DNA fingerprinting.
- Typically DNA content of about 100,000 cells or about 1 microgram is sufficient.
- The procedure of DNA fingerprinting involves the following major steps :

(i) DNA is isolated from the cells in a high-speed refrigerated centrifuge.

(ii) If the sample of DNA is very small, DNA can be amplified by Polymerase Chain Reaction (PCR).

(iii) DNA is then cut up into fragments of different length using restriction enzymes.

(iv) The fragments are separated according to size using gel electrophoresis through an agarose gel. The smaller fragments move faster down the gel than the larger ones.

(v) Double stranded DNA is then split into single stranded DNA using alkaline chemicals.

(vi) These separated DNA sequences are transferred to a nylon or nitrocellulose sheet placed over the gel. This is called ‘Southern Blotting’ (after **Edward Southern**, who first developed this method in 1975).

(vii) The nylon sheet is then immersed in a bath and probes or makers that are radioactive synthetic DNA segments of known sequences are added. The probes target a specific nucleotide sequence which is complementary to VNTR sequences and hybridizes them.

(viii) Finally, X-ray film is exposed to the nylon sheet containing radioactive probes. Dark bands develop at the probe sites which resemble the bar codes used by grocery store scanners to identify items.

(b) **Applications of DNA fingerprinting**

This technique is now used to :

(i) Identify criminals in forensic laboratories.

(ii) Settle paternity disputes.

(iii) Verify whether a hopeful immigrant is, as he or she claims, really a close relative of already an established resident.

(iv) Identify racial groups to rewrite biological evolution.

(vi) **Gene therapy :** The use of bioengineered cells or other biotechnology techniques to treat human genetic disorders is known as gene therapy. Gene therapy is the transfer of normal genes into body cells to correct a genetic defect. It can be used to treat genetic diseases like sickle-cell anaemia and Severe Combined Immuno Deficiency (SCID). It (SCID) is caused by a defect in the gene for the enzyme adenosine deaminase (ADA). SCID patients have no functioning T lymphocytes and one treated with the injections of their white blood cells that have been engineered to carry the normal ADA alleles.

(vii) **Transgenics :** A gene that has been introduced into a cell or organism is called a transgene (for transferred gene) to distinguish it from endogenous genes. The animal carrying the introduced foreign gene is said to be transgenic animal and the possessor called Genetically Modified Organisms (GMOs). Most of the transgenic animals studied to date were produced by microinjection of DNA into fertilized eggs. Prior to microinjection, the eggs are surgically removed from female parent and fertilized *in vitro* then DNA is microinjected into the male pronucleus of the fertilized egg through a very fine-tipped glass needle. The integration of injected DNA molecules appears to occur at random sites in the genome.

The first transgenic animal produced was the 'supermouse' by the incorporation of the gene for human growth hormone by **Richard Palmiter** and **Ralph Brinster** in 1981.

(viii) **Genomics and human genome project :** The term genome has been introduced by **Winkler** in 1920 and the genomics is relatively new, coined by **Thomas Rodericks** in 1986. Genomics is the subdiscipline of genetics devoted to the mapping, sequencing and functional analysis of genomes. Genomics is subdivided into following types:

(a) **Structural genomics :** It is the study of genome structure deals with the complete nucleotide sequences of the organisms.

(b) **Functional genomics :** It is the study of genome function which includes transcriptome and proteome. Transcriptome is a complete set of RNAs transcribed from a genome while proteome is a complete set of proteins encoded by a genome and aims the determination of the structure and function of all the proteins in living organisms. The human genome project, sometimes called "biology's moon

shot", was launched on october 1, 1990 for sequencing the entire human genome of 2.75 billion (2.75×10^9 or 2750000 bp or 2750000 kilobase pairs or 2750 megabase pairs) nucleotide pairs.

Two important scientist associated with human genome are **Francis Collins**, director of the Human Genome Project and **J. Craig Venter**, founding president of Celera genomics. The complete sequencing of the first human chromosome, small chromosome 22, was published in December 1999.

Genome of Model organisms

S. No.	Organism	No. of base pair	No. of genes
(1)	Bacteriophage	10 thousand	–
(2)	E. coli	4.7 million	4000
(3)	*Saccharomyces cerevisiae*	12 million	6000
(4)	*Caenorhabditis elegans*	97 million	18,000
(5)	*Drosophila melanogaster*	180 million	13,000
(6)	Human	3 billion	30,000
(7)	Lily	106 billion	–

Prospects and implications of human genome :

(1) The genome project is being compared to the discovery of antibiotics.

(2) Efforts are in progress to determine genes that will revert cancerous cells to normal.

(3) The human genome sequencing not only holds promise for a healthier living. It also holds the prospects of vast database of knowledge about designer drugs, genetically modified diets and finally our genetic identity.

Important Tips

- Pallindromic DNA is a segment of DNA in which the base pair sequence reads the same in both directions from a point of symmetry.
- Western blotting is the technique used to detect specific proteins.
- Northern blotting is the technique used to blot transfer of RNAs.
- Recombinant DNA is also called chimeric DNA.

- Eli Lily (American company) in 1983 produced genetically engineered insulin called humulin with the help of E. coli plasmid clone.
- **DNA foot printing :** It determines the location and lengths of binding sites of various proteins that bind to DNA.
- Hargovind Khorana is associated with genetic engineering. He synthesized ‘gene’ artificially in a test tube (1969).
- Polymerase Chain Reaction (PCR) was developed by Kary Mullis in 1983 and got Nobel prize for chemistry.
- Southern blotting technique is used for separating DNA fragments and identification of cloned genes.
- Gel electrophoresis and autoradiography are employed in nucleic and blotting.
- Delayed ripening is possible by reducing the amount of cell wall degrading enzyme ‘Polygalacturonase’ responsible for fruit softening.

ASSIGNMENT

CHROMOSOMES

Basic Level

1. The term '*gene*' was first used by
(a) Johannsen (b) Mendel (c) Lemark (d) Cuvier

2. The prokaryotic genetic system contains
(a) DNA and histones (b) Either DNA or histones
(c)DNA but no histones (d) Neither DNA nor histones

3. '*One gene one enzyme'* theory was given by
(a) Beadle and Tautum (b) Watson and Crick (c) Beadle and Morgan (d) Morgan and Muller

4. In an organism that has 44 chromosomes i.e. 22 homologous pairs; at the end of first meiotic division the daughter cell will have
(a) 44 chromosomes (b) 11 chromosomes
(c) 22 chromosomes (d) Any number between 44 and 22

5. The somatic chromosome complement in all human being is
(a) 21 pairs of autosomes and one pair of heterosomes
(b) 23 pairs of autosomes and one pair of heterosomes
(c) 22 pairs of autosomes and one pair of heterosomes
(d) 22 pairs of autosomes and one pair of XY chromosomes

6. Relative morphologies of chromosome of an individual indicates his/her
(a) Genotype (b) Phenotype (c) Pedigree chart (d) Karyotype

7. The smallest portion of a gene which is responsible for mutation is called as
(a) Operon (b) Codon (c) Recon (d) Muton

8. Each chromosome carries a distinct region which plays a fundamental role in chromosome movements during mitosis is
(a) Centromere or Kinetochore (b) Telomere
(c) Centriole (d) Chromatid

9. The terminal end of a chromosome is called
(a) Centromere (b) Chromomere (c) Telomere (d) Metamere

10. Where is the genetic information in body contained
(a) Structural proteins (b) Enzymes (c) DNA (d) Enzymes and DNA

11. Wilkins *X*-ray diffraction showed the diameter of the DNA helix as
(a) 10 Å (b) 20 Å (c) 30 Å (d) 40 Å

12. The number of autosomes in man is
(a) 22 pairs (b) 11 pairs (c) 43 pairs (d) 23 pairs

13. A normal metaphase chromosome with a midial centromere is
(a) Metacentric (b) Sub-metacentric (c) Acrocentric (d) Telocentric

14. Lampbrush chromosomes are found inside
(a) Salivary glands of *Drosophila* (b) Salivary glands of silk moth
(c) Oocytes of frog (d) Nucleus of man

15. Depending upon size and centromere position the 46 chromosomes have been divided into a number of groups
(a) 6 (b) 5 (c) 7 (d) 10

16. Chromosome number is
(a) Fixed for a species (b) Fixed for an ecosystem
(c) Fixed for a community (d) Fixed for a biosphere

17. The twenty third pair of chromosomes in man is known as
(a) Chromatid (b) Heterosome (c) Autosome (d) Gene

18. The careers of hereditary material are
(a) Chromosomes (b) Gene (c) Gametes (d) Gametocytes

19. In man the normal number of chromosomes is
(a) 42 (b) 44 (c) 46 (d) 48

20. The genetic material of virus is
(a) DNA (b) RNA (c) DNA and RNA (d) DNA or RNA

21. The chromosomes as thread like structures in nucleus was first described by
(a) Mendel (b) Strasburger (c) Darwin (d) Levitzky

22. Genes are made up of
(a) Histones (b) Lipoproteins (c) Polynucleotides (d) Hydrocarbons

23. The function of chromosomes of carrying the genetic information from one cell generation to another is performed by
(a) RNA (b) DNA (c) Histones (d) Calcium

24. In chromosomes, the material controlling heredity is
(a) DNA (b) Histones (c) Chromocentres (d) RNA

25. The term '*gene*' refers to
(a) A portion of RNA (b) A linkage group
(c) A Portions of DNA (d) A sequence of amino acids

26. The structure of the chromosome to which spindle fibre is attached is
(a) Chromatid (b) Telomere (c) Centromere (d) Chromomere

27. Who used the word "*chromosome*"
(a) Huxley (b) Flemming 1888 (c) Kollikar 1888 (d) Waldeyer 1888

28. Polytene chromosomes were first observed by
(a) Batanetzky-1980 (b) Heitz and Bauer-1935 (c) Balbiani-1881 (d) Stevens and Wilson-1905

29. The structure present over chromosome is
(a) Nucleolus (b) Centromere (c) Centrochrome (d) Golgi bodies

30. A genome is the

(a) Diploid set of chromosomes (b)Haploid set of chromosomes

(c) Triploid set of chromosomes (d)All the above

31. Noble prize to Kornberg and Ochoa was given for

(a) Artificial synthesis of genes (b) Chemistry of DNA and RNA

(c) 'One gene one enzyme' hypothesis (d) Artificial synthesis of DNA

32. The chromosomes except those relating to sex are known as

(a) Heterosomes (b) Autosomes (c) Allosomes (d) Cytosomes

33. Chromosome set in zygote is

(a) 2*n* (b) 1*n* (c) 3*n* (d) 4*n*

34. Number of autosomes in a normal female is

(a) 21 (b) 22 (c) 23 (d) 44

35. Homologous chromosomes which are present in male and female both are known as

(a) Heterosomes (b) Replosomes (c) Androsomes (d) Autosomes

36. Number of chromosomes present in gorilla is

(a) 46 (b) 44 (c) 48 (d) 50

37. Allosomes are the name of

(a) Sex chromosomes (b) Swellings on the chromosomes

(c) Chromosomes other than the ones which determine sex

(d) Nucleolus organising regions or chromosomes.

38. In humans, the sex chromosome complement is

(a) XX-XY (b) XX-XO (c) ZO-ZZ (d) ZW-ZZ

39. Diploid chromosome number in humans is

(a) 46 (b) 44 (c) 48 (d) 42

40. A chromosome with sub-terminal centromere is

(a) Acentric (b) Acrocentric (c) Metacentric (d) Telocentric

41. A chromosome with centromere near the middle is called

(a) Metacentric (b) Submetacentric (c) Acrocentric (d) Telocentric

42. X-chromosome is

(a) Telocentric (b) Submetacentric (c) Acrocentric (d) Acentric

43. Y- chromosome is

(a) Acrocentric (b) Telocentric (c) Submetacentric (d) Acentric

44. A giant chromosome with a number of chromonemata is

(a) Lampbrush chromosome (b) Heterochromosome

(c) Supernumerary chromosome (d) Polytene chromosome

45. Chromatid is

(a) One half of chromosome (b) Haploid chromosome

(c) Complete chromosome (d) Duplicate chromosome

46. Centromere is that part of chromosome where

(a) Nucleoli are formed (b) Crossing over takes place

(c)Chromatids are attached (d) Nicking occurs

47. Separation will occur in two genes if they are

(a) Dominant alleles (b) Recessive alleles

(c) Present on homologous chromosome (d) Present on two separate chromosome

48. The polygenic genes show

(a) Different genotype (b) Different phenotype

(c) Different Karyotype (d) None of the above

49. Unit of distance between genes on the chromosomes is

(a) C.DNA (b) Morgan (c) Centimorgan (d) Chisquare

50. Total number of autosomes in a fertilized egg of human beings is :

(a) 44 (b) 22 (c) 46 (d) 23

51. A gene is said to be dominant, if

(a) It is never expressed in any condition

(b) It is expressed only in heterozygous

(c) It expresses its effect only in, homozygous stage

(d) It is expressed both in homozygous and heterozygous condition

52. Laws of segregation and dominance were given by

(a) Darwin (b) Morgan (c) Mendel (d) De Vries

53. Segregation of alleles takes place during

(a) Cleavage (b) Meiosis (c) Fertilization (d) Non-disjunction

54. The sum of genes in a population is called

(a) Genotype (b) Karyotype (c) Gene pool (d) Lethal gene

55. If two opposite alleles come together, one finding morphological expression masking the other, the fact is described as law of

(a) Inheritance (b) Dominance (c) Limiting factor (d) Segregation

56. A cross used to ascertain whether a dominant is homozygous or heterozygous is termed

(a) Monohybrid (b) Reciprocal (c) Back cross (d) Linkage cross

57. Organisms phenotypically similar but genotypically different are said to be

(a) Heterozygous (b) Monozygous (c) Multizygous (d) Homozygous

58. An individual receiving like genes for the same characters from its two parents, is known as

(a) Allelomorphic (b) Homozygous (c) Heterozygous (d) Azygous

59. When dominant and recessive alleles express themselves together, it is called

(a) Dominance (b) Co-dominance (c) Amphidominance (d) Pseudodominance

60. An organism which receives identical alleles of a particular gene from both parents is

(a) Heterozygote (b) Holometabolous (c) Homosapiens (d) Homozygote

61. When an allele fails to express itself in presence of the other allele, the former is said to be

(a) Recessive (b) Dominant (c) Codominant (d) Complementary

62. A pair of contrasting character is termed as

(a) Allelomorphs (b) Homozygous (c) Heterozygous (d) Polymorphs

63. The haploid condition is found in

(a) Amoeba (b) Bacteria (c) Ovum (d) Zygote

64. The character which appears physically in an animal is

(a) Genotype (b) Phenotype (c) Heterotype (d) Morphozygous

65. The frequency of a character is found to be increasing when

(a) It is dominant (b) It is recessive (c) It is adaptable (d) It is inheritable

66. Epistasis implies

(a) One pair of genes can completely mask the expression of another pair of gene

(b) One pair of genes independently controls a particular phenotype

(c) One pair of genes enhances the phenotype expression of another pair of gene

(d) Many genes collectively control a particular phenotype

Advance Level

67. Whereas the number of chromosomes is reduced to half in first reduction division of meiosis, then what is the need for second mitotic division

(a) For the segregation of replicated chromosomes

(b)For equal distribution of haploid chromosomes

(c) For the formation of four gametes

(d) For the equal distribution of genes on chromosomes

68. The polytene chromosomes were discovered for the first time in

(a) *Chironomus* (b) Fruitfly (c) *Drosophila* (d) House fly

69. The point at the which the polytene chromosomes appear to be attached together is known as

(a) Centriole (b) Chromocentre (c) Centromere (d) Chromomere

70. What is the chromosome number of plasmodium

(a) 18 (b) 14 (c) 10 (d) 9

71. How many nucleosomes are found in helical coil of 30 *nm* chromatin fibre

(a) 10 (b) 12 (c) 06 (d) 09

72. If the number of chromosomes in most body cells of a mammal is 40, the cells in the seminiferous tubule will have

(a) 40 chromosomes (b) 20 chromosomes

(c) 10 chromosomes (d) While some other will have 20

73. Balbiani discovered special type of chromosome from the salivary gland of *chironomus* larva which are recognized by the presence of

(a) Bands (b) Loops (c) Both bands and loops (d)All of the above

74. A cistron is

(a) Structural unit of gene (b)Functional unit of RNA

(c) Functional unit of gene (d)Replication unit of gene

75. Genetic information are carried in form of long chain of molecules made up of

(a) Amino acids (b) RNA (c) Nucleotides (d) Polypeptides

76. The human chromosomes are divided into 7 groups, *B*-chromosomes are

(a) 6-12 (b) 1-2 (c) 13-15 (d) 4-5

77. The condensation of the chromosomes are maximal with visible centromeres at which phase of cell cycle

(a) G_1 phase (b) *S* phase (c) G_2 phase (d) *M* phase

78. Gene can be defined as

(a) Unit of segregation (b) Unit of physiological activity

(c) Unit of recombination (d) Unit of function

79. Lamp-brush chromosome is found in

(a) *Drosophila* (b) *Ascaris* (c) *Hydra* (d) None of the above

80. In recent past human chromosomes have been studied by a technique using specific, often fluorescent dyes, known as

(a) Dyeing technique (b) Banding technique (c) Ultra dyeing technique (d)Karyotyping technique

81. Arrangement of chromosomes in the order of decreasing length is termed

(a) Pedigree (b) Eugenetics (c) Idiogram (d) Dysengenics

82. Traits controlled by genes located on autosomes are said to be

(a) Sex affected (b) Sex influenced (c) Sex linked (d) Genetic traits

83. Tjio and Levan's contribution is very significant because they

(a) Gave the number of human chromosomes (b) Pointed out mutational changes

(c) Identified Barr bodies (d) Detected sex linkage

84. Polytene or giant chromosomes are found in

(a) Salivary glands of man (b)Salivary glands of woman

(c) Salivary glands of all animals (d) Salivary glands of *Drosophila*

85. Telocentric chromosome differs from acrocentric chromosomes in that

(a) The former has a subterminal centromere whereas the later has a centrally located centrosome

(b) The centromere in the former is terminal and in the later is subterminal

(c) The former has a terminal centromere and the later has a medially located centromere

(d) None of the above

86. Each chromosome at the anaphase stage of a bone marrow cell in our body has

(a) Two chromatids (b) No chromatids (c) Only one chromatid (d) Several chromatids

87. A complete set of chromosomes inherited as single unit from one parent is known as

(a) Gene pool (b) Genotype (c) Genome (d) Genoid

88. Centromere is a part of chromosome which helps in the

(a) Division of centrosomes (b)Formation of spindle fibres

(c) Movement of chromosomes (d)Formation of nuclear spindle

89. The puffs and rings are associated with the

(a) Endoplasmic reticulum (b)Polytene chromosomes

(c) Golgi bodies (d)Nucleus

90. Lampbrush chromosomes are visible

(a) In diplotene of meiosis (b)In prophase of meiosis

(c) In interphase (d)In mataphase of meiosis

91. The grouping of human chromosomes is based on

(a) Secondary constrictions alone (b)Dot-like satellites alone

(c) Banding patterns alone (d)All the above

92. The gene which increases the frequency of mutation in other is referred to as

(a) Mutator gene (b) Mutagene

(c) Hypostatic gene (d) Complementary gene

93. Two allelic genes are located on

(a) The same chromosomes (b) Two homologous chromosomes

(c) Two non-homologous chromosomes (d) Any two chromosomes

94. The frequency of an allele in an isolated population may change due to

(a) Gene flow (b) Mutation (c) Genetic drift (d) Natural selection

95. Number of autosomes in human sperm is

(a) 11 (b) 22 (c) 44 (d) 45

96. Banding pattern of present man and chimpanzee is nearly the same. It indicates that both have
(a) Similar gene pool (b) Similar number of chromosomes
(c) Evolved from a common stock (d) Developed brain and memory

97. Chromosomes of all races of human are
(a) Different (b) Similar
(c) Different in banding only (d) Similar in banding only

98. The banding pattern indicates that
(a) Gibbon, chimpanzee, gorilla and human are fundamentally the same
(b) Chromosome material is highly concerned throughout their evolution
(c) Differences in banding pattern is due to inversion, translocation etc.
(d) All the above

99. Chromosome were first seen by
(a) Hofmeister (b) Strasburger (c) Flemming (d) Waldeyer

100. As per latest information human genome has
(a) 3,00,000 genes (b) 30,000 genes (c) 3,000 genes (d) 300 genes

101. Foetal sex can be determined from cells present in amniotic fluid by looking for
(a) Kinetochores (b) Chiasmata
(c) Barr bodies and sex chromosomes (d) Autosomes

102. Asymmetric karyotype is
(a) Advanced feature (b) Very primitive feature
(c) Primitive feature (d) Without any evolutionary significance

103. Asymmetric karyotype is the one which has
(a) Fewer metacentric chromosomes
(b) Large scale difference between large and small chromosomes
(c) Both (a) and (b) (d) Chromosomes with varied shape

104. Puffs or balbiani rings in salivary gland chromosomes are sites of
(a) DNA replication (b) DNA duplication (c) RNA synthesis (d) Protein synthesis

105. More than 200 chromosomes occur in
(a) Chicken (b) Dog (c) Amoeba (d) Gorilla

106. If two genes are present at the same locus and after interacting with each other produce different effect are called
(a) Codominance (b) Dominance (c) Epistasis (d) None of the above

107. A complete set of chromosomes inherited from parent to offspring is called
(a) Genome (b) Allele (c) Diploid (d) Gamete

108. Which of the following is used to define the karyotype of a species (In this item one or more answers given may be correct)

1. The length of chromosome
2. The position of centromere
3. The number of chromosomes

Answer codes :

(a) 1, 2 and 3 are correct (b)Only 1 and 2 are correct

(c) Only 2 and 3 are correct (d) Only 1 and 3 are correct

109. To make a karyotype, chromosomes are photographed during

(a) Interphase (b) Fertilization (c) Mitotic metaphase (d) Meiotic metaphase

MULTIPLE ALLELISM

Basic Level

110. Inheritance of ABO blood group system is an example of

(a) Multiple allelism (b) Partial dominance (c) Epistasis (d) Dominance

111. Who discovered *Rh* factor

(a) Huxley (b) Landsteiner

(c) Landsteiner and Weiner (d) Weiner

112. A person with angtigens '*B*' in RBC and antibodies '*a'* in the plasma belongs to the blood group

(a) *A* (b) *B* (c) *AB* (d) *O*

113. Persons of blood group A contain

(a) Antigen *A* and antibodies *b* (b) Antigen *A* and antibodies *a*

(c) Antigen *A* and *B* and no antibodies (d) No antigens and both *a* and *b* antibodies

114. *Rh* factor may be responsible for

(a) Turner's syndrome (b) AIDS (c)Sickle-cell anaemia (d)Erythoblastosis foetails

115. The problem due to *Rh* factor arises when the blood two (Rh^+ and Rh^-) mix up

(a) In a test tube (b) Through transfusion (c) During pregnancy (d) (a) and (c) both

116. The antigen 'A' is present in the

(a) Blood plasma of 'B' blood group person (b) RBC of 'B' blood person

(c) Blood plasma of 'A' blood group person (d) RBC of 'A' blood group person

117. A person with antigens *A* and *B* and not antibodies belongs to blood group **or** In which blood group antibodies are absent

(a) *A* (b) *B* (c) *AB* (d) *O*

118. If a man Rh^+ marries a lady Rh^- then

(a) First child will die (b) First child will survive

(c) No child will be born (d) None of the above

119. At what temperature the blood is stored in bottles

(a) 4°*C* (b) 37°*C* (c) 0°*C* (d) 25°*C*

120. In how much maximum period the stored blood should be transfused

(a) 7 days (b) 10 days (c) One month (d) 15 days

121. The stored blood must be free from

(a) Impurities only (b) Viruses only

(c) Infectious biotic potentialities (d) Bacteria only

122. Blood group of an individual in determined by

(a) Shape of RBC (b) Combination of RBC and WBC

(c) Genetic material carried by individual (d) Nature of haemoglobin

123. A person having blood group O can receive blood

(a) Group O, B and AB (b) Group A, B and AB (c) Group B and AB (d) Group 'O' only

124. A child's blood group is '*O*'. The parents blood groups cannot be

(a) AB and O (b) B and O (c) A and B (d) A and A

125. Blood groups are named because of the agglutinogen *A* and *B* present in

(a) Plasma (b) RBC (c) WBC (d) Platelet

126. Which of the following is genetically dominant in man

(a) Colour blindness (b) Rh positive (c) Haemophilia (d) Albinism

127. Which of the following is an inherited trait in man

(a) FSH (b) LH (c) TSH (d) Rh

128. Rh factor is named after

(a) Man (b) Rat (c) Monkey (d) Chimpanzee

129. A child of a mother with blood group A and a father with blood group AB may have any one of the following blood group except

(a) *A* (b) *B* (c) *AB* (d) *O*

130. A patient has type of '*A*' blood, he needs blood transfusion, Type 'A' blood is not available which of the following blood type could be substituted without causing harm to the patient

(a) *AB* (b) *O* (c) *B* (d) *AB* and *O*

131. *Rh* factor is concerned with

(a) Blood groups (b) Blood clotting

(c) Carbohydrates metabolism (d) Eugenics

132. In erythroblastosis foetalis, which factors of the mother pass through placenta into the foetus

(a) *Rh* antigens (b) *Rh* antibodies (c) ABO antibodies (d) Agglutinins

133. Who was scientist to introduced ABO blood groups

(a) Wiener (b) Levine (c) Fisher (d) Landsteiner

134. If blood group of parent are AB and O, children have

(a) O^- group (b) AB (c) AB and O (d) A or B

135. The blood of *AB* group donor can be transfused to a person with the blood group

(a) A (b) B (c) AB (d) O

136. A person with blood group 'A' can be given blood of which group

(a) A and B (b) B and O (c) A and O (d) A, B, AB and O

137. For a child having blood group B, if father has blood group A. What may be the blood group of the mother

(a) O and A (b) O (c) B or AB (d) A

138. Donors and recipients in a blood transfusion process can be

(a) Only father and son (b) Only brother and sister

(c) Only maternal and niece (d) All the above

139. If a certain patient with blood group B requires immediate blood transfusion, the following type can be given to him

(a) O and B (b) O and AB (c) A and AB (d) B and AB

140. Universal donors have no antigens in RBC and have both *a* and *b* antibodies. They belongs to blood group

(a) *A* (b) *B* (c) *AB* (d) *O*

141. A person meets with a accident and great loss of blood has occurred. There is no time to analyze his blood group. It is safe to transfer blood of group

(a) AB, Rh^+ (b) AB, Rh^- (c) O, Rh^- (d) O, Rh^-

142. Universal donor is

(a) *A* blood group (b) *B* blood group (c) *AB* blood group (d) *O* blood group

143. Antisera used to detect Rh blood group

(a) Anti A (b) Anti B (c) Anti C (d) Anti D

144. To store blood some anticoagulant is added. It can be

(a) Sodium chloride (b) Sodium oxalate (c) Potassium chloride (d) Thromboplastin

145. Which of the blood group does not contain any antigen **or** In which blood group antigens are absent

(a) Blood group O (b) Blood group A (c) Blood group B (d) Blood group AB

146. Which one of the following blood groups belongs to the category of universal recipient

(a) A (b) AB (c) B (d) O

147. The blood group of father is 'A' and that of mother is 'B' what will be the blood group of their children

(a) *A, B, O, AB* (b) *AB* (c) *AO* (d) None of the above

148. Genotype of blood group 'A' will be

(a) I^AI^A (b) I^BI^B (c) I^AI^A or I^AI^O (d) I^AI^O

149. Blood group 'B' will have alleles

(a) *ii* (b) I^AI^A (c) I^BI^B (d) I^BI^B or *Ii*

150. Which of the following are most abundant types of antibodies

(a) *IgA* (b) *IgE* (c) *IgG* (d) *IgM*

151. Multiple Allelism control inheritance of

(a) Blood group (b) Phenylketonuria (c) Colour blindness (d) Sicke cell anaemia

152. Which one of the following is hereditary character of blood

(a) Blood group (b) Haem (c) Nucleus (d) None of the above

153. Which of the following would result in haemolysis of foetus

(a) Rh incompatibility (b) BO incompatibility (c) AB incompatibility (d) AO incompatibility

154. During blood typing agglutination indicates that the :

(a) RBCs carry certain antigens (b) RBCs carry certain antibodies

(c) Plasma contains certain antigens (d) Plasma contains certain antibodies

155. The genotype of Rh positive person could be :

(a) RR (DD) (b) Rr (Dd) (c) rr (dd) (d) Both (a) and (b)

156. **Assertion** (a) **:** Person with blood group AB can take blood from any other person.

Reason (R) : Blood group incompatibility is due to antigen antibody reaction. Blood group AB has no antibody and thus the antigen of other group is not affected.

(a) Both A and B are true and R is the correct explanation of A

(b) Both A and B are true but R is not the correct explanation of A

(c) A is true but B is false

(d) Both A and B are false

Advance Level

157. *MN* factor are due to

(a) Two co-dominant genes *M* and *N* (b) Recessive genes *m* and *n*

(c) *Mn* and *Nm* (d) None of the above

158. If a child has *O* type of blood group and the father *B* type, the genotype of the father will be

(a) I^OI^O (b) I^AI^B (c) I^OI^B (d) I^BI^B

159. Detection of blood group is done by agglutinisation test using antiserum. According to this

(a) If the blood shows coagulation with antiserum B the blood group is B

(b) If the blood shows coagulation with both antiserum A and B, the blood group is O

(c) If the blood shows coagulation with antiserum A, the blood group is AB

(d) None of the above

160. In term of *ABO* system of blood grouping a transfusion reaction is likely to follow administration of

(a) Group A blood to a group A person (b) Group B blood to a group AB person

(c) Group O blood to a group AB person (d) Group A blood to a group O person

161. Between persons of which two blood groups is the transfusion is not possible

(a) O and AB (AB recipient) (b) O and A (O donor)

(c) O and B (O donor) (d) O and AB (AB donor)

162. If a human mother has 'O' blood group, the foetus would die if the blood of foetus is

(a) A

(b) B

(c) AB

(d) Would remain unaffected by blood group whether it is A, B or AB

163. The second pregnancy of a woman terminates due to anaemia of the foetus. She has never had a blood transfusion. On the basis of this, which of the following is correct

(a) Child from the first pregnancy is Rh^{+ve} (b) The husband of the woman is Rh^{+ve}

(c) The woman is Rh^{-ve} (d) All the above

164. A man with blood group 'AB' marries a woman with 'O' blood group. In this situation

(a) The blood group of their children will be the same as that of the mother

(b) The blood group of the children differs from both the parents

(c) While 50% of children will have father's blood group, the remaining will have mother's blood group

(d) None of the above

165. When whole blood is stored with an anticoagulant at $4°C$, the K^+ ions move out from the RBC into the plasma. The most likely reasons for this is that

(a) RBC haemolyses and hence leakage of K^+ ions

(b) K^+ ions become more mobile at $4°C$

(c) Active transport ceases resulting in ionic equilibrium

(d) The anticoagulant attracts the K^+ ions into the plasma

166. In a medico-legal case of accidental interchange between two babies in a hospital, the baby of blood group A could not be rightly given to a people

(a) With both husband and wife if group O (b) Husband of group O and wife of group A

(c) Husband of group A and wife of group O (d) Both husband and wife of group A

167. If one parent belongs to 'A' blood group and the other of 'O' blood group, their children possibly represent

(a) A and B groups only (b)AB only (c) A and O groups only (d)All four groups

168. Which one of the following blood group system is determined by genes on the *X* chromosome

(a) *Yt* (b) *ABO* (c) *Xg* (d) *MNSs*

169. Even though a donor *X* and a recipient Y belongs to the same blood group, transfusion of blood leads to aggulutination. This is because

(a) *X* is Rh^+ and *Y* is Rh^- (b) Haemoglobin of *X* and *Y* is different

(c) *X* is Rh^- and *Y* is Rh^+ (d) Both are Rh^+

170. Blood bank of the body or reservoir where the blood is stored and can be mobilized, is

(a) Heart (b) Liver (c) Bone marrow (d) Spleen

171. A woman of blood group 'O' presented a baby of blood group 'O' which she claimed as her child. She brought a suit against a man of 'AB' group as the father of the child. Which statement is correct as per your judgement

(a) The father and mother claimed are the true persons

(b) Father is true and mother is not true person

(c) Both the parentage claims are false

(d) Mother is the true person and father claimed is not true

172. The probability of having a child with blood group *O* to parents with blood groups *A* and *B* is

(a) 4 out of 4 (b) 3 out of 4 (c) 2 out of 4 (d) 1 out of 4

173. Parents of blood O and AB cannot have a child of group AB because

(a) Gene O is dominant over gene A (b) Gene O is dominant over gene B

(c) Gene A or B is absent in one of the parents

(d) Gene A and B are absent in one of the parents

174. In case of disputed parentage, the blood group analysis of the mother, child and alleged father can

(a) Definitely prove a man to be the father (b) Only prove that he can not be the father

(c) Not be of any use (d) None of the above

175. During serological test in which anti-human serum is mixed with blood of another animal, blood of which animal gives the thickest precipitate

(a) Gibbon (b) Chimpanzee (c) Dog (d) Mule

176. A human female with blood group 'A' has

(a) Antibody-anti-*B* on the red blood cells and antigen *A* in the serum

(b) Antigen *A* on the red blood cells and antibody-anti-*B* in the serum

(c) Antigen *B* on the red blood cells and antibody-anti-*B* in the serum

(d) Antigen *A* on the red blood cells and antibody-anti-A in the serum

177. Biological marriage of one of the following should be avoided **or** After examining the blood groups of a couple, the doctor advised them not to have more than one child. The blood group of the couple are likely to be

(a) Rh^+ male and Rh^- female (b) Rh^+ male and Rh^+ female

(c) Rh^- male and Rh^+ female (d) Rh^- male and Rh^- female

178. How many possible phenotypes are there for ABO blood groups

(a) 4 (b) 6 (c) 8 (d) 16

GENETIC VARIATION

Basic Level

179. Number of the chromosomes in Klinefelter's syndrome is
(a) 44 (b) 47 (c) 45 (d) 46

180. Edward's syndrome, Patau's syndrome and Down's syndrome are due to
(a) Mutation due to malnutrition (b)Change in sex chromosomes
(c) Change in autosomes (d)Change in both sex chromosomes and autosomes

181.A person who is trisomic for twenty first pair of chromosomes is
(a) Klinefelter's syndrome (b)Down's syndrome (c) Turner's syndrome (d) None of the above

182. Turner's syndrome in human is caused by
(a) Autosomal aneuploidy (b)Sex chromosome aneuploidy
(c) Polyploidy (d)Point mutation

183. Which of the following is genetic disease
(a) Phenylketonuria (b) Blindness (c) Cataract (d) Leprosy

184. Webbed neck is characteristic of
(a) XXY male (b) YY male (c) XO female (d) XXX female

185. The number of chromosomes in Down's syndrome is
(a) 23rd pair with one less = 45 (b) 21st pair with one more = 47
(c) 17th pair with one more = 47 (d)One extra sex chromosome = 47

186. A person who has 47 chromosomes due to an extra *Y* chromosome is affected by
(a) Turner's syndrome (b) Klinefelter's syndrome (c)Super female (d) Down's syndrome

187. The exchange of one part of a chromosome to the other part of same or another chromosome is called
(a) Inversion (b) Mutation (c)Translocation (d) Linkage

188. Down's syndrome is
(a) Autosomal abnormality (b)Sex chromosome abnormality
(c)Sex-linked disease (d) None of the above

189. A human with chromosome number 44+*XXY* is male, Such a person suffers from
(a) Turner's syndrome (b) Klinefelter's syndrome
(c) Down's syndrome (d) Patau's syndrome

190. Genotype of a Down's syndrome is
(a) 45+*XX* (b) 44+*XY* (c) 44+*XXY* (d) 22+*XY*

191. Mongolism syndrome is caused by
(a) One extra chromosome (b) One extra sex chromosome
(c) One extra chromosome in 21st pair (d) One less sex chromosome

192. If mother and father both are without pigments (albinism), then
(a) All the offsprings will be albinism (b) Half of the offsprings will be albinism
(c) 75% offsprings will be albinism (d) No offspring will be albinism

193. Sickle cell anaemia is due to

(a) Hormones (b) Viruses (c) Genes (d) Bacteria

194. The cause of Turner's syndrome in man is

(a) Incomplete sex linkage (b) Sex-linked inheritance

(c) Autosomal abnormality (d) Sex-chromosomal abnormality

195. In Klinefelter's syndrome, what is generally the set of sex chromosome

(a) XX (b) XY (c) XXY (d) XYY

196. The condition in which there are more than two complete set of chromosome is called

(a) Polytene (b) Monoploidy (c) Polyploidy (d) Aneuploidy

197. Extra 18^{th} autosomal chromosome result in

(a) Edward syndrome (b) Patau's syndrome (c) Down's syndrome (d) None of the above

198. In Down's syndrome (Mongolism) each cell has how many chromosomes

(a) 21^{st} pair having one less (b)23^{rd} pair with one less

(c)45 (d) 47

199. An abnormal human male phenotype involving an extra *X* chromosome (*XXY*)is a case of

(a) Down's syndrome (b) Intersex

(c) Edward syndrome (d) Klinefelter syndrome

200. The reduction of one pair of chromosome in human is due to

(a) Fusion of two chromosomes into one (b) Elimination

(c) Mutation (d) Reproductive isolation

201. The monosomic condition in human beings depicted as XO is referred to as

(a) Criminal syndrome (b) Down's syndrome

(c) Klinefelter's syndrome (d) Turner's syndrome

202. Edward syndrome is on account of .....

(a) 45 chromosomes instead of 46

(b) Presence of three chromosomes on 18th pair of autosome

(c) Presence of three chromosomes on 21st pair of autosome

(d) Presence of three pair of sex chromosomes

203. Number of sex chromosomes is normal in

(a) Super female (b) Turner's syndrome

(c) Klinefelter's syndrome (d) Down's syndromes

204. Which of the following is not related to chromosomal aberration

(a) Euploidy (b) AIDS

(c) Aneuploidy (d) Klinefelter's syndrome

205. Mongoloid condition is related to **or** In mongolism a patient shows

(a) Monosomy (b) Trisomy (c) Nullisomy (d) None of the above

206. Change in the sequence of nucleotide in DNA is

(a) Mutation (b) Isolation (c) Polyploidy (d) Sexual reproduction

207. Gene mutation is caused by

(a) Reproduction (b) Linkage

(c) Change in the sequence of nitrogenous base (d) Change in the sequence of genes in DNA

208. Discontinuous variation are

(a) Mutation (b) Acquired characters

(c) Essential features (d) Nonessential features

209. The functional unit of mutation is

(a) Gene (b) Muton (c) Recon (d) Cistron

210. Mutation is

(a) Sudden change in morphology (b)Change in characters

(c) Change in heritable characters (d)None of the above

211. Mutations occur

(a) Mainly in haploid cells (b) Mainly in diploid cells

(c) Whenever cells are exposed to X-rays (d) In any cell of the body exposed to radiation

212. The reason of fault in gene duplication is

(a) Transformation (b) Translocation (c) Mutation (d) None of the above

213. To be evolutionary successful, a mutation must be

(a) Germplasm DNA (b) Somatoplasm DNA (c) Cytoplasm (d) RNA

214. Which of the following is the main category of mutation

(a) Genetic mutation (b) Zygotic mutaion (c) Somatic mutaion (d) All of the above

215. Hugo de Vries formulated the "Mutation theory" based on the experiments he conducted on

(a) *Althea rosea* (b) *Pisum sativum*

(c) *Drosophila melanogaster* (d) *Oenothera lamarckiana*

216. Chromosome aberration occurs due to

(a) Aneuploidy (b) Polyploidy (c) Physical effects (d) All the above

217. Trisomy has chromosome complement or

(a) $2n-1$ (b) $2n-1-1$ (c) $2n+1+1$ (d) $2n+1$

218. A strong mutagen is

(a) Cold (b) Heat (c) Water (d) X-ray

219. Mental retardation associated with sex chromosomal abnormality is due to

(a) Reduction in X-complement (b) Increase in X-complement

(c) Moderate increase in Y-complement (d) Large increase in Y-complement

220. Mutation is change that is

(a) Never inherited (b) Inherited only in F_2 generation

(c) Inherited (d) Responsible for plant growth

221. An inborn error of metabolism which eventually affects mental development is

(a) Albinism (b) Phenylketonuria (c) Anaemia (d) Bleeder's disease

222. Monosomics are

(a) n (b) $2n+1$ (c) $2n-2$ (d) $2n-1$

223. Mutation are responsible for

(a) Extinction of organisms (b) Variations in population

(c) Increase in population (d) Maintaining genetic continuity

224. In human beings 45 chromosomes/single X/XO abnormality causes

(a) Down's syndrome (b) Klinefelter's syndrome

(c)Turner's syndrome (d) Edward's syndrome

225. Numerical change in chromosome number which is not be exact multiple of haploid genome is

(a) Triploid (b) Allopolyploid (c) Autopolyploid (d) Aneuploid

226. Mutation are commonly

(a) Dominant (b) Codominant (c) Recessive (d) Incomplete

227. A change in chromosomal number is called

(a) Polyploidy (b) Aneuploidy

(c) Chromosomal mutation (d) Somatic mutation

228. Deletion of certain genes cause

(a) Gene mutation (b) Chromosome mutation (c)Gene modification (d)Aneuploidy

229. Albinism and phenylketonuria are disorders due to

(a) Recessive autosomal genes (b) Dominant autosomal genes

(c) Dominant sex genes (d) Recessive sex genes

230. Name the mutagen

(a) SO_2 (b) CO_2 (c) CO (d) HNO_2

231. Mutation are

(a) Always harmful (b) Rarely useful (c) Mostly useful (d) Always useful

232. Point mutation is

(a) Loss of gene (b) Change in a base of gene

(c) Addition of a gene (d) Deletion of a segment of gene

233. Exact multiple of haploid number is

(a) Euploid (b) Aneuploid (c) Heteroploid (d) Hyperploid

234. Ultimate source of organic variation is

(a) Mutations (b) Natural selection (c) Isolation (d) Hormonal activity

235. Polyploidy means occurrence of

(a) Haploid sets of chromosomes (b) Diploid sets of chromosomes

(c) More than diploid sets of chromosomes (d) All the above

236. Which of the following causes point mutation

(a) Deletion (b) Inversion (c) Transition (d) All the above

237. Individual with Turner's syndrome is

(a) Normal female (b) Normal male

(c) A female with rudimentary ovaries and undeveloped breasts

(d) A male with rudimentary testes and undeveloped penis

238. A supermale has a genetic constitution of

(a) XY (b) XXY (c) XXYY (d) XYY

239. The risk of Down's syndrome offspring is more to mothers at the age of

(a) 20 years (b) 25 years (c) 30 years (d) 35 years

240. The mutation which returns to the original state is called

(a) Reversible mutation (b)Lethal mutation (c) Backward mutation (d) Abnormal mutation

241. The term mutation was coined by

(a) Morgan (b) Beadle and Tatum (c) Hugo de Vries (d) H. J. Muller

242. Mutagens are

(a) The genes which can mutate (b) The organism which show mutation

(c) Genes that regulate mutations (d) The agent which cause mutation

243. Cystic fibrosis is caused by

(a) Recessive autosomal allele (b) Dominant autosomal allele

(c) Recessive sex linked allele (d) Dominant sex linked allele

244. Huntington's disease is

(a) Autosomal dominant disease (b) Autosomal recessive disorder

(c) Sex-linked recessive disorder (d) Sex-linked dominant disease

245. A sudden or spontaneous change in the structure and action of a particular gene is called

(a) Linkage (b) Variation (c) Mutation (d) Allelomorph

246. Aneuploidy is the term applied for the

(a) Gene mutation

(b) Chromosomal mutation

(c) Chromosomal mutation involving the addition or loss of one or more chromosome

(d) Chromosomal mutation involving the addition of one or more complete set of chromosomes

247. What term is applied to the gene mutation where a base is replaced by another base

(a) Duplication (b) Aneuploidy (c) Euploidy (d) Substitution

248. An exchange of segments between the two non-homologous chromosomes is called

(a) Polyploidy (b) Chromosomal aberration (c)Translocation (d) Inversion

249. If a part of a chromosome get separated and reattached in reverse position to the same chromosome, the mutation is called

(a) Inversion (b) Transversion (c) Translocation (d) Gene mutation

250. If a part of chromosome gets detached and lost during the cell division, the mutation so produced is called

(a) Deletion (b) Euploidy (c) Inversion (d) Transcription

251. Huntigton's chorea is characterised by

(a) Incongruant muscle movement

(b) Disordered muscle movement and mental deterioration

(c) Weak eye sight and hearing power (d) Inability to speak

252. A person having Klinefelter's syndrome is chracterised by

(a) Male with some secondary sexual characters of female

(b) Female with some secondary sexual characters of male

(c) Having both male and female sex organs

(d) Female internal sex organs and male external sex organs

253. The child affected with Down's syndrome has

(a) Flattened nasal bridge, open mouth with protruding tongue

(b) Small forehead, bulging eyes and raised nasal bridge

(c) Habitually open mouth with long protruding tongue, bulging eyes and small forehead

(d) Large forehead, raised nasal bridge and long included tongue.

254. In cystic fibrosis there is

(a) Failure of chloride ion transport (b) Mucous clogging of lungs

(c) Defective functioning (d) All the above

255. Albinism in man has been reported in :

(a) Negroes (b) Europeans

(c) Both negroes and europeans (d) None of the above

256. A normal woman whose father was albino marries an albino; what proportion of normal and albino can be expected among their offspring

(a) All albino (b) One normal : one albino

(c)All normal (d) Two normal : one albino

257. Which of the following is not heritable

(a) Point mutation (b) Somatic mutation

(c) Gene mutation (d) Chromosomal mutation

258. Genetic variation arises by

(a) Recombination (b) Mutation (c) Nucleolus (d) Both (a) and (b)

259. Multiple genes are involved in the inheritance of

(a) Colourblindness (b) Phenylketonuria (c) Sickle cell anaemia (d) Skin colour

260. The inheritance of two recessive alleles for sickle cell anaemia is

(a) Epistatic (b) Lethal (c) Both (a) and (b) (d) Pleiotropic

261. Sudden changes appear in the organism due to

(a) Gene recombination (b) Chromosomal aberrations

(c) Spontaneous mutations (d) Polyploidy

262. Variations that involve change in number of chromosomes are

(a) Euploidy (b) Aneuploidy (c) Both (a) and (b) (d) None of the above

263. More specific biological effects are caused by

(a) *X*-rays (b) Gamma rays (c) Alpha particles (d) UV light

264. When one chromosome is lacking in a diploid set it is called

(a) Monosomic (b) Nullisomic (c) Trisomic (d) Pentasomic

265. Mutations which occurs in non-reproductive cells are

(a) Gametic mutation (b) Somatic mutations

(c) Point mutation (d) Chromosomal mutations

266. If a trait is passed from man to all of his sons only

(a) Gene for that trait is dominant (b) Gene is located on any chromosome

(c) Both (a) and (b) (d) None of the above

267. Albinos have pigment absent in

(a) Skin (b) Hairs (c) Eyes (d) All the above

268. Which one is a hereditary diseases

(a) Cataract (b) Blindness (c) Leprosy (d) Phenylketonuria

269. Which of the following is related to the high frequency of Down's syndrome

(a) Maternal age (b) Paternal age (c) Both (a) and (b) (d) None of the above

270. Gaucher's disease is associated with abnormal metabolism of

(a) Fat (b) Nucleic acid (c) Protein (d) Carbohydrate

271.'Epicanthus' is the symptom of

(a) Haploidy (b) Hetroploidy (c) Down's syndrome (d) Turner's syndrome

272. Patients suffering from alkaptonuria have an abnormal compound in urine known as

(a) Uric acid (b) hydrochloric acid (c) Phenylalanine (d) Homogentisic acid

273. Which of the following is a correct match

(a) Haemophilia : *Y*-Chromosome (b)Sickle-cell anaemia : *X*-Chromosom

(c) Down's syndrome : 21st Chromosome (d) Parkinson's disease : *X* and *Y*-Chromosome

274. An example of molecular mutations is

(a) Anaemia (b) Sickle-cell anaemia

(c) Haemophilia (d) Erythroblastosis faetalis

275. Which one of the following is a genetic trait

(a) Thalassemia (b) Grave's disease

c) Cushing's syndrome (d) Parkinson's disease

276. Amyloid β protein deposits damages the brain of patients suffering from

(a) Tay-sachs disease (b) Cystic fibrosis (c) Alzheimer's disease (d) Huntington disease

277. Progressive degeneration of brain cells results from

(a) Cystic fibrosis (b) Marfan syndrome (c) Thalassemia (d) Huntington disease

278. 'Von Recklinghausen disease is another name of

(a) Gaucher's disease (b) Alzheimer's disease (c) Neurofibromatosis (d) Sickle-cell disease

279. Hexosaminidase deficiency results in

(a) Tay-Sachs disease (b) Huntington disease (c) Sickle-cell disease (d) Marfan syndrome

280. Which of the following is a lethal genetic disease due to an autosomal recessive mutation

(a) Haemophilia (b) Cystic fibrosis (c) Neurofibromatosis (d) Huntington disease

281. In humans, Philadelphia chromosome results from the reciprocal translocation between chromosome numbers

(a) 10 and 20 (b) 3 and 11 (c) 9 and 21 (d) 9 and 22

282. Which of the following diseases is due to deletion of chromosome

(a) Down's syndrome (b) Patau's syndrome

(c) Cri-du-chat-syndrome (d) Edward's syndrome

283. Deletion of short arm of chromosome $4(4p-)$ result in

(a) Patau's syndrome (b) Edward's syndrome

(c) Klinefelter's syndrome (d) wolf-Hirschhorn's syndrome

284. Albinism is known to be due to an autosomal recessive mutation. The first child of a couple with normal skin pigmentation was an albino. What is the probability that their second child will also be an albino

(a) 100% (b) 25% (c) 75% (d) 50%

285. Albinism is a congenital disorder resulting from the lack of the enzyme

(a) Catalase (b) Fructokinase (c) Tyrosinase (d) Xenthine oxidase

286. Which one in man is a wholly genetic trait

(a) Diptheria (b) Leucoderma (c) Albinism (d) Tuberculosis

287. Albinism is a

(a) Hereditary disease (b) Deficiency disease (c) Contagious disease (d) Sex linked disease

288. If an albino man marries with a normal woman and 50% offsprings are albino and 50% are normal the woman is

(a) Heterozygous normal (b)Homozygous normal

(c)Heterozygous carrier (d)None of the above

Advance Level

289. A person may have one gene for normal hemoglobin and one gene for sickle cell haemoglobin. This heterozygous condition is called

(a) Genome (b) Anaemia (c) Gene trait (d) Sickle cell trait

290. If haploid chromosome number in a cell is 12, then the monosomic number will be

(a) 25 (b) 22 (c) 26 (d) 23

291. Monosomy and trisomy can be represented as

(a) $2n+1,\ 2n+3$ (b) $2n-1,\ 2n-2$ (c) $2n,\ 2n+1$ (d) $2n-1,\ 2n+1$

292. Phenylketonuria (PKU) is an inherited disease which refers to

(a) Decrease in phenylalanine in tissue and blood

(b) Increase in phenyl pyruvic acid in tissue and blood

(c) Elimination of sugar in urine

(d) Elimination of gentisic acid in urine

293. In man sometime during gametogenesis sex chromosomes are not separated themselves on account of which chromosome number becomes 45, 47 or 48. In this condition which of the following genotype and phenotype is correct

(a) 22 pairs + XXY males

(b)22 pairs + XX females

(c)22 pairs + XXXY females (d) 22 pairs + Y females

294. Down's syndrome is caused by an extra copy of chromosome number 21. What percentage of offspring produced by an affected mother and a normal father would be affected by this disorder

(a) 25% (b) 100% (c) 75% (d) 50%

295. Meta-females have

(a) XX (b) XXO (c) XXXX (d) XXXXXX

296. Symptoms representing a particular disease due to chromosomal abnormalities are referred to

(a) Sex mosaic (b) Syndrome (c) Lethal (d) Pedigree

297. Trisomic condition of Down's syndrome arises due to

(a) Triploidy (b) Translocation

(c) Non-disjunction (d) Dicentric bridge formation

298. Condition of sex chromosomes in a male child of Down's syndrome will be

(a) XY (b) XXY (c) XX (d) XO

299. Persons with the following syndrome have a tendency of tall structure, mental defects and a strong antisocial behavior

(a) XYY syndrome (b) Down's syndrome

(c) Klinefelter's syndrome (d) Turner's syndrome

300. Euploidy is best explained by

(a) Exact multiples of a haploid set of chromosomes

(b) One chromosome less than the haploid set of chromosome

(c) One chromosome more than the haploid set of chromosomes

(d) One chromosome more than the diploid set of chromosomes

301. Sometimes chromosome number increase or decrease due to

(a) Non-disjunction of chromosome (b)Genetic repeate

(c) Mutation (d) All of the above

302. If a diploid cell is treated with colchicine, then it becomes

(a) Tetraploid (b) Diploid (c) Triploid (d) Monoploid

303. The formation of multivalents at meiosis in diploid organism is due to

(a) Monosomy (b) Inversion

(c) Deletion (d) Reciprocal translocation

304. Alkaptonuria is caused due to

(a) Dominant autosomal gene (b) Recessive autosomal gene

(c) X linked recessive gene (d) X linked dominant gene

305. What would be the number of chromosome in the ovum (Fertilized by a normal sperm) that resulted in the appearance of Klinefelter's syndrome in the offspring

(a) 23 (b) 22 (c) 21 (d) 24

306. In *Drosophila melanogaster*, 3 pairs of autosomes and sex chromosomes of XO type give rise to a normal male in appearance. In man, 22 pairs of autosomes and XO type of sex chromosomes form

(a) Normal female (b) Normal male

(c) Klinefelter's syndrome (d) Turner's syndrome

307. In Turner's syndrome, people are

(a) Externally females, chromatin negative (b) Externally males, chromatin positive

(c) Externally males, chromatin negative (d) Externally females, chromatin positive

308. In Klinefelter syndrome, people are

(a) Externally females, chromatin positive (b) Externally females, chromatin negative

(c) Externally males, chromatin positive (d) Externally males, chromatin negative

309. Match list I with list II and select the correct answer using code given below

List I (syndrome)

(1) Patau's syndrome

(2) Klinefelter's syndrome

(3) Down's syndrome

(4) Turner's syndrome

List II (Chromosomal abnormality)

(A) $44 + XXY = 47$

(B) $44 + X = 45$

(C) $46 + 1 = 47$ Chromosome 13^{th}

(D) $46 + 1 = 47$ Chromosome 21^{st}

Code

(a) $\begin{matrix}1&2&3&4\\A&B&C&D\end{matrix}$ (b) $\begin{matrix}1&2&3&4\\D&C&B&A\end{matrix}$ (c) $\begin{matrix}1&2&3&4\\C&B&D&A\end{matrix}$ (d) $\begin{matrix}1&2&3&4\\C&A&D&B\end{matrix}$

310. "Philadelphia chromosome" is found in the patient suffering from

(a) Insomnia (b) Leukaemia (c) Hepatitis (d) Albinism

311. The genotype of a boy having sexual characters of a girl is

(a) XXX (b) XXY (c) XO (d) XYY

312. Disorders of amino acid metabolism results in

(a) Alkaptonuria (b) Phenylketonuria (c) Albinism (d) All of the above

313. The concept of sudden genetic change which breeds true in an organism is visualised in the principle of

(a) Natural selection (b) Heredity (c) Variations (d) Mutations

314. The most striking example of point mutation is found in a diseases called

(a) Night blindness (b) Thalassemia (c) Down's syndrome (d) Sickle-cell anaemia

315. Normally DNA molecule has A-T, G-C pairing. However, these bases can exist in alternative valency status, owing to rearrangements called

(a) Point mutation (b) Analogue substitution

(c) Frame-shift mutation (d) Tautomerisational mutation

316. The process of genetic mutation is

(a) Reversible (b) Irreversible (c) Partially reversible (d) Continuous

317. Which of the following discoveries resulted in a Nobel Prize

(a) Genetic engineering

(b) *X*-rays induce sex-linked recessive lethal mutations

(c) Cytoplasmic inheritance (d) Recombination of linked genes

318. Identify the following point mutations in mRNA: UAU ACC UAU to UAU AAC CUA and UUG CUA AUA to UUG CUG AUA

(a) Transition and frame shift respectively (b) Transversion and frame shift respectively

(c) Frame shift and transversion respectively (d) Frame shift and transition respectively

319. Mutations are responsible for

(a) Increasing the population rate (b) Variations in organisms

(c) Constancy in organisms (d) For beneficial changes in organisms

320. Which one of the following is not a mutagen

(a) 5-bromouracil (b) Acetic acid (c) Nitrous acid (d) Gamma radiation

321. The frequency of a mutant gene in a population is expected to increase, if the gene is

(a) Recessive (b) Dominant (c) Sex linked (d) Favourably selected

322. Mutation rates are affected by

(a) Temperature (b) *X*-rays

(c) Gamma and beta radiation (d) All of the above

323. Transition type of gene mutation is caused when

(a) GC is replaced by TA (b)CG is replaced by GC

(c)AT is replaced by CG (d)AT is replaced by GC

324. The reason why some mutations which are harmful do not get eliminated from the gene pool, is that

(a) They have future survival value

(b)They are recessive and carried by heterozygous individuals

(c) They are dominant and show up more frequently

(d) Genetic drift occurs because of a small population area

325. In man the mutation disease, aniridia (Congenital absence of iris) occurs due to

(a) Dominant mutation (b) Recessive mutation (c) Lethal mutation (d) Iso-alleles

326. The point mutations *A* to *G*, *C* to *T*, *C* to *G* and *T* to *A* in DNA are

(a) Transition, transition, transversion and transversion respectively

(b) Transition, transversion, transition and transversion respectively

(c) Transversion, transversion, transition and transition respectively

(d) All four are transition

327. Which one of the following mutation partially or fully reverses the harmful effects of previous mutation

(a) Indirect suppression (b) Intergenic mutation

(c) Intragenic mutation (d) Suppressor mutations

328. Muller was awarded Nobel prize in 1946 for his work on

(a) Protein synthesis (b) Chemistry of nucleic acids

(c) Cancer (d) *X*-ray induced mutations

329. Hyperchromism is presence of

(a) Some chromosome more than once (b) Some type of chromosome less than once

(c) Variable chromosomes in nucleus (d) None of the above

330. Mutation refers to sudden change in

(a) Phenotype (b) Maturation time (c) metabolic rate (d) Genetic make up

331. Match the columns

Column I	Column II
a Down's syndrome	*p* An additional sex chromosome
b Cri-du-chat syndrome	*q* Loss of a part of chromosome 5
c Klinefelter's syndrome	*r* Absence of sex chromosome
d Turner's syndrome	*s* Presence of an extra chromosome
	t Presence of two extra chromosomes

(a) $a-s, b-q, c-p, d-r$ (b) $a-t, b-s, c-p, d-q$ (c) $a-s, b-q, c-q, d-r$ (d) $a-s, b-q, c-r, d-p$

332. Mutation caused by a mutagen is

(a) Induced (b) Natural (c) Spontaneous (d) Chemical mutation

333. *X*-rays cause mutation by

(a) Transition (b) Transversion (c) Deletion (d) Base substitution

334. The gene that controls the rate of mutation of another gene is

(a) Regular gene (b) Inducer gene (c) Mutable gene (d) Mutator gene

335. Huntington's chorea is

(a) Common in Korea

(b) Nervous degeneration causing involuntary shaking of legs, arms, head

(c) Disease of kidney

(d) Related to diabetes

336. Replacement of a purine with another purine or a pyrimidine with another pyrimidine is

(a) Transition (b) Transversion (c) Insertion (d) Deletion

337. A point mutation comprising substitution of purine with pyrimidine is

(a) Transition (b) Transversion (c) Deletion (d) Translocation

338. Frame shift mutation occurs when

(a) Base is deleted or added (b) Base is added

(c) Base is deleted (d) Anticodons are not present

339. In gene mutation, adenine is replaced by guanine. It is known as

(a) Substitution (b) Point mutation (c) Transition (d) Transversion

340. Nullisomy is the term used for the condition when an organism has

(a) An additional chromosome (b) One chromosome less than normal

(c) A complete set of chromosomes except one homologous pair

(d) None of the above

341. Which one of the following is responsible for mental abnormalities in humans

(a) XXX and XY (b) XX and XXX (c) XO and XXX (d) XX and XO

342. Down's syndrome is due to trisomy of 21^{st} chromosome caused by

(a) Nondisjunction during egg formation (b) Nondisjunction during sperm formation

(c) Addition of extra chromosome during mitosis of zygote

(d) Either (a) or (b)

343. Rearrangement of genes occurs due to

(a) Translocation and duplication (b)Translocation and deficiency

(c) Deletion and deficiency (d) Translocation and inversion

344. One of the following is a random process

(a) Variations (b) Adaptations (c) Evolution (d) Mutations

345. Haploid number is 10. What shall be tetrasomic number

(a) 22 (b) 40 (c) 20 (d) 19

346. Recessive mutation is not expressed in

(a) Homozygous male (b) Heterozygous male

(c) Heterozygous female (d) Homozygous female

347. Fertilization between *22A + XX* egg and *22A +Y* sperm will result in
(a) Down's syndrome (b) Patau's syndrome
(c) Turner's syndrome (d) Klinefelter's syndrome

348. Marfan's syndrome is characterised by
(a) Dislocation of eye lens
(b) Hypermobility of joints
(c) Slender body, elevated limbs and susceptibility to heart diseases
(d) All the above

349. Allele for cystic fibrosis occurs over
(a) 21 Chromosome (b) 14 Chromosome (c) 7 Chromosome (d) 4 chromosome

350. Gene of sickle cell anaemia is carried by
(a) Sex cell (b) Sex chromosome (c) Autosome (d) Bone cell

351. The possibilities of the hereditary and evolutionary changes are greatest in the species, which are reproduced by
(a) Fission (b) Budding (c) Asexual means (d) sexual means

352. Pleiotropy is the term that refers to the situation in which a gene influences more than one trait. Its example is given by
(a) Sickle cell anaemia (b) Haemophilia (c) Colour blindness (d) Only (a) and (b)

353. Which one of the following conditions though harmful in itself, is also a potential saviour from a mosquito borne infectious disease
(a) Pernicious anaemia (b) Leukaemia (c) Sickle-cell anaemia (d) Thalassemia

354. Tay-Sach's disease is an example of
(a) Dominant *X*-linked trait (b) Autosomal recessive trait
(c) Autosomal dominant trait (d) Recessive sex-linked trait

355. 'Philadelphia chromosome' is reported from the patients suffering from
(a) PKU (b) CML (c) Liver dysfunction (d) Kidney stone

356. In sickle-cell anaemia, which of the following amino acids is substituted
(a) Glutamic acid by valine in β-chain (b) Valine by glutamic acid in β-chain
(c) Glutamic acid by valine in α-chain (d) Valine by glutamic acid in α-chain

357. In man albinism is due to non-synthesis of melanin in the absence of
(a) Lysine (b) Tyrosinase (c) Luciferase (d) Melanase

358. An albino lady marries a normal man having one gene for albinism. They have 3 daughters and 1 son. The son will be
(a) Normal
(b)Either normal or albino
(c) Half albino half normal

(d)Normal in childhood but turns albino on becoming adult

359. Knowing that albinism is determined by a recessive gene in man, presence of albinism in children born to a couple proves that

(a) Both the father and the mother are heterozygous for albinism

(b) The father is homozygous normal but the mother is heterozygous or vice versa

(c) The father is homozygous for albinism but the mother is heterozygous or vice versa

(d) (a) and (c) are correct

SEX DETERMINATION

Basic Level

360. In man, sperms contain autosomes and

(a) Only Y chromosome (b)Only X chromosome

(c) Both X and Y chromosomes (d) Either X or Y chromosomes

361. Sex of the unborn mammal can be predicted by

(a) Placental biopsy (b) Examining the chorion

(c) Amniocentesis (d) Examining the mother's blood

362. A family has five girls and no son, probability of son as the 6th child will be

(a) 50% (b) 75% (c) Full (d) No chance

363. The average ratio of male to female individuals based on XX and XY type of sex determination, in total world of human population is

(a) 3 : 1 (b) 1 : 3 (c) 1 : 4 (d) 1 : 1

364. Total number of autosomes in fertilized egg of man is

(a) 44 (b) 22 (c) 23 (d) 46

365. In man, the composition of female destined zygote is

(a) 22 + *X* (b) 44 + *XX* (c) 22 + *Y* (d) 44 + *XY*

366. An unfertilized human egg contains

(a) Two X chromosomes (b)One X and Y chromosome

(c) One Y chromosome only (d) One X chromosome only

367. A male child would be born to parents if

(a) Father is healthier than mother

(b) Genetic chromosomal constitution of child is *XX*

(c) Mother feeds well during pregnancy

(d) Genetic chromosomal constitution of child is *XY*

368. *Drosphila* flies with one half of the body male and other half female is referred to as

(a) Gynandromorph (b) Hermaphrodite (c) Super female (d) Intersex

369. Male child will be born if

(a) Father is sexually more excited

(b)Sperm of male with *Y* chromosome fertilized the egg

(c) Sperm of male with *X* chromosome fertilized the egg (d)None of the above

370. Genic balance theory was proposed by
(a) Bridges (b) Morgan (c) Bovery (d) Lyon

371. The formation of a male child depends on the sperms because
(a) Sperms may be X and Y (b) Sperms are all Y
(c) The eggs from the other ovary may be Y (d) Sperms are more active

372. In human chromosomal condition of male is
(a) 44 AA + XO (b) 44 AA + XX (c) 44 AA + XY (d) 44 AA + XXY

373. Genetic identity of a human male is determined by
(a) Autosome (b) Nucleolus (c) Sex chromosome (d) Cell organelles

374. How many chromosomes are present in an unfertilized egg
(a) 22 autosomes and one sex chromosome
(b) 22 pairs of autosomes and one sex chromosome
(c) 44 autosomes and one pair of sex chromosome (d)Total 22 chromosomes

375. The male human is represent by sex chromosomes
(a) XX (b) XO (c) XY (d) YY

376. Sex of a human child is determined by
(a) Size of the egg at the time of fertilization (b) Size of sperm at the time of fertilization
(c) Sex chromosome of father (d) Sex chromosome of mother

377. The sex determination pattern in honeybee is called
(a) Female haploidy (b) Haplodiploidy (c) Gametic diploidy (d) Gametogony

378. Free martin condition is found in
(a) Man (b) Sheep, goat etc. (c) Rabbit (d) Frog

379. The chromosome in females are
(a) XX (b) XY (c) YY (d) XXY

380. The chromosomes responsible for the determination of sex are called
(a) Autosomes (b) Allosomes (c) Multiple alleles (d) Heterosis

381. In human zygote, the male sex is determined by whether
(a) Mother gets good nutrition (b) Father is stronger than mother
(c) Strength of male chromosome (d) The presence of Y chromosome

382. Determination of sex of a child depends upon
(a) Nature of sperm (b) Nature of egg (c) Health of father (d) Age of mother

383. The theory where ratio between the number of X-chromosomes and number of complete sets of autosomes will determine the sex is known as
(a) Chromosome theory of sex determination
(b) Genic balance theory of sex determination
(c) Hormonal balance theory of sex determination
(d) Environmental sex determination theory

384. There are five daughters and no son in a family. It may be due to
(a) Father produced only X containing sperms (b) Father produced no sperms at all
(c) Y type sperms are weaker and not effective
(d) By chance each time X sperms fertilized the egg

385. Free martin is an example of
(a) Hormonal control of sex (b) Sex reversal by gene
(c) Environmental control of sex (d) None of the above

386. The theory relevant to organism possessing inherited potential to transform male to female is
(a) Chromosomal theory (b) Genic balance theory
(c) Chromosomal and gene theory (d) Hormonal theory

387. Lethal genes are those which
(a) Cause death of the individual in which they are present
(b) Cause death of homozygous infant being formed
(c) Cause death of heterozygous infant being formed
(d)None of the above

388. Recessive character are expressed
(a) Only when they are present on X-chromosomes of male
(b) Only when they are present on both the X-chromosomes of female
(c) No any autosome (d)On one the chromosomes of female

389. Hinny and mule are the example of
(a) Test cross (b) Dihybrid cross (c) Back cross (d) Reciprocal cross

390. A fruitfly exhibiting both male and female traits is
(a) Heterozygous (b) Gynandromorph (c) Hemizygous (d) Gynander

391. What is the true in case of Honey Bee
(a) Male diploid, female haploid (b)Male diploid, female diploid
(c) Male haploid, female haploid (d)Male haploid , female diploid

392. An autosome is
(a) Chromosome half (b) Sex Chromosome
(c) Chromosome other than sex (d) None of the above

393. In human zygote male sex is determined by
(a) Strength of father (b) Nutrition of mother
(c) Composition of required chromosome pair (d) None of the above

394. Genetic identity of human male is known by
(a) Nucleolus (b) Cell organelles (c) Autosomes (d) Sex chromosomes

395. In humans, sex is determined by
(a) Y- chromosome (b) X- chromosome
(c) A and X-chromosomes (d) A and Y-chromosomes

396. Allosomes are the name of

(a) Sex chromosomes

(b) Swellings on the chromosome

(c) Chromosomes other than the ones which determine sex organising regions of chromosomes

(d) Nucleolus

397. Theory of heterogamesis for sex determination was proposed by

(a) Morgan (b) Darwin (c) Correns (d) Bridges

398. Which one is homogametic

(a) Human child (b) Human embryo (c) Human male (d) Human female

399. An animal where male carries half the chromosomes present in female is

(a) *Amoeba* (b) *Gorilla* (c) Honey Bee (d) Geometrid Moth

400. Sex is determined in human beings

(a) By ovum (b) At time of fertilization

(c) 40 days after fertilization (d) Before fertilization

401. Barr body in mammals represents

(a) All the hetero chromatin in female cell

(b) One of the two X chromosomes in somatic cells of female

(c) All the hetero chromatin in male and female cells

(d)The chromosome in somatic cells of male

402. Lyon hypothesis deals with

(a) Centromere position (b)Genetic compatibility

(c)Genetic incompatibility (d)Number of barr bodies

403. Barr body and drumsticks are of what significance of geneticists and biologists

(a) They indicate the presence of abnormal sex cells

(b) They indicate the presence of more than one X chromosome in the cells

(c) They indicate male cells

(d) They signify the presence of sex linked traits

404. Barr bodies (seem in saliva test in Olympic games) are found in human and are associated with

(a) Male autosome (b) Female autosome

(c) Female sex chromosome (d) Male sex chromosome

405. Barr bodies are

(a) Chromatin negative (b) Not influenced by stains (c)Chromatin positive (d)Poorly staining

406. How many barr bodies are there in XXY

(a) One (b) Two (c) Three (d) Four

407. Sex chromosomes are found in the cells of :

(a) Testes (b) Ovaries (c) Kidney and liver (d) All of the above

408. The number of Y chromatin corresponds to :

(a) Number of X-chromosomes (b) Number of Y-chromosomes

(c) One less than number of X-chromosomes (d) One more than number of X-chromosomes

409. Sex determining chromosomes are called :

(a) Heterosomes (b) Autosomes (c) Centrosomes (d) Spherosomes

410. Barr body is found in :

(a) Male somatic cells (b) Male germinal cells

(c) Female somatic cells (d) Female germinal cells

411. According to Lyon hypothesis, one of the two X-chromosomes in each female somatic cell is known as

(a) Barr body (b) Karyotypic body (c) Phenotypic body (d) Genotypic body

412. Balance theory of sex determination holds good for :

(a) Humans (b) *Drosophila* (c) Grasshoppers (d) *Allium cepa*

413. In *Drosophila,* the sex is determined by :

(a) The ratio of pairs of X-chromosomes to the pairs of autosomes

(b) Whether the egg is fertilized or develops parthenogenetically

(c) The ratio of number of X-chromosomes to the sets of autosomes

(d) X and Y-chromosomes

414. Super male and super female type of determination of sex in *Drosophila* is based on :

(a) Biodiversity (b) Genic balance (c) Uniformity (d) Oxygen balance

415. According to genic balance theory, $\frac{X}{A}$ =1.5 *Drosophila* individual will be :

(a) Male (b) Female (c) Intersex (d) Super female

416. If somatic cells of a human male contain single Barr body, the genetic composition of the persons would be

(a) XYY (b) XXY (c) XO (d) XXXY

417. A female with XXXX chromosome will have the following number of Barr bodies

(a) One (b) Two (c) Three (d) Four

Advance Level

418. For determination of sex, body cells are taken from

(a) Buccal epithelium (b) Buccal epithelium and root of hair

(c) Gonads (d) Root of hairs

419. Gynandromorphs develop in *Drosophila* when the two cells in the two-celled proembryo will have one of the following chromosomal sets

(a) $2A + XX$ in one cell and $2A + X$ in the other (b) $2A + X$ in both the cells

(c) $2A + XXX$ in both the cells (d) All of the above

420. Loss of a *X* chromosome in a particular cell during its development, results into

(a) Diploid individual (b) Triploid individual (c) Gynandromorphs (d) (a) and (b) both

421. The male of grasshoppers and moths posses two sets of autosomes and
(a) X and Y chromosomes (b) Only X chromosome
(c) Only Y chromosome (d) Neither X nor Y chromosome

422. Mule is an offspring of
(a) Male and female donkey (b) Cow and ox
(c) Male ass and mare (d) Male horse and female ass

423. Animal which remains male initially, then changes to female (*Tapeworm proglottides*) is called
(a) Protandrous (b) Apomixis (c) Profixation (d) None of the above

424. In melandrium the sex determination type is
(a) XX-XY type (b) XX-XO type (c) ZZ-ZW type (d) XY-XO type

425. In human beings XX-genotype is
(a) Always female (b) Can be male or female
(c) Always male (d)Commonly female with 5 per lakh being male.

426. Frequency of XY-genotype being female in human beings is
(a) 1.0% (b) 0.1% (c) 0.001% (d) 0.00001%

427. In crocodiles male sex is predominant at
(a) Low temperature (b) Intermediate temperature
(c) High temperature (d) Ratio of genetically determined

428. In common Turtles the females are predominant at
(a) Below 28°C (b) Above 33°C
(c) Between 28°-33°C (d) Sex is genetically determined

429. The males of roundworms have
(a) One Y-chromosome (b) One chromosome less than female
(c) Two similar sex chromosomes (d) Distinct sex chromosomes

430. XX-XO sex chromosome complement occurs in
(a) Cockroach (b) Honey Bee (c) Human beings (d) Chimpanzee

431. Role of chromosomes in sex determination was proposed by
(a) Sutton and Boveri (b) Henking (c) Mc Clung (d) Morgan

432. Chromosome theory of XY sex determination was proposed by
(a) Henking (b) Wilson and Stevens (c) Johannsen (d) Punnet

433. In *Drosophila* the XXY constitution determines
(a) Maleness (b) Femaleness (c) Intersex (d) Both A and C

434. Which chromosome set is found in male Grasshopper
(a) XY (b) X (c) YY (d) XX

435. Larva of *Bonellia* settling near proboscis of adult female develops into male due to
(a) Substances secreted by proboscis (b) Electrolytes in water
(c) Oxygen in environment (d) Carbon dioxide in environment

436. According to genic balance theory, $\frac{X}{A} = 1.5$ will make the individual
(a) Male (b) Metafemale (c) Intersex (d) None of the above

437. In *Drosophila*, XXY is female. In human it represents an abnormal male because
(a) Y-chromosome induces male traits in humans
(b) Y-chromosome is essential for female sex-in *Drosophila*
(c) Y-chromosomes is not essential for male sex in humans (d) None of the above

438. Probability of all four sons to a couple is
(a) 1/4 (b) 1/8 (c) 1/16 (d) 1/32

439. XY sex chromosomes were discovered by
(a) Gregor Johann Mendel (b)M.J.D. White (c) Nettie Stevens (d) Robert Brown

440. *Drosophila melanogaster* possesses
(a) 3 pairs autosomes + 1 pair sex chromosomes
(b) 2 pairs autosomes + 2 pairs sex chromosomes
(c) 1 pair autosomes + 3 pairs sex chromosomes
(d) 2 pair autosomes + 1 pair sex chromosomes

441. Male XX and female XY develop sometimes due to
(a) Hormonal imbalance (b) Aneuploidy
(c) Deletion (d) Transfer of segment between X and Y

442. Barr body is absent in normal female in
(a) Skin cells (b) Leucocytes (c) Oogonia (d) Secretory cells

443. The males of grasshoppers and bugs possess two sets of autosomes and :
(a) Only Y-chromosome (b) Only X-chromosome
(c) X and Y-chromosome (d) Neither X nor Y- chromosome

444. A pregnant woman who has done amniocentesis test finds an extra Barr body in her embryo. The syndrome which is likely to be associated with the embryo is :
(a) Edward's syndrome (b) Down's syndrome (c) Klinefelter's syndrome (d)Patau's syndrome

445. A certain human abnormal individual showing two Barr bodies in somatic cells would be :
(a) Only a male with one X-chromosome
(b) Only a female with two X-chromosomes
(c) Only a male having two X-chromosomes
(d) Either a male or a female having three X-chromosomes

446. In recent years, DNA sequence (nucleotide sequence) of mt-DNA and Y-chromosome were considered for the study of human evolution, because:

(a) Their structure is known in greater detail

(b) They can be studied from the samples of fossil remains

(c) They are small, and therefore, easy to study

(d) They are uniparental in origin and do not take part in recombination

447. Match List I and List II and select the correct answer using the codes given below the list :

List I (peculiarity of male-determining sperm)		**List II** (Organism in which it is seen)
(A)	No sperm is needed at all	(1) Grasshopper
(B)	Necessarily with a Y-chromosome	(2) Honeybee
(C)	With haploid set of autosomes	(3) Birds
(D)	With W-chromosome	(4) *Drosophila*
		(5) Human

Answer codes :

(a) A = 2, B = 1, C = 3, D = 4 (b)A = 5, B = 2, C = 4, D = 3

(c) A = 3, B = 5, C = 1, D = 4 (d)A = 2, B = 5, C = 1, D = 3

448. In human female, Barr bodies are formed by

(a) Inactivation of mother's *X* chromosome (b) Inactivation of father's *X* chromosomes

(c) Inactivation of both mother's and father's *X* chromosomes

(d) Inactivation of either mother's or father's *X* chromosome

449. A medical technician while observing a human blood smear under the microscope notes the presence of Barr body close to the nuclear membrane in the WBC. This indicates that persons under investigation is **[Kerala CET (Med.) 2003]**

(a) Colour blind (b) Haemophilic (c) Normal female (d) Normal male

450. In the buccal cavity slide cell of an individual we get a dark stained body near nuclear membrane. The genotype of the individual may be **[DPMT 1983]**

(a) XX (b) XY (c) XYY (d) XXX

451. The number of Barr bodies in Turner syndrome is

(a) 0 (b) 1 (c) 2 (d) 3

452. A woman has a child with klinefelter's syndrome. Number of barr bodies present in the child is **[Har. PMT**

(a) One (b) Two (c) Three (d) None of the above

453. A normal woman whose father was albino marries a man who is albino. What proportion of normal and albino can be expected among their offsprings **[CBSE PMT 1994]**

(a) 1 normal : 1 albino (b) All albino (c) 2 normal : 1 albino (d) All normal

Basic Level

454. If a normal woman marries a colourblind man, their **[KCET 1994; MP PAT 1995; MP PMT 2002]**

(a) All son will be colourblind and daughters normal (b) All daughters will be colourblind and sons normal

(c) All children will be normal (d) All children will be colourblind

455. A haemophilic man marries a normal homozygous woman. What will be the probability of his sons to be haemophilic **[CPMT 1999]**

(a) 0% (b) 25% (c) 100% (d) 50%

456. The genes, which are confined to differential region of *Y* chromosome only, are called **[AIIMS 1998; MP PMT 2000; CBSE PMT 1994; CPMT 2003; MP PAT 1995]**

(a) Mutant (b) Autosomal (c) Holandric (d) Completely sex-linked

457. Usually the recessive character is expressed only when present in a double recessive condition. However, a single recessive gene can express itself in human beings when the gene is present on **[AIIMS 19**

(a) Any autosome (b) X chromosome of female

(c) X chromosome of male (d) Either on autosome or X chromosome

458. If a character is always transmitted directly from a father to all his sons and from their sons to all their sons, then which chromosome carries the gene for the character **[MP PMT 1997, 2000]**

(a) Autosomes (b) X chromosome (c) Y chromosome (d) None of the above

459. A colurblind daughter is born when **[MP PMT 1998; KCET 2002]**

(a) Father is colourblind, mother is normal (b) Mother is colourblind, father is normal

(c) Mother is carrier, father is normal (d) Mother is carrier, father is colourblind

460. Which disease is genetically linked **[MP PMT 1999]**

(a) Haemophilia (b) Dysentery (c) Plague (d) Tuberculosis

461. Genes for colour blindness in man are located on **[CPMT 1979, 84; MP PMT 2003]**

(a) X chromosome only (b) Y chromosome only (c) Either X or Y chromosome (d) Both X and Y chromosome

462. If a colour blind lady marries a normal man, their children will be **[BHU 1986; CBSE PMT 1994, 99; MP**

(a) Normal daughters and normal sons (b) Normal sons and carrier daughters

(c) Colour blind sons and carrier daughters (d) Colour blind sons and colour blind daughters

463. Which one is hereditary trait **[CPMT 1981]**

(a) Night blindness (b) Baldness (c) Colour blindness (d) Beri-beri

464. The traits control by the genes located on X chromosome of human beings are said to be **[CPMT 1981; AF**

(a) Sex linked (b) Sex influenced (c) Sex limited (d) None of the above

465. Sex linked disease is

[CPMT 1978, 82, 87, 90, 95, 99 2003 AFMC 1985,96, 2001; BHU 1984, 85, 86; Pb PMT 2000; J&K CE MP PMT 1994, DPMT 1985]

(a) Haemophilia (b) Colourblindness (c) Sickle-cell anaemia (d) Both (a) and (b)

466. Doctor's son **[CPMT 1972]**

(a) Will always be a doctor (b) Will never be a doctor (c) Can be a doctor (d) Will not be a doctor at all

467. The character of organism is said to be sex linked when its gene is carried on **[CPMT 1982]**

(a) Y chromosomes (b) X chromosome of male or female

(c) X and Y chromosomes (d) A particular autosome

468. Haemophilia is a disease which is **[CPMT 1975, 78, 82]**

(a) Hereditary and sex linked (b) Caused by deficiency of calcium

(c) Caused by deficiency of blood (d) None of the above

469. Red green colour blindness appears due to **[CPMT 1971; DPMT 1982]**

(a) Excessive drinking of alcohol (b) Inheritance through X chromosome

(c) Over activity of adrenal (d) Vitamin A deficiency

470. In which of the following colour blindness is inherited **[MP PMT 2000]**

(a) In males only (b) In female only (c) In both males and females (d) None of the above

471. A black dog heterozygous for the colour is crossed with white bitch, recessive homozygous. Progeny will show black to white offsprings in the ratio of **[DPMT 1983]**

(a) All black (b) All albino (c) 1 : 1 (d) 3 : 1

472. In human beings, sex is determined **[MP PMT 1990]**

(a) Before fertilization of ovum

(b) During 6th week of foetal life when androgens are produced

(c) At the time of fertilization of ovum

(d) During 7th and 8th weeks of foetal life when gonads differentiate into testis and ovary

473. Multiple genes are involved in the inheritance of **[CBSE PMT 1999]**

(a) Colourblindness (b) Phenylketonuria (c) Sickle-cell anaemia (d) Skin colour

474. If a boy's father has haemophilia and his mother has one gene for haemophilia; what is the chance that the boy will inherit the disease

[CPMT 1980, 2000; DPMT 1992; AIIMS 1999; Pb PMT 1999]

(a) 25% (b) 50% (c) 75% (d) 100%

475. In human the inheritance of sex linkage takes place through **[MP PMT 2003]**

(a) Autosome (b) Y- chromosome (c) X- chromosome (d) Both (b) and (c)

476. A man receives his *X* chromosomes from

(a) His mother only (b) His father only

(c) Both his mother and father and his father (d) Either his mother

477. Given is : $\bar{x}$ is the chromosome with gene for haemophilia and X is the chromosome with normal gene, Which of the following individual will act carrier for haemophilia **[MP PMT 1992]**

(a) XY (b) $\bar{x}Y$ (c) $\bar{x}\bar{x}$ (d) $\bar{x}X$

478. The female children of a haemophilic man and a carrier woman are likely to be **[MP PMT 1992]**

(a) All haemophilic (b) Half haemophilic and half carriers

(c) All carriers (d) Half normal and half carriers

479. The daughter born to haemophilic father and normal mother could be **[AIIMS 1992]**

(a) Normal (b) Carrier (c) Haemophilic (d) None

480. What is the cause of haemophilia **[MP PMT 1998]**

(a) Chromosomal aberration (b) Somatic mutation (c) *X*-linked mutation (d) All the above

481. A man who carries a sex linked gene on his 'Y' chromosome will transmit this gene to **[NCERT 1977]**

(a) Half of his sons (b) Half of his daughters (c) All his sons (d) All his daughters

482. Female rarely experience the physiologic defect of haemophilia because they do so only when they are **[MP PMT 1990]**

(a) Heterozygous for the defect (b) Homozygous for the defect

(c) Carrier for the defect (d) Wives of haemophilic husbands

483. A pleiotropic gene is one which

(a) Affects one character (b) Affects more than one characters

(c) (a) and (b) both (d) None of the above

484. Example of quantitative inheritance is **[CPMT 1995]**

(a) Colour of skin (b) Colourblindness (c) Klinefelter's syndrome (d) Alkaptonuria

485. Haemophilic man marries a normal woman. Their offsprings will be **[CBSE PMT 1999; MP PMT 1993, 9**

(a) All girls haemophillic (b) All normal (c) All haemophillic (d) All boys

486. If a haemophilic man marries a woman carrier (heterozygous) for haemophilia, what would be the possibility that their daughter would be haemophilic **[BHU 1990]**

(a) 100% (b) 75% (c) 50% (d) 0%

487. Persons who are colour blind cannot distinguish **[KCET 2000]**

(a) Red and green (b) Black and yellow (c) Green and blue (d) Yellow and white

488. Colour blindness is caused by a single **[CPMT 1990]**

(a) Dominant gene in woman (b) Dominant gene in man (c) Recessive gene in man (d) Recessive gene in woman

489. In human beings, the colour of skin is controlled by **[BHU 1983; KCET 2002]**

(a) Multiple alleles (b) Lethal genes (c) Polygenic effect (d) None of the above

490. Colour blindness in man is **[NCERT 1976, 79]**

(a) Due to deficiency of vitamin A (b) Due to absence of visual purple of retina

(c) Due to absence of rods in retina (d) A sex linked abnormality

491. Sex linked inheritance was discovered by **[CPMT 1990]**

(a) Mc Clung (b) Mendel (c) Landsteiner (d) Morgan

492. One of the following is not true to haemophilia **[KCET 2003]**

(a) Royal disease (b) Bleeder's disease (c) X-linked disease (d) Y-linked disease

493. Haemophilia is **[CPMT 1992, 93]**

(a) Autosomal (b) *Y*-linked (c) *Z*-linked (d) *X*-linked

494. If mother is carrier for colour blindness and father is normal, then in the offsprings this disease may be seen in **[CPMT 1992; DPMT 1993; CBSE PMT 1999]**

(a) All the sons (b) All the daughters

(c) 50% sons and 50% daughters (carrier) (d) All the sons and not in daughters

495. Which one of the following diseases belongs to the same category as colourblindness in man **[DPMT 1986;**

(a) Nightblindness (b) Presbyopia (c) Diabetes incipidus (d) Haemophilia

496. Which one is ineffective against antibiotics **[NCERT 1976]**

(a) Bacterial infected wound (b) Bacterial infected throat

(c) Haemophilia (d) Bacterial infected gonorrhoea

497. When a single gene influences more than one trait it is called **[CBSE PMT 1998]**

(a) Pleiotrophy (b) Epistasis (c) Pseudo dominance (d) None of the above

498. Sex linked characters are generally **[CPMT 1980; DPMT 1985]**

(a) Lethal (b) Recessive (c) Dominant (d) Not inherited

499. Haemophilia is a genetic disorder in which **[AFMC 1998]**

(a) Blood clots in blood vessels (b) There is delayed coagulation of blood

(c) Blood is coagulate (d) Blood cell count falls

500. Blood does not stop coming out of wound in **[AIIMS 2000]**

(a) Tetanus (b) Malaria (c) Haemophilia (d) AIDS

501. An *X*-linked recessive trait is **[CPMT 1997]**

(a) Colour blindness (b) Hunter's syndrome (c) Sickle-cell anaemia (d) Leishman's syndrome

502. An example of sex influenced inheritance is **[APMEE 2002]**

(a) Haemophilia (b) Baldness (c) Colourblindness (d) Down's syndrome

503. A child gets sex-linked traits from **[Bih. PMT 1994]**

(a) Father (b) Mother (c) Both father and mother (d) None of the above

504. A single recessive allele which can express its effect should occur on **[AIIMS 1992]**

(a) Any autosome (b) Any chromosome (c) X- chromosome of female (d) X-chromosome of male

505. A colour blind son is born to normal parents. It shows that

(a) The father was heterozygous for colour blindness (b) The mother was genotypically homozygous

(c) The mother was heterozygous for colour blindness (d) Both parents carried a recessive gene for the disorder

506. A character is transmitted from father to daughter and from there to grandson. It is

(a) Holandric inheritance (b) Holongenic inheritance (c) Criss cross inheritance (d) Dominant inheritance

507. Colour blindness is disease in which the factor is usually transmitted to children by woman. It is because the factor is located on

(a) An autosome (b) X-chromosome (c) Y-chromosome (d) Cytoplasm

508. The sex linked characters are those

(a) Which are related to sexual physiology (b) The genes of which are present on sex chromosomes

(c) Which appear either in male or in female (d) Which are controlled by sex hormones

509. In protanopia, a person cannot distinguish

(a) Green colour (b) Red colour (c) Blue colour (d) Blue and green colour

510. The 'Christmas disease' patient lacks antihaemophilic : **[KCET 2003]**

(a) Factor IX (b) Factor XI (c) Factor VIII (d) Homogentisic acid oxidase

511. Which of the following will be colour blind **[MP PMT 1991]**

(a) XY (b) X^CX (c) XX (d) X^CX^C

512. One of the genes present exclusive on the X-Chromosome in human is concerned with **[AIIMS 2003]**

(a) Baldness (b) Night blindness

(c) Red-green colour blindness (d) Facial hair/Moustaches in male

513. Turner's syndrome is **[Har. PMT 2000]**

(a) Trisomy of 18th chromosome (b) Trisomy of 21st chromosome

(c) Absence of one sex chromosome (d) An autosomal recessive condition

514. Colour blindness results from **[MP PMT 1996; AFMC 1997]**

(a) Inverted retina (b) Absence of rods (c) Absence of eye lids (d) Abnormal cones

515. In 1956, an XXXY type of abnormality was seen in there patients which is **[DPMT 1996]**

(a) Male phenotype (b) Female genotype (c) Female phenotype (d) Gynandromorph

516. A haemophilia is more common in males because it is a **[CBSE PMT 1990]**

(a) Recessive trait carried by X-chromosome (b) Recessive trait carried by Y-chromosome

(c) Dominant trait carried by X-chromosome (d) Dominant trait carried by Y-chromosome

517. A man who carries a holandric gene will transmit it to

(a) All his male offspring (b) $1/2$ his male offspring (c) All his female offspring (d) $1/2$ his female offspring

518. An example for holandric inheritance is **[EAMCET 2002]**

(a) Epidermolysis (b) Turner's syndrome (c) Haemophilia (d) Webbed toes

Advance

519. A diseased man marries a normal woman. They get three daughters and five sons. All the daughters were diseased and sons were normal. the gene of this disease is **[CBSE PMT 2002]**

(a) Autosomal dominant (b) Sex linked recessive (c) Sex limited character (d) Sex linked dominant

520. Mental retardation in man, associated with sex chromosomal abnormality is usually due to **[CBSE PMT 1**

(a) Reduction in *X* complement (b) Increase in *X* complement

(c) Moderate increase in *Y* complement (d) Large increase in *Y* complement

521. Pattern baldness, moustaches and beard in human males are examples of **[CBSE PMT 2003]**

(a) Sex-determining traits (b) Sex-linked traits (c) Sex limited traits (d) Sex differentiating traits

522. A child receives **[CPMT 1977; CBSE PMT 1995]**

(a) 25% genes form his father (b) 50% genes form his father (c) 75% genes form his father (d) 100% genes from his father

523. A husband and wife have normal vision but fathers of both of them were colour blind. Probability of their first daughter to be colour blind **[CBSE PMT 1990]**

(a) 25% (b) 50% (c) 75% (d) 0%

524. One way of determining sex-linked inheritance is **[CPMT 1973]**

(a) Sons resemble mother and daughter resemble father (b) Both sons and daughter resemble father

(c) Sons resemble father and daughter resemble mother (d) Both sons and daughters resemble mother

525. What are all the chances of colour blind daughter and sons being born in a marriage of normal man marrying a normal woman, whose father was colour blind **[KCET 2003]**

(a) All sons are normal and all daughters are colourblind

(b) Both the sons and daughters are colourblind

(c) All the sons are colurblind and all daughters are normal

(d) 50% sons are colourblind and all daughters are phenotypically normal

526. Colour blindness is found more in males than in females because **[MP PMT 1992]**

(a) The males containing the single affected X chromosome are colour blind

(b) Heterozygous females are colour blind

(c) Males having affected Y chromosome are colour blind

(d) Affected X chromosome has much high affinity to Y chromosome as compared to unaffected X chromosome

527. A man homozygous for brown colour marries a lady heterozygous for brown colour dominant. What will be the fate of their children **[BHU 1986]**

(a) All brown (b) Three brown (c) Two brown (d) None of the above

528. A fruit fly is heterozygous for sex-linked genes when meted with normal female fruit fly, the males specific chromosome will enter egg cell in the proportion **[CBSE PMT 1997]**

(a) 1 : 1 (b) 2 : 1 (c) 3 : 1 (d) 7 : 1

529. Haemophilia A is due to the absence of

(a) Antihaemophilic globulin (b) Calcium (c) Plasma thromboplastin (d) X chromosome

530. As a result of marriage of curly hair mother and straight hair father, 8 children are born. The ratio of curly and straight haired will be **[CPMT 1980; MP PMT 2001]**

(a) 6 : 2 (b) 2 : 6 (c) 4 : 4 (d) 3 : 5

531. When can a women be colourblind **[CPMT 1991]**

(a) If her father is colourblind and mother is carrier (b) If her father is normal and mother is carrier

(c) If her father is colourblind and the mother is normal (d) If her father is normal and the mother is colourblind

532. Holandric genes are

(a) Genes located on *X* chromosomes

(b) Genes located on the homologous segment of *Y* chromosome

(c) Genes located on non-homologous segment of *Y* chromosome

(d) Genes located on autosomes

533. Transmission of *X* chromosomes of male to the female in F_1 generation and to males in F_2 generation is called as

(a) Mendelian inheritance (b) Criss-cross inheritance

(c) Sex linked inheritance (d) Extra-chromosomal inheritance

534. A sex linked recessive gene C produces red green colour blindness in humans. A normal woman whose father was colourblind marries a colourblind man. Of all the girls born to these parents what percentage is expected to be colourblind **[JIPMER 2002]**

(a) 25 percent (b) 50 percent (c) 75 percent (d) 100 percent

535. A normal woman whose father was colourblind marries a normal man. What kinds of children would be expected and in what proportion

[MP PMT 1997]

(a) Daughters normal, 50% of sons colourblind (b) Daughters normal, all sons colourblind

(c) 50% of daughters colourblind, all sons normal (d) All daughters colourblind, sons normal

536. Genes of colour blindness are carried in **[BHU 1985]**

(a) Eye cells (b) Sex cells (c) Body cells (d) Heterosomes

537. A colourblind man has a colourblind sister but a normal brother than phenotype of its parents is **[CPMT 199**

(a) Father colourblind and mother normal (b) Father normal and mother colourblind

(c) Father and mother both are colourblind (d) Father and mother both are normal

538. Haemolytic jaundice is caused due to a dominant gene but only 10% of the people actually develop the disease. A heterozygous man marries a homozygous normal woman; what proportion of the children would be expected to develop the haemolytic disease **[AIIMS 1982]**

(a) 1/5 (b) 1/10 (c) 1/15 (d) 1/20

539. A man known to be victim of haemophilia marries a normal woman whose father was known to be bleeder. Then it is expected that

[AIIMS 1985; Pb PMT 1999; CBSE PMT 2000]

(a) All their children will be bleeders (b) Half of their children will be bleeders

(c) One fourth of their children will be bleeders (d) None of their children will be bleeder

540. Expected children of a blue-eyed (recessive) woman and brown-eyed (dominant) man who had a blue-eyed mother are likely to be

[CBSE PMT 1991]

(a) All brown-eyed (b) One blue-eyed and one brown-eyed

(c) All blue-eyed (d) Three blue-eyed and one brown-eyed

541. All the sons are haemophilic and daughters are normal of a haemophilic mother and normal father. This character is **[CBSE PMT 1996]**

(a) *X*-linked recessive (b) *Y*-linked recessive (c) *X*-linked dominant (d) *Y*-linked dominant

542. Marriages between close relative and cousins is not advisable because **[NCERT 1976; MP PMT 1990]**

(a) More mutation can occur (b) More recessive defects are likely to appear

(c) More chances are there for *Rh* blood group anomalies (d) More chances are there for multiple births

543. All sons of a couple are colourblind because **[CPMT 1980]**

(a) Mother is homozygous colourblind (b) Mother is heterozygous and father normal

(c) Mother is heterozygous and father colourblind (d) Mothers is normal and father colourblind

544. A colour blind son will born when **[CPMT 1992, 93]**

(a) Mother is normal and father normal (b) Mother is colour blind and father normal

(c) Mother is normal and father is colour blind (d) All the cases are correct

545. A colour blind man marries the daughter of a colour blind person. Then in their progeny **[AIIMS 1983, 92;**

(a) None of their daughters are colour blind (b) All the sons are colour blind

(c) All the daughters are colour blind (d) Half of their sons are colour blind

546. Person whose father is colourblind marries a lady whose mother is daughter of a colourblind man. Their children will be **[CPMT 1984; DPMT 1993]**

(a) All normal (b) All colour blind

(c) All sons colour blind (d) Some sons normal and some colour blind

547. Normally all genes occur in pairs occupying position on the X chromosome and not on Y chromosome. This indicates that **[CPMT 1979]**

(a) Y chromosome is larger than X chromosome

(b) Entire set of gene on X is different from those on Y chromosome

(c) X chromosome is larger than Y chromosome

(d) X chromosome is dominating with Y chromosome

548. The expression of genes for the production of milk in only females is a **[AIIMS 1993]**

(a) X linked character (b) Y linked character (c) Sex limited genes (d) Sex influenced genes

549. Haemophilia is caused due to lack of **[AIIMS 1992]**

(a) ADH (b) AHF (c) STH (d) ACTH

550. A boy is colour blind, in his two sisters one is colour blind and one is carrier (normal). Then who is colour blind in his family **[CPMT 1994]**

(a) Father (b) His grand father and mother (c) Mother (d) His grand father

551. Hypertrichosis (hairy pinnae) is trait linked to **[APMEE 1999]**

(a) X-chromosome (b) Y-chromosome (c) Autosomes (d) None of the above

552. Of a normal couple, half the sons are haemophilic while half the daughters are carriers. The gene is located on **[CBSE PMT 1993]**

(a) X-chromosome of father (b) Y-chromosome of father

(c) One X-chromosome of mother (d) Both the X-chromosomes of mother

553. Christmas disease is another name of **[CBSE PMT 2003, CET Chd. 2003]**

(a) Sleeping sickness (b) Down's syndrome (c) Hepatitis (d) Haemophilia B

554. Colour blindness is due to one **[CPMT 1990]**

(a) Recessive allele in females (b) Dominant allele in females (c) Dominant allele in males (d) Recessive allele in males

555. Deficiency of VIII factor leads to **[Har. PMT 2001]**

(a) Haemophilia A (b) Haemophilia B (c) Haemophilia C (d) Haemophilia D

556. In *Drosophila*, white eye colour is recessive X-linked trait while red eye colour is dominant. A white eyed female is crossed with red eyed male. The female offspring with red eye colour would be **[CPMT 199**

(a) 100% (b) 50% (c) 25% (d) Zero

557. A diseased man marries a normal woman. The couple has 3 daughters and 5 sons, the daughters are diseased while the sons are normal. The gene of the disease is **[CBSE PMT 2002]**

(a) Sex-linked recessive (b) Sex linked dominant (c) Autosomal character (d) Sex limited character

558. Queen Victoria of England was **[KCET 2000]**

(a) Haemophilic carrier (b) Colour blind (c) AIDS patient (d) Deaf

559. A colour blind girl is rare because she will be born only when **[CBSE PMT 1991]**

(a) Her mother and maternal grand father were colour blind

(b) Her father and maternal grand father were colour blind

(c) Her mother is colour blind and father has normal vision

(d) Parents have normal vision but grand parents were colour blind

560. In colour blindness red, green and other colours appear

(a) White (b) Yellow (c) Grey (d) Pink

561. Colour blindness in which all colour appear grey is

(a) Monochromatism (b) Dichromatism (c) Protanopia (d) Deuteronopia

562. A cross between white eyed female and red eyed male *Drosophila* gives red eyed females and white eyed males. Rarely the cross gives rise to white eyed females and red eyed males. This is due to **[BHU 1994**

(a) Loss of sex chromosome (b) Mutation in female fly

(c) Nondisjunction of two X-chromosomes in female (d) None of the above

563. Laws of inheritance were given by **[AFMC 2000]**

(a) Doron (b) Donoran (c) Mendel (d) Morgan

564. A heterozygous individual carrying recessive sexlinked gene is called

(a) Carrier (b) Crossing over (c) Transmitter (d) Albino

565. The fact that baldness is more common in men than in women could be explained on the basis that **[CBSE P**

(a) Genes for baldness are located on X-chromosome only

(b) Genes for baldness are located on Y-chromosome only

(c) Baldness in dominant in males and recessive in females

(d) Genes are not involved and baldness is due to male hormones only

566. A normal woman is married to a man having hypertrichosis. They got one daughter and one son. What is the possibility of this daughter to show hypertrichosis condition

(a) 0% (b) 25% (c) 50% (d) 75%

567. One child is heamophilic (sex-linked trait) whereas his fraternal twin brother is normal. Which one of the following information is most appropriate **[CBSE PMT 1999]**

(a) The haemophilic child is male (b) The child is a monozygotic twin

(c) The mother must have been heterozygous (d) The other child is a female and the father is haemophilic

568. The errors in meiosis that produces a 47,XYY karyotype is best described by

(a) Meiosis division I of maternal oogenesis (b) Meiosis division II of maternal oogenesis

(c) Meiosis division I of paternal spermatogenesis (d) Meiosis division II of paternal spermatogenesis

569. In humans, male XXY and female XXXX occur due to **[CBSE PMT 2000]**

(a) Euploidy (b) Aneuploidy (c) Autosomal syndrome (d) None of the above

570. Ram has hairy ears (hypertrichosis), a trait carried by a gene in his Y-chromosome. What is the chance that his grandson will inherit the trait from him

(a) 0% (b) 25% (c) 50% (d) 100%

571. Match List I and List II and select the correct answer using the codes given below the list :

List I (Character of man)	**List II** (Example)
(A) Sex-linked	(1) Baldness
(B) Sex-influenced	(2) Acquired immune deficiency syndrome
(C) Sex-limited	(3) Kilinefelter's syndrome
	(4) Haemophilia
	(5) Beard in man

Answer codes :

(a) A = 4, B = 1, C = 5 (b) A = 5, B = 3, C = 2

(c) A = 5, B = 1, C = 3 (d) A = 4, B = 3, C = 2

572. Red-green colour blindness in humans is governed by sexlinked recessive gene. A normal woman whose father was colour blind marries a colour blind man. What proportion of their daughters is expected to be colour blind **[CBSE PMT 1999]**

(a) $\frac{1}{4}$ (b) $\frac{1}{2}$ (c) $\frac{3}{4}$ (d) All

573. A woman with two genes for haemophilia and a gene for colour blindness on one of the X-chromosomes marries a normal man. How will the progeny be **[CBSE PMT 1998]**

(a) Haemophilic and colour blind daughters

(b) All sons and daughters haemophilic and colour blind

(c) 50% haemophilic daughters and 50% colour blind daughters

(d) Among sons 50% haemophilic and 50% haemophilic colour blind

574. Mr. Kapoor has *Bb* autosomal gene pair and *d* allele sex-linked. What shall be the proportion of *Bd* in sperms **[CBSE PMT 1993]**

(a) 0 (b) $\frac{1}{4}$ (c) $\frac{1}{8}$ (d) $\frac{1}{2}$

575. Haemophilia is most likely originated as a result of

(a) A gene mutation in the X-chromosome (b) The crossing over of two sex chromosomes

(c) A nondisjunction of chromosome number 21 (d) The separation of two homologous chromosomes

576. Chromosomal analysis reveals a 47,XXY Karyotype. Which of the following descriptions best fits this abnormality

(a) Autosomal trisomy (b) Sex chromosome triploidy

(c) Sex chromosome aneuploidy (d) A female with turner's syndrome

577. It is well known that Queen Victoria of England was a carrier for haemophilia. Since this is an X-linked disease, it can be predicted that

[CPMT 1993]

(a) All of her sons would have disease

(b) All her daughters would have been carriers

(c) Her father must definitely have had haemophilia

(d) Haemophilia would have occurred in more of her male than her female descendents

578. If a man and a woman both having colour blind fathers marry, the percentage probability of their first daughter to be colour blind is

[Wardha 2003]

(a) 25% (b) 50% (c) 100% (d) 0%

Pedigree analysis

Basic Level

579. Most studies on human genetics have been made through **[MP PMT 1990]**

(a) Genetic engineering (b) Eugenics

(c) Microscopic studies of chromosomes (d) Pedigree charts

580. The symbol of empty circles used in pedigree analysis represents

(a) Normal female (b) Normal male (c) Affected female (d) Affected male

581. Brachydactyly signifies **[CET BV Pune 1998]**

(a) Abnormally short knees (b) Abnormally long fingers (c) Abnormally long toes (d) Abnormally short fingers

582. Syndactyly refers to **[CET BV Pune 1998]**

(a) Split fingers (b) Fused fingers (c) Split toes (d) fused toes

583. Presence of extra finger or toe is known as **[Haryana PMT 1994]**

(a) Marfan's syndrome (b) Polydactyly (c) Brachydactyly (d) None of the above

584. Polydactyly in man due to **[J & K CET (Med.) 2002]**

(a) Autosomal dominant gene (b) Autosomal recessive gene (c) Sex linked dominant gene (d) Sex linked recessive gene

585. Which one of the following is a dominant trait

(a) Albinism (b) Sickle cell anaemia (c) Phenylketonurea (d) Polydactyly

586. An abnormality not due to recessive genes is **[AMU 2001]**

(a) Phenylketonurea (b) Alkeptonurea

(c) Polydactyly (d) Tay-sach's syndrome

Twins & I.Q.

Basic Level

587. Differences in twins suggest **[NCERT 1983]**

(a) Incomplete dominance (b) That phenotype is influenced by many genes

(c) That single gene may produce multiple effects (d) That they develop from two different eggs

588. Two offspring developed in the same uterus but from fertilization of two different ova are **[AFMC 2002]**

(a) Dizygotic twins (b) Monozygotic twin (c) Fraternal twins (d) Both (a) and (c)

589. Identical twins are produced when **[BHU 1982; MP PMT 2001]**

(a) One fertilized egg divided into 2 blastomeres and both separate (b) One sperms fertilized two eggs

(c) One egg fertilized with two sperms (d) Two eggs are fertilized

590. Fraternal twins are produced when **[NCERT 1976; CPMT 1990, 91]**

(a) A fertilized egg divided into two (b) An egg is fertilized by two sperm

(c) A divided egg has two set of chromosomes (d) Two eggs are fertilized simultaneously

591. If in a child of 10 years ages of the intelligence is of 14 years child that I.Q. of this child would be **[CPMT 1**

(a) 140 (b) 100 (c) 160 (d) 110

592. Sometimes the separation of twins is incomplete and these are borne attached are remain so even after. Such twins are known as

(a) Fraternal (b) Dizygotic (c) Identical (d) Siamese

593. I.Q. is the ratio of mental age to **[CPMT 1983]**

(a) Chronological age multiplied by ten (b) Chronological age plus ten

(c) Chronological age plus 100 (d) Chronological age multiplied by 100

594. The I.Q. of a genius ranges from **[CPMT 1980]**

(a) 70-89 (b) 90-109 (c) 110-139 (d) 140-more

595. Identical twins are **[AFMC 1986]**

(a) Heterozygous (b) Homozygous (c) Monozygotic (d) Dizygotic

596. Genetically identical progeny is produced when an individual **[AFMC 1994]**

(a) Practices self-fertilization (b) Produces identical gametes

(c) Practices reproduction (d) Practices in breeding without meiosis

597. Conjoint twins are also known as **[CBSE PMT 1988]**

(a) Fraternal twins (b) Siamese twins (c) Dizygotic twins (d) None of the above

598. Monozygotic twins are formed when **[CPMT 1996]**

(a) No cleavage takes place to the zygote

(b) Two ova are fertilized at the same time

(c) Incomplete cleavage of zygote takes place

(d) The cells formed from first cleavage of zygote become independent

599. Twins are born because **[AIIMS 1996]**

(a) Two sperms fertilize four ova (b) Two sperms fertilize two ova

(c) Two sperms fertilize single ovum from two sites (d) None of the above

600. Free-martins are common in

(a) Birds (b) *Drosophila* (c) Cattle (d) Human beings

601. Free-martins condition is observed in

(a) Dizygotic twins (b) Monozygotic twins (c) Both of these (d) None of the above

602. In free-martin condition **[CBSE PMT 1994]**

(a) Both female and male are sterile (b) Both female and male are normal

(c) Female is sterile and male is normal (d) Male is sterile and female is normal

603. Free martin is an example of **[MP PMT 2000]**

(a) Sex reversal (b) Transformer gene (c) Hormonal control of sex (d) Both (a) and (c)

Eugenics, Euphenics, Euthenics

Basic Level

604. Improvement of human race by improving the environment is called **[MP PMT 1998; CBSE PMT 1990; C**

(a) Euphenics (b) Eugenics (c) Euthenics (d) None of the above

605. 'Eugenics' pertains to **[CPMT 1984, 86; CBSE PMT 1990]**

(a) Improvement of mankind by improving his heredity (b) Preservating human sperms for artificial insemination

(c) Study of human genetics (d) Controlling size of a human family

606. Euphenics is **[DPMT 1992; MP PMT 1993]**

(a) Improvement of human race by better environment (b) Improvement of human race by genetic engineering

(c) Treatment of communicable diseases (d) Treatment of inheritable diseases

607. Improvement of genetic characters and present day generation on the basis of best nutrition and training is called **[MP PMT 1995]**

(a) Eugenics (b) Euphanics (c) Euthenics (d) Gerontology

608. Which of the following fungus is used in experiments to study the genetics **[RPMT 1998]**

(a) *Rhizopus* (b) *Mucor* (c) *Neurospora* (d) *Claviceps*

609. This organism has been used very much in genetics **[KCET 1991]**

(a) Rana tigrina (frog) (b) Domestic fly

(c) Domestic lizard (d) Fruit fly (*Drosophila melanogaster*)

610. Who is called 'father of eugenics'

(a) Galton (b) Griffith (c) Garrod (d) Goldschmidt

611. The process of improving human race genetically is called **[Orissa JEE 1992; BCECE 1996; AFMC 2000]**

(a) Eugenics (b) Euphenics (c) Euthenics (d) All of the above

612. Eugenics is the study of **[CBSE PMT 1992]**

(a) Evolution (b) Human genetics (c) Development (d) Modern genetics

613. Branch of biology dealing with heredity and variation is called **[MP PMT 1998]**

(a) Palaeontology (b) Evolution (c) Genetics (d) Ecology

Advance

614. The genetic ratio is termed as **[DPMT 1982]**

(a) Dominant (b) Genotype (c) Phenotype (d) Alleles

615. Sir Archibald is associated with

(a) Eugenics (b) Euthenics (c) Genetics (d) Human genetics

616. The best method of improve the genetic quality of mankind is **[CPMT 1974; AFMC 1976]**

(a) Marriage restriction (b) Sterilization

(c) Control of immigration (d) Sexual separation of defectives

617. Who of the following is concerned with biochemical genetics

(a) Beadle (b) Galton (c) Garrod (d) Mendel

618. Under certain conditions, the scientists have obtained cell-like structures but not true organization of a cell, these are referred as **[MP PMT 1994]**

(a) Microbes (b) Coacervates (c) Eobionts (d) Protists

619. Random genetic drift in a population probably results from **[CBSE PMT 2002, 03]**

(a) Large population size (b) Highly genetically variable individuals

(c) Interbreeding within small isolated population (d) Constant low mutation rate

620. In a random mating population in equilibrium, which of the following brings about a change in gene frequency in a non-directional manner **[CBSE PMT 2003]**

(a) Migration (b) Mutation (c) Random drift (d) Selection

621. Curing of genetic diseases is studied under

(a) Genetics (b) Eugenics (c) Euphenics (d) Dysgenics

622. Genetic drift **[CBSE PMT 1992; KCET (Med.) 1998]**

(a) Is an orderly change in gene frequencies (b) Products greatest fluctuations in large population

(c) Is the random change in gene frequencies (d) Has nothing in common with inbreeding

623. An allele is said to be dominant if **[CBSE PMT 1992]**

(a) It is expressed only in both homozygous and heterozygous condition

(b) It is expressed only in second generation

(c) It is expressed only in heterozygous combination

(d) It is expressed only in homozygous combination

624. In order to determine whether variation of a character in a population were genetically controlled, the most appropriate procedure will be to

[AIIMS 1983]

(a) Count chromosomes and find out the variations in number in the population

(b) Examine DNA and see if the population shows any variation

(c) Measure the variation and see if they are continuous or discontinuous

(d) Cross individuals of both extremes and see if the offsprings and parents show the same range of variations

625. Genetic drift operates only in **[CBSE PMT 1998]**

(a) Island population (b) Smaller population (c) Larger population (d) Mendelian population

Genetic engineering and tissue culture

Basic Level

626. Genetically engineered insulin can be obtained by

(a) Recombinant DNA technique with the help of *E. coli*

(b) Two coded insulin genes separated then incorporated into bacteria

(c) The extraction of cow's and pig's pancreas

(d) Technique not developed till now

627. Trade name of genetically engineered insulin is

(a) Anulin (b) Beta insulin (c) Humulin (d) Gilbert's insulin

628. Which of the following organelles is related with genetic engineering

(a) Golgi apparatus (b) Lysosomes (c) Mitochondria (d) Plasmids

629. Restriction endonuclease is used in

(a) Genetic engineering (b) Tissue culture (c) Cell fractionation (d) Regeneration of tissues

630. The term genetic engineering is used for **[MP PMT 2003]**

(a) Blotting technique (b) RNA reaction technique

(c) Protein synthesis technique (d) Recombinant DNA technique

631. Genetic engineering is the

(a) Formation of new gene artificially (b) Formation of RNA from DNA artificially

(c) Modification of genes artificially from non DNA material (d) Formation of DNA

632. The interferon of other animals

(a) Cannot be used in human (b) Can be used in human

(c) Can only be used in that particular animal (d) (a) and (c) both

633. The genetic study of human beings is done by **[CPMT 1994]**

(a) Genetic chart (b) Genetic engineering (c) Eating of food (d) Vertebral column

634. The viruses infect the host cells. By the induction of viruses the host cells produce the

(a) Antigens (b) Oncogens (c) Interferon (d) Carcinogens

635. Gene synthesis is related to

(a) V. Baer (b) H.G. Khorana (c) L. Pasteur (d) C. Linnaeus

636. Who among the following scientists is associated with the discoveries in genetic engineering

(a) Khorana (b) Watson (c) Crick (d) Messelson

637. Which of the following enzyme is used to join DNA fragments

(a) Terminase (b) Endonuclease (c) Lygase (d) DNA polymerase

638. Father of DNA finger printing is

(a) Sunder Lal Bhuguna (b) Wishwanath (c) Jeffreys (d) Rockfeller

639. For DNA finger printing, DNA is obtained from

(a) White blood corpuscles (b) Hair root cells (c) Body secretion (d)

640. It is now possible to breed plants and animals of desired characters through

(a) Tissue culture (b) Genetic engineering (c) Ikebana technique (d) Chromosome engineering

641. Genetic engineering is **[DPMT 1996]**

(a) Plastic surgery (b) Addition or removal of genes (c) Study of extra nuclear genes (d) All the above

642. DNA finger printing can resolve **[MP PMT 1998]**

(a) Identification of a person (b) Paternity dispute (c) Maternity dispute (d)

643. Restriction enzyme are used in genetic engineering because they **[CBSE PMT 1995; BHU 2001; Orissa 2002]**

(a) Can join DNA fragments (b) Cut DNA at specific base sequence

(c) Cut DNA at variable sites (d) Are proteolytic enzymes which degrade harmful proteins

644. Blunt ends of passenger and vehicle DNAs are joined by

(a) DNA polymerase I (b) RNA polymerase (c) DNA ligase (d) DNA polymerase III

645. Chemical knives/molecular scissors of DNA are **[CBSE PMT 1998, 2001; KCET 2000; Wardha 2001**

(a) Endonucleases (b) Polymerases (c) Ligases (d) Trascriptases

646. First successful animal clone was

(a) Dolly goat (b) Dolly sheep (c) Molly goat (d) Molly sheep

647. Arber, Smith and Nathans are famous for discovery of

(a) Gene therapy (b) Restriction enzyme (c) Humulin (d) Second generation vaccine

648. Genetically engineered bacteria are being used in commercial production of **[CBSE PMT 1996; DPMT**

(a) Melatonin (b) Testosterone (c) Human insulin (d) Thyroxine

649. When the genotype of an organism is improved by the addition of foreign genes the process is called **[AFMC 1999]**

(a) Biotechnology (b) Tissue culture (c) Genetic engineering (d) Genetic diversity

650. "Tissue culture" means **[MP PMT 1993]**

(a) Cultivation of tissue in laboratory through formation of new cells

(b) Introduction of new tissue in an animal body

(c) A technique for maintaining fragments of cells alive after their removal from an organism

(d) Maintaining tissue alive by immersing it partially in a nutrient fluid

651. Chromosomal abnormality of an unborn baby (while in mother's womb) can be found out by a technique called **[MP PMT 1990, 95]**

(a) Amniocentesis (b) CAT scanning (c) Ultrasound (d) Tissue culture

652. The primary biological importance of sex in organism is that it **[MP PMT 1990]**

(a) Is essential for organismic reproduction (b) Is essential for cellular reproduction

(c) Causes new mutation to occur in offspring (d) Promotes genetic variability in offspring

653. A genetic marker is

(a) An enzyme used to cut DNA (b) A radioactive probe used to find out a gene

(c) A nucleotide sequence near a particular gene (d) A place where a restriction enzyme cuts DNA

654. Gene library refers to **[Orissa JEE 2003]**

(a) DNA fragments maintained in agarose gel

(b) Photographs of DNA fragments printed in books

(c) DNA sequence information maintained in data bank

(d) DNA fragments of a genome maintained by cloning in cultured cells

655. Cryopreservation refers to germplasm protection by **[AMU 2002]**

(a) Low temperature treatment (b) Breeding with wild varieties

(c) Energy flow through each tropic level (d) Both (b) and (c)

656. *Escherichia coli* is used in biological researches because **[MP PMT 1993]**

(a) It is easy to handle (b) It is easily available

(c) It can be easily cultured (d) It can easily multiply in the host

657. Southern blot technique is related to **[Kerala PMT 2002]**

(a) Blood test (b) DNA profiling (c) ELISA test (d) Sonography

658. Southern blotting is used to identify

(a) DNA in a cell (b) RNA in a cell (c) A protein in a cell (d) Antibiotic resistance in a cell

659. Southern blots, but not colony blots, require which of the following

(a) Autoradiography (b) Membrane filter (c) DNA hybridization (d) Gel electrophoresis

660. Plasmids are **[EAMCET 1996; CBSE PMT 2002]**

(a) Outgrowth of mitochondria membrane (b) Outgrowth of cell

(c) Outgrowth of nuclear membrane circular material (d) Extrachromosomal

661. One of the most useful methods for identifying a specific gene is the

(a) Southern blot (b) Western blot (c)Northern blot (d) None of the above

662. A collection of an organism's DNA fragments that are stored in a host organism is called a

(a) Plasmid (b) DNA clone (c) DNA library (d) DNA restriction site

663. DNA fingerprint cannot be prepared from

(a) RBC (b) Sperm (c) WBC (d) Inner lining of cheek

664. The organism which is used for gene transfer in higher organism is **[DPMT 2003]**

(a) *E. coli* (b) *Acetobacter*

(c) *Bacillus thuringiensis* (d) *Agrobacterium tumefaciens*

665. How are RFLPs detected

(a) By doing standard Mendelian cross (b) By amplifying the DNA using PCR

(c) By observing DNA of different lengths on a gel (d) By looking at the chromosome in the microscope

666. Which of the following is the best way to determine paternity **[AIIMS 2000]**

(a) Gene counting (b) Protein analysis (c) DNA fingerprinting (d) Chromosome counting

667. Thermal cycler is used in this reaction **[KCET 2001]**

(a) Radioactivity (b) Chemical reaction (c) Polymerase chain reaction (d)

668. Which of the following enzymes is used in polymerase Chain Reaction (PCR)

(a) Taq polymerase (b) Vent polymerase (c) Both of these (d) None of the above

669. Which of the following is a recent application of genetic engineering in diagnostic technique **[BHU 200**

(a) PCR (b) ELISA test (c) Gravidex test (d) ABC blood groups

670. Polymerase chain reaction is concerned with **[Pb PMT 2003; CPMT 2003]**

(a) DNA amplification (b) DNA repairing (c) DNA proof reading (d) DNA replication

671. The transfer of genetic material of one bacterium to another is called **[CPMT 1996]**

(a) Replication (b) Translation (c) Transcription (d) Transduction

672. The process which cannot take place in the absence of viruses **[KCET 2002]**

(a) Conjugation (b) Transduction (c) Translocation (d) Transformation

673. Transfer of DNA from bacterium to another through cell to cell contact is known as **[Kerala PMT 2002**

(a) Conjugation (b) Transduction (c) Transcription (d) Transformation

674. The uptake of naked DNA by a bacterium is called

(a) Cloning (b) Conjugation (c) Transduction (d) Transformation

675. Genetically engineered bacteria are being used in commercial production of **[CBSE PMT 1996; DPMT**

(a) Melatonin (b) Thyroxine (c) Testosterone (d) Human insulin

676. A clone is a group of individuals obtained through **[JIPMER 1999; AFMC 1999]**

(a) Hybridization (b) Self pollination (c) Cross pollination (d) Vegetative propagation

677. For producing the world's first animal clone Dolly, which cells were used **[BHU 2003]**

(a) Sperm cells (b) Brain cells (c) Udder cells (d) Blood cells

678. The technique by which 'Dolly' the sheep obtained is termed as **[CBSE PMT 1999]**

(a) Cloning by gene transfer (b) Cloning by chromosome transfer

(c) Cloning by nuclear transfer (d) None of the above

679. A new method of harvesting stem cells is known as **[JIPMER 2002]**

(a) Cloning (b) Sporogony (c) Entrapping (d) Schizogony

680. Clonal cell lines can be obtained by **[MP PMT 2000]**

(a) Tissue system (b) Tissue culture (c) Tissue fractionation (d) Tissue homogenization

681. How many genes are there in a human sperm cell

(a) 23 (b) 46 (c) 30,000 (d) 5,000-10,000

682. An extrachromosomal DNA which can be used as vector in gene cloning is called **[JKCMEE 2003]**

(a) Axon (b) Intron (c) Plasmid (d) Transposon

683. The basis of DNA fingerprinting is **[CBSE PMT 1996]**

(a) Availability of cloned DNA

(b) Knowledge of human karyotype

(c) Phenotypic difference between individuals

(d) Occurrence of RFLP (Restriction Fragment Length Polymorphism)

684. RFLPs distributed throughout human genome are useful for

(a) Gene mapping (b) DNA fingerprints (c) Both of these (d) None of the above

685. In genetic engineering, the term vector is applied for **[DPMT 2003]**

(a) Plasmid (b) Bacteria (c) Sources of DNA (d) Cell which receive

686. Genes which confer antibiotic resistance on bacteria are located on **[MP PMT 1998]**

(a) RNA (b) Plasmid (c) Polysome (d) Circular DNA molecule

687. Restriction endonucleases cut **[BHU 1995; Orissa JEE 2003]**

(a) Double stranded DNA (b) Single stranded DNA (c) Single stranded RNA (d) Double stranded RNA

688. Generally, plasmids carry which type of genetic material

(a) Essential genes (b) Useless genes (c) Nonessential genes (d) Metabolic genes

689. Which of the following forms chemical scissors **[Kerala PMT 1997]**

(a) *Eco* RI (b) *Hind* III (c) *Bam* HII (d) All of the above

690. Electrophoresis is used to

(a) Clone genes (b) Cut DNA into fragments

(c) Separate fragments of DNA (d) Match gene with its function

691. Genetic engineering is **[BHU 2003]**

(a) Making artificial genes

(b) Production of alcohol by using microorganisms

(c) Hybridization of DNA of one organism to that of others

(d) Making artificial limbs, diagnostic instruments such as ECG, EEG, etc

692. The approximate number of genes contained in the genome of Kalpana Chawla was **[AIIMS 2003]**

(a) 80,000 (b) 40,000 (c) 1,00,000 (d) 30,000

693. The enzymes commonly used in genetic engineering are **[KCET 2003]**

(a) Restriction endonuclease and polymerase (b) Restriction endonuclease and Ligase

(c) Endonuclease and Ligase (d) Ligase and Polymerase

694. The technique which involves addition of deletion of genes is **[AFMC 2002]**

(a) Gene therapy (b) Gene splicing (c) Genetic engineering (d)Artificial synthesis

695. Apart from DNA in the bacterial nucleoid, there is a circular extrachromosomal DNA called**[AMU 200**

(a) Plasmid (b) Mesosome (c) Chromosome (d) None of the above

696. In plasmid R gene is responsible for **[AMU 2001]**

(a) Exchange of genetic material between two partners (b) Drug resistance

(c) Locomotion (d) All of the above

697. Introduction of foreign genes for improving genotype is called **[Pb PMT 2003]**

(a) Vernalization (b) Tissue culture (c) Biotechnology (d) Genetic engineering

698. Advancement in genetic engineering has been possible due to **[BCECE 2001]**

(a) Oncogenes (b) Transposons (c) Exonucleases (d) Endonucleases

699. Manipulation of DNA in genetic engineering became possible due to the discovery of**[CBSE PMT 200**

(a) Primase (b) DNA ligase (c) Transcriptase (d) Restriction endonuclease

700. Which of the following produce DNA fragment with "sticky ends"

(a) DNA ligase (b) Restriction enzymes (c) DNA polymerase (d) All of the above

701. Which is a genetic vector **[AFMC 1997]**

(a) Plastid (b) Plasmid (c) Mosquito (d) All of the above

702. 'Cloning' is meant **[AFMC 1997]**

(a) To preserve the genotype of the organism (b) To produced hGH gene in *E. coli*

(c) To replace the original one (d) All of the above

703. ……. is the transfer of normal genes into body cells to correct a genetic defect

(a) Gene therapy (b) Gene mutation (c) Reverse transcription (d)Nucleic acid hybridizat

704. Plasmid **[RPMT 1998]**

(a) Is a component of cell wall of bacteria (b) Is a structure which helps in respiration

(c) Consists of genes found inside the nucleus (d) Is the genetic part in addition to DNA in microorganisms

Advance

705. Genetic engineering means **[CMC Vallore 1993]**

(a) Manipulation of cell contents (b) Test tube babies

(c) Manipulation of cell cytochromes (d) Manipulation of genes

706. The process of joining together different DNA fragments is often referred to as **[MP PMT 1994]**

(a) Transcription (b) Cloning (c) Gene splicing (d) DNA amplification

707. The transfer of protein from electrophoretic gel to nitrocellulose membrane is known as **[MP PMT 200**

(a) Transferase (b) Northern blotting (c) Western blotting (d) Southern blotting

708. Which of these is widely used in genetic engineering **[CPMT 2003]**

(a) Anopheles (b) Dragon fly (c) Dragon lizard (d) Fruit fly

709. Recombinant DNA technology is related with **[BHU 1999]**

(a) C. Darwin (b) Stanley Cohen (c) Herbert Boyer (d) Both (b) and (c)

710. ANDI is cloned **[KCET 2002]**

(a) Sheep (b) Bull (c) Monkey (d) Cat

711. How many amino acids are present in the human insulin

(a) 21 (b) 30 (c) 31 (d) 51

712. In human type(s) of interferon present is (are)

(a) Leucocytic (b) Fibroblastic (c) Immune interferon (d) All of the above

713. The interferon which is synthesised by WBC is known as

(a) Fibroblastic interferon (b) Immune interferon (c) Leucocytic interferon (d) None of the above

714. Which of the following correctly defines a transgenic animal **[CBSE PMT 1995]**

(a) An animal which has foreign DNA and RNA in some of its cells because of an injection of DNA and RNA into the nucleus of the zygote from which it is developed

(b) An animal which has foreign DNA in all its cells because of an injection of DNA into the nucleus of the zygote from which it is developed

(c) An animal which has foreign DNA in some of its cells because of an injection of DNA into the nuclei of some of the cells of the blastocyst

(d) An animal which has foreign DNA in all its cells because of an injection of DNA into the nuclei of some of the cells in adulthood

715. Hargobind Khorana is known for **[JIPMER 1996; Manipal 1997]**

(a) Discovery of DNA (b) Discovery of DNA ligase (c) Discovery of *t*RNA(d)

716. Giant Mouse has been produced through **[CBSE PMT 2000]**

(a) Tissue culture (b) Gene differentiation (c) Gene manipulation (d) All the above

717. The first mammal clone 'Dolly' was created by **[Kerala 2002]**

(a) Gregor Mendel (b) Thomas King (c) Robert Briggs (d) Ian Wilmut

(e) T.H. Morgan

718. Humulin is

(a) Carbohydrate (b) Fat (c) Hybridoma (d) Protein

719. Which one of the following pairs of terms/names mean one and the same thing **[AIIMS 2003]**

(a) Gene pool - genome (b) Codon - gene

(c) Cistron - triplet (d) DNA finger printing – DNA profiling

720. VNTR is employed for **[AMU 2002]**

(a) Protoplasmic culture (b) DNA finger printing

(c) Regulation of plant growth hormones (d) Enhancing photosynthesis in desert plants

721. DNA probes used in finger printing are

(a) Highly sensitive electron microscope (b) UV beams

(c) DNA segments having radioactive isotopes (d) X-ray scanners

722. Process used for amplication or multiplication of DNA for finger printing is

(a) Polymerase chain reaction (b) Nesslerisation (c) Southern blotting (d)

723. Dermatoglyphics is connected with

(a) Skin disease (b) Care of skin (c) Cosmetics (d) Finger printing

724. Construction of recombinant DNA involves **[KCET 2002]**

(a) Cleaving and rejoining DNA segments with 'endonuclease' alone

(b) Cleaving DNA segments with 'endonuclease' and rejoining them with 'ligase'

(c) Cleaving DNA segments with 'ligase' and rejoining them with 'endonuclease'

(d) Cleaving and rejoining DNA segments with ligase alone

725. A plasmid is a **[MP PMT 1992; CPMT 1994]**

(a) Bacteriophage

(b) DNA molecule present in mitochondria

(c) DNA molecule incorporated in the bacteria chromosome

(d) A small circular DNA molecule capable of self replication and that can carry genes into host organism

726. The plasmid pBR 322 used in biotechnology is **[EAMCET 2002]**

(a) Yeast (b) M_{32} phage (c) Parasite (d) Cloning vehicle

727. What is the function of a vector

(a) Helps to amplify the DNA (b) Allows cells to take up foreign DNA

(c) Destroys cells that do not contain cloned DNA (d) Carries cloned DNA, enabling it to replicate in host cells

728. The offsprings produced through which of the following processes are not exactly similar to their parents **[AFMC 2002]**

(a) Sexual reproduction (b) Dizygotic twins (c) Asexual reproduction (d) Parthenogenesis

729. In DNA segment the probe blinds is identified by its size by using a technique called **[AMU 2002]**

(a) DNA probe (b) DNA denaturation (c) DNA polymorphism (d)None of the above

730. Which one of the following can give a complementary and palindromic sequence **[EAMCET 2003]**

(a) 5'-ATATCC-3' (b) 5'-CCGAAT-3' (c) 5'-GAATTC-3' (d) 5'-AGGTTC-3'

731. What are true of plasmids **[CBSE PMT 2001]**

(a) They are found in viruses (b) They are main part of chromosomes

(c) They are widely used in gene transfer (d) They contain gene for vital activities

732. Restriction enzymes are present in several microorganisms cut foreign DNA at specific sites and destroy them. The enzymes do not destroy the cellular DNA because **[AMU 2003]**

(a) The cellular DNA does not have the specific sites

(b) The susceptible specific sites are masked by protein

(c) The restriction enzyme susceptible sites are modified by cellular enzymes

(d) The restriction enzymes and DNA occupy different compartments

733. In transgenics, expression of transgene in target tissue is determined by **[CBSE PMT 2004]**

(a) Reporter (b) Enhancer (c) Transgene (d) Promoter

734. DNA fingerprints used as evidence in a murder trial look something like supermarket bar codes. The pattern of bars in a DNA fingerprint shows

(a) The order of bases in a particular gene

(b) The order of genes along particular gene

(c) The exact location of a specific gene in a genomic library

(d) The presence of various-sized fragments from choppedup DNA

735. A restriction enzyme *Eco* RI from *E. coli* is expected to cleave DNA at following sequence**[Kerala PM**

(a) GAATTC (b) AAGTTC (c) AAGCTT (d) GTATATC

736. The specific DNA sequence where *Eco* RI cuts is **[Kerala PMT 2003]**

(a) ATTCGA / TAAGCT (b) GAATTC / CTTAAG (c) GCTTAA / CGAATT (d) GTTCAA / CAAGTT

737. Which one of the following pairs of terms/names mean one and the same thing **[AIIMS 2003]**

(a) Codon - Gene

(b) Cistron - Triplet

(c) Gene pool - Genome

(d) DNA fingerprinting - DNA profiling

738. Restriction endonucleases **[CBSE PMT 2004]**

(a) Are synthesized bacteria as part of their defense mechanism

(b) Are present in mammalian cells for degradation of DNA when the cell dies

(c) Are used in genetic engineering for ligating two DNA molecules

(d) Are used for *in vitro* DNA synthesis

739. Which statement is correct for bacterial transduction **[CBSE PMT 2002]**

(a) Bacteria obtain its DNA directly

(b) Bacteria obtain DNA from other external source

(c) Transfer of genes from one bacterium to another bacterium by conjugation

(d) Transfer of some genes from one bacterium to another bacterium through virus

740. A biologist isolated a gene from a human cell, attached it to a plasmid and inserted the plasmid into a bacterium. The bacterium made a new protein, but it was nothing like the protein normally produced in a human cell. Why

(a) The gene contained introns (b) The gene did not have sticky ends

(c) The biologist should have cloned the gene first (d) The bacterium had undergone transformation

741. Scientists have produced a smallpox virus that contains genes from several other disease-causing microorganisms. They hope to use the virus

(a) As a compact genomic library (b) In a vaccine against several diseases

(c) As a gene vector for human gene therapy (d) To perfect a germ-warfare weapon with no antidote

742. DNA fingerprinting refers to **[CBSE PMT 2004]**

(a) Techniques used for identification of fingerprints of individuals

(b) Molecular analysis of profiles of DNA samples

(c) Analysis of DNA samples using imprinting devices

(d) Techniques used for molecular analysis of different specimens of DNA

743. The following can be described as clones **[AMU 2003]**

(a) The mother of the sheep Dolly and Dolly

(b) Identical twins arising out of a single egg

(c) A colony of bacteria derived from a single cell by a sexual reproduction

(d) All of the above

ANSWER

ASSIGNMENT (BASIC & ADVANCE LEVEL)

1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20
a	c	a	c	c	d	d	a	c	c	b	a	a	c	c	a	b	a	c	d
21	22	23	24	25	26	27	28	29	30	31	32	33	34	35	36	37	38	39	40
b	c	b	a	c	a	d	c	b	b	d	b	a	d	d	c	a	a	a	b
41	42	43	44	45	46	47	48	49	50	51	52	53	54	55	56	57	58	59	60
b	b	a	d	a	c	c	b	c	a	d	c	b	c	b	c	a	b	b	d
61	62	63	64	65	66	67	68	69	70	71	72	73	74	75	76	77	78	79	80
a	a	c	b	d	a	a	a	b	c	c	d	c	c	c	d	d	c	d	b
81	82	83	84	85	86	87	88	89	90	91	92	93	94	95	96	97	98	99	100
c	d	a	d	b	c	c	c	b	a	c	a	b	c	b	c	d	d	a	b
101	102	103	104	105	106	107	108	109	110	111	112	113	114	115	116	117	118	119	120
c	a	c	d	c	a	a	a	c	a	c	b	a	d	b	d	c	b	a	c
121	122	123	124	125	126	127	128	129	130	131	132	133	134	135	136	137	138	139	140
c	c	d	a	b	b	d	c	d	b	a	b	d	d	c	c	c	d	a	d
141	142	143	144	145	146	147	148	149	150	151	152	153	154	155	156	157	158	159	160
c	d	d	b	a	b	a	c	c	c	a	a	a	a	d	a	a	c	a	d
161	162	163	164	165	166	167	168	169	170	171	172	173	174	175	176	177	178	179	180
d	d	d	b	b	a	c	c	a	d	d	d	d	b	b	b	a	a	b	c
181	182	183	184	185	186	187	188	189	190	191	192	193	194	195	196	197	198	199	200
b	b	a	b	b	b	c	a	b	a	c	a	c	d	c	c	a	d	d	b
201	202	203	204	205	206	207	208	209	210	211	212	213	214	215	216	217	218	219	220
d	b	d	b	b	a	c	a	b	c	c	c	a	a	d	d	d	d	b	c
221	222	223	224	225	226	227	228	229	230	231	232	233	234	235	236	237	238	239	240
b	d	b	c	d	c	c	b	a	d	b	b	a	a	c	d	c	d	d	a
241	242	243	244	245	246	247	248	249	250	251	252	253	254	255	256	257	258	259	260
c	d	a	a	c	c	d	c	a	a	b	a	a	d	c	b	b	d	d	b
261	262	263	264	265	266	267	268	269	270	271	272	273	274	275	276	277	278	279	280
c	c	a	a	b	d	d	d	a	a	c	d	c	b	a	c	d	c	a	b
281	282	283	284	285	286	287	288	289	290	291	292	293	294	295	296	297	298	299	300
d	c	d	b	c	c	a	c	d	d	d	b	a	d	c	b	c	a	a	a
301	302	303	304	305	306	307	308	309	310	311	312	313	314	315	316	317	318	319	320
a	a	d	b	d	d	a	c	d	b	b	d	d	d	d	b	b	d	b	b
321	322	323	324	325	326	327	328	329	330	331	332	333	334	335	336	337	338	339	340
d	d	d	b	a	a	d	d	a	d	a	a	c	d	b	a	b	a	c	c

341	342	343	344	345	346	347	348	349	350	351	352	353	354	355	356	357	358	359	360
c	d	d	d	a	c	d	d	c	c	d	a	c	b	b	a	b	a	d	d
361	362	363	364	365	366	367	368	369	370	371	372	373	374	375	376	377	378	379	380
c	a	d	a	b	d	d	a	b	a	a	c	c	a	c	c	b	b	a	b
381	382	383	384	385	386	387	388	389	390	391	392	393	394	395	396	397	398	399	400
d	a	b	d	a	b	b	b	d	b	d	c	c	d	a	a	c	d	c	b
401	402	403	404	405	406	407	408	409	410	411	412	413	414	415	416	417	418	419	420
b	d	b	c	c	a	d	b	a	c	a	b	c	b	d	b	c	b	a	c
421	422	423	424	425	426	427	428	429	430	431	432	433	434	435	436	437	438	439	440
b	c	a	a	d	c	c	b	b	a	c	b	b	b	a	b	a	c	c	a
441	442	443	444	445	446	447	448	449	450	451	452	453	454	455	456	457	458	459	460
d	c	b	c	d	d	d	a	c	a	a	a	a	c	a	c	c	c	d	a
461	462	463	464	465	466	467	468	469	470	471	472	473	474	475	476	477	478	479	480
a	c	c	a	d	c	b	a	b	c	c	c	d	b	d	a	d	b	b	c
481	482	483	484	485	486	487	488	489	490	491	492	493	494	495	496	497	498	499	500
c	b	b	a	b	c	a	c	c	d	d	d	d	c	d	c	a	b	b	c
501	502	503	504	505	506	507	508	509	510	511	512	513	514	515	516	517	518	519	520
a	b	c	d	c	c	b	b	b	a	d	c	c	d	a	a	a	d	d	b
521	522	523	524	525	526	527	528	529	530	531	532	533	534	535	536	537	538	539	540
c	b	d	a	d	a	a	a	a	c	a	c	b	b	a	d	a	d	b	b
541	542	543	544	545	546	547	548	549	550	551	552	553	554	555	556	557	558	559	560
a	b	a	b	d	d	c	c	b	a	b	c	d	d	a	a	b	a	b	c
561	562	563	564	565	566	567	568	569	570	571	572	573	574	575	576	577	578	579	580
a	c	c	a	c	a	c	d	b	d	a	b	d	b	a	c	d	a	d	a
581	582	583	584	585	586	587	588	589	590	591	592	593	594	595	596	597	598	599	600
d	b	b	a	d	c	d	d	a	d	a	d	d	d	c	b	b	d	b	c
601	602	603	604	605	606	607	608	609	610	611	612	613	614	615	616	617	618	619	620
a	c	d	c	a	b	c	c	d	a	a	b	c	b	d	d	a	b	c	c
621	622	623	624	625	626	627	628	629	630	631	632	633	634	635	636	637	638	639	640
c	c	a	d	b	a	c	d	a	d	c	d	b	c	b	a	c	c	d	b
641	642	643	644	645	646	647	648	649	650	651	652	653	654	655	656	657	658	659	660
b	d	b	c	a	b	b	c	c	a	a	d	c	d	a	c	b	a	d	d
661	662	663	664	665	666	667	668	669	670	671	672	673	674	675	676	677	678	679	680
a	c	a	d	c	c	c	c	a	a	d	b	a	d	d	d	c	c	a	b
681	682	683	684	685	686	687	688	689	690	691	692	693	694	695	696	697	698	699	700
c	c	d	b	a	b	a	c	d	c	c	d	b	c	a	b	d	d	d	b
701	702	703	704	705	706	707	708	709	710	711	712	713	714	715	716	717	718	719	720
b	a	a	d	d	c	c	d	d	c	d	d	c	b	b	c	d	d	d	b
721	722	723	724	725	726	727	728	729	730	731	732	733	734	735	736	737	738	739	740
c	a	d	b	d	d	d	a	d	c	c	c	d	d	a	b	d	a	d	a
741	742	743																	
b	b	d																	